Unmet healthcare needs, health-related quality of life and assistive technology for cognition in Huntington’s disease

A population-based cross-sectional study

Regina Marleen Van Walsem

Doctoral thesis

The Faculty of Medicine, University of Oslo
Department of Neurohabilitation, Oslo University Hospital, Ullevål

2017
Unmet healthcare needs, health-related quality of life and assistive technology for cognition in Huntington’s disease.

Series of dissertations submitted to the Faculty of Medicine, University of Oslo

ISBN 978-82-8377-054-4

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http://www.duo.uio.no/

Cover: Hanne Baadsgaard Utigard.
Print production: Reprosentralen, University of Oslo.
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PAPERS I - III
Var det dit hun skulle?


Å sette spor etter seg i verden. Å være mer enn sykdommen sin.

Was this where she was going?

Huntingtonland. Unaware that the disease existed. A mark in the forehead, the fall of man, the path of kins. I can’t help it. Truly, I can’t help it.

This is how life turned out, and how life must be lived. Exploring the world through language. I write, thus I am. Therefore I am. Confronting the disease. Animal metaphors: The elephant that dwells in my body, in company with a flapping crow. Others comprehend. To laugh at oneself - and others, we are so incredibly comical as we come paddling and waving.

Magical human encounters. Professionals and other people with the disease, a choreographed dance with side steps.

To leave traces in the world. Being more than one’s disease.

Written by Tove Berg, October 23rd 2016
FOREWORD

Whilst studying for my degree in psychology with specialization in clinical neuropsychology, and specifically during my internship at Leiden University Medical Centre in 2002, where I first met patients affected by Huntington’s disease (HD), I realized a number of things that made me determined to want to work with HD in my future professional life. First of all, I realized that basically all aspects of psychology possibly imaginable and all that I had learned during my studies came together in this one disease. This made HD extremely interesting. Moreover, I realized that the complex nature of HD makes it devastating for anyone affected by it; the patient, the family, parents, children, spouses and friends. I realized that in the absence of a curative or disease modifying treatment, further efforts needed to be directed at developing non-pharmacological interventions, and that providing adequate healthcare and attending to the complex and changing aspects of HD across the whole disease spectrum is of vital importance to those affected by HD.

Almost a decade later, in August 2011, I started working as a clinical psychologist at the Department of Neurohabilitation, at the Oslo University Hospital. During my work there, it seemed that many patients with HD (as well as patients with other complex neurological diseases), did not receive the comprehensive, multidisciplinary healthcare they needed and were advised to receive after being carefully and comprehensively assessed by a multidisciplinary team. It made me question my experience and wonder whether patients with HD really did receive the comprehensive multidisciplinary care that has been established as the gold standard.

My hope is that this thesis will shed light on the healthcare needs that are not met and that results in the longer run may contribute to improving healthcare for patients with HD. Furthermore, that HD may serve as a case for improving healthcare for other patient populations with complex neurological (rare) conditions seen in the Norwegian healthcare system, specifically in the habilitation services.

Even though a disease modifying treatment may be available in the future, there is no curative or disease modifying treatment yet. Patients affected by HD in the present and for several years to come, will be dependent on tailored comprehensive care that can help them maintain function and quality of life.
ACKNOWLEDGEMENTS

This work was conducted from October 2013 to December 2016. It was part of one of the five umbrella projects conducted at the Center for Habilitation and Rehabilitation Models and Services (CHARM), at the Institute of Health and Society at the University of Oslo. The research was carried out in collaboration with the Oslo University Hospital, the Department of Neurohabilitation, Department of Neurology, Department of Medical Genetics, and the Center for Rare Disorders, Vikersund Rehabilitation Center, as well as with the Norwegian HD Association (Landsforeningen for Huntington sykdom) and the professional network for Huntington’s Disease (Huntington fagnettverk).

I would like to express my extensive gratitude to everyone who has contributed to making this work happen. There are a number of people that I would like to thank in particular.

First of all, I wish to express my sincere gratitude to my main supervisor Nada Andelic. It has been invaluable and an honor to have been supervised by her, perfectly balancing criticism and support. Working with her has been an ongoing inspiration and she gave me the necessary confidence when I doubted myself the most. Secondly, I would like to greatly thank my co-supervisor Jan Frich. His knowledge and experience with Huntington’s disease and health services have been vital to this work. His calmness and clear feedback has helped me tremendously at the times I was confused and stressed. Both of you have made this journey a positive experience. Your supervision has taught me about my strengths and weaknesses as a researcher. This will be invaluable in the future.

I would also like to specifically thank Emilie Howe, who from being a driven and meticulous student, became a true colleague and friend during the past years. Especially during the data collection for this work we got to know each other well, during our many travels through urban and rural Norway. Further, I would like to thank Hanne Ladt Fossmo, another ambitious and meticulous student for the help with the recruitment and follow up on some data as part her master thesis. I also want to thank Nancy Borgerød, Ragnhild Wehus, Kristin Iversen, Gunvor Aslaksen Ruud, and Arvid Heiberg for excellent collaboration with identifying and recruiting participants to the present study.

I further would like to thank all CHARM colleagues. In particular, I would like to thank the leaders of CHARM, Marit Kirkevold and Cecilie Røe for their continuous support during this work. I also would like to specifically thank Gunvor Klevberg, fellow Ph.D. candidate, for the
good times exchanging thoughts about our work and life. All my colleagues at the Department of Neurohabilitation deserve a sincere thank you, in particular my fellow psychologists and Anne-Brit Hunsrød, for their patience and ongoing interest in my work. Specifically, I would like to thank Nils Olav Aanonsen, Head of the Department of Neurohabilitation for his support during my work, for enabling me to continue clinical work and most importantly; without him I would have missed out on this opportunity.

Professor Kjetil Sundet also deserves a special thank you. He was the first neuropsychologist and researcher I met in Norway more than 12 years ago. His contribution to my first article has been of invaluable importance for my motivation to pursue becoming a researcher. I also owe thanks to Bodil Stokke, for being a continuous inspiration and a friend through our long talks and discussions.

I owe great thanks to all my friends in Norway and the Netherlands, who all supported me tremendously while carrying out this work and for their treasured friendships. In particular, I thank Sanne for always listening, the invaluable talks and for understanding the challenges and victories along the process of completing this work. I would like to thank Florence, Carlijn and Mirjam, specifically for their positivity and lasting interest despite distances and motherhood.

I could not have conducted the present work with such pleasure without invaluable family support. Especially, I would like to thank my parents and my sister for their continuous encouragement and interest throughout carrying out this work, and my parents and sister in law, Anne, Rob and Susanne, for their cheering every time a new step closer to the goal was made. A special thank you needs to be expressed to my opa and oma, who have always been listening, asking questions, and for being there.

This work would not have been possible without the participation of the patients and their families and professional caregivers. Thank you all so much, for allowing me to become so close to your personal lives, for your time and patience and for sharing with me. You are all a continuous inspiration to me, and meeting you all, has been an experience I will never forget. Specifically, I would like to thank Tove Berg, for writing the poem at the beginning of this thesis.

I would like to acknowledge the Research Council of Norway for funding this work as part of the general project the Center for Habilitation and Rehabilitation Models and Services (CHARM).
Last but not least, I would like to thank my wonderful husband Robert-Jan and my lovely energetic daughter Julia. Words cannot describe the happiness you both bring into my life. Thank you both for your unconditional love and support, your patience, and positive spirits, which have given me the strength to complete this work.

Oslo, January 2017

Marleen van Walsem
ABSTRACT

There is a shortage of studies systematically investigating healthcare needs and social support in a representative population of patients with Huntington’s Disease (HD). In order to understand the extent of the problems and to identify the population at risk for unmet health care needs and how unmet need may be associated with health-related quality of life (HRQoL), it is necessary to assess such a population in the geographical regions of interest. This knowledge may be helpful when planning improvement of health care delivery in patients with HD.

The general aim of the present thesis was to describe the levels and prevalence of unmet needs for healthcare and social support services among patients with HD in the south-east part of Norway, which factors are associated with these unmet healthcare needs, particularly the association with HRQoL, and to specifically investigate the use of assistive technology for cognition (ATC) among patients with HD and associations with HRQoL as a case of a supportive unmet healthcare needs.

The study findings suggest substantial unmet needs for healthcare and social support services, overall, for the subdomains of health and personal care and of social and supportive care services, as well as for different types of healthcare and social support services, primary for the sub-scales Rehabilitation and Family and social support. Moreover, substantial levels and high proportions of unmet healthcare needs were found at all five disease stages (I-V), but most profoundly among patients with HD in the middle phase (disease stage III). Being in this phase increased the odds of having high levels of total unmet healthcare needs and of the domain Health and personal care substantially. Level of education and whether information on healthcare services received was obtained from the patient alone were other factors connected with overall level of unmet healthcare needs and the Health and personal care domain. Our results further revealed that higher levels of unmet healthcare needs were linked with lower HRQoL. Studying the effect of ATC, a specific supportive healthcare service, on HRQoL, an association between use of the service and HRQoL was not observed. Yet the only significant association was found for functional ability as measured by Total Functional Capacity (TFC). At the same time, the use of ATC, information, needs assessment and training were seen to be reported relatively infrequently, especially given the clinical picture of HD, which includes the progressive development of cognitive impairment as a hallmark symptom.
Our results indicate that it is essential to systematically evaluate, record and track patients’ needs with regards to healthcare and social support services, and follow up on whether those needs are met besides performing clinical evaluations. Specific attention should be directed to healthcare service delivery that targets patients in the middle phase of HD (stage III). Our findings regarding ATC suggest that increased awareness of and knowledge on the processes for what entails successful delivery is necessary. Further, they propose that delivery of adequate comprehensive healthcare aimed at improving function and HRQoL across the whole disease spectrum of HD, is facilitated by collaborative relationships with patients’ family caregivers, and understanding of the patients’ social context.
LIST OF PAPERS

This thesis is based on the following papers, which are referred to by their roman numbers:


# LIST OF ABBREVIATIONS

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
</tr>
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<tbody>
<tr>
<td>AD</td>
<td>Alzheimer’s Disease</td>
</tr>
<tr>
<td>ADL</td>
<td>Activities of Daily Living</td>
</tr>
<tr>
<td>ALS</td>
<td>Amyotrophic Lateral Sclerosis</td>
</tr>
<tr>
<td>ANOVA</td>
<td>Analysis of Variation</td>
</tr>
<tr>
<td>AT</td>
<td>Assistive Technology</td>
</tr>
<tr>
<td>ATC</td>
<td>Assistive Technology for Cognition</td>
</tr>
<tr>
<td>BMI</td>
<td>Body Mass Index</td>
</tr>
<tr>
<td>CAG</td>
<td>Cytosine, Adenine, Guanine, (the amino-triplet which is repeated in the gene causing HD)</td>
</tr>
<tr>
<td>CRD</td>
<td>Centre for Rare Disorders</td>
</tr>
<tr>
<td>CPNQ</td>
<td>Cancer Patient Needs Questionnaire</td>
</tr>
<tr>
<td>EHDN</td>
<td>European Huntington’s Disease Network</td>
</tr>
<tr>
<td>EQ-5D-3L</td>
<td>EuroQol Five Dimensions Questionnaire Three-level</td>
</tr>
<tr>
<td>FAS</td>
<td>Functional Assessment Scale</td>
</tr>
<tr>
<td>HD</td>
<td>Huntington’s Disease</td>
</tr>
<tr>
<td>HDQoL</td>
<td>The Huntington’s Disease health-related Quality of Life questionnaire</td>
</tr>
<tr>
<td>HDQLIFE</td>
<td>The Huntington Disease Health-Related Quality of Life</td>
</tr>
<tr>
<td>HRQoL</td>
<td>Health-related Quality of Life</td>
</tr>
<tr>
<td>HTT-gene</td>
<td>Huntingtin gene</td>
</tr>
<tr>
<td>ICD-10</td>
<td>International Classification of Diseases, tenth version</td>
</tr>
<tr>
<td>IS</td>
<td>Independence Scale</td>
</tr>
<tr>
<td>LTNC</td>
<td>Long-term Neurological Conditions</td>
</tr>
<tr>
<td>MS</td>
<td>Multiple Sclerosis</td>
</tr>
<tr>
<td>Abbreviation</td>
<td>Description</td>
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<tr>
<td>NLWA</td>
<td>Norwegian Labour and Welfare Administration (NAV in Norwegian)</td>
</tr>
<tr>
<td>NPCs</td>
<td>Needs and Provision Complexity Scale</td>
</tr>
<tr>
<td>OR</td>
<td>Odds Ratio</td>
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<tr>
<td>OUS</td>
<td>Oslo University Hospital</td>
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<tr>
<td>PD</td>
<td>Parkinson’s Disease</td>
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<tr>
<td>QoL</td>
<td>Quality of Life</td>
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<tr>
<td>SD</td>
<td>Standard Deviation</td>
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<tr>
<td>SF-36</td>
<td>36-Item Short Form Survey</td>
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<tr>
<td>SIP</td>
<td>Sickness Impact Profile</td>
</tr>
<tr>
<td>TBI</td>
<td>Traumatic Brain Injury</td>
</tr>
<tr>
<td>TFC</td>
<td>Total Functional Capacity</td>
</tr>
<tr>
<td>UHDRS</td>
<td>Unified Huntington’s Disease Rating Scale</td>
</tr>
<tr>
<td>VAS</td>
<td>Visual Analogue Scale</td>
</tr>
<tr>
<td>WHO</td>
<td>World Health Organization</td>
</tr>
<tr>
<td>REGISTRY</td>
<td>European observational multinational multicenter study on HD of the European Huntington’s Disease Network (<a href="http://www.euro-hd.net/registry">www.euro-hd.net/registry</a>)</td>
</tr>
<tr>
<td>PREDICT-HD</td>
<td>US multicenter observational study of the earliest signs of HD (<a href="http://www.predict-hd.net">www.predict-hd.net</a>)</td>
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1 INTRODUCTION

In the absence of a curative or disease modifying treatment, it is widely acknowledged that the clinical care of patients with HD is dependent on disease management, alleviating symptoms and maintaining functional ability and health-related quality of life (HRQoL) by means of comprehensive multidisciplinary care using pharmacological and non-pharmacological interventions. This knowledge is documented in the literature regarding HD and through the publishing of clinical standards of care guidelines (1-4). Yet, despite anecdotal information from patients and their caregivers, as well as experiences from expert clinicians working with HD suggesting that the needs of patients with HD and their families are not always recognized or met, literature investigating unmet healthcare needs for patients with HD and their families is still scant. A small number of studies evaluating HD families’ experiences with healthcare services found a lack of knowledge on HD (5, 6). One very recent study looked into healthcare delivery for patients with HD (7). Two other studies have researched utilization and needs of health and social care services in patients with HD (8, 9). To date no studies have systematically investigated unmet needs in terms of healthcare and social support services in patients with HD across the five clinical disease stages, which based on anecdotal knowledge and the few studies available may be expected to exist.

Research on HRQoL and HD has been directed at investigating the impact of HD and the related clinical symptoms of HD on HRQoL as well as on developing disease-specific measurements for HRQoL in the population with HD (10-15). To date there are no investigations that have reviewed the potential effect of unmet needs with respect to healthcare services on HRQoL.

Over the last two decades a tremendous amount of invaluable knowledge has been garnered on the hallmark symptoms of cognitive decline in HD, resulting in the understanding of much more about the development of HD across the whole disease spectrum, including prodromal HD, how cognitive impairment develops, how changes in the brain are related to different clinical symptoms of cognitive impairment, and regarding sensitive instruments for identifying and measuring cognitive changes over time (16-21). This has led to a relatively novel interest through targeting cognitive impairment in pharmacological and non-pharmacological treatments, including rehabilitation and attempting to tap into the plasticity and compensative mechanisms of the brain (22-24). However, a long-known supportive service aimed at ameliorating the negative effects of cognitive impairments among patient
populations developing cognitive disabilities, assistive technology for cognition (ATC), has not yet been investigated as an option for potentially enhancing functional abilities and HRQoL in patients with HD.
2 BACKGROUND

2.1 HUNTINGTON’S DISEASE (HD)

2.1.1 Cause and prevalence

Huntington’s disease (HD) is a rare neurodegenerative disease characterized by a triad of three core symptoms - movement disorders, psychiatric disturbances and cognitive decline resulting in dementia. HD is caused by an expanded CAG repeat in the HTT-gene located on chromosome 4, and inheritance is autosomal dominant (25). This means that each child of a parent with HD has a 50% chance of inheriting the gene and developing HD. Grandchildren have an initial risk of 25% if the status of their parent is unknown. Individuals with a CAG repeat of over 39 will inevitably develop HD during their lifetime, while an individual with a CAG repeat of 36 - 39 carries the HD gene but may or may not live long enough to manifest into a clinical diagnosis of HD. If a child of an affected parent does not carry the HD gene, HD no longer exists in that branch of the family (26). With the discovery of the HD gene in 1993, it became possible to genetically test individuals with 100% certainty whether they had HD (confirmative testing) or for carrying the HD gene prior to showing clinical symptoms (predictive testing) (27).

The prevalence of HD varies across countries and has been reported to be 5 – 10 per 100,000 within the Caucasian population (28). This prevalence varies across countries and has not been stable over time (29). A rising occurrence in Australia, North America, the United Kingdom and Western Europe may be because of increased awareness of HD, reduced stigma and also increased mutation rates (30). No study has yet formally investigated the prevalence of HD in Norway, but in a guide of the Centre for Rare Disorders from 2011, it was estimated that there are about 350 individuals with clinical HD, and approximately 700 individuals are thought to be at risk for developing HD in Norway (31).

2.1.2 Clinical symptoms, diagnosis and progression

The motor disturbances, including chorea, rigidity, dystonia and psychiatric disturbances comprising i.e., depressive symptoms, apathy, anxiety, irritability and cognitive decline resulting in global dementia, constitute the most characteristic symptoms of HD. However, a number of others further contribute to the complex clinical picture. Weight loss, sleep
disturbances and dysregulation of the autonomic nervous system as well as difficulty communicating, swallowing difficulties (dysphagia) and incontinence, which become most pronounced in advanced stages of disease, are common (26, 28, 32-34). Reduced or lack of self-awareness is another frequent complicating feature (35). Clinical symptoms of HD usually become apparent between 30 and 50 years of age. It typically takes about 15 - 20 years from the signs of the first clinical symptoms to complete care dependency and death (28, 36).

A formal clinical diagnosis of HD today is given to an individual based on the criteria of a) the presence of unequivocal motor symptoms objectified by neurological examination and b) a family history of HD or genetically confirmed presence of an expanded CAG repeats on the HD gene (28, 37). However, subtle motor signs, psychiatric symptoms and cognitive changes may be present many years prior to clinical diagnosis (38-42). The expanding knowledge on symptom development and natural history of HD has resulted in investigations adjusting the currently used criteria based only on the core symptom of motor impairment. Till now none of these proposals are formally adapted into clinical practice or diagnostic systems (37, 43).

HD is characterized by progressive functional decline caused by its symptoms. Yet, progression varies greatly between individuals in terms of rate of disease progression, and of presentation and severity of clinical symptoms (44). The advancement in functional decline is used to categorize patients in five functional disease stages based on the Shoulson and Fahn Functional Capacity Scale (TFC), a scale developed specifically for HD, as one part of the Unified Huntington’s Disease Rating Scale (UHDRS) (45, 46). During the early disease phase, where patients are still mostly independent from others, are represented by stages I and II. Stage III signifies the middle phase of the disease, that is characterized by becoming increasingly functionally dependent on the aid from others. The advanced disease is represented by stages IV and V, during which patients require full-time care, often being transferred from receiving care at home to moving into long-term care facilities as care provision at home becomes increasingly difficult. In the final stage, patients often eventually die of secondary illnesses (i.e., pneumonia, choking) (1, 2, 47).

### 2.1.3 Cognitive impairment

One of the hallmark symptoms of HD is progressive cognitive impairment. It has been shown to be one of the most debilitating symptoms, compared to movement disorders, such as
chorea. Cognitive impairment early during disease progression results in reduced functioning in certain abilities, like working and driving (48, 49). Cognitive symptoms have also been indicated to greatly impact HRQoL adversely, both with respect to patients and their carers (13, 48, 50, 51). They have even been indicated as the factor with the most negative effect on HRQoL (11, 13). Moreover, subtle signs of cognitive decline become apparent more than a decade prior to clinical diagnosis (based on motor impairment) (16, 18-21). Changes in brain function, structure and connectivity in individuals with pre-manifest and manifest HD have been demonstrated by neuro-imaging studies and relationships between neuro-imaging measures and poor performance in cognitive tasks have been observed (17, 19, 20, 52). While alterations in cognition vary from individual to individual, cognitive impairments are usually most pronounced in the cognitive domains of psychomotor speed, executive functions and memory (specifically visuo-spatial memory), with relatively specific and mild impairments during the earlier stages (I and II), development into moderate impairments in the middle stage, and then progressing till global cognitive impairment and dementia in advanced HD stages (stages IV and V) (20, 26, 53).

2.1.4 Clinical management

Despite extensive efforts, at present, there is no curative or disease-modifying treatment for HD. Disease management, alleviating symptoms and maintaining functional ability and HRQoL are the emphasis of clinical care provided to patients with HD (28). This involves pharmacological treatment by means of medications to relieve symptoms, including antidepressants. It also involves a variety of non-pharmacological interventions, such as physiotherapy and comprehensive multidisciplinary rehabilitation programs (1, 23, 25, 54, 55).

Studies have indicated that patients in the early to middle stages of HD are able to benefit from various non-pharmacological interventions and from coordinated healthcare services, provided by different healthcare professionals (3, 23, 56, 57). These include neuropsychological testing as part of cognitive evaluation, counseling by (neuro)psychologists (26), rehabilitation programs (23, 24, 54), active physiotherapeutic interventions (55, 58), speech therapist training (25, 26, 59) and occupational therapy (26, 60). Additionally, in cases of advanced disease, even though mostly being dependent on full-time personal care, patients may still reap therapeutic merits from receiving assistance from various healthcare professionals, such as respiratory physiotherapy and involvement of
occupational therapists for environmental adjustments along with personal care (1, 60). Clinical guidelines for standards of care for various professions have been published with the goal of becoming the groundwork for further research in the field and to assess the care provided (4, 59, 60). This supports the general contention that in order to maintain functional ability and quality of life, clinical care in HD should comprise comprehensive coordinated healthcare provision entailing multidisciplinary, proactive, flexible and regularly monitored healthcare, all centered around the needs of the HD patient, and their family members’, that experience challenges and need support and guidance (1-3, 26, 57, 61). A number of clinics adapting a comprehensive care model have emerged in the United States, Scotland and Australia (26, 57, 62). However, it is unknown to what degree guidelines for standards of care addressing the needs of patients with HD and their family members have been formally recognized and implemented systematically in healthcare delivery internationally or nationally.

2.1.5 Unmet healthcare needs in Huntington’s disease

Providing adequate care may be compromised by the chronic, changing and complex clinical picture of HD. It is pivotal to understand the unmet needs of patients with HD in order to be able to establish whether they receive long-term multidisciplinary care that could lead to improved service delivery. This requires systematic assessment of patients’ needs and understanding of the services. Besides that, provision of long-term comprehensive multidisciplinary care is agreed upon to be the gold standard for HD healthcare, though little research has addressed healthcare delivery, utilization of and the needs for healthcare services. Health and social care utilization was studied by Busse et al. - they found that the majority of the patients received formal primary and community care services and, to a lesser extent formal hospital-based services, but there was a large reliance on informal care in the home (9). Most studies aiming to identify needs have been performed on carers of patients with HD, yet only one survey study included patients with HD along with carers. This study determined a number of unmet needs related to medical healthcare, functional and physical ability and social support (8). A recent global study report on health care delivery in HD found that the majority of clinics take a multidisciplinary care approach, having multiple professional specialties at their clinics and offer various relevant services. Yet how the care delivery is organized, i.e., to what degree the care is coordinated, remains unclear (7). None of the studies have systematically established the healthcare and support needs patients with HD
have based on a systematic assessment of need conducted through a normative approach, which healthcare they receive and their unmet needs, and equally including disease stages I – V (early to advanced HD).

### 2.1.6 HRQoL in HD

HRQoL has risen as an increasingly important patient and clinician reported outcome measure next to other endpoints of symptom ratings for HD (63, 64). Large-scale international observational studies, such as REGISTRY and PREDICT-HD have contributed to a rapidly developing body of research on HRQoL with regards to HD (43, 65). The attention paid in studies of HRQoL and HD thus far has been directed in three ways: 1) describing the HRQoL of premotor manifest patients, manifest patients and carers (13); 2) investigating which disease-related aspects are associated mostly with HRQoL (10, 11, 13, 14); and 3) developing sensitive disease-specific HRQoL outcome measures, with the goal of being used as outcome measures in future clinical trials (12, 15). Findings have demonstrated that patients with a clinical diagnosis of HD report a lower HRQoL compared to HD premotor manifest patients, individuals at risk of HD and their partners, and that HRQoL is reduced in patients in middle to advanced disease stages compared to the early disease phase (13). Additionally, functional ability and psychiatric symptoms, including depression and cognitive impairment have been shown to be the strongest determinants of HRQoL across a variety of studies (10, 11, 13, 14). The variation in the most influential disease-specific factors may be because of different measurements employed as well as unique sub-populations within the HD disease spectrum. Two disease-specific patient HRQoL measurements have been developed - the Huntington’s Disease Quality of Life (HDQoL) measurement and, more recently, the HDQLIFE (12, 15, 66). In recent years there has emerged increased efforts expanded on HRQoL in health services research, by examining and using HRQoL in relation to (unmet) healthcare needs (67, 68). However, this still must become an interest in the field of HD. Both HRQoL and healthcare services that include patients with HD, among patients with long-term neurological conditions (LTNC) such as Multiple Sclerosis (MS), Amyotrophic Lateral Sclerosis (ALS), Parkinson’s Disease (PD) has only been investigated in one study (69). According to the results of this study, patients with complex neurological conditions do not receive the healthcare services that might positively influence their HRQoL (69).
2.1.7 Assistive technology for cognition in Huntington’s disease

Assistive technology for cognition (ATC), briefly defined as external devices aimed at supporting cognitive function, is a type of supportive service, widely used to support individuals with cognitive disabilities in order to maintain or improve functional ability and quality of life (70, 71). It has been shown to be beneficial in several conditions of progressive and non-progressive acquired brain injury, such as traumatic brain injury (TBI) and Alzheimer’s disease (AD) (72-78). Generally, professionals working with patients with HD are aware of ATC. Nonetheless, the use of ATC to address cognitive impairment and ameliorate functional ability and quality of life deterioration has rarely been a topic in HD research despite the quickly increasing knowledge on cognitive decline in HD and the increasing focus on interventions specifically targeting cognition. Two studies investigating the use of talking mats as a way to support communication in nine patients with HD thus far has been the only relevant work in the area of ATC. The studies by Ferm et al. and Hallberg et al. showed that talking mats enabled patients with HD to have better structured one-to-one conversations compared to patients without communication aids, and to engage in more effective conversations reflected through asking more questions in group conversations (79, 80).

However, there is greater potential for other ATCs in HD on the basis of the various types of cognitive impairment (e.g., planning and organization, need for structure, reduced psychomotor speed) (53). For example, aids supporting structure and memory or support planning, organization and attention, may contribute to stabilizing functional capacity and independence (77). In practice, family caregivers often increasingly help patients by for instance remembering appointments. This can be a burden on both the patient and family member, while aforementioned examples of ATC may replace or supplement the patients’ relief, removing a burden from both patient and family member (77). While based on the potential of the effects of ATC in other similar populations, or patients with comparable cognitive impairments, the efficacious utility of formal ATC as an intervention is challenged by other symptoms of the disease, such as motor impairments, which may make using a touch screen on a smartphone difficult; apathy, possibly expressed as a lack of motivation to actually use a device despite the potential for relieving suffering, or reduced awareness, perhaps resulting in a patient not noticing the need for an ATC as they do not experience cognitive problems. Furthermore, the clinical picture of the profile of cognitive impairment changes over time, requiring focus on different ATCs or parts of ATC programs as HD
progresses, compared to those ATCs that may have been effective earlier on in the disease. As well, personal demographic factors, such as age, may be significant, as patients from younger generations may be more comfortable with or motivated to use an app than an older patient less familiar with technological advances. This underlines that establishment of the use of ATCs in a beneficial way is multifaceted and necessitates more than ascertaining that an ATCs can be beneficial for the patient and prescribing it. It requires identifying individual patient needs and preferences and matching the ATC with this so that an individual actually uses it and can thereby effectively enhance their cognitive functioning to maintain or improve participation in daily life as well as Quality of Life (QoL) (70, 72, 81).

2.1.8 Healthcare for HD in Norway

The healthcare of patients with HD is delivered in specialized healthcare services at hospitals and in primary care in the municipalities. Patients almost always receive healthcare from several departments at specialized hospital services, including the genetics department, the neurology department, the psychiatry department and, department of habilitation. With the development and evaluation of the effects of rehabilitation programs specifically for patients with HD in Norway, they also obtain care from two specialized rehabilitation centers (23, 56, 82). Further, the Center for Rare Disorders (in Norwegian: Senter for sjeldne diagnoser), which is a national competence center with the nationwide responsibility for a variety of rare disorders including HD, offers advice to patients and family members affected by HD as part of these specialized hospital services (83).

Primary care services include all healthcare services patients with HD may need as part of their chronic and incurable condition, and include community (re)habilitation, vocational rehabilitation, day care, and provision of personal care at home or within a care facility, including (specialized) nursing homes, respite care, and daytime care. Moreover, assistance from a personal assistant (“brukerstyrt assistant”) and procuring and implementation of the use of technological aids is the duty of primary healthcare services (84, 85). Patients with HD may also have a coordinated care plan (individuell plan) and an interdisciplinary coordination group (ansvarsgruppe) based at the primary care level aiming to ensure coordinated and tailored care that patients with long-lasting needs from chronic disease are regulated to receive (86, 87).
Taken together, patients with HD, in accordance with Norwegian regulations and guidelines, as well as the fact that Norway being a welfare state, in theory, have access to the care they require during the course of their disease regardless of the rising and changing needs, which is multidisciplinary and coordinated in line with the clinical care that these patients should be provided based on the literature. There are clinical guidelines for care in HD and there are specific Norwegian guidelines for healthcare provision to patients with long-lasting needs due to complex disabilities and chronic conditions, and for patients with dementia (86, 87), which apply to the population with HD, but we do not know in what extent these are integrated within healthcare service delivery.

2.2 IDENTIFYING UNMET HEALTHCARE NEEDS

2.2.1 The concept of healthcare needs

Identifying unmet needs requires understanding the concept of ‘needs’ in the context of health and healthcare. This is challenging - needs and health both are complex and vast concepts. As health covers a broad range of issues, including education, being able to participate in leisure activities and employment, transport, etc. healthcare needs should entail a broad range of care, including health education, disease prevention, diagnosis, treatment, social care, rehabilitation and more (88-91). It is unrealistic to attain a robust state of health in all areas, and so the goal of healthcare needs therefore is to attain an “optimal state of health” that entails safeguarding both functional capacity and quality of life, through the receipt of healthcare (89, 90, 92).

Despite this, even when placing needs in the common context of healthcare, no consensus on the concept of healthcare needs exists. There are many approaches to defining healthcare needs, and several definitions have been used in health service research and health needs assessment. Healthcare needs defined on the basis of the societal taxonomy by Bradshaw has been used frequently in healthcare service research (89, 93-95). Therein, needs are defined the following four ways:

- *normative* needs, needs as evaluated by healthcare professionals;
- *felt* needs, representing wants, wishes and desires by the individual;
- *expressed* needs, vocalized needs or the way people use healthcare; and
- *comparative* needs, or needs that can be compared between similar groups, for example, for severity, size etc.
These four types interact with one another. For instance, a need can be wished for by an individual (felt need) and expressed by contacting a healthcare professional (expressed need), but the professional may not agree that the wish of the individual is indeed a need (normative need). Another definition of healthcare needs is that they are those needs that can benefit from healthcare provision (88, 89, 93). This definition includes the aspect of cost containment and is preferred by health economists and policy makers that are bestowed with the task of organizing and distributing healthcare. It is perhaps the most commonly used definition and is often used when performing health needs assessment. The intricacy of the concept of healthcare needs is further underscored, in relation to the concepts of demand and supply, which often overlap each other (88). Demands comprise expressed needs, and are influenced by knowledge of interventions and services available as well as by expectations about services and interventions from the patients and their caregivers. Supply includes the healthcare received or utilized by patients, and is dependent on accessibility, acceptance of patients, the healthcare services made available by policy makers taking into account (financial) resources, and on the knowledge and interests of healthcare professionals and policymakers. Thus, the concept of healthcare needs is also a changing and dynamic concept - healthcare needs will change depending on alterations to demand and supply. New technology may make new interventions available that might give rise to new expectations and possibilities (88, 89).

2.2.2 Health needs assessment

Identifying unmet healthcare needs is part of the comprehensive process of health needs assessment which seeks to improve healthcare delivery. Performing health needs assessment includes needs assessment as well as gauging unmet needs (88, 90, 96). Wright et al. define health needs assessment as “the systematic method of identifying unmet health and healthcare needs of a population and making changes to meet these unmet needs” (88). Different topics and questions require varied methodologies comprising qualitative methods, including individual semi-structured and structured and standardized and non-standardized interviews, group interviews, and quantitative methods including indexes, and measures generating scores, as well as different approaches, such as comparative approaches, comparing healthcare received in one population versus another or one area compared to another area and epidemiological and cost effectiveness approaches (88, 90, 96, 97). Complete health needs assessment requires a combination of qualitative and quantitative methodology and pursuing several approaches. Determining health needs and/or unmet health needs, as part of
this process, also often combines qualitative with quantitative methods. Until 2003 as well as over the past decade, a number of tools and measurements for determining health needs and unmet needs have been developed (90). They include various types of qualitative and quantitative measures or an amalgamation thereof. Tools consist of interviews, either individual or group, semi-structured and structured, non-standardized and standardized, along with checklists and clinician and self-administered questionnaires. They are aimed at a variety of populations, and include disease-specific tools, such as the Cancer Patient Needs Questionnaire (CPNQ) or tools directed at specific healthcare services (i.e. rehabilitation services) (98, 99). In addition, tool are directed at obtaining information from patients (self-reports or perceived), caregivers and healthcare professionals (normative) (100). Furthermore, the comprehensive process of health needs assessment and the various components of this process, including the determination of health needs and unmet health needs, should be performed among various populations, though identifying and monitoring health needs and / or unmet health needs should also be conducted at the individual patient level and should have clinical relevance (88, 89). Finally, assessing health needs and unmet needs, should not be used interchangeably with outcome assessment, which seeks to assess the effects of an intervention (67, 90).

2.2.3 Unmet healthcare needs

Unmet healthcare needs can be operationally defined as “the differences (gaps), if any, between the services judged necessary and the services actually received” (97, 101-103). The services judged necessary represent expressed healthcare needs, either determined by patients and/or caregivers using self-reports, or in a normative way based on knowledge about health status (identification of the health problem(s), function) and about healthcare services and interventions contributing to better health through professionals. In light of the nature of the disease and taking a normative approach unmet healthcare needs in patients with HD can be understood as needs for healthcare and social support services which should generate “optimal health” in terms of enabling stable or better function and / or quality of life. The needed services which are actually received are the healthcare services that the patients are provided with and utilize, gauged based on interviewing the patient and / or caregiver by experienced healthcare professionals. Figure 1 outlines the operationalization of unmet healthcare needs presented here as a “gap-model” for unmet health needs.
Based on the complexity of health needs and the range of methods, perspectives and approaches that can be applied to assess health needs and unmet health needs different definitions have been employed depending on the question and methods chosen for the research. For example, one study investigating unmet health care needs of people with disabilities in Canada defined unmet needs as difficulties receiving service in response to problems that significantly interfered with daily life and assessed unmet health needs with a single yes/no question to participants. Van de Port et al. described unmet needs as the invisible factors significantly affecting health and well-being but that go undetected and unattended by the formal healthcare system (104, 105). Unmet needs have also been referred to as an absence of service use among people with a similar health problem (106).

2.2.4 Factors associated with unmet healthcare needs

The literature uses different methods and approaches, as per Chapter 2.2.2 “Health needs assessment”, and information obtained from different perspectives and unmet healthcare needs research is conducted in many types of populations. As a result, findings vary, and it is not easy to obtain a clear overview. Research conducted in the area of unmet health needs has identified several factors that are associated with unmet healthcare needs. For instance, a Canadian study observed higher rates of unmet needs in the general populations was associated with socio-demographic factors of urban residency, gender (being female),
younger age, more education, and medical factors including poorer health status and having a chronic condition (106). Education, age and gender were also found to affect unmet health needs for primary healthcare services in a Greek population sample. A lower level of education and younger age was observed to be associated with higher levels of and more unmet health needs (101). Education and age have also been revealed to be associated with unmet needs for social support (101, 107). Studies on unmet needs in specific populations found that clinical factors were more strongly associated with needs for healthcare services (i.e. receiving drugs, rehabilitation services) (104, 107-109). Socio-economic factors of lower income and not having a pharmaceutical insurance were associated in the Canadian study and a Norwegian study on the general population uncovered socio-economic inequalities with respect to utilization of private medical specialists and outpatient hospital services despite theoretical equal access based on need instead of prosperity (106) (110).

2.3 HRQoL

2.3.1 HRQoL

HRQoL can be said to be the cross-section between two broad and conceptually complicated viewpoints - QoL and health. QoL has been conceptualized as subjective, multi-dimensional and including both positive and negative dimensions by the World Health Organization (WHO). The WHO defined QoL as the “individuals’ perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concern” (91). This concept incorporates individuals’ physical health, psychological status, level of independence, social relationships, personal beliefs and environmental factors, all of which can be influenced by cultural context, time and place.

Health, which is also difficult to define and for which there is also a consensual definition lacking, is defined by the WHO as “the state of complete physical, psychological, and social well-being and not simply the absence of disease of infirmity” (67, 111). Importantly, this definition implies that health is not only the absence of disease, but that it can also be understood that robust health is a state where all physical, psychological and general well-being is optimal simultaneously, potentially posing unrealistically high expectations to being healthy (112). With this the definition can be seen as moving beyond the framework of health services, with which there is the ultimate objective of generating better health. General QoL has been shown to be more reflective of the state and experience of how several dimensions
and contexts affect an individuals’ life, while an evaluation of health is more comprehended from a biological point of view, reflected in function and capacity (113). QoL can be argued to therefore provide a less specific picture of the effect of disease on an individual’s life, while health alone may be unable to adequately capture the experience of the effect of disease on a patient’s life. As a result, the construct, HRQoL, was introduced as to be an overlap area between these two concepts (see Figure 2). It also is a subjective multi-dimensional concept, reflecting how symptoms of disease and/or its treatment impacts on an individuals’ physical, psychological/emotional and social well-being (114). As such the concept is still affected by a broad range of factors from both QoL and health approaches, but with the intent of capturing the impact of disease symptoms of and features of an individual’s QoL. With the increasing interest in applying HRQoL measurements as an outcome for evaluating treatment interventions (63), the term ‘disease-specific HRQoL’ has emerged, which entails capturing the impact of symptoms characteristic for a specific diagnosis on an individual’s QoL, and has been proposed to be better able at describing changes in QoL as a consequence of an intervention on symptoms characteristic for the specific patient group (12, 15, 67). Disease-specific HRQoL can thought of as a specific aspect or sub-area of the HRQoL concept (see Figure 2).

Figure 2. Simplified depiction of the connection between quality of life, health and HRQoL, including disease-specific HRQoL (modified from Rand-Hendriksen og Augestad (115).
2.3.2 Factors associated with HRQoL

In accordance with nature of HRQoL as the multi-dimensional, subjective and dynamic (changing) concept, many factors can impact HRQoL to various degrees. The number of factors underpinning to a greater or lesser extent an individual’s HRQoL can best be illustrated by a theoretical and empirically based model of HRQoL. The model of Ferrans et al. has been proposed as to present the most useful model (116, 117). It is a modification of the model developed by Cleary, which combines the biological approaches of the health concept with the more psychological approach of the broader QoL concept, and includes individual and environmental characteristics (118). The model features five levels, from more basic biological to the advanced general level of HRQoL, as well as individual and environmental traits. The five domains are: a) biological and physiological variables, including genetics, cell and organ function; b) symptom status, affected by biological and physiological influences, like the individual’s physical, emotional and cognitive status; c) functional state, representing the individual’s ability to perform activities in the physical, social, psychological and cognitive areas and is again affected by the domains of the previous levels; d) general health perceptions, reflecting the subjective evaluation of the individual affected by all the previous factors, and e) overall QoL, primarily resulting from the four previous health related factors in addition to individual and environmental characteristics. It has been clarified that all five levels including the most basic biological rooted level, each are in different ways influenced by individual characteristics, which include demographic factors, such as age, gender, developmental aspects (in which stage in life is the individual), psychological factors like motivation and emotional response patterns, and other biological elements including body mass index, and by environmental characteristics such as relational situations, living arrangements and geographical area. The relationships described here are the predominant relationships, cumulatively from the most basic level of biological function to overall HRQoL at the other end, and from the individual and environmental characteristics directly affecting each of the five levels described. As well, Ferrans et al. explain that relationships in their model are also reciprocal (117).
2.3.3 Measuring HRQoL

HRQoL has been used increasingly as an important outcome measure during clinical care, as end points in clinical trials and effectiveness research of interventions (116, 118). It further has been suggested as an important measure in assessment of healthcare needs and quality of care in health service research (67, 68, 119, 120). Especially in conditions that are complex, chronic and life-long lacking curative treatment, HRQoL has become an important measure (12, 64). As HRQoL is primarily a subjective concept, and not one that can be directly observed, it is typically measured using self-report questionnaires developed to capture HRQoL as experienced by the individual. HRQoL can be determined using generic measures or disease-specific measures. Generic measurements often combine questions about health-related states or symptoms, such as pain and ability to execute activities of daily living (ADL), for which often composite scores can be generated, in addition to single questions reflecting overall HRQoL. They allow comparison of health and effects of treatment across diseases. Additionally, the are usually employed to investigate the economic burden of diseases and health problems by enabling cost and cost-effectiveness calculations (112). More recently, they have also been proposed for use in health needs assessment (67, 68). Disease-specific measures are those specifically developed for utility in specific diseases, capturing function and burden of specific characteristic symptoms for an individual. A disadvantage with generic measures is that they may not be sufficiently sensitive to record changes as a consequence of treatment interventions in specific diseases, while disease-specific measures challenge comparison of results between different diseases and often are not validated, having unknown or inadequate psychometric qualities (15). Establishing the impact of symptoms of disease and of treatment interventions on HRQoL using generic and / or disease-specific measures, may specifically contribute to guidelines for healthcare provision. This is specifically important for patient populations suffering from incurable long-term conditions like HD, where clinical care is concentrated on alleviating symptoms and at maintaining or improving function and HRQoL.

Both generic and disease-specific measures of HRQoL are, by their definitions self-reports based on the subjective character of the concept. This poses challenges for populations where cognitive impairment and reduced disease awareness, may develop, such as neurological diseases as Alzheimer disease (AD), PD and HD, traumatic brain injury (TBI) as well as within the elderly in normal populations (121). A clear-cut solution is not yet available, but certain studies have looked into what extent ratings performed by proxies, i.e. family of
professional caregivers who know the individual well, can be comparable or assumed representative for patient-rated HRQoL (66, 122). As for HD one work demonstrated comparable ratings of HRQoL between patients with advanced HD generally having moderate to severe cognitive impairment, and their proxies (66).

2.4 ATC

2.4.1 ATC, formal ATC and informal ATC

ATCs can be defined as any item that increases, maintains or improves functional capabilities for individuals with cognitive changes that limit their effective participation in daily activities (70). ATC are external aids that can address disabilities in memory, executive functions such as planning, organization and attention, in addition to reduced psychomotor speed (70, 71, 123). They can be aids accessible and employed by anyone, like smartphones, calendars and post-it notes, to more complex devices (high-tech) that require an application, like software programs developed for the purpose of supporting cognitive disabilities. Based on the literature on ATCs, a differentiation between formal and informal ATCs can be made (70). Informal ATCs comprise external aids introduced by the patients themselves in order to compensate for self-experienced reduced cognitive abilities and to maintain their daily functioning. These are generally familiar to the patient and readily accessible. For instance, if individuals notice they have difficulties remembering appointments, they might begin using an (electronic) calendar with an alarm or consciously start using post-it notes. Formal ATCs on the contrary, require an implementation process and training. They entail assessment of the patients’ cognitive impairment and an application requesting the devices. These devices are specially designed to compensate or lessen the consequences of cognitive disabilities. Examples include apps and devices such as MEMOplanner to assist remembering appointments, and software programs like COGNITASS, a more advanced program for structuring appointments and activities in Norway (124, 125).

During the last three to four decades, development of ATCs have been further facilitated with the arrival of computer technology, which has enabled the production of more advanced computer-based and electronic devices (high-tech ATCs) and increased the possibilities for ATCs tremendously (70, 72, 126). Two important approaches to ATCs are often taken in the research literature – one is based on the relationship between ATCs and specific user groups
or disease populations and the other focusing on the relationship between the ATC and different types of cognitive impairments, i.e., ATCs that support structure and memory impairment, planning, organization and attention, and communication (77). However, among patients with the same diagnosis a large proportion of heterogeneity may exist in the manifestation of cognitive impairments and consequential functional decline. Moreover, one ATC in reality often supports more than one specific cognitive problem.

2.4.2 Factors associated with successful ATC implementation and use

ATCs must be used if individuals are to benefit from them. All individuals have a distinctive combination of abilities, needs and preferences, emotional reaction patterns and social networks. Similarly, individuals within different conditions developing cognitive impairment, have unique combinations of cognitive disabilities and physical, psycho-social and sensory impairments, which all may change over time (i.e., in neurodegenerative conditions) and have unique ways of impacting an individual’s daily functioning and QoL. In order to be properly implemented into and accepted within individual patient’s daily life, thereby achieving optimal benefits from ameliorating negative effects of cognitive impairment, it is vital that an ATC is tailored, to suitably match the individuals’ needs on the basis of their (cognitive) disabilities and preferences as well as their environment (70, 123).

The literature suggests that various factors contribute to successful implementation and use of ATCs, such as the patients’ detailed patterns of cognitive weaknesses and strengths, unique issues related to the natural history of the disorder affecting cognitive profile and symptom change over time requiring similar adaptation of ATCs over time. Further, the presence or development of other physical disabilities that may interfere with cognitive function, the pre-morbid and current system of psychosocial support, important for defining the potential of caregiver involvement with implementation by practice using the ATC, lifestyle preferences and technical complexity of the ATC also come into play (72, 127). From previous work in the field, the various influences associated with ATCs have been divided into three main areas: a) personal influences; b) environmental influences; and c) ATC influences. These all need to be taken into account and assessed in order for ATC to be employed successfully (71, 81, 123, 127). If the patient and ATC are well suited, particular in a social context the ATC will be used and foster positive interactions with the patient. Examples of environmental, personal and ATC factors associated with successful ATC use in the literature are listed in Table 1.
Table 1: Examples of personal, environmental ATC influences affecting the use and no-use of ATC.

<table>
<thead>
<tr>
<th>Person</th>
<th>Environment</th>
<th>ATC</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Patients’ pattern of cognitive weaknesses and strengths</td>
<td>• Culture</td>
<td>• Complexity of the ATC device</td>
</tr>
<tr>
<td>• Unique issues associated with the natural history of the condition</td>
<td>• Financial support</td>
<td>• Dissemination about ATC</td>
</tr>
<tr>
<td>• Emotional and behavioural changes associated with the condition</td>
<td>• Access to ATC</td>
<td>• Design of the ATC</td>
</tr>
<tr>
<td>• Premorbid and current personality and attitudes</td>
<td>• Availability of support from social environment (i.e. family members)</td>
<td>• User friendliness the device</td>
</tr>
<tr>
<td>• Expectations from ATC</td>
<td>• Setting the ATC is used in</td>
<td>• Flexibility of adaptation to additional/secondary (physical, sensory) disabilities.</td>
</tr>
<tr>
<td>• Previous experience with ATC</td>
<td>• Availability of training</td>
<td></td>
</tr>
<tr>
<td>• LIFESTYLE and interests</td>
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</table>

Favorable delivery of ATCs within supportive healthcare services implies considering a wide variety of factors, including personal ones of the individual, the environment and the devices. It is a process reliant on the knowledge of ATCs (providing adequate information about ATC characteristics and possibilities), needs assessment in order appropriately connect the individual with the right ATC, and implementing and maintaining the ATC usage, including training the user (81).

2.5 HEALTHCARE IN NORWAY

Norway, like the other Scandinavian countries, and Canada is thought of as a welfare state with a long tradition of organization and resource allocation within the healthcare systems for patients with long-lasting complex disabilities. The majority of healthcare services are either free of charge (government funded) or subsidized and therefore only partially paid by patients at the point of delivery.

Healthcare in Norway is structured in semi-decentralized manner. This means that the government is responsible for specialized care in hospitals and the municipalities are responsible for primary healthcare. All citizens need to be registered with a general practitioner (GP), who plays an important role in accessing and collaborating with the
specialized healthcare segment, and acts as the gatekeeper for those specialized healthcare services. Healthcare in the municipalities is offered by all types of healthcare professionals at the patients’ home or within institutional facilities, like supportive accommodation, and nursing homes (84). Healthcare services within the municipalities, (primary care/community care) include a wide variety of services, including (re)habilitation comprised of physiotherapy, speech therapy, occupational therapy, (specialized) nursing, day care, respite care, vocational and participation care services and the coordination of care through individual care plans (85). The responsibility of coordinating care through individual care plans and interdisciplinary coordination groups (ansvarsgrupper) has moved increasingly towards primary care in the municipalities, with the white paper on Norwegian Coordination Reform and its implementation in 2012 (128). This reform was engineered to improve the quality and effectiveness of healthcare service delivery by increasing resources to municipalities and imposing on them such a coordinating role. The interdisciplinary coordination groups (ansvarsgrupper) are the proposed way to work with the individual care plans, and have the task of enduring coordinated care and facilitating the communication between disciplines and services (129).

The municipalities are also responsible for the provision of assistive technology (AT), including ATC (86). The Norwegian Labor and Welfare Administration (NLWA) (In Norwegian: NAV, arbeids- og velferdsforvaltning) has the task to assist the municipalities in doing so. The NLWA has a special central, the Central for Assistive Technology (Hjelpemiddelsentralen), which has the responsibility to assist communities with adequate dissemination of ATC to users. This entails acquisition through formal applications and implementation through assessment and training (130).

Healthcare for rehabilitation and habilitation include multidisciplinary healthcare provided both through specialized and in primary healthcare. For example, acute rehabilitation after a TBI or stroke is provided in specialized healthcare facilities and follow-up in the municipalities is based on directives from the specialized institution. In Norway this distinction is made where rehabilitation includes mainly populations with late acquired disabilities, such brain injuries while habilitation includes a wide variety of populations with congenital disabilities or early acquired (before the age of 18 years) disabilities including, individuals with mental disabilities of various origins, i.e., Down syndrome, but also (rare) genetic conditions causing complex disabilities, as well as progressive neurological diseases. Similar to rehabilitation, habilitation in specialized healthcare provides advice to the
municipalities on healthcare and support services after conducting multidisciplinary assessments, generally with the objective of maintaining functional ability and QoL (87). Specialized healthcare services also include national competence centers. These centers provide nationwide information and advice on specific areas, i.e., rare disorders, or specific neuropsychiatric conditions (e.g., competence center for Attention Deficit Hyperactivity Disorder (ADHD), Tourette’s syndrome and narcolepsy). Professionals with different backgrounds provide this advice, develop courses, etc. These centers have no responsibility to provide clinical care.

2.6 RATIONALE FOR THIS THESIS

The present thesis addresses a number of the challenges mentioned in the Introduction. We do not have accurate documentation on the number of individuals with a clinical HD diagnosis living in Norway, or more specifically south-east Norway, but a register does exist at the Center for Rare Disorders at the Oslo University Hospital. By seeking to reach as many patients with a clinical HD diagnosis and including as many patients in all disease stages including those with advanced HD who frequently end up being excluded from research precisely because of their advanced disease, as possible, the present work will contribute to prevalence estimates of the population with HD in Norway.

Further, to the best of our knowledge, no studies to date have investigated healthcare and social needs in populations with HD in Norway, or other Scandinavian countries. In order to understand the extent of the problems and to identify the specific population at risk for unmet health care needs, it would be necessary to assess such a population in the geographical region of interest. This knowledge may be helpful when planning improvement of health care delivery for patients with HD.

As such, the present research aims to contribute to the literature by investigating the level of unmet needs among patients with HD and the use of ATCs as an example of an unmet health need for supportive healthcare as well as factors associated with unmet needs. The measures of outcomes in the populations with chronic disabilities are incomplete if the factors that contribute to HRQoL are not considered. Moreover, the importance of inclusion of HRQoL measurements in healthcare needs assessment has been proposed (67, 68). As is best currently
known, no work conducted in HD has included unmet needs for healthcare and social services as a factor of relevance for HRQoL. With this, the association between the level of unmet needs in patients with HD and HRQoL is investigated herein.
3 AIMS OF THE STUDY

The overall aim of this study was to identify and describe gaps between needs for and provision of healthcare and support services and related factors including health-related quality of life (HRQoL), among patients with HD.

3.1 PAPER I

This paper looked into the following three aims: a) investigate the level of unmet needs for healthcare and social support services among patients with HD; b) examine how the levels of unmet needs are divided across HD disease phases; and c) determine the association between socio-demographic and clinical characteristics and levels of unmet needs. These aims can be further divided based upon addressing the specific research questions:

- Are there unmet needs for healthcare and social support services among patients with HD in south-eastern Norway?
- What are the levels and amount of unmet needs for patients with HD?
- How are levels of unmet needs among patients with HD divided across various types of healthcare services?
- How are unmet needs for healthcare and social support services divided across the HD disease spectrum (stage I-V)?
- Which socio-demographic, clinical and disease factors are associated with the level of unmet needs among patients with HD?

3.2 PAPER II

The aims of paper II were: a) to describe the health status (HRQoL) in a Norwegian cohort of patients with HD; and b) to assess the association between unmet needs for healthcare and social support services and HRQoL, specifically through the following research questions:

- What do the health status profiles of patients with HD in south-eastern Norway look like?
- What is the overall self-reported HRQoL among patients with HD across the disease phases?
- Is the level of unmet needs for healthcare and social support services among patients with HD associated with overall HRQoL?

3.3 **PAPER III**

The purpose of this paper was to a) describe the use of ATC across the disease stages in a Norwegian cohort of patients HD, and to b) investigate the association between ATC and HRQoL, addressing the following specific research questions:

- How can the use of ATC and related aspects be described for patients with HD across the disease stages I-V?
- Is formal and/or informal ATC use associated with improved HRQoL among patients with HD in south-eastern Norway?
4 SUBJECTS AND METHODS

4.1 PARTICIPANTS AND RECRUITMENT PROCEDURE

For the present cross-sectional population-based study, all patients who were a) residing in the south-eastern region of Norway (a population of 2.7 million inhabitants), and b) had a clinical diagnosis of HD were eligible for participation in the study. No exclusion criteria were defined as the goal was to recruit as many patients with HD as possible.

A total of 158 eligible patients, corresponding to a prevalence of 5.9/100,000, were identified and invited to participate through the regional academic center, Oslo University Hospital (OUS), at the Department of Neurohabilitation, Department of Neurology and the Department of Medical Genetics and the National Advisory service, Centre of Rare Disorders, offering guidance to patients, families and healthcare professionals, and the Vikersund Rehabilitation Centre. They each composed a list of eligible patients. Each of these lists was first carefully reviewed by the contact person(s) at the respective departments. Patients that: a) had died; b) were residing outside the health-care region of south-eastern Norway; and c) patients without a clinical diagnosis of HD were removed from the list. Next the lists were verified by the PhD researcher for double appearance of eligible patients - patients may have been in contact with more than one department - to avoid eligible patients contacted by several departments. The department at which the patient was being treated/followed up with at the time of the study was set as the primary contact for recruitment. A written invitation, enclosing information on the study and an informed consent form with a pre-paid reply envelope was sent to all eligible patients. Patients received a written reminder to participate when no response was sent to all eligible patients. Patients received a written reminder to participate when no response was received after six weeks. When no reply came after an additional four weeks, patients were followed up with by telephone up to two times by the respective departments. To further attempt to reach all eligible patients with HD in south-eastern Norway, we cooperated with the national professional network for community care, Huntington fagnettverk, and the Norwegian HD association (Landsforeningen for Huntington sykdom), by providing details about the study at meetings as well as for an announcement on the webpage of the HD association. This did not generate additional eligible patients.
During the study period 88 patients gave their informed consent and were included. To ensure the clinical diagnosis of two of these patients, their medical records were carefully reviewed by a medical expert HD clinician. This resulted in exclusion of two patients as they did not have sufficient clinical symptoms to have been formally diagnosed with HD at the time of inclusion. A final number of 86 patients (54.4%) out of 158 eligible patients were included in the study. Of the 70 patients who did not participate, 27 declined participating in the study while 43 patients did not respond. Furthermore, of the 86 included in Paper I, two patients did not return the EQ-5D-3L questionnaire for HRQoL leading to 84 patients included in Papers II and III. For an overview of the patient recruitment flow see Figure 3.

Figure 3. Flow chart of recruitment process

Total number of invited participants: N = 158

- Excluded: N = 2
- Did not participate: N = 70
  - Did not reply: N = 43
  - Did not wish to participate: N = 27

Included: N = 86

Survey interviews conducted: N = 86

EQ-5D-3L questionnaires returned: N = 84
  - Including EQ-5D VAS: N = 82

Paper I

Paper II, III

Socio-demographic and clinical disease characteristics for the complete sample of 86 patients are presented in Table 2 and 3. Of the complete sample of 86 patients included, 54.7 patients were male. Patients’ years of age and years of education showed median values and interquartile ranges (IQR) of 58 and 15, and 12 and 5 respectively. There was an equal division between manual (n = 41 (48%)) and non-manual (n = 42 (49%)) occupation. The majority of the patients lived at home.
Table 2: Socio-demographic statistics for complete sample and divided across V disease stages

<table>
<thead>
<tr>
<th>Variables</th>
<th>Categories</th>
<th>Complete sample (N=86)</th>
<th>Stage I (n=12)</th>
<th>Stage II (n=23)</th>
<th>Stage III (n=23)</th>
<th>Stage IV (n=15)</th>
<th>Stage V (n=17)</th>
<th>Sign.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age*</td>
<td>Median (IQR)</td>
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<td>49 (20)</td>
<td>54 (21)</td>
<td>58 (11)</td>
<td>58 (8)</td>
<td>59 (16)</td>
<td>0.104</td>
</tr>
<tr>
<td>Education (years)/§</td>
<td>Median (IQR)</td>
<td>12 (5)</td>
<td>13 (5)</td>
<td>12 (7)</td>
<td>11 (2)</td>
<td>12 (6)</td>
<td>11 (6)</td>
<td>0.168</td>
</tr>
<tr>
<td>Gender</td>
<td>Female</td>
<td>39 (45)</td>
<td>5 (42)</td>
<td>9 (39)</td>
<td>7 (37)</td>
<td>8 (53)</td>
<td>10 (59)</td>
<td>0.625</td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td>47 (55)</td>
<td>7 (58)</td>
<td>14 (61)</td>
<td>12 (63)</td>
<td>7 (47)</td>
<td>7 (41)</td>
<td>0.168</td>
</tr>
<tr>
<td>Education</td>
<td>Lower (≤ 12 years)</td>
<td>52 (60.5)</td>
<td>5 (42)</td>
<td>12 (52)</td>
<td>15 (79)</td>
<td>9 (60)</td>
<td>11 (65)</td>
<td>0.424</td>
</tr>
<tr>
<td></td>
<td>Higher (&gt; 12 years)</td>
<td>34 (39.5)</td>
<td>7 (58)</td>
<td>11 (48)</td>
<td>4 (21)</td>
<td>6 (40)</td>
<td>6 (35)</td>
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<tr>
<td>Civil status</td>
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<td>4 (33)</td>
<td>7 (30)</td>
<td>9 (47)</td>
<td>8 (53)</td>
<td>8 (47)</td>
<td>0.666</td>
</tr>
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<td>Married</td>
<td>50 (58)</td>
<td>8 (67)</td>
<td>16 (70)</td>
<td>10 (53)</td>
<td>7 (47)</td>
<td>9 (53)</td>
<td>0.587</td>
</tr>
<tr>
<td>Occupation*</td>
<td>Manual</td>
<td>41 (48)</td>
<td>5 (42)</td>
<td>10 (43.5)</td>
<td>12 (63)</td>
<td>6 (40)</td>
<td>8 (47)</td>
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</tr>
<tr>
<td></td>
<td>Non-manual</td>
<td>42 (49)</td>
<td>7 (58)</td>
<td>13 (56.5)</td>
<td>7 (37)</td>
<td>8 (53)</td>
<td>7 (41)</td>
<td>0.001</td>
</tr>
<tr>
<td>Occupational status</td>
<td>Employed</td>
<td>14 (16)</td>
<td>11 (92)</td>
<td>3 (13)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>0.001</td>
</tr>
<tr>
<td></td>
<td>Unemployed</td>
<td>72 (84)</td>
<td>1 (8)</td>
<td>20 (87)</td>
<td>19 (100)</td>
<td>15 (100)</td>
<td>17 (100)</td>
<td>0.001</td>
</tr>
<tr>
<td>Informant</td>
<td>Patient</td>
<td>27 (31)</td>
<td>9 (75)</td>
<td>14 (61)</td>
<td>4 (21)</td>
<td>0 (0)</td>
<td>15 (100)</td>
<td>0 (0)</td>
</tr>
<tr>
<td></td>
<td>Patient &amp; informant/informant only</td>
<td>59 (69)</td>
<td>3 (25)</td>
<td>9 (39)</td>
<td>15 (79)</td>
<td>0 (0)</td>
<td>15 (100)</td>
<td>17 (100)</td>
</tr>
<tr>
<td>Housing situation</td>
<td>Living at home</td>
<td>54 (63)</td>
<td>12 (100)</td>
<td>23 (100)</td>
<td>13 (68)</td>
<td>6 (40)</td>
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<tr>
<td></td>
<td>Not living at home</td>
<td>32 (37)</td>
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<td>6 (32)</td>
<td>9 (60)</td>
<td>17 (100)</td>
<td>0.878</td>
</tr>
<tr>
<td>Residence</td>
<td>Rural</td>
<td>13 (15)</td>
<td>1 (8)</td>
<td>4 (17)</td>
<td>2 (10.5)</td>
<td>3 (20)</td>
<td>3 (18)</td>
<td>0.001</td>
</tr>
<tr>
<td></td>
<td>Urban</td>
<td>73 (85)</td>
<td>11 (92)</td>
<td>19 (83)</td>
<td>17 (89.5)</td>
<td>12 (80)</td>
<td>14 (82)</td>
<td>0.001</td>
</tr>
</tbody>
</table>

Note: IQR: Interquartile range; Group comparison across the five disease stages performed using Chi-square tests for independent samples (categorical values). * normally distributed and therefore reported result from ANOVA). § not normally distributed therefore performed and reported Kruskal-Wallis test. # 3 responses missing (1 in stage IV and 2 in stage V). Remaining proportions and comparisons are crosstabs / Chi-square. IQR: Interquartile range.

The median value and IQR for disease duration for the complete sample was 6 and 7 years with a range of less than 1 year to 19 years. Of the total included patients 12 were in disease stage I, 23 in stage II (22 in Papers II and III), 19 in stage III, 15 in stage IV (14 in Papers II and III) and 17 in stage V.
Table 3: Sample Clinical Characteristics

<table>
<thead>
<tr>
<th>Variables</th>
<th>Complete sample (N = 86)</th>
<th>Stage I (n = 12)</th>
<th>Stage II (n = 23)</th>
<th>Stage III (n = 19)</th>
<th>Stage IV (n = 15)</th>
<th>Stage V (n = 17)</th>
<th>Sign.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disease duration</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Median (IQR)</td>
<td>6 (7)</td>
<td>2 (2)</td>
<td>5 (6)</td>
<td>7 (5)</td>
<td>8 (7)</td>
<td>10 (8)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Total FAS score</td>
<td>15 (17)</td>
<td>24 (2)</td>
<td>20 (2)</td>
<td>15 (4)</td>
<td>5 (2)</td>
<td>0 (3)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Independence score</td>
<td>70 (35)</td>
<td>97 (9)</td>
<td>80 (5)</td>
<td>65 (10)</td>
<td>45 (20)</td>
<td>20 (6)</td>
<td>P &lt; 0.001</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Comorbid conditions</th>
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<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>None</td>
<td>50 (58)</td>
<td>7 (8)</td>
<td>10 (12)</td>
<td>9 (10)</td>
<td>11 (13)</td>
<td>13 (15)</td>
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<td>0 (0)</td>
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<td></td>
</tr>
<tr>
<td>Heart and vessels</td>
<td>8 (9.3)</td>
<td>2 (2.3)</td>
<td>2 (2.3)</td>
<td>2 (2.3)</td>
<td>2 (2.3)</td>
<td>0 (0)</td>
<td></td>
</tr>
<tr>
<td>Lung</td>
<td>2 (2.3)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>2 (2.3)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td></td>
</tr>
<tr>
<td>Cancer</td>
<td>4 (4.8)</td>
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<td>1 (1.2)</td>
<td>1 (1.2)</td>
<td>1 (1.2)</td>
<td>1 (1.2)</td>
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</tr>
<tr>
<td>Musculoskeletal</td>
<td>5 (5.8)</td>
<td>0 (0)</td>
<td>3 (3.5)</td>
<td>2 (2.3)</td>
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</tr>
<tr>
<td>Other</td>
<td>9 (10.5)</td>
<td>2 (2.3)</td>
<td>2 (2.3)</td>
<td>2 (2.3)</td>
<td>0</td>
<td>3 (3.5)</td>
<td></td>
</tr>
<tr>
<td>Multiple</td>
<td>7 (8.1)</td>
<td>0 (0)</td>
<td>5 (5.8)</td>
<td>1 (1.2)</td>
<td>1 (1.2)</td>
<td>0 (0)</td>
<td></td>
</tr>
</tbody>
</table>

Note: FAS: Functional Assessment Scale; IQR: Interquartile range; Group comparisons are completed using Chi-square tests for categorical variables and Kruskall-Wallis for continuous / interval variables, as none of the continuous variables were normally distributed.

4.2 DATA COLLECTION PROCEDURES

Data were collected during study visits as either outpatient visits or at the patients’ home from January – August 2014. Study visits were scheduled with the patient and/or primary carer upon receiving the informed consent form. All study visits were conducted by two trained clinical psychologists with experience in the field of HD (RMvW and EH). Data were collected by means of a survey interview and patient ratings mainly by using standardized measurements. Data collection was performed in the same order at each study visit, starting with collection of patients’ socio-demographic, clinical and disease-specific characteristics followed by an evaluation of cognitive impairment and functional ability, and lastly,
evaluation of the patients’ needs for healthcare and social services along with recording to what extent these needs were being met through the provision. Information from medical records was used to estimate the years of education and level of education for four patients, to determine the type of occupation for three patients and to estimate disease duration (number of years with clinical HD diagnosis) for three patients. The CAG repeat length in the HTT-gene, which served as confirmatory information of HD diagnosis was lacking for three patients and could not be retrieved from the patients’ medical records. At the end of each visit, patients were requested to complete a generic self-report questionnaire for HRQoL. Based on the characteristics of HD, a number of patients were unable to complete the questionnaire independently (i.e. as a consequence of motor impairment). They were assisted by their primary carers who were family members or healthcare personnel involved with the patient daily. The primary carers were explicitly instructed to help the patient complete the form in order to obtain patient ratings. The questionnaire was completed by the primary carer on behalf of eight patients with advanced HD. As a self-report questionnaire was employed, the carers were specifically instructed to attempt to reflect the health status experienced by the patient to the best of their capability and to leave the question unanswered if they considered themselves unable to do so. Open questions became missing values. If the questionnaire was not filled out during the study visit, the patient and/or carer was provided with a pre-paid response envelope to return the questionnaire after the study visit. When questionnaires were not returned, patients or their caregivers were followed up with by telephone. The same procedures for data collection were followed for all patients included in the three studies in this thesis. The data used in paper I-III varied depending on the aims investigated in the papers.

4.3 DATA COLLECTED AND MEASUREMENTS/ASSESSMENTS USED

Table 4 provides an overview of the study variables, featuring the data collected and measurements used for the studies this thesis entailed. It is further indicated whether the variables were employed as independent or dependent variables. Certain variables, namely variables of specific interest (related to healthcare needs and HRQoL), were utilized both as independent and as dependent variables for different studies.
<table>
<thead>
<tr>
<th>Variables and measurements</th>
<th>Paper I</th>
<th>Paper II</th>
<th>Paper III</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Socio-demographics; independent variables</strong></td>
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<td></td>
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<tr>
<td>Age at time of inclusion</td>
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<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Gender</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Education (years)</td>
<td>X</td>
<td>X</td>
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<tr>
<td>Educational level</td>
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</tr>
<tr>
<td>Occupation (type)</td>
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</tr>
<tr>
<td>Employment</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Civil status at time of inclusion</td>
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<td>X</td>
</tr>
<tr>
<td>Informant</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Housing situation at time of inclusion</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Residence at time of inclusion</td>
<td>X</td>
<td>X</td>
<td>X</td>
</tr>
<tr>
<td><strong>Clinical and disease-specific characteristics; independent variables</strong></td>
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<td></td>
<td></td>
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<tr>
<td>Comorbid conditions</td>
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<td>X</td>
<td>X</td>
</tr>
<tr>
<td>Disease duration at time of inclusion</td>
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<td>X</td>
</tr>
<tr>
<td>Total Functional Assessment Scale (FAS) score (UHDRS)</td>
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<tr>
<td>Independence score (UHDRS)</td>
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<tr>
<td>Total Functional Capacity (TFC) (UHDRS)</td>
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<td>Disease phase</td>
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<td>Level of cognitive impairment</td>
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<td><strong>Variables of interest Health care needs, provision and unmet needs – independent variables</strong></td>
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<td>Health Needs variables (Needs and Provision Complexity Scale, NPCS)</td>
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<td>NPCS total needs</td>
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<tr>
<td>NPCS total provision</td>
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<tr>
<td>NPCS total unmet needs</td>
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<tr>
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<tr>
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<td>NPCS corresponding sub-scales, needs, provision and unmet needs*</td>
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<td>Overall health-related quality of life (EQ-5D-3L VAS score)</td>
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<tr>
<td>EQ-5D-3L dimension mobility</td>
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<tr>
<td>EQ-5D-3L dimension self-care</td>
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</tr>
<tr>
<td>EQ-5D-3L dimension usual activities</td>
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<td>EQ-5D-3L dimension pain/discomfort</td>
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<td>EQ-5D-3L dimension anxiety/depression</td>
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<tr>
<td><strong>Assistive technology for cognition (ATC)</strong></td>
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<tr>
<td>ATC formal / informal</td>
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</tr>
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<td>ATC use</td>
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<td>ATC training</td>
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<td><strong>Variables of interest – dependent variables</strong></td>
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<td>Total level of unmet needs (NPCS total score)</td>
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<td>Total level unmet needs for domain: health and personal care (total NPCS domain score)</td>
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<td>Total level of unmet needs for domain: social care and support (total NPCS domain score)</td>
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<tr>
<td>Overall health-related quality of life (EQ-5D-3L VAS score)</td>
<td>X</td>
<td>X</td>
<td></td>
</tr>
</tbody>
</table>

*See appendix 1 for an overview of the sub-scales included as part of the NPCS clinician version, needs (Needs), provision (Gets) and unmet needs, where unmet needs are calculated based needs and provision.
4.3.1 Data collected

4.3.1.1 Socio-demographic characteristics and clinical characteristics (Papers I - III)

The socio-demographic and clinical disease characteristics recorded for the purpose of the present study (Papers I, II and III) served as independent variables (for an overview see table 4). This information was further classified as follows: marital status - single (single, separated, widowed) vs. married (married/ living together), residence - rural vs. urban, educational level - lower education (≤ 12 years education) vs. higher (> 12 years education), occupation - manual vs. non-manual, employment - into employed vs. unemployed, housing situation - living at home vs. not living at home (patients in care facilities, and assisted living facilities), and how the information was obtained (informant) - patient alone vs. patient with informant and informant only. In paper II, the latter category was further separated into patients with informant vs. informant only, resulting in three categories. Comorbid conditions were divided into seven categories, being none, neurological, heart and vessels, lung, cancer, musculoskeletal, other and multiple, for paper I and as having comorbid conditions (yes) vs. having no comorbid conditions (no(ne)) for papers II and III. The disease-specific characteristic, disease duration in years, was calculated as the date of formally obtained clinical diagnosis of HD subtracted by the date of the conducted study visit (Papers I, II and III) and the level of cognitive impairment (mild, moderate, severe; Paper III) was evaluated based on a scoring key. Functional ability was measured by the UHDRS, Functional Assessment Scale (FAS), Independence Scale (IS) and Total Functional Capacity (TFC) (Papers I, II and III), further serving as independent variables of disease-specific characteristics and described in more detail in the following.

4.3.1.2 General evaluation of cognitive impairment (Paper III)

A general clinical evaluation of cognitive impairment was performed during each study visit, using a pre-defined scoring key, describing three categories reflecting three levels of cognitive impairment being mild, moderate, and severe cognitive impairment along with a fourth category, unable to evaluate (Paper III). The information on which the general cognitive evaluation was based on, included: a) information concerning cognitive function and symptoms as conveyed by the patient; b) information on cognitive functioning and symptoms relayed by the patients’ relative and/or professional caregiver if available; and c) the clinician’s observation during the evaluation. The four categories were defined as follows:
1) mild cognitive impairment: a slight reduction in one or more cognitive domains causing changes in or impaired ability to perform daily activities and the need for minor adjustments in order to be able to perform everyday activities; and next-of-kin begins noticing changes in the patient

2) moderate cognitive impairment: overt cognitive impairment in more than one cognitive domain as compared to premorbid function, with clear need for support/adjustments in order to continue carrying out daily activities and no longer being able to perform complex tasks, changes evident to next-of-kin

3) severe cognitive impairment: severe cognitive dysfunction in all domains, impaired communication, no longer being able to carry out daily activities or maintain self-care and in need of around-the-clock care

4) unable to evaluate: the rater clinicians are in doubt about the patients’ cognitive function as a consequence of lack of comparative information from next-of-kin or primary healthcare professional. The result of the evaluation regarding the level of cognitive impairment was recorded as part of the survey interview form.

4.3.1.3 Information on ATC (Paper III)

For the purpose of Paper III, information on ATC was included in the survey interview and recorded on the studies’ survey form. The following information on ATCs was collected: a) whether patients had ATC and whether these were informal or formal ATCs. Based on the literature, a distinction can be made regarding external devices such that patients with cognitive impairment may commence use of their own in order to compensate for impairment in cognitive functioning, and which incorporate main stream products such as cell phones, calendars, alarm clocks, etc. which are defined as informal ATCs. On the other hand, external devices that are specifically developed and designed to support patients with cognitive impairment and that can only be acquired through a formal process of application and implementation are defined as formal ATCs; b) whether the patients used ATCs; c) whether they had been informed about the possibility of ATCs; d) whether a formal ATC needs assessment was undertaken; and e) whether they had received training for their ATC.
4.3.2 Measurements used

4.3.2.1 UHDRS, Functional assessment (Papers I – III)

The UHDRS Functional assessment scales were used in order to assess patients’ functional abilities (Paper I, II and III). The UHDRS is a standard tool developed especially for evaluation of patients with HD that is comprised of four domains of assessment, including motor, cognitive, behavioral and functional assessment (45, 46). The functional assessment, employed herein, makes use of three scales. Firstly, there is a scale to establish TFC, rating the ability to engage in occupation, finances management, domestic chores and perform ADL. The TFC has a scoring range of 0 – 13 and classifies patients with HD into five functional disease stages. The convention is as follows (45, 131, 132):

- Stage I represents a TFC score of 11 – 13;
- Stage II represents a TFC score of 7 – 10;
- Stage III represents a TFC score of 3 – 6;
- Stage IV represents a TFC score of 1 – 2; and
- Stage V represents a TFC score of 0.

In the present study UHDRS TFC was employed to divide the participating patients with HD into disease stages I – V in accordance with this convention. Secondly, the UHDRS functional assessment features a daily living checklist of 25 tasks that are rated as either being capable (1) or being incapable (0) to perform the task, namely the FAS. This scale has a scoring range of 0-25. The third scale incorporated into the UHDRS functional assessment is the IS, where the clinician indicates the level of independence on a scale of 5 – 100. Of note is that the FAS and IS are included as disease-specific characteristics. For all three scales higher scores indicate better functioning (45).

4.3.2.2 Needs and Provision Complexity Scale (NPCS) (Papers I, II)

The clinician version of the NPCS was used to identify unmet needs for healthcare and social support services among the HD patients included in this study (Paper I and II) (133, 134). Besides the clinician version, a caregiver and a patient report version of the NPCS have also been developed. However, these latter two are not utilized in the present study. The NPCS clinician version is a 15-item measure comprising six sub-scales and a total scoring range of 0 – 50 that covers low and high levels of needs, and is completed by the clinician in
collaboration with the patient. It consists of two parts: Part A, *NPCS Needs*, and Part B, *NPCS Gets*. Part A (Needs) is a clinical evaluation of the patients’ need for health and social and support care services while Part B (Gets) is a mirror image of NPCS Needs and is an evaluation of the healthcare and social care services provided to the individual. Additionally, the NPCS has two main domains, the domain *Health and personal care needs* (score 0 – 25), comprising of the sub-scales: *Healthcare* (score 0 - 6), *Personal care* (score 0 - 10) and *Rehabilitation* (0 - 9), and the domain *Social care and support needs* (score 0-25), constituting subscales *Social and family support* (score 0 -13), *Equipment* (score 0 – 3) and *Environment* (score 0 - 9). The total level of *unmet needs* is derived by subtracting the total Gets score from the total Needs score. The levels of unmet needs for the two domain scores and for each of the six subscales were calculated in the same manner. Higher scores indicated greater levels of needs, gets and unmet needs. The scale also allows recording of qualitative information, though this was not included in the data analyses of this thesis.

The NPCS was originally developed in the United Kingdom specifically for detecting healthcare and social support needs and to what extent those needs were met among patients with LTNC. It has shown robust psychometric qualities (134, 135). The scale can be employed at the individual level where it can identify and monitor the changing healthcare and social support needs and those for provided services over time along the care pathway, while at the population level, the scale can inform on the gaps between healthcare service needs and provision of services. The NPCS Needs and Gets evaluation were performed on the basis of clinical and functional information (needs) attained as part of the survey (e.g., functional capacity assessment) and the information supplied by the patient and / or family or professional caregiver (gets) during the survey.

For the purpose of the study the NPCS was translated by native speakers from English to Norwegian and then back-translated to English to verify accuracy. Researchers and clinicians in the field of healthcare services and HD reviewed the translations. The NPCS clinician version can be found in the Appendix (133, 134).

4.3.2.3 4.3.2.3 EQ-5D-3L (Papers II, III)

As a generic measure of HRQoL, the EQ-5D-3L questionnaire was used (Papers II and III). It is a generic self-report measure developed by the EuroQol Group (136). The measure
included two parts. The first part can aid in the description of health profiles and to generate (global) health indices of populations with a weighted total value for HRQoL (Paper II). It contains five single-item dimensions of health - mobility, self-care, usual activities, pain/discomfort and anxiety/depression rated on a severity scale of three levels being no problem (1), slight problem (2) and major problem (3). The second part is often a general measure for HRQoL (Papers II and III). This part is a Visual Analogue Scale (VAS) with a scoring range of 0 - 100, 0 indicating the worst health state and 100 the best health state. The EQ-5D-3L has been applied to several conditions, including HD (12, 66, 137), and has been found to be valid for use in the Norwegian population (138). In this study health profiles for the five HD stages were described using the level scores and the VAS scores were used as an overall measure of perceived HRQoL.

4.4 STATISTICAL ANALYSES

All statistical analyses were conducted using Statistical Package for Social Sciences (SPSS), version 21.0 (SPSS Inc., Chicago, USA). The data were screened for extreme values, defined as ≥ 3 standard deviations from the mean value and for missing data before conducting the statistical analyses (Papers I, II, and III). Certain extreme values were identified among the sample characteristics and the NPCS scores. They were verified against the original data material. They were found to be true values and therefore, they were kept and included in the data analyses. Certain missing data were identified as well. The majority of these could be determined based on information from the patients’ medical records, except for the occupation of three patients (< 4%). Further, the CAG repeat number of three patients (< 4%) could not be recovered from their medical files. Additionally, information on one item of the NPCS scale was missing for one patient (< 2%) (Paper I and II). As a result, we were unable to calculate scores for a number of the NPCS scales for one patient. Furthermore, explorative analyses investigating the distribution of independent and dependent continuous variables and large-scale variables of age, years of education, disease duration, Total FAS score, IS score, NPCS total score and total domain scores and the EQ-5D VAS score were conducted. This demonstrated that disease duration, total FAS score (Papers I, II and III), total NPCS score (Papers I and II) and NPCS total domain scores (Paper II) were not normally distributed.

Descriptive statistics calculated in Papers I - III included mean values and standard deviation (SD) for normally distributed variables and median values and interquartile range (IQR) for
non-normally distributed variables. Frequencies and proportions (%) were computed for the nominal variables described in Papers I - III. All descriptive analyses presented in Papers I - III, were performed for the entire sample and across disease stages I - V, and classified using the TFC score. Overall group differences between the five HD stages for all variables in Papers I, II and III, were calculated with One-way analysis of variance (ANOVA) for normally distributed variables, Kruskal-Wallis for k-samples for non-normally distributed variables, and Chi-square cross-tabulations for overall group differences between nominal variables.

The investigation of associations between independent variables and dependent variables in Papers I - III was performed with different approaches of multivariate regression analyses. In Paper I, multivariate logistic regression analyses were conducted in order to determine the sociodemographic, clinical and disease factors associated with the level of total unmet needs for healthcare and social support services and with the levels of unmet needs for the health and personal care and the social care and support services domains. The regression analyses were performed on the group of patients with unmet needs for these three variables. The median value was employed to categorize the total NPCS and the NPCS domain scores, Health and personal care and Social care and support into two groups of high levels vs. low levels of unmet needs (respectively with median values 1 – 6 = low level vs. median value ≥ 6 = high level; median value 1-3 = low level vs. > 3 high level; 1 & 2 = low level vs. > 2 = high level). The five disease stages were collapsed into three groups of early (stage I & II), middle (stage III) and advanced (stage IV and V) phases because of the small patient number. Results from the logistic multivariate analyses were presented with odds ratios (OR) with 95% confidence intervals. Hosmer-Lemeshow goodness of fit statistics were calculated. In Paper II multivariate linear regression analyses using the hierarchical approach were carried out to examine the relationship between unmet needs and patients’ self-reported HRQoL. Independent variables were entered in three blocks, starting with unmet needs for healthcare and social support services, followed by clinical and disease-related variables and then socio-demographic variables. Results were reported in adjusted R², R² change, and standardized Beta (β) values with confidence intervals. Paper III examined the relationship between use of formal and informal ATCs on self-reported overall HRQoL using standard multivariate regression analyses. As the disease-related variables, TFC and overall cognitive impairment, were highly correlated with one another (> 0.09) and both were known to be important for both variables interest (ATC use) and for the dependent variable (HRQoL), they were entered
as disease-specific control variables in separate models. Adjusted $R^2$, standardized Beta ($\beta$) values with confidence intervals and partial $r^2$ were utilized to present the results. The Variance Inflation Factor (VIF) was computed to establish multicollinearity between independent variables when performing the multivariate regression analyses in Paper II and III.

Which independent variables were the most important predictors and control variables to be entered in the multivariate regression models, in addition to the independent variables of interest (socio-demographic, clinical and disease factors, total NPCS score (total level of unmet needs) and formal and informal ATC use for Papers I, II and III respectively, was based on the results of simple logistic (Paper I) and simple linear (Papers II and III) regression analyses between independent variables and dependent variables (NPCS total and domain scores and EQ-5D VAS) in Paper I and Papers II and III respectively, and on clinical importance and existing literature. Simple regression analyses were conducted prior to performing the multivariate regression analyses. Inspection of violations for performing the multivariate regression analyses resulted in a logarithmical transformation of the NPCS total score in Paper II and of disease duration in Papers II and III. Residual analyses were carried out and influential data points were investigated using Cook’s distance (Papers II and III), uncovering no outliers or influential data points among variables included in the analyses. Variables with correlations of $> .70$ were not part of the multivariate regression analyses. All statistical tests performed in Papers I - III were two-sided with the significance level set at 0.05.

### 4.5 Ethical Considerations

The study was conducted in accordance with the ethical guidelines of the Helsinki declaration. Approval for the study was obtained from the Regional Committee for Medical and Health Research Ethics (ref. 2013/2089). Prior to inclusion in the study, as mentioned earlier, informed consent was obtained for all patients.

Regarding signing the informed consent, specific considerations had to be taken into account with regards to clinical characteristics of patients with HD. First for all, patients may be unable to sign informed consent based on their movement disorder, while still having sufficient mental and cognitive abilities. If this was the case informed consent was signed by the patients’ primary caregiver, for the most part a close family member. Secondly, as a
consequence of the evolving cognitive impairment during the course of the disease causing
general cognitive impairment (dementia) in advanced disease, as well as because of the
reduced or lacking communication skills, capacity to provide informed is often reduced or
absent in patients with advanced HD. This poses challenges in HD research, and often results
in these patients being unable to participate in studies. This is obviously very unfortunate, and
it is important to attempt to also include patients with advanced HD. Regarding this specific
study, it was significant to examine the unmet needs for healthcare services throughout the
entire disease spectrum. Specific effort was expended into including patients with advanced
HD despite the challenges related to obtaining informed consent to attain a representative
sample of the HD population in south-eastern Norway. With HD, usually, family members are
closely involved in the patients’ care, and therefore they can be considered to be able to make
an informed decision regarding study participation on behalf of those patients. Additionally,
patients with advanced HD frequently have an appointed legal representative, especially if in
advanced care and when lacking a close relative. When patients were unable to provide
informed consent themselves, informed consent was obtained from their primary caregiver or
legal representative.

All identifying information was removed for the data analyses and for reporting results in
scientific publications and presentations in order to prevent the possibility of linking
information to particular participating patients. Yet, as HD is a rare disease, extra caution had
to be taken regarding the possibility of connecting data to individual participating patients,
even if it was anonymized and limited for reporting only on group levels. For example, there
are areas in Norway where there are living several patients with HD and there are also regions
with nearly no patients existing, or just one, and thus including certain demographic
information that may have been of interest to the study had to be considered on a case-by-case
basis.

Overall, the project was not considered to cause any risk for the patients. Furthermore, the
study data and procedures for collecting the study data were believed not to cause any harm or
significant discomfort to the patients included. In light of this, it was necessary to account for
the burden to the patient and/or their carer if included in the study and choices and
compromises regarding the type and amount of data collected had to be made. All patients
were informed that they could withdraw from the study at any time, without giving the reason
for their withdrawal and without consequences to the treatment they were receiving.
During study visits, if patients were found to be in need of healthcare they were not receiving, the relevant healthcare institution was contacted by the study clinicians.
5 MAIN RESULTS

5.1 Paper I

The goals of this paper were to examine the gaps between needs and provision of healthcare and social support services (unmet needs) among patients with early to advanced HD (stages I – V) living in south-eastern Norway, how the levels of those unmet needs were divided across disease stages and which socio-demographic and clinical disease features were associated with levels of unmet needs.

It was hypothesized that there would be identification of substantial levels of unmet needs for healthcare and social support services across all disease phases and that the advanced disease phase (stages IV and V) would be strongly associated with levels of unmet needs.

Generally, increasing median values were found across disease stages I – IV with stable median values for stages IV and V for Needs, Gets and Unmet Needs for the total NPCS score, for the two domains and for the sub-scales Personal care and Accommodation. Similar results were obtained for the proportion of patients with unmet needs, and unmet needs were most frequent for sub-scales Rehabilitation and on Social and family support. The levels of unmet needs were highest and most common for patients in the middle phase (stage III) of the disease. For the total level of unmet needs odds for increased levels of unmet needs increased by 3.5 (OR = 3.5) and 1.4 (OR = 1.4) fold in the middle phase (stage III) and the advanced phase (stages IV and V) respectively, and for the domain Health and personal care the odds ratio (OR) increased by 2.77 (OR = 2.77) and 2.20 (OR = 2.20) fold. Both for the total level of unmet needs and for the domain, Health and personal care, it was observed that information obtained from the patient alone tended to decrease the level of unmet needs (OR = 0.52 and OR = 0.57 respectively). Higher education seemed to diminish the level of unmet needs for the domain of Health and personal care (OR = 0.48), whereas higher education tended to increase the odds of having higher levels of unmet needs on the domain Social care and support (OR = 1.4). Additionally, the presence of comorbid conditions tended to reduce the level of unmet needs for the Social care and support domain (OR = 0.65).

Briefly, it was concluded that there indeed were considerable levels of unmet needs across all disease stages, but that levels of unmet needs were most pronounced among patients in the middle phases (stage III) of HD rather than the advanced phase (stages IV and V). Yet, also whether information was obtained with an informant present, educational level and
comorbidity variously played in the levels of unmet needs for total unmet needs and the two main domains. In order to provide adequate healthcare across the disease spectrum not only clinical but also systematic assessment and monitoring of needs for healthcare and support services should be carried out.

5.2 Paper II

The intent of this study was to describe the health status (HRQoL) among patients with HD and to study the association between the level of unmet needs for healthcare and support services and overall HRQoL. It was hypothesized that a higher level of unmet needs was associated with a lower overall HRQoL.

The mean overall HRQoL (EQ-5D-3L VAS) for the complete sample of HD patients was found to be 52.1 (SD = 26.1). Patients with advanced disease (stages IV and V) reported the lowest overall HRQoL (mean = 35, SD = 25.5 and mean 38.3, SD = 20.9, respectively). However, the health profile for the five EQ-5D health dimensions was most variable in patients in the middle phase (stage III). Regarding levels of unmet needs and HRQoL, patients with higher levels of unmet needs generally seemed to have slightly lower average HRQoL.

The model resulting from hierarchical regression analyses, where total levels of unmet needs, clinical disease characteristics and demographic control variables were entered consecutively, explained 42% of the total variance of HRQoL. Total level of unmet needs had a significant effect explaining 9.2% of the total variance. The model improved by 30.2% when clinical disease-specific characteristics were accounted for, but only TFC exhibited a significant effect. Demographics only added 2.6% of variance. A negative β-value was found for level of unmet needs (β = -2.228; p = 0.018), indicating that a higher level of unmet needs was associated with lower HRQoL, while a positive β-value was elicited for TFC (β = 0.564; p < 0.001), suggesting that better functioning was associated with higher HRQoL.

It was concluded that the averages for HRQoL among patients with HD were below average as early as stage III, in line with other studies on HRQoL and HD. Further, the variation regarding the health profiles in stage III may have reflected the variation in symptoms during this transitional disease stage. Although TFC was observed to have the largest impact on HRQoL, the total level of unmet needs for healthcare and social support services on its own
significantly contributed to self-reported HRQoL. The results here propose that receiving adequate healthcare and social support services may increase the HRQoL in patients with HD.

5.3 Paper III

The objective of this study was to describe the use of ATCs, a health support service, among patients with HD and examine the association between ATCs and HRQoL.

The results showed that just approximately one-third (36.9%) of patients with HD had ATCs with slightly less actually using their ATCs (35.7%) at the time of the study. Patients that used ATC were found to be at stages I - III and having mild to moderate cognitive impairments. ATC training was provided to only a fifth of the patients (20.2%), mostly in stage III, and less than one-third of the patients (32.1%) underwent ATC needs assessment. A little less than half of all patients (44%) had received information about ATCs at some point in time. While we found a significant positive β-value for formal ATC use (β-value = 0.356, β 95% CI: 8.45 – 34.1, p = 0.001), indicating that informal ATC use contributes to higher HRQoL by means of simple regression analyses, this effect disappeared with multivariate regression controlled for clinical disease and demographic factors. The multivariate model explaining 30% of the variance, only identified TFC to bear significant impact on HRQoL (β value = - 0.564, β 95% CI 1.47 – 5.34, r² = 0.142, p = 0.001).

It was concluded that the low values regarding all aspects of ATC (use) and the absence of an association between ATC use and HRQoL in the multivariate regression analyses may primarily be a sign of the dearth of awareness and knowledge about the availability and implementation of ATCs among healthcare professionals. These findings exhibit the significance of a thorough needs assessment and implementation process, in order to be able to discern the potential enhancement of maintenance of HRQoL through ameliorating cognitive impairment and elevating functional capacity in patients with HD.
6 DISCUSSION

6.1 METHODOLOGICAL CONSIDERATIONS

When interpreting the results of the three studies (Paper I – III) comprising this work, several methodological issues need to be taken into consideration. For one, we had to make choices when planning and conducting the study, while we at the same time look to optimize the possibilities of drawing valid inferences based on this research’s results. Herein, these methodological challenges will be discussed in more detail.

6.1.1 Study population

The study population in all three studies featured in this work included patients who had received a clinical diagnosis of HD. No other inclusion or exclusion criteria were applied as we aimed to establish the prevalence of patients with a clinical diagnosis of HD within south-eastern Norway, and to cover the whole clinical disease spectrum.

All patients were recruited through the regional academic hospital, Oslo University Hospital, through the Department of Neurohabilitation, the Department of Neurology, the Department of Medical Genetics, and the National Competence Center, the Center for Rare Disorders, in addition to Vikersund Rehabilitation Center. As the Oslo University Hospital had regional coverage and the Vikersund Rehabilitation Center was the only rehabilitation center that provided an intensive rehabilitation program for HD patients in the south-eastern Norway, one could assume that the overall majority of patients with HD had been or was in contact with one of these centers as part of the diagnostic process or for treatment purposes.

During the inclusion period, a total of 158 eligible patients with a clinical diagnosis of HD were identified. This corresponds to a prevalence of 5.9 per 100,000 inhabitants (based on a population of 2.7 million inhabitants in south-eastern Norway), which is similar with the prevalence reported in the study of Pringsheim et al. (29). However, establishing the true prevalence of HD is challenging because of biases related to disease characteristics. One such issue is the population bias as a result of the clinical picture of HD, which is often part and parcel with reduced self-awareness. Patients may not perceive themselves as ill and, consequently, may not be in contact with healthcare institutions, thus are not recognized as eligible participants.
6.1.2 Patients (subjects)

Of the 158 eligible patients that had received a clinical diagnosis of HD during the inclusion period, 86 were included in the analyses of Paper I and 84 in Paper II and III, covering patients across all five disease stages. These numbers correspond to response rates of just over half of the number of identified patients, specifically of 54.4% in Paper I and 53.2% respectively in Paper II and III.

Response bias was likely to stem from patients with reduced self-awareness declining to participate as they perceived themselves as not yet having clinical HD yet. Additionally, cognitive impairment among patients with HD may have meant that patients forget to reply to the invitation or to lose the invitation and unable to find it, but also psychiatric symptoms, such as apathy, a common symptom in HD may have fostered patients’ non-response based on not reading, answering or mailing their response to participate. In the advanced disease phase, dependency of patients with HD on family and / or professional caregivers, who are likely to be constrained by time and energy to follow-up on the study invitation, may also have reduced the number of patients being included in the study.

Although these response rates reduced the statistical power of the data analyses, they could be considered acceptable and representative of the HD population. Response rates in HD studies are scarcely reported because of different study designs and aims (12). Much scientific work has been based on prospective observational cohort studies, such as the previously mentioned REGISTRY and PREDICT studies (16, 65, 139). Studies that indicate a response rate have lower response rates (< 50 %) (64, 140, 141). With respect to the sub-population of patients in the advanced phase of HD (stages IV and V) in particular, the response rate here can actually be considered relatively high. This contributes to the present sample featuring a relatively representative sample that covered the whole course of disease compared to other clinical studies in the field of HD (9, 139). As a result of study visits often being conducted as part of outpatient clinical care and inclusion of in-depth symptom assessments in the majority of clinical and observational studies, patients with advanced HD are often too ill to participate in studies. In those studies from the European wide REGISTRY study, patients with advanced HD are generally underrepresented (9, 139, 142). The fact that patients with advanced HD were not underrepresented in the present study was considered a strength of the current work. Further, it is worth mentioning that the limitations in terms of limited response rate and the types of response bias were general and hence equivalent for interpreting results of clinical research in the field of HD.
6.1.3 Study design

The present work has a cross-sectional study design preventing us from drawing any causal inferences regarding the associations between independent variables and dependent variables investigated in Papers I - III. As well, the results describing the prevalence of the variables reported across the disease stages should not be interpreted as reflective of a development during the course of the disease in individual patients. The results merely indicate a pattern of prevalence during each of the five disease stages.

Using a cross-sectional study design was considered appropriate for the purposes of the studies in the present work. The aim was to establish the prevalence of certain phenomena in the HD population, and to explore relationships between independent variables and these phenomena. The findings of the study consequently serve the purpose of generating hypotheses and informing future research directions on these topics (143).

Additionally, one has to be cognizant of the fact that even though this study design was perceived most suitable for carrying out the present studies, it assumes that who was included in the studies was random and that there did not exist a relationship between the variables representing the phenomena (i.e., unmet needs for healthcare and social services) and the patients not included. When interpreting the results of cross-sectional studies, including this one, one must consider that this cannot be entirely assured. For example, in the present study, there may exist a relationship between patients not participating and their perceptions of the presence of unmet healthcare needs, resulting in patients with met needs for healthcare and social services not participating in the study because it would be of less interest to them, thereby leading to a greater presence of patients with unmet needs for healthcare and social services and thus an increased prevalence of patients with unmet needs.

However, there are two things that deserve to be pointed out with regards to this. It is likely that patients who chose not to participate were, as noted previously, those who believed themselves to not be ill because of diminished disease awareness and cognitive impairment and those dependent on caregivers who may be restricted in terms of time to support participation in studies. This does not automatically describe to what extent their healthcare needs were met - this evaluation was based on a normative assessment of their healthcare needs and not on reports of healthcare needs as perceived by the patients themselves (self-reported or subjective healthcare needs), as is the case in a large number of other studies on
unmet healthcare needs (101, 102, 106, 107, 144). Again, such could highlight the relative validity of the current study results.

Besides, the carrying out of this work came with the anticipated time and resource restrictions, while this type of study design has been proposed to be able to generate valid and reliable scientific knowledge in line with the aims and the topics of the present studies on which scientific work currently is scant.

Rather, the findings from this work should be extrapolated further in future studies that employ a longitudinal design to better explore the associations and causal relationships between the independent and the dependent variables. In addition, replicative studies featuring a cross-sectional design, are warranted to verify the present findings.

### 6.1.4 Data collection procedures

Data collection procedures are important for the reliability of study results and may also contribute to the representativeness the sample data are collected for.

The data collection procedure was the same for all three Papers (I – III). The data collected as part of this work were all obtained by the same two experienced clinical raters and this may have introduced variation because of differences in ratings consequently affecting the reliability to the data. In order to optimize the quality and comparability of the data obtained by each rater, a number of precautions were taken, namely: a) all variables collected and reported in the study had been discussed prior to the study start; b) scoring keys were developed for the general clinical evaluation of cognitive impairment and ATC use; and c) a consensus document was drafted by the clinicians in order to ensure similar interpretation and rating of the NPCS with consensus meetings held specifically to this end. Yet, these precautions could not guarantee robust inter-rater reliability and were thus unable to remove the potential influences of personal factors on rating, such as the demeanor of the rater and consequent reaction of the patients, like being more or less motivated to answer all the questions and undergo assessments. We therefore additionally independently assessed twelve randomly selected patients in order to calculated inter-rater reliability using Cohen’s kappa statistics on the NPCS Needs sub-scales (Paper I and II). Kappa coefficients were satisfactory and ranged from 0.55 – 0.71 (Healthcare: 0.58, Personal care: 0.71, Rehabilitation: 0.67, Social and family support: 0.55, Equipment: 0.65 and Accommodation: 0.65). We also
calculated inter-rater reliability using Cohen’s Kappa statistic for the general evaluation of cognitive impairment in these patients (paper III), which yielded a coefficient of 1.00.

Data for this work were collected from the patient alone or with an informant present. Informants were primary caregivers that were family members or healthcare professionals involved with the patient daily. With this socio-demographic and clinical information may have been remembered erroneously by the patient and caregivers. In order to make certain the data was correct and decrease missing data as a result of this, socio-demographic and clinical data were systematically corroborated through the patients’ medical records. In a number of cases socio-demographic or clinical information was unknown and in such cases the data were collected through the patients’ medical records only. The self-report measurement for HRQoL, the EQ-5D-3L and VAS scale, was completed with assistance from an informant for nearly half the patients included in this study, as for instance motor impairment may have compromised filling out the questionnaire, and in eight cases the questionnaire was completed by an informant only. It is unknown what a combined approach of patient and proxy ratings may imply for the ratings. In order to best guarantee that the data obtained were comparable within the sample, we specifically instructed the informants to only assist and not complete the questionnaire on behalf of the patients, as seen in another study on HRQoL and HD, which also indicated that proxy ratings are comparable with patient ratings in advanced HD (66). In future studies collecting data from both patients and proxies may be advised to engender reliability of the data obtained.

Finally, the data were collected both from outpatient visits and where the patient lived in order to increase response rates and for participation of patients with advanced disease unable to travel or for those living in rural areas, or far from Oslo for whom travelling was a substantial burden. Consequently, we were able to attain a representative sample of patients with clinical HD, which is a relative strength in the present study as mentioned previously.

6.1.5 Data collected

As stated a number of times, socio-demographic and clinical data were collected as part of the survey study. Much of this was from the recording of information, while other data were gathered from rating or evaluation. Reliability and validity concerns pertaining to a number of these variables deserve more in-depth consideration.
First of all, disease duration, calculated as the number of years between the date a clinical diagnosis was formally obtained and the date of inclusion in the study, was used in Papers I – III. The operationalization of this variable we selected is one of two main approaches used. The other approach defines disease duration as the date of the study visit minus the date of estimated first occurrence of clinical symptoms, as reported by patient, family or healthcare professional (139). It seeks to capture to the true number of years a person has been ill and is not based on the criteria for HD diagnosis, and includes the prodromal phase of HD. It is of particular interest for targeting disease-modifying treatment options. The difficulty with this approach is that it is prone to leading to a definition of the development of disease as too early - it can be hard to distinguish symptoms from normal variation. Our approach meanwhile may have led to an underestimation of disease duration - patients may have been ill with symptoms several years before they received formal clinical diagnosis. The present work targeted the population with clinical HD and we desired to ensure that patients included in the study indeed had clinical HD. Therefore, we calculated disease duration based on the data from clinical diagnosis obtained from the patient and / or informant as part of the survey interview and as corroborated by the patients’ medical journal. Disease duration was used to describe the population of this study and was included as a factor for all three regression analyses.

For the purpose of Paper III a general evaluation of cognitive impairment was performed as a disease-specific clinical assessment. The evaluation was conducted using a scoring key developed specifically for the present study, where the degree of cognitive impairment, predefined as mild, moderate and severe was assessed, and a fourth category indicating that it was judged such that a sufficiently reliable evaluation could not be made, i.e., because of severe communication problems, and not using standardized cognitive measures. Although this may be a less precise assessment, a high correlation (> 0.9) between the evaluation for cognitive impairment and TFC suggests a satisfactory reliability. The evaluation was performed as mentioned previously by two trained clinical raters with experience in the field of HD and neuropsychological testing. The inter-rater reliability has been reported earlier as part of the methodological discussion and was 1.00 corresponding to complete agreement.

With respect to aspects of validity for assessing cognitive impairment (adequately measuring that which one aims to measure) in general and in the context of HD, several factors must be considered. General factors threatening the validity of evaluating cognitive impairment involve tiredness, fatigue, pain, hunger, use of medication. For instance, fatigue may cause difficulties in concentrating and slowing of thought processes, negatively affecting cognitive
abilities. In the case of HD, symptoms specific for HD, including communication difficulties as a result of dysarthria and involuntary movements, like eye movements and psychiatric symptoms, may have interfered with ratings and assessments of cognitive impairment. These factors could jeopardize both established standardized cognitive assessments known to be used in patients with HD as well as non-standardized global evaluations, such as those performed as part of this thesis. Broad standardized cognitive assessments would have likely negatively impacted the response rate - patients may perceive cognitive testing as burdensome and confrontational, while patients with advanced HD would have been considerably limited in their abilities or would be unable to perform the assessment, negatively affecting reliability and validity of the data for this group of patients. Hence, while the use of standardized instruments for determining cognitive impairment would have yielded more specific information on cognitive impairment, they would have influenced the response rate and representativeness of the sample considerably while simultaneously not ensuring enhanced reliability and validity of results obtained.

6.1.6 Measurements used

Internationally acknowledged and standardized measurements, with good reliability and validity, were employed in order to assess the variable of functional ability, levels of needs, the use of and unmet needs for healthcare and social support services and HRQoL. Yet, there were limitations related to the selections opted that deserve reflection.

6.1.6.1 UHDRS Functional assessment

In all three studies (Paper I - III) the UHDRS functional capacity scale, one of the four domains covered by UHDRS assessment that was developed specifically for use in HD, was utilized in order to assess functional ability (45). The UHDRS as a whole has become the standard for assessment of clinical symptoms in patients with HD as it has shown reliability and consistency (28, 46). The functional assessment and independence scale were used to describe the sample while the TFC developed by Shoulson and Fahn and included in the UHDRS was employed for staging the patients (stages I - V) and as the independent variable of overall functional ability for the regression analyses. It was shown that there is robust inter-correlation between the three parts of the functional assessment of the UHDRS (46). As well, the TFC score has been commonly used for staging clinical HD (45, 131, 139). Furthermore,
the TFC has shown good reliability and validity in several studies and has turned into the common standard in international research on HD (131, 132).

The UHDRS, including the functional assessment, has also been widely used in Norway, both clinically and in research, like as part of the EHDN REGISTRY study. As of yet, however, a specific validation of the assessment has not been carried out in Norway. Yet, Norwegian data collected using the TFC has been applied in international research studies (9, 65, 139). Additionally, in our studies, we observed excellent inter-correlations with the functional assessments scales of the UHDRS (> 0.9).

6.1.6.2 NPCS

Assessment of needs for healthcare and social support services in Papers I and II was conducted using the NPCS (133, 134). The scale was recently developed in the United Kingdom to determine healthcare and social support needs among patients with LTNC, including a subsample of progressive neurological conditions. A few matters regarding the choosing to use the NPCS in this work merit discussion.

In terms of psychometric characteristics in patients with LTNC the NPCS has exhibited value (133-135). However, the psychometric characteristics of the Norwegian version of the scale are unknown - a validation study on how the scale performs in Norwegian healthcare services has not yet been performed. For the purpose of the present study the NPCS was carefully translated into Norwegian using a comprehensive process of translation and back-translations followed by validation of the translation by experts in the field of healthcare research and HD. Further, a consensus document was drafted documenting the interpretations for rating of items of the scale to best ensure similar ratings, by clarifying how to rate the needs for healthcare and social support services in the Norwegian and HD contexts. It was used as a scoring guide for the raters.

In the present study, the NPCS clinician version was utilized. In this version, Needs (Part A) are rated by healthcare professionals, and entail what services patients should receive in order to garner the best health outcomes (function and quality of life). Provision (Gets, Part B) are the services the patients actually receive, reported by the patient and / or family / professional caregiver. The unmet needs defined as the gap between needs for healthcare and social support services and the healthcare and social support services received in our work thus
largely depended on determining patients’ normative needs, based on Bradshaw’s societal taxonomy of needs (89, 93-95, 97, 101-103). Normative needs may be considered relatively objective and reliably (repeatable) reflect true healthcare needs. However, they rely upon judgments of professionals and may be influence by differences in background, knowledge, skills and areas of interest (93). Meanwhile, assessing healthcare needs and unmet needs based on the patients’ judgment, as gauged by the patient, corresponding to expressed and/or felt needs also has challenges which need to be taken into account. Taking this perspective, the patient (carer) is able to judge to what degree health issues pose a problem for which help or care is necessary. Yet this is also affected by the patients’ knowledge and understanding regarding the consequences of health issues or about available interventions (93). Research assessing healthcare needs in a normative way may be criticized for not sufficiently taking into account the patients perspectives and reduced clinical relevance for a disease population (89), and hence might overlook patients’ needs resulting in fewer healthcare needs or even unmet needs. Nevertheless, assessment of healthcare needs and unmet needs based on the patient / carer perspective alone (expressed / felt need) is likely to reflect the wishes from patients, thereby bringing about increased healthcare needs (88, 89). Certain studies investigated both patients’ perspectives (expressed / felt needs) and professionals’ perspectives (normative needs) and indicated a discrepancy between these perceptions (108, 145). To date, the few studies on needs for healthcare services within the field of HD feature patients / carers perspectives (5, 6, 8). These considerations are important to take into account when performing an assessment of healthcare needs as part of research or in the clinical setting. Moreover, they are fundamental when interpreting the results of this work. Combining patient / carer and professional’s perspectives in future work is warranted.

Formally, the NPCS has been presented as designed for evaluating individuals’ needs for community healthcare and rehabilitation (133, 134). As such, there is a reduced focus on patients’ healthcare needs within specialized healthcare services. However, clinical HD is a complex and chronic disease, with clinical care to a large extent and over a long time period provided in the municipalities by community care services. Consequently, the NPCS was found most suitable to be able to cover best the areas of potential needs and unmet needs for healthcare and social care services, despite including a very global medical healthcare services that features specialized healthcare services. It is important to be aware that the results with respect to unmet needs for healthcare and social support service not adequately
assess the needs for healthcare services delivered as part of specialized healthcare services, such as participating in unique HD rehabilitation programs.

6.1.6.3  **Self-report measure of HRQoL, the EQ-5D**

In Papers II and III, a generic self-report measure of HRQoL, the EQ-5D-3L was employed. The EQ-5D is widely used internationally and in Norway in HD studies and those with other populations (12, 66, 137, 146). A formal Norwegian version of the EQ-5D-3L is available through EuroQol Group, and has been shown valid in the Norwegian population (138). Within the HD population, the scale has not undergone specific reliability and validity studies. One study investigated the reliability and validity of two other generic instruments, the Short Form-36 health survey questionnaire (SF-36) and the Sickness Impact Profile (SIP), where the SF-36 was determined to be the better instrument for use based on construct validity and test-retest reliability in addition to being the shorter instrument of the two (64). Despite this we opted for the EQ-5D, mainly because of the scale being a much less complicated measure versus the SF-36, which was considered of specific importance in this work specifically including patients with advanced HD.

The use of a self-report measure for HRQoL in HD and other patient groups with moderate to severe cognitive impairment and lack of self-awareness and insight their disease, endangers the reliability of this study, as these symptoms affect patients’ abilities to evaluate their situations. Yet, the concept of HRQoL is, in addition to being multifactorial and dynamic by itself, subjective (12, 15, 91). Collecting HRQoL data based on proxy ratings would also threaten reliability - it is known that family members of patients are also burdened by HD, which may have a part in their evaluation of HRQoL (141). Exactly, how reliable data are, is hard to determine for a subjective concept like HRQoL. Knowledge of this challenge and its potential impact on these types of data and the results of the present work is required.

There are both generic and disease-specific measurements of HRQoL, such as HDQoL and HDQLIFE for HD patients (12, 15). Choosing to use a disease-specific measure (or both), might have impacted our results in several ways. When selected the measure there are a number of aspects that need to be taken into account when interpreting the results. Where generic measures often have evidence of reliability and validity and are expected to generate relatively valid data, the psychometric characteristics of disease-specific HRQoL are often lesser known, as there is a shortage of such studies. However, disease-specific measures may
capture more precise information related to the disease investigated (12, 15, 112, 147). Further, and in contrast, disease-specific measurements more often are longer and more difficult to complete, especially in populations where cognitive impairment occurs, risking representativeness of the sample and reliability.

6.1.7 Statistical analyses

Although a response rate of a little over half of the invited patients, corresponding to 86 and 84 participants respectively, in paper I and Papers II and III, can be thought to be satisfactory given HD being a rare disorder with a complex clinical picture, this response rate is statistically low, undoubtedly reducing the statistical power of the results in the present study. Consequently, interpretations of the results should be made with caution.

In studies with low response rates and low numbers of participants, low and high scores of data and measurements have relatively large effects when calculating statistics, such as mean values. Moreover, larger standard deviations (variations) are often the result. In the present work, we therefore tried indemnify this by calculating median values and IQR, instead of the mean values and standard deviations for any variables that were non-normally distributed (and in Paper I for all variables). Further, we were strict with regards to the number of variables included in the multivariate regression models, using one variable per 10 participants, in order increase reliability / validity. Meanwhile, a more limited number of variables that could still supply interesting information may have been included in our regression models. Selecting which variables to include in our models was based on a combination of theoretical and clinical knowledge as well as on statistical analyses via simple regression analyses.

It is worth mentioning that a relative strength in this work was that there were less than four percent missing data (< 2 - < 4%) just described earlier (see page 41- 42).

6.2 General Discussion of the Results

6.2.1 Unmet needs for healthcare and support services in HD (Paper I)

At the time of planning this research, no studies had systematically investigated unmet needs for healthcare and social support services in patients with HD. One study on healthcare
services utilization was previously conducted, and another had assessed the unmet healthcare needs in a specific demographic area in the United Kingdom (8, 9). Taken together, the results of these studies indicated healthcare and support needs related to medical care, physical abilities, social and carer support as unmet. As expected, the results of our study showed there was a considerable degree of unmet needs for healthcare and support services of various types and across the whole HD disease spectrum as assessed by the normative approach (89, 93-95). Further, several factors were linked with the level of unmet needs for healthcare and social support services.

6.2.1.1 Needs, Gets and Unmet healthcare needs

The findings of this study highlighted the considerable gaps between needed and received services, i.e. unmet health care needs (97, 101-103). The levels of Needs and Gets (utilized healthcare) for the total level and levels of the domains, Health and personal care and Social care and support, rose steadily across disease stages I - V. In terms of the levels of types of healthcare services (NPCS subscales) Needs and Gets correspondingly increased for Personal care, Specialist equipment (which records ATC utility) and Accommodation. The remaining healthcare sub-scales, including Healthcare, Rehabilitation and Social and family support care, remained stable in the advanced disease phase (stages IV and V). This pattern was in accordance with our expectations based on the progressive nature of HD, and the type of healthcare services. As individuals become more ill, they generally need more healthcare and support services in order to manage and alleviate disease symptoms, and as a result, they will also receive more care. Healthcare services that remain stable in advanced disease are typically those where less gains are is expected, i.e., Social and family support care needs and utilization may drop as patients move to a long-term care facility and relieve family from informal care burdens and through the transition to the formal care. Similarly, benefits from rehabilitation may become limited to primarily those from physiotherapy for respiratory purposes and occupational therapy for AT. Despite this pattern of rising healthcare service utilization alongside increasing needs, patients’ needs for health and social support care were not met. Overall, the levels of unmet needs for the five disease stages were observed to be highest for Rehabilitation and Social and family support. Similar to the results for levels of unmet needs, a high frequency of unmet needs and gaps in the provision of health care needs overall, for domains Health and personal care and Social care and support were seen. Further, for the sub-scales Rehabilitation and Social and family support, the largest gaps in services provision and needs were recognized with more than half of all patients having unmet needs
in each of the five disease stages. Our findings are suggested to represent real needs of patients with HD as they are generally consistent with the findings of the study in the United Kingdom surveying unmet healthcare needs as well as the European study on healthcare utilization (8, 9). Furthermore, studies based on information from both patients and at risk and not at risk family members, indicate a large perceived lack of sufficient family support, additionally supporting the present results, specifically regarding unmet needs for family and social support (5, 6). In addition, our results are equivalent to the results of studies on patients with LTNC using the same measurement to assess unmet needs for healthcare and social support (133, 135).

6.2.1.2 Unmet healthcare needs across HD disease stages

As opposed to our hypothesis that the gaps between unmet healthcare needs and received services for healthcare and social support needs would be most substantial among patients in advanced disease stages, the results indicate that the most sizeable gaps regarding both levels of unmet needs and frequencies of patients with unmet needs are found in the middle phase of HD (disease stage III). More specifically, the highest levels of unmet needs were observed for stage III for all sub-scales except for Healthcare, and more than half of the patients in stage III had unmet needs for all sub-scales except for Healthcare and Accommodation. One explanation for why our findings did not support our hypothesis may be that the middle phase represents a heterogenic group of patients because of higher variation in symptom presentation and in the rate of disease progression. The middle phase can be assumed to be a phase of transition, where many patients convert from being mostly functionally independent to becoming significantly functionally dependent. An additional explanation for these findings may be that symptoms are more overt in patients with advanced disease (stages IV and V). These patients often clearly exhibit functional disabilities and are unable to independently perform the activities of daily living. This may result in a more heightened awareness of their needs for healthcare and support services by healthcare professionals, family and even the patients themselves. Patients with advanced HD, therefore, may receive a larger amount of healthcare services, better addressing their needs and resulting in lower levels and frequencies of unmet needs despite having greater healthcare and support needs compared to the patients in the middle phase (stage III). This is supported by experience from clinical practice indicating that patients with HD in later disease stages are more likely to be taken care of by professional caregivers and are more commonly cared for in care facilities, such as assisted
living and nursing homes. The results are further in line with research on long-term neurological conditions (LTNC) and traumatic brain injury. It was found that patients with LTNC reporting met needs for rehabilitation 12 months after discharge from the hospital were more dependent, compared to those with unmet rehabilitation needs, and patients with more visible needs after traumatic brain injury had a higher degree of met needs (135, 144).

6.2.1.3 Factors associated with unmet healthcare needs

The results of the present study have further demonstrated a substantial association between disease phase and overall level of unmet needs and for the Health and personal care domain. Specifically, being in the middle phase of disease (stage III) substantially increased the likelihood for increased levels of overall unmet needs and of unmet needs for the Health and personal care domain. As explained previously, the middle phase of HD can be considered a transitional phase that features more heterogenic patients. Patients in this phase progress from being largely independent to exceedingly dependent for daily functioning. However, which symptoms contribute most to functional declines varies among patients. Both symptom presentation and progression rate impact the complex and varying character of patients in this phase. In certain patients, cognitive decline primarily brings about functional declines and dependency, while in others, motor symptoms may be the main cause of functional decline.

Other factors associated with unmet needs included educational level and whether the information obtained during the survey interview was collected from the patient individually. A lower level of education was likely to decrease the probability of higher levels of overall unmet needs in the Health and personal care domain. This may be understood as people with higher education being more aware of the healthcare and social support services available to them and to which they are entitled, and by being more resourceful to follow up on receiving the necessary services. These results are in line with the trends found in the general population (106, 148). Information for the survey interview obtained from the patient only also tended to diminish the odds of having higher overall levels of unmet needs and levels of unmet needs in this domain. This may both be a consequence of underreporting regarding healthcare utilization and also reflect the patients’ reduced disease awareness potentially in the form of denial by the patient regarding the required healthcare needs as assessed by a healthcare professional or family member. However, proxies may overestimate patients’
disabilities and therefore overestimate patients’ needs for healthcare and social services (66, 141).

The results of this study did not reveal significant associations between level of unmet needs for the social care and support domain and socio-demographic and clinical factors. Yet, contrary to the results for the levels of overall unmet needs and of unmet needs for the Health and personal care domain, higher education had a tendency for raising the likelihood of having greater levels of unmet needs for social care and support. This was not expected as higher education overall has been associated with receiving higher levels of social support (148). Based on our study data, we are unable to further explain this phenomenon. In addition, a tendency indicating that the presence of comorbid conditions reduced the odds for higher levels of unmet needs for social care and support was observed. Based on comorbidity generally being associated with poorer social functioning, this may be somewhat unanticipated, as this would probably cause an increase in the probability of a higher level of unmet social care and support needs. On the other hand, potentially, the presence of comorbid conditions may contribute to increased cognizance of needs for social care and support, resulting in more adequate provision of these services.

6.2.2  HRQoL and unmet healthcare needs in HD (Paper II)

When planning the present study, several studies had already been conducted in the area of HRQoL and HD, and several more have emerged since (10, 12-15, 64, 66, 140). However, this is the first study that included the potential impact of unmet needs for healthcare and social services on HRQoL in the context of HD. Additionally, in most research, advanced stages of HD are underrepresented. Finally, HRQoL studies to date have reported index scores of generic measures for HRQoL (12, 66), while profile descriptions of perceived health status may supply more detailed information and reflect on symptom related and functional status characteristics of health in need of healthcare services and in this way contribute to overall HRQoL (117, 118, 146).

6.2.2.1  HRQoL in patients with HD in south-eastern Norway

Our findings have shown that health status is rated poorest for all five health dimensions among patients with moderate to advanced disease stages (stages III -V). Additionally, patients with advanced HD (stage IV and V) reported the largest decrement in overall HRQoL. Regarding health status profiles and overall HRQoL, disease stage III stands out. Patients at this stage exhibited the most wide-ranging health profile, with a number of patients
reporting no problems while others described major problems. Further, these patients’ average for overall HRQoL was reported below the average of the total sample included in the present study. These findings emphasize the substantial burden of HD on HRQoL, in agreement with previous research on HRQoL in HD (10, 13, 66, 69, 149). More specifically, an overall average score for HRQoL below the average of the study sample, as early as stage III, was also described by Hocaoglu et al (66). The present results, particularly the health profiles reported for disease stage III, further expand upon the notion that this disease stage signifies a largely heterogeneous group of patients with a greater variation in symptom presentation and disease progression, thus pose additional challenges when it comes to adequate healthcare and social support delivery.

6.2.2.2 Association between unmet healthcare needs and HRQoL

The present study revealed significant associations between TFC, which was the strongest association, and the total level of unmet needs for healthcare and social support services and HRQoL.

The multidimensional and complex character of the HRQoL concept makes it difficult to study, as it is subject to the influence of many factors (91, 114, 116, 117). In the present work we accounted for various socio-demographic and clinical, disease-specific factors. At the same time our dataset was limited to the number variables that could be accounted for, and over half of the variance remained unexplained. This could be because the present study did not include a more detailed standardized assessment of the triad of HD core symptoms (motor, psychiatric and cognitive). These disease characteristics have been shown to be associated with HRQoL in other research (11, 13, 14), but these studies did not include factors that are included in the present study, such as comorbidity and in particular the level of unmet needs for healthcare and social support services. Besides these disease-specific factors, and the level of unmet needs, there were also other potential explanatory environmental factors related to healthcare provision which have not included, comprised of factors on the organization, the costs and the quality of healthcare provision, as well as socio-economic inequalities (i.e., income) (106). Social dissimilarities in specialized healthcare service utilization has been demonstrated in the general Norwegian population, even though Norwegian public healthcare system is organized according to needs-based equal access for all citizens and not according to wealth (110). Information on healthcare utilization and
healthcare needs and on HRQoL and HD is scant. A Canadian qualitative study proposed ways to improve quality of care by, for instance, better education, as the results investigating experiences with healthcare services among HD affected families, revealed complex needs for healthcare services and a lack of knowledge on HD among family physicians.

Although the strongest association was determined between functional capacity (TFC) and HRQoL, which is in agreement with existing studies on HRQoL and HD (10, 14), the hypothesis that higher levels of unmet needs for healthcare and social support services were associated with a reduction in HRQoL, was supported. Information about the needs for healthcare and social services in the general Norwegian population or in individuals with disabilities to our knowledge is absent. Yet, research from Canada, which has a healthcare system similar to Norway, demonstrated that adults from ages 20 - 64 with physical, sensory or cognitive impairments reported three fold more unmet healthcare needs compared to aged matched non-disabled individuals, supporting the existence of considerable unmet healthcare needs among populations with disabilities in welfare states (105). Our study results are further supported by research on unmet healthcare needs and HRQoL in other patient groups, including patients with coronary heart disease, cancer, mental illness and dementia, further strengthening the study findings (92, 103, 108, 109, 150, 151). Physical and social needs are associated with HRQoL in coronary heart disease (92). Cancer research has demonstrated that unmet needs for supportive services in young adolescents and young adults with cancer were connected with lower overall HRQoL and that cancer survivors with unmet supportive care needs in the physical, psychological and patient care domains had poorer HRQoL (103, 109). Studies on persons with mental illness established associations between unmet healthcare needs (108, 150), and basic, social and functioning needs were found to be the strongest predictors (150). Research involving patients with dementia in residential care showed that needs related to sensory and physical disabilities, mental health and social needs were frequently unmet (151). Moreover, a study on a sample of patients with LTNC, including a small number of HD patients, proposed that these patients’ do not receive the healthcare and social support services they need. The authors suggest that this may reflect insufficient coordinate care (69).
6.2.3 ATC and HRQoL in HD (Paper III)

ATC is a healthcare support service, aimed at improving functioning in daily activities through compensating for cognitive impairment. HD patients are commonly assisted by AT, mostly because of their movement disorder. In Norway AT targeted at relieving cognitive disabilities in the form of ATC has also been provided to HD patients. The scientifically based knowledge in the area of cognition in HD has advanced tremendously over the years, raising interest in specific interventions for cognitive symptoms. Despite this development and ATC exhibiting promising influences on function and HRQoL in other similar conditions, to date, no other studies besides the present work have thoroughly reviewed ATC in HD (72-77). Based on the research within this thesis, considerable unmet needs have been identified among healthcare support services, including specialist equipment, which also comprises ATC. Specifically, within stage III, there was a large proportion of patients that had unmet needs pertaining to specialist equipment. Further, the results of this work indicated that having unmet needs overall may negatively impact HRQoL.

6.2.3.1 The use of ATC

Given that all patients with HD develop cognitive impairment, the present study has shown there is a relatively infrequent use of ATC - about one-third of all patients that participated used ATC. This usage was predominantly observed amongst patients in disease stages I – III with mild to moderate cognitive impairment. Further, it appeared that informal ATCs were mostly utilized at disease stages I and II while formal ATCs were most prevalent at disease stages II and III. These findings may be explained by the differences between informal and formal ATCs. Informal ATCs are introduced by the patients themselves with the purpose of compensating for self-perceived and often less overt reductions in cognitive function, and as a way to maintain daily activity. The ATCs are often well-known and readily accessible. Formal ATCs, mean the ATC is specialist equipment requiring formal application and implementation, including assessments and training. The characteristics of the cognitive disabilities between patients in these disease phases may further elucidate the results here. Patients at disease stages I and II generally feature earlier symptoms and cognitive impairment often in more specific cognitive domains, commonly milder and less pronounced. Taken together with a higher functional capacity means that these patients are more likely to be able to recognize and find compensatory strategies for their problems by means of familiar aids. As the severity of their cognitive impairment increases with disease progression, the
needs for more complex aids might arise, and in line with the finding that formal ATC usage is most frequent in disease stages II and III, particularly stage III.

We further observed that less than 50% of all patients received information on ATC, with only one-third undergoing ATC needs assessment and just a fifth of patients being trained with ATC. These results are in agreement with other studies on patients with cognitive disabilities. Scherer et al. and Adolfsen et al. both provided evidence that patients are not properly endowed with information on the possibilities of ATC, pointing towards a lack of knowledge regarding ATC among health professionals and a deficit in trained professionals to implement ATC appropriately (123, 126). In line with formal ATC being most frequently used in HD disease stages II and III, these patients also most commonly obtained ATC information, underwent needs assessment and received training. While the use of formal ATC in these stages maybe understood as a consequence of the need for formal ATCs arising later in the course of the disease because of more severe cognitive impairment, it may additionally be based on the deficiency in awareness for and knowledge of what an adequate process of formal ATC implementation involves among professionals working with HD. Resultantly, performance of a needs assessment and implementation of formal ATC may take place later. As the progressive nature of the cognitive impairment in HD reduces the capacity for learning to use ATCs through training, late initiation of ATC needs assessment and integration into daily living may in turn give rise to diminished abilities to benefit from ATC usage or potentially significantly reducing the period patients may be able to effectively use it. Despite the fact that there is a near absence of ATC employment among patients with advanced disease (stages IV and V), this actually may be expected because patients with advanced HD often have global cognitive impairment (dementia) and severe motor disturbances, concomitant with great difficulties in applying ATCs, though this could be reversed by enhancing knowledge and with the timely initiation of assessment and implementation. Well-times initiation of the ATC provision process may enable prolonged and effective use of ATC, and facilitate ATC usage across the transition from one disease stage to the next. These findings and challenges are in line with a report from the Norwegian Directorate of Health regarding the current status of healthcare services to patients with dementia and challenges for improvement of these services, and with research on how patients with dementia use ATC and factors associated with ATC use in Nordic countries (152, 153). Among other, one of the challenges was that assessment of dementia is conducted at the specialized healthcare levels, while the responsibility of delivering the adequate ATC lies at the primary care level with the
municipalities (85, 130, 153). This requires the need for good collaboration. The development of dementia is a hallmark of HD, and thus these results may also be applicable to patients with HD.

6.2.3.2 The association between ATC use and HRQoL

In the present research no association between ATC and HRQoL was observed. Only TFC was seen to be a significant factor associated with HRQoL, which was also demonstrated in earlier studies (10, 13, 14). The absence of an association between ATC and HRQoL here may be explained in several ways. For one, prior studies on ATCs have identified factors vital for their success. They have underscored the necessity of comprehensive assessment of individual characteristics, including personal factors, factors in individuals’ environments and of the device to attain the best match between the patient and their cognitive disabilities and the ATC (70, 72, 81, 123). Hence, ATCs as a fruitful intervention is fostered when interactions between the device and the everyday environment of the patient are taken into account. When this process is not executed satisfactorily, ATCs fail to positively affect cognitive impairment and daily functioning. The present results may thus reflect that ATCs were not successfully implemented and did not favorably impact on HRQoL. Other studies on ATCs in populations with cognitive impairments have shown promise in terms of cognitive function and demonstrated improvement in QoL (74, 76, 78, 154-157). Secondly, the effect of ATCs on HRQoL is thought to be the result of the amelioration of the negative effects of cognitive impairment and improving functional ability. For the patients included in this study that used ATCs, their effects could have emanated from enhanced functional capabilities as reflected by TFC scoring. With inclusion of TFC as a disease-specific factor in our model, the effect of ATC on HRQoL may be incorporated in the significant association between the TFC and HRQoL. Thirdly, using a generic instrument for assessing HRQoL may have reduced sensitivity when it comes to determining the link between ATC and HRQoL. Investigating this can be considered examining the effect of an intervention (ATC) on an outcome of HRQoL in a specific patient population. It has been proposed that for such purposes, a disease-specific measure of HRQoL would be more suitable while a generic measure would be more appropriate for use as part of health needs assessment as per Paper II (67, 90, 147).
7 CONCLUSION AND IMPLICATIONS

This thesis has provided knowledge on the prevalence of HD in south-eastern Norway, addressed the gaps between needs for healthcare and social support services and received services (i.e., unmet needs) among patients with HD, use of ATC, as a case of a supportive unmet healthcare need, and reviewed the association of unmet needs with HRQoL.

7.1 CONCLUSION

Overall, this research suggests that unmet needs for healthcare and social support services are common across all five disease stages (I-V), but are most profound among patients with HD in the middle phase (disease stage III). High levels of unmet healthcare needs were associated with lower HRQoL. Further, the use of ATC, information, needs assessment and training were found to be reported relatively infrequently, especially given the clinical picture of HD, which includes the progressive development of cognitive impairment being an inevitable hallmark symptom.

Our findings of there being clear unmet needs for almost all patients at least at one time-point during the course of the disease, might indicate that healthcare delivery in the population of interest is rather fragmented versus coordinated. This is contrary to the proposed guidelines for clinical care (Physicians Guide) and standards of care promoting coordinated multidisciplinary tailored care centered around the patient with HD and their family across the whole disease spectrum. Furthermore, this is in contrast to the intention of the Coordination Reform in Norway that was intended to enhance coordination between two main health sectors - primary health and long-term care on the one hand, and hospitals and specialist services on the other, as a way to improve health care delivery. Our results might also suggest that patients with HD do not receive the comprehensive and tailored care that can influence patients HRQoL. This is significant in light of guidelines / directives for care of people with dementia and chronic conditions where HRQoL is an important outcome in the absence of curative treatment.

The findings also suggest there are considerable unmet healthcare and supportive needs, and the negative association between unmet needs and healthcare and social support services and HRQoL in our study, may be reflective of our findings on ATC usage as a case of an unmet
supportive healthcare need, underscoring the lack of awareness and the importance of a thorough delivery process that could positively influence patients’ independence and HRQoL.

7.2 IMPLICATIONS AND FUTURE RESEARCH

An important implication of the results from this thesis is that patients with HD should be evaluated clinically and also be systematically assessed with regards to their needs for healthcare and social support services during the entire course of their disease. Further, these needs should be recorded and followed upon regularly because of their dynamic character. Implementing systematic healthcare needs assessment and follow-up may be one way towards improvement of coordinated multidisciplinary tailored healthcare delivery in line with the intention of the Norwegian Coordination Reform. Building cooperative relationships with family and/or professional carers may facilitate this process. Such partnerships can be helpful when establishing prioritization of the identified needs and unmet needs for health and supportive care services. Increased awareness about the possibilities of the competence center, the Center Rare Disorder can be a resource towards improving care through dissemination of knowledge about HD, including knowledge about availability and accessibility of healthcare services. Furthermore, there may be a larger potential to exploit the interdisciplinary coordination groups (ansvarsgrupper) and coordinated care plans (individuelle planer) to facilitate tailored and coordinated healthcare delivery and to ground development and investigation of care models upon.

The findings of this thesis regarding ATC use and HRQoL, seem to primarily call for increased awareness and delivery along with implementation. Yet, further research into ATC as a potentially effective intervention and supportive health service is warranted in order to tease apart to what degree our findings indeed reflect the lack of adequate delivery of ATCs to address unmet need, or whether they show that ATC may not be such an effective support service for HD patients, by employing both generic and disease-specific measurements of HRQoL.

Further, as to what has been observed through this research, patients with HD in the middle phase (stage III) stand out in terms of having particularly large unmet healthcare and supportive needs, wide-ranging health profiles and regarding aspects related to ATC use. Emphasis should be placed on patients in the middle phase of HD (stage III), taking into
account the transitional character, the changing and complex clinical picture because of large variation in disease symptoms and progression, often with less overt symptoms compared to stages IV and V. It is necessary for healthcare professionals working with HD patients at all healthcare service levels to become conscious about this through increased focus on stage III patients in clinical care guidelines and standards of care.

The studies included in this thesis were based on a cross-sectional design. Future studies should make use of longitudinal designs to further determine the strongest predictors of unmet healthcare and supportive needs and of HRQoL, including disease-specific factors covering the cognitive, psychiatric and motor impairment triad of HD symptoms, in addition to functioning, and include more factors related to healthcare delivery, such as socio-economic characteristics. International studies involving countries with different healthcare organization would also be valuable in order to further verify our findings. Moreover, larger samples and longitudinal designs would contribute to validating the particular challenges related to providing adequate clinical care to patients with HD in the middle phase (stage III). With regards to the assessment of healthcare and social support needs, the present study did not take into account the patients’ / cares’ perspectives (expressed / felt healthcare needs). Future studies would best seek to identify both normatively evaluated and expressed /felt healthcare needs, as both types may lead to more accurate healthcare needs assessment - the “true healthcare needs” likely lie somewhere in between as a result of the relativeness of both concepts based on the influence of a variety of factors. In addition, the healthcare needs assessment tool used in this study, the NPCS, does not cover healthcare needs primarily addressed by specialized healthcare services. Future studies could also look into needs as they may be relevant specifically for individuals without a clinical diagnosis of HD. Furthermore, validating the Norwegian translation of the NPCS in the Norwegian context is necessary.

Taken together, the knowledge that can be gained from the proposed implications and directions for future research based on the results discussed in this thesis may contribute to development of robust models and guidelines, improve the delivery of tailored and coordinated care to patients with clinical HD and other groups with complex chronic and / or progressive neurological conditions, and positively affect patient’s outcomes of daily functioning and HRQoL.
8 REFERENCES


APENDIX

Needs and Provision Complexity Scale (NPCS) – clinician version
The Needs & Provision Complexity Scale (NPCS) for LTNC

Extended version

The NPCS can be used and copied freely,

*but please acknowledge the originators in all publications*

Further information and advice may be obtained from:

Professor Lynne Turner-Stokes DM FRCP  
Herbert Dunhill Chair of Rehabilitation, King's College London.

Regional Rehabilitation Unit,  
Northwick Park Hospital,  
Watford Road,  
Harrow, Middlesex.

HA1 3UJ

Tel: +44 (0) 208-869-2800;  
Fax: +44 (0) 208-869-2803  
Email: lynne.turner-stokes@dial.pipex.com
Background

The National Service Framework (NSF) for Long Term Neurological Conditions (LTNC) promotes joined-up services to provide holistic, person-centred care (Department of Health, 2005).

It includes 11 quality Requirements covering the care pathway from diagnosis to death. Critically, integrated care planning (QR1) provides the backbone to the NSF recommendations.

It is recognised, however, that resources to support integrated care planning are currently very limited, and this presents a major threat to implementation of the NSF recommendations.

It is vital therefore to be able to evaluate service provision in relation to need, both at an individual and at a population level, in order to focus future service development efficiently.

LTNC are themselves a highly diverse group of conditions, and within those, people have widely different needs for services, against which the adequacy of service provision must be judged.

Diagnosis is a poor determinator of need in this context, and we require some other way of defining need for services. The Needs and Provision Complexity Scale (NPCS) was designed to measure needs for community care and rehabilitation and to assess provision against these needs.

Over view of the Needs and Provision Complexity Scale (NPCS)
The NPCS is an ordinal scale with five main domains and fifteen subscales which are summarized in the table below. It has a total range of 0-50

<table>
<thead>
<tr>
<th>Domains</th>
<th>Range</th>
<th>Items</th>
<th>Code</th>
<th>Score Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Healthcare</td>
<td>0-6</td>
<td>Medical care needs</td>
<td>M</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Skilled nursing needs</td>
<td>N</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td>Personal care</td>
<td>0-10</td>
<td>Number of carers</td>
<td>CN</td>
<td>0-2 0-2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Care frequency</td>
<td>CF</td>
<td>0-5 0-5</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Personal assistant / enabler</td>
<td>PA</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td>Rehabilitation</td>
<td>0-9</td>
<td>Therapy disciplines</td>
<td>TD</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Therapy Intensity</td>
<td>TI</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Vocational support/rehabilitation</td>
<td>VR</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td>Social and family</td>
<td>0-13</td>
<td>Social work case management</td>
<td>S</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td>Support</td>
<td></td>
<td>Family carer support</td>
<td>FC</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Respite - residential</td>
<td>RR</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Respite - day care</td>
<td>RD</td>
<td>0-2 0-2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Advocacy</td>
<td>AD</td>
<td>0-2 0-2</td>
</tr>
<tr>
<td>Environment</td>
<td>0-12</td>
<td>Equipment</td>
<td>E</td>
<td>0-3 0-3</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Accommodation</td>
<td>AC</td>
<td>0-9 0-9</td>
</tr>
<tr>
<td>Total</td>
<td>0-50</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
The NPCS is divided into two parts

**Part A: NPCS-Needs** – defines the needs for input under the different headings
(The NPCS records both professional and personal perspectives on what the patient needs (professional view) and what they would like (personal choice).

**Part B: NPCS- Gets (or ‘Provision’)** – defines the current level of input or service currently provided within the same structure.

Unmet needs – are therefore recorded as the difference between the two scales.

Broadly, the rating levels under each item are designed to determine
a) the number of staff required and
b) the frequency of needs/intervention.
Level descriptors give a rough guide as to what might be “occasional”, “regular” or “frequent” , but these are not designed to be strictly defined cut-off points.

The summary score sheet includes a check list of the specific services required under each heading
It also includes a set of boxes to record the reasons for variance (unmet need) which may be:

a) **Service not available** – ie there is no service available, or it has not been offered
b) **Service declined** – ie service has been offered, but declined by the pt / carer (this will often be because they consider that which has been offered to be unsuitable) – there is space to record the specific circumstances if desired.

c) **Other** - some other reason

‘Needs wants and gets’

There is also the option to score ‘what the patient wants’.

This is particularly relevant in two scenarios:

a) where professionals consider that the patient requires a certain service, but the patient does not accept it.

* A typical example would be where the professionals believe that an individual requires care for the purpose of safety monitoring but the individual refuses this, either because they lack insight into their difficulties, or because they wish to maintain their independence and autonomy

b) where professionals consider that the patient does not require a certain service, because there is no clinical potential to benefit it, but the patient and or family wants that service because they feel that it may help.

* A typical example would be the situation where an individual has permanent physical disability which does not have the potential to change with physiotherapy, but a maintenance programme is prescribed. However, the individual and/or their family cling strongly to the believe that, with enough therapy, they will regain the lost ability

In both situations, the patient /family has a valid view-point which should be recorded but, for various reasons, the services they want cannot be given.
# The Needs & Provision Complexity Scale (NPCS) for LTNC

## PATIENT IDENTIFICATION

| Name: | NHS No: | Date of score:.../.../...... |

## PART A - NEEDS: For each subscale, circle highest level applicable

### 1. MEDICAL CARE NEEDS – requiring intervention from a doctor for investigation, monitoring or treatment
- Specialist Medical input may be from any medical specialty

| M 0 | GP occasional | no regular contact – self-initiated visits to GP as required |
| M 1 | GP active monitoring | regular monitoring/ treatment by GP solely |
| M 2 | Low level specialist support | eg for largely stable condition, ongoing monitoring/ treatment by GP with occasional specialist advice / review |
| M 3 | Active specialist medical intervention required | eg for changing/unstable condition or for unresolved symptoms. Investigation or treatment requiring frequent contact with specialist medical team |

### Types of medical care
- Neurology
- Rehab medicine
- Neuropsychiatry
- Palliative care
- Other

### 2: SKILLED NURSING NEEDS – intervention required from trained and/or specialist nursing staff
- eg district nursing or Specialist nurse (E.g. for wound care, bladder / bowel management / medication monitoring / specialist advice/support/counselling)

| N 0 | No needs for skilled nursing |
| N 1 | Occasional intervention (eg monthly or less) |
| N 2 | Regular intervention | eg every 1-2 weeks |
| N 3 | Frequent intervention on a daily basis, or Several times a week |

### Types of nursing care
- District nurse
- Specialist nurse
- Neurology
- Mental Health
- Palliative care
- Other...

## 3. PERSONAL CARE - In and around the home.

### 3a: Number of Carers: Required to help with basic self-care

| CN 0 | No carers | required for basic care activities |
| CN 1 | Requires help from 1 person | for most basic care needs |
| CN 2 | Requires help from ≥2 people | for most basic care needs |

### Who provides this help?:
- Informal family care
- Formal paid carers
- Other........................................

### 3b: Care frequency: Frequency of care for help with basic self-care, including maintaining safety

| CF 0 | No need | for help with self care. |
| CF 1 | Occasional need | – less than daily for help with self care, or extended activities of daily living |
| CF 2 | Requires regular help once daily |
| CF 3 | Requires regular help | 2-3 times a day – could be met by an intermittent visiting care package |
| CF 4 | Frequent or unpredictable care needs, requiring the presence of someone most of the time |
| CF 5 | Requires constant supervision | - unable to be left alone in the house, even for short periods AND/OR requires waking night care – needs > 2 interventions at night |

Able to be left safely for >4 hours and does not require care / supervision at night

Cannot be left safely for >4 hours or requires care / supervision at night (but not waking night care)
3c: Personal assistant/enabler  
**Frequency of assistance for participation in day time community activities**

<table>
<thead>
<tr>
<th>PA</th>
<th>No need for assistance with community activities</th>
</tr>
</thead>
<tbody>
<tr>
<td>PA 1</td>
<td>Occasional need – 1-2 days per week</td>
</tr>
<tr>
<td>PA 2</td>
<td>Frequent need – 3-5 days/week</td>
</tr>
<tr>
<td>PA 3</td>
<td>Daily – 6-7 days/week</td>
</tr>
</tbody>
</table>

4. THERAPY NEEDS – including outpatient, community-based and vocational rehabilitation

<table>
<thead>
<tr>
<th>Number of Therapy Disciplines:</th>
<th>required to be actively involved in treatment (ie at least 1 hr per month)</th>
</tr>
</thead>
<tbody>
<tr>
<td>TD 0</td>
<td>0</td>
</tr>
<tr>
<td>TD 1</td>
<td>Single discipline only</td>
</tr>
<tr>
<td>TD 2</td>
<td>Individual disciplines, not co-ordinated</td>
</tr>
<tr>
<td>TD 3</td>
<td>Co-ordinated interdisciplinary team</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Therapy Intensity:</th>
<th>Overall intensity of trained therapy intervention required</th>
</tr>
</thead>
<tbody>
<tr>
<td>TI 0</td>
<td>No need for trained therapy intervention</td>
</tr>
<tr>
<td>TI 1</td>
<td>Requires occasional review or maintenance programme – OR requires Group therapy solely</td>
</tr>
<tr>
<td></td>
<td>Eg Rehab needs met by family/care staff or self-exercise, supervised by therapist eg 1-2 hrs total/month</td>
</tr>
<tr>
<td>TI 2</td>
<td>Regular intervention for maintenance / treatment eg every 1-2 weeks: OP or domiciliary treatment</td>
</tr>
<tr>
<td>TI 3</td>
<td>Requires frequent intervention involving several sessions per week</td>
</tr>
</tbody>
</table>

5. VOCATIONAL /EDUCATIONAL SUPPORT NEEDS

| VR 0 | No need for vocational/educational support |
| VR 1 | Requires vocational assessment / advice or educational statementing |
| VR 2 | Requires on-going vocational /educational support eg Access to work scheme, or withdrawal from work |
| VR 3 | Requires formal vocational / educational rehab eg work prep, work re-training, supported placements |

6. SOCIAL WORK AND CASE MANAGEMENT — support / intervention to co-ordinate care / services

| S 0  | No needs for social work or case management |
| S 1  | Requires occasional intervention or available for advice when needed eg contact 2-3 x per year |
| S 2  | Requires regular intervention or contact eg every 1-2 months |
| S 3  | Requires frequent intervention or contact eg every 1-2 weeks |

7. FAMILY / CARER SUPPORT / RESPITE NEEDS

<table>
<thead>
<tr>
<th>7a: Family career support</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>FC 0</td>
<td>No needs for family / carer support</td>
</tr>
<tr>
<td>FC 1</td>
<td>Assessment required for family / carer</td>
</tr>
<tr>
<td>FC 2</td>
<td>Time-limited family/carer support required eg for skills training</td>
</tr>
<tr>
<td>FC 3</td>
<td>On-going family/carer support required eg for emotional support</td>
</tr>
</tbody>
</table>
### 7b. Respite - residential and day care centre:

<table>
<thead>
<tr>
<th>Type of respite care</th>
</tr>
</thead>
<tbody>
<tr>
<td>□ Home-based temporary live-in care</td>
</tr>
<tr>
<td>□ Residential home</td>
</tr>
<tr>
<td>□ Nursing home</td>
</tr>
<tr>
<td>□ Specialist nursing home</td>
</tr>
<tr>
<td>□ Hospice</td>
</tr>
<tr>
<td>□ Other</td>
</tr>
</tbody>
</table>

#### RESIDENTIAL RESPITE

| RR 0 | No need for residential respite care |
| RR 1 | Requires **occasional residential respite** — eg to cover holidays etc. |
| RR 2 | Requires **regular planned residential respite**, but not very frequent (eg 1-2 weeks per 6 months) |
| RR 3 | Requires **frequent planned residential care** (eg every 4-6 weeks) AND/OR **back-up support at times of crisis** |

#### DAY CARE

<table>
<thead>
<tr>
<th>Type of day care</th>
</tr>
</thead>
<tbody>
<tr>
<td>□ Community day centre</td>
</tr>
<tr>
<td>□ Specialist day centre</td>
</tr>
<tr>
<td>□ Hospice</td>
</tr>
</tbody>
</table>

| RD 0 | No need for day care |
| RD 1 | **Occasional day care** — 1-2 days per week |
| RD 2 | **Frequent day care** — 3-5 days/week |

### 8. ADVOCACY NEEDS —

| AD 0 | No needs for advocacy |
| AD 1 | **Mental capacity assessment** required |
| AD 2 | **Independent advocacy** required |

### 9. SPECIALIST EQUIPMENT — *Eg Special seating, assistive technology, ventilation equipment*

<table>
<thead>
<tr>
<th>Type of Equipment</th>
</tr>
</thead>
<tbody>
<tr>
<td>□ Basic lifting handling equipment</td>
</tr>
<tr>
<td>□ Seating/wheelchair</td>
</tr>
<tr>
<td>□ Standing/postural support</td>
</tr>
<tr>
<td>□ Electronic Assistive technology</td>
</tr>
<tr>
<td>□ Communication aid</td>
</tr>
<tr>
<td>□ Assisted ventilation</td>
</tr>
<tr>
<td>□ Other..</td>
</tr>
</tbody>
</table>

| E 0 | No specialist equipment required |
| E 1 | **Basic equipment required** (eg from social services equipment store eg kitchen aids, commode, bed, hoist etc) |
| E 2 | **Specialist equipment required** — equipment requiring professional assessment and provision (eg seating, standing frames) |
| E 3 | **Highly specialist equipment required** — bespoke equipment requiring professional prescription (eg environmental control, communication aids, ventilatory support) |

### 10. ACCOMMODATION NEEDS

| AC 0 | No need for special accommodation |
| AC 1 | **Restricted accommodation options** (eg requires ground floor or lift access accommodation) |
| AC 2 | Requires partially adapted accommodation (eg rails, ramps etc) |
| AC 3 | Requires fully adapted accommodation (eg fully wheelchair accessible) |

#### SHELTERED AND RESIDENTIAL CARE

| AC 4 | Requires **sheltered living accommodation** (eg warden controlled) |
| AC 5 | Requires supervised living arrangement eg small group home |
| AC 6 | Requires residential care home setting |
| AC 7 | Requires **nursing home care** |
| AC 8 | Requires **specialist nursing home** |
| AC 9 | Requires **Hospice care** |
Part B: The Inputs provided

Part B is intended to mirror Part A, except that it records what the person actually gets – and so identifies **unmet need**
# PART B – The Inputs Provided

## PATIENT IDENTIFICATION

| Name: | NHS No: | Date of score:....../....../....... |

For each subscale, circle **highest level** applicable

### 1. MEDICAL CARE PROVISION – intervention from a doctor for investigation, monitoring or treatment

- Specialist Medical input may be from **any** medical specialty

| M 0 | GP occasional — no regular contact – self-initiated visits to GP as required | Types of medical care:
- Neurology
- Rehab medicine
- Neuropsychiatry
- Palliative care
- Other |
| M 1 | GP active monitoring - regular monitoring/treatment by GP solely |
| M 2 | Low level specialist support eg for largely stable condition On-going monitoring/treatment by GP with occasional specialist advice/review |
| M 3 | Active specialist medical intervention eg for changing/unstable condition or for unresolved symptoms. Investigation or treatment requiring frequent contact with specialist medical team |

### 2: SKILLED NURSING PROVISION – intervention from trained and/or specialist nursing staff

eg district nursing or Specialist nurse (E.g. for wound care, bladder/bowel management / medication monitoring / specialist advice/support/counselling)

| N 0 | No provision of skilled nursing |
| N 1 | Occasional intervention from a trained or specialist nurse (eg monthly or less) |
| N 2 | Regular intervention from a trained nurse or specialist nurse eg every 1-2 weeks |
| N 3 | Frequent intervention from a trained nurse or specialist nurse on a daily basis, or Several times a week |

### 3. PERSONAL CARE - In and around the home.

#### 3a: Number of Carers: provided to help with **basic self-care**

| CN 0 | No carers for basic care activities | Who provides this help?:
- Informal family care
- Formal paid carers
- Other................................. |
| CN 1 | 1 carer |
| CN 2 | 2 carers |

#### 3b: Care frequency: Frequency of care for help with **basic self-care**, including maintaining safety

| CF 0 | No provision for help with self care. |
| CF 1 | Occasional care visits – less than daily for help with self care, or extended activities of daily living |
| CF 2 | Once daily care visit 1-2 hours |
| CF 3 | 2-3 care visits per day – (or 3-6 hours care per day in total) - no night time care |
| CF 4 | Live-in or all day care package - >6 hours |
| CF 5 | 1:1 care throughout the day AND/Or waking night care |
3c: Personal assistant/enabler - Assistance for participation in day time community activities

| PA 0 | No provision for assistance with community activities |
| PA 1 | Occasional assistance provided – 1-2 days per week |
| PA 2 | Frequent assistance provided – 3-5 days/week |
| PA 3 | Daily assistance provided – 6-7 days/week |

4. THERAPY PROVISION — including outpatient, community-based and vocational rehabilitation

Number of Therapy Disciplines: - actively involved in treatment (ie at least 1 hr per month)

| TD 0 | 0 |
| TD 2 | Individual disciplines, not co-ordinated |
| TD 3 | Co-ordinated interdisciplinary team |

Tick therapy disciplines involved:
- Physio
- O/T
- SLT
- Dietetics
- Orthotics / Prosthetics
- Psychology
- Counselling
- Mental health
- Other:

Therapy Intensity: - Overall intensity of trained therapy intervention

| TI 0 | No therapy intervention (or <1 hr per month) |
| TI 1 | Occasional review or maintenance programme — about 1-2 hours/month in total – OR attends for Group therapy solely |
| TI 2 | Regular intervention for maintenance / treatment eg every 1-2 weeks: |
| TI 3 | Frequent intervention involving several sessions per week (may be from different disciplines) |

5. VOCATIONAL / EDUCATIONAL SUPPORT PROVISION

| VR 0 | No provision for vocational/educational support |
| VR 1 | Received/ing vocational /educational assessment / advice or statementing |
| VR 2 | Receives on-going vocational/educational support eg Access to work scheme, or withdraw from work |
| VR 3 | Receives formal vocational / educational rehabilitation eg work preparation, work re-training, supported placements |

6. SOCIAL WORK AND CASE MANAGEMENT — support / intervention to co-ordinate care / services

| S 0 | No provision of social work or case management – or very inconsistent (ie effectively none) |
| S 1 | Occasional intervention or contacts for advice when needed eg 2-3 times per year |
| S 2 | Regular intervention or contact eg every 1-2 months |
| S 3 | Frequent intervention or contact eg every 1-2 weeks |

7. FAMILY / CARER SUPPORT / RESPITE PROVISION

7a: Family career support

| FC 0 | No provision for family / carer support |
| FC 1 | Received/ing assessment for family / carer |
| FC 2 | Received/ing family/carer support eg for skills training |
| FC 3 | Receives on-going family/carer support eg for emotional support |
7b. Respite - residential and day care centre:

**RESIDENTIAL RESPITE**

- **RR 0** No provision for residential respite care
- **RR 1** Occasional residential respite provision – eg to cover holidays etc.
- **RR 2** Regular planned residential respite provision, but not very frequent (eg 1-2 weeks per 6 months)
- **RR 3** Frequent planned residential care (eg every 4-6 weeks) AND/OR back-up support at times of crisis

**DAY CARE**

- **RD 0** No provision for day care
- **RD 1** Occasional day care provided – 1-2 days per week
- **RD 2** Frequent day care provided – 3-5 days/week

8. ADVOCACY PROVISION –

- **AD 0** No provision for advocacy
- **AD 1** Received/ing mental capacity assessment
- **AD 2** Receiving Independent advocacy

9. SPECIALIST EQUIPMENT – Eg Special seating, assistive technology, ventilation equipment

- **E 0** No specialist equipment /provision inadequate
- **E 1** Basic equipment provided (eg from social services equipment store eg kitchen aids, commode, bed, hoist etc)
- **E 2** Specialist equipment provided – equipment requiring professional assessment and provision (eg seating, standing frames)
- **E 3** Highly specialist equipment provided – bespoke equipment requiring professional prescription (eg environmental control, communication aids, ventilatory support)

10. ACCOMMODATION PROVISION

- **AC 0** No provision for special accommodation
- **AC 1** Restricted accommodation options met (eg requires ground floor or lift access accommodation)
- **AC 2** Has partially adapted accommodation (eg rails, ramps etc)
- **AC 3** Has fully adapted accommodation (eg fully wheelchair accessible)

SHELTERED AND RESIDENTIAL CARE

- **AC 4** Has sheltered living accommodation (eg warden controlled)
- **AC 5** Has supervised living arrangement eg small group home
- **AC 6** Has residential care home setting
- **AC 7** Has nursing home care
- **AC 8** Has specialist nursing home
- **AC 9** Has Hospice care
**Validation of the NPCS.**

Preliminary evaluation of the NPCS has been undertaken and findings were presented at the World Congress in Neuro-rehabilitation, Melbourne, May 2012.

A first evaluation in a cohort of 423 patients discharges from specialist in-patient rehabilitation services across the London region demonstrated that the NPCS was easily understood and completed by both clinicians and patients.

- Needs were rated by the treating clinicians on discharge.
- Patients and/or their family carers recorded the levels of services provision in relation to those needs by self report postal questionnaire (with follow-up telephone interview where necessary).

Exploratory factor analysis indicated two primary factors, reflecting needs for ‘Health and Personal Care’ and ‘Social and Family Support’.

- Full scale reliability was excellent with Cronbach’s α=0.94.
- Test-retest reliability for self-report was encouraging with ICCs for the six subscales ranging from 0.61-0.85.
- Item-by item agreement, rated by quadratic-weighted Cohen’s kappa coefficient ranged from 0.47-0.93.

*(Paste link to the poster here – WCNR poster no 49 (pdf) – factor structure)*

Application of the NPCS in the same series of patients demonstrated significant gaps between needs and service provision, especially with respect to on-going community rehabilitation, equipment and social support.

- By contrast, needs for medical, nursing and personal care were relatively well met.
- Provision of support for personal care above the level of predicted need suggested deterioration of independence for some patients after discharge, possibly as a result of the failure to meet needs for rehabilitation and social support.

*(Paste link to the poster here – WCNR poster no 48 (pdf) – unmet needs)*

Formal psychometric evaluation, including assessment of inter-rater reliability, has been submitted for publication Dec 2012.

**Creating a costing algorithm within the NPCS**

The Needs and Provision Complexity Scale (NPCS) provides an ordinal scale for estimating met and unmet need. A costing algorithm has been developed to express the impact of met and unmet needs directly in terms of cost.

- Data gathered using the Client Service receipt inventory (CSRI) data provided information on the number and duration of contacts for each type of service.
- These data were analysed within NPCS items to derive a set of costing assumptions for each scoring level.
- Where CSRI data was insufficient, intuitive assumptions were made based on clinical experience and tested within a peer group of clinicians experienced in the planning and provision of community services.
Costs were computed with reference to Curtis, 2011 and adjusted where necessary to reflect the costs of specialist care (with helpful further personal communication from Lesley Curtis). The 2011 unit costs were used because these data will be used prospectively in contrast to the retrospective application of 2010 costs to CSRI data, which reflected the year during which services were received by study participants.

(The NPCS costing algorithm)

References


PAPER I
Abstract

Background: In order to plan and improve provision of comprehensive care in Huntington’s disease (HD), it is critical to understand the gaps in healthcare and social support services provided to HD patients. Research has described utilization of healthcare services in HD in Europe, however, studies systematically examining needs for healthcare services and social support are lacking. This study aims to identify the level and type of met and unmet needs for health and social care services among patients with HD, and explore associated clinical and socio-demographic factors.

Methods: Eighty-six patients with a clinical diagnosis of HD living in the South-Eastern region of Norway were recruited. Socio-demographic and clinical characteristics were collected. The Needs and Provision Complexity Scale (NPCS) was used to assess the patients’ needs for healthcare and social services. Functional ability and disease stage was assessed using the UHDRS Functional assessment scales. In order to investigate factors determining the level of total unmet needs and the level of unmet needs for Health and personal care and Social care and support services, multivariate logistic regression models were used.

Results: A high level of unmet needs for health and personal care and social support services were found across all five disease stages, but most marked in disease stage III. The middle phase (disease stage III) and advanced phase (disease stages IV and V) of HD increased odds of having a high level of total unmet needs by 3.5 times and 1.4 times respectively, compared with the early phase (disease stages I and II). Similar results were found for level of unmet needs in the domain Health and personal care. Higher education tended to decrease odds of high level of unmet needs in this domain (OR = 0.48) and increase odds of higher level of unmet needs in the domain of Social care and support (OR = 1.3). Patients reporting needs on their own tended to decrease odds of having unmet needs in Health and personal care (OR = 0.57).

Conclusions: Needs for healthcare and social services in patients with HD should be assessed in a systematic manner, in order to provide adequate comprehensive care during the course of disease.

Keywords: Huntington’s disease, Healthcare services, Social support services, Healthcare needs
Background

Many rare diseases, such as Huntington’s disease (HD), are chronic and complex, and are associated with physical, mental or neurological disabilities. Systematic assessment of patients’ needs for healthcare and social services may identify gaps that could lead to improved service provision [1, 2].

HD is an autosomal dominant neurological disease caused by an expanded CAG repeat in the Huntingtin gene. The disease is characterized by progressive functional decline and motor, psychiatric and cognitive symptoms, in addition to weight loss, sleep disturbances and dysregulation of the autonomic nervous system [3–6]. A clinical diagnosis of HD is given when unequivocal motor symptoms are present, but subtle motor signs, psychiatric symptoms and cognitive changes may be present years prior to clinical diagnosis [7–11]. Clinical symptoms of HD usually develop during adult life between 30 and 50 years of age and disease duration from first clinical symptoms to complete care dependency and death is approximately 15–20 years [4].

At present, there is no curative treatment for HD, and treatment is aimed at alleviating symptoms, maintaining and improving function and quality of life [12]. HD patients in early to middle stages of the disease need coordinated multidisciplinary healthcare services, including assessment of cognitive function and counselling by (neuro)psychologists [4], rehabilitation programs [13, 14], active physiotherapeutic interventions [15, 16], speech therapist training [4, 17, 18] and occupational therapy [4]. Patients in advanced stages of the disease are usually dependent on full-time personal care, but may still benefit from multidisciplinary care [19]. Family members experience challenges and need support and guidance [20].

Although there is general agreement that comprehensive, multidisciplinary care is needed [4, 19, 21–24], the complex and changing clinical picture may be a challenge for health professionals. Standards of care, aimed at separate groups of healthcare professionals (i.e., speech and language therapists) have been published, with the purpose of being a foundation for further research and evaluation of provided care [18, 25, 26]. A few clinics adapting comprehensive care models have emerged in the US, Australia and Europe [4, 24, 27].

In order to plan and improve provision of long-term care in HD, it is essential to understand what healthcare and social services HD patients receive, and what unmet healthcare needs they may have. A few studies have assessed health and social care utilization and needs in HD and results showed a number of unmet needs related to body functions, activities and level of participation as well as carer support [28, 29].

To date, research has not addressed healthcare needs of HD patients in Norway. Norway, like the other Scandinavian countries, is a welfare state with equal access to health and social care services, and services are either free or subsidized at point of delivery. Thus, the Scandinavian studies on HD patients and delivery of healthcare services may be of international interest.

The aims of the present study are threefold: a) to investigate the level of unmet needs for healthcare and social support services among HD patients, b) to investigate how the level of unmet needs are divided across disease phases, and c) to investigate the association between sociodemographic and clinical disease characteristics and levels of unmet needs.

We anticipated considerable levels of unmet needs for healthcare and social support services across all phases of the disease. Furthermore, we anticipated the advanced disease phase to be highly associated with the level of unmet needs for healthcare and social support services.

Methods

Participants and participant recruitment

Patients with a clinical diagnosis of HD residing in the South-Eastern region of Norway, a region with a population of 2.7 million, were invited to participate in a survey of healthcare needs and utilisation of healthcare services. Eligible patients were identified through the Department of Neurology, Department of Neurorehabilitation, and Department of Medical Genetics at Oslo University Hospital, the regional academic medical center. In addition, patients were recruited through the Centre for Rare Disorders at Oslo University Hospital, a national advisory service for HD that offers guidance to patients, families and healthcare professionals. In a further effort to reach all eligible patients, we collaborated with a Norwegian professional network for community care in HD (Huntington fagnettverk) and the Norwegian HD lay association (Landsforeningen for Huntingtons sykdom). The Vikersund Rehabilitation Centre, which runs a rehabilitation program for HD patients, were informed about the study and issued invitations to additional patients.

All eligible patients received a written invitation, enclosing information about the study and an informed consent form. Following return of the consent form, the patient/carer was contacted and an appointment for a study visit was made.

We identified a total of 158 eligible patients (which correspond to a prevalence of 5.9/100,000) who were invited to participate in the study, of which 88 patients gave their consent to participate and were included. Among the 70 patients who were not included, 27 declined to participate and 43 did not reply (see Fig. 1 for flow chart illustrating patient recruitment). An expert HD clinician reviewed medical records if there was any doubt about the diagnosis. Two patients did not have
sufficient symptoms to formally have been given a clinical diagnosis of HD and were therefore excluded. Finally, 86 (54.4%) out of the 158 potential participants were included in the data-analysis.

Ethics
The study was approved by the Regional Ethical Committee (ref. 2013/2089). Informed consent was obtained for all patients. For patients who were unable to give informed consent themselves, consent was obtained from the primary caregiver or legal representative.

Data collection
Data were collected through interviews conducted by two experienced clinical raters, either during an outpatient study visit (38.0%) or at the patients’ home (62.0%). We recorded with whom the interview was performed, i.e., patient alone, patient with primary informal and/or formal carer, or informant only. Socio-demographic information and clinical functional assessment was recorded at the beginning of the visit. Co-morbid conditions not related to HD were also recorded. Furthermore, patients were rated regarding functional ability and needs for healthcare and social services. Patients’ medical records were reviewed if further information was needed.

Description of measurements
Unified Huntington’s disease rating scale – functional assessment
The UHDRS-Functional assessment comprises three scales: a) the Total Functional Capacity Scale (UHDRS-TFC), rating ability to engage in occupation, manage finances and domestic chores, and perform activities of daily living (ADL), with a score range of 0 – 13. The scale is used to classify patients into five functional disease stages, using the following convention: A TFC score of 11–13 represents Stage I, a TFC score of 7–10 represents Stage II, TFC score of 3–6 represents Stage III, a TFC score of 1–2 represents Stage IV and a TFC score of 0 represents Stage V. b) the Functional Assessment Scale (FAS), a daily living checklist with scoring range 0–25, and c) the Independence scale (IS) with range 10 to 100. Higher scores indicate better functioning [30].

The Needs and Provision Complexity Scale (NPCS)
The NPCS was specifically developed to identify healthcare and social support needs among patients with long term neurological conditions in the UK [2]. It is a brief and practical tool for measuring the needs for healthcare and social support of an individual, and the extent to which those needs are met through service provision. At the individual level, the NPCS can be used to monitor the changing needs of patients over time and services provided to support them along the care pathway, while at a population level it can identify gaps of service provision. The NPCS clinician version consists of a 15-item measure with six sub-scales and a total scoring range of 0-50 covering “low” to “high” levels of needs. It has two parts: a) Part A (NPCS-Needs) which is completed by the clinician to evaluate each patients’ need for health and social care, b) Part B (NPCS-Gets) is a mirror image of the same instrument to evaluate the services that have been provided. Furthermore, the NPCS consists of two main domains a) Health and Personal care needs (score range 0–25), including the following subscales: Healthcare (score 0–6), Personal care score (0–10), Rehabilitation (0–9), and b) Social care and support needs (score range 0–25), including subscales Social and family support (score 0–13), Equipment (score 0–3) and Environment (score 0–9). For an overview of the NPCS, see Additional file 1.

The NPCS was translated by native speakers from English to Norwegian and then back-translated to English to check for accuracy. The translation was reviewed by expert researchers and clinicians in the field of healthcare services and HD.
Statistical analyses

Outliers and missing values

Prior to analysis, the data were screened for extreme values and missing data. Some outliers were identified in the data on sample characteristics and NPCS scores, and checked with the original data material. These were true values and therefore kept in the statistical analyses. Initially, four missing values for variables years of education, level of education (lower vs. higher), and three for occupation type (manual vs. non-manual) were identified and later estimated from the patients’ medical records. For four patients, we were unable to collect information about disease duration (number of years with clinical diagnosis of HD) during the survey interview, and we used clinical information that was available through medical records to estimate disease duration. For three patients we were unable to attain CAG repeat number. Additionally, information on one item in the NPCS scale was missing (<2 %) for one participant.

Descriptive analyses

Descriptive statistics were used (proportions, mean values, standard deviations (SD) or median values with interquartile range, (IQR)) in order to describe the socio-demographic and clinical disease characteristics of the complete sample and across the five disease stages. Cross tabulations’ Chi-square tests were used in order to compare nominal socio-demographic and clinical characteristics across disease stages. Group comparisons across disease stages for continuous variables, were calculated using Non-parametric Kruskal-Wallis k-sample tests, as the data were not normally distributed (with exception of participants age).

Descriptive analyses for NPCS needs, gets and unmet needs

Scores for levels of unmet needs (representing gaps between patients’ needs for healthcare and social support services and provision of these services) were calculated as the discrepancy between the scores for level of the patients’ Needs and Gets (Score on NPCS Needs – Score on NPCS Gets = NPCS Unmet needs). The level of unmet needs were calculated for the NPCS Total score, for the domain scores Health and personal care and Social care and support, as well as for the six corresponding subscales. Descriptive statistics presented by median values with interquartile scores were used in order to present the level of patients’ needs, the level of provision (Gets), and the level of unmet needs for the total sample as well as for disease stage I-V. Additionally, frequencies and proportions of patients with unmet needs on each of the NPCS scores were calculated for the total sample and five disease stages. Group comparisons across the five disease stages for levels of needs, gets and unmet needs and for frequencies were made using Kruskal-Wallis k sample test. The p-value was set at 0.05.

Evaluating effects on the level of unmet needs

In order to investigate the factors determining the level of total unmet needs, and the level of unmet needs for Health and personal care and Social care and support services, multivariate logistic regression models were used. Regression analyses were performed on the group of patients having unmet needs on the NPCS total score and the two main domains health and personal care and social care and support. The NPCS total score and the domain scores were categorized into two groups based on median value: low level of unmet needs vs. high level of unmet needs. This resulted in the following categories: NPCS Total unmet needs: low level (1–6) vs. high level of unmet needs (>6); NPCS Health and personal care unmet needs: Low level (1–3) vs. high level (>3); NPCS social care and support unmet needs: Low level (1 & 2) vs. high level (>2). Due to the small number of patients, the five disease stages were collapsed into three disease phases: early phase (disease Stage I & II), middle phase (Stage III) and advanced phase (Stages IV & V) in the regression analyses. Statistically significant factors (level of education (lower vs. higher), informant (patient alone vs. patient and informant/informant alone) and disease phase (early, middle or advanced) from the univariate analyses including simple logistic regression were included in the multivariate models in order to investigate their impact on the total level of unmet needs and unmet needs for health and personal care and social care and support. We applied similar multivariate models to demonstrate that certain factors are common and consistently important. To control for the heterogeneity in the included sample, all models were adjusted for age (years), disease duration (years), and comorbidity (yes/no). Results from the multiple logistic regression analyses are presented with odds ratios (OR) with 95 % confidence intervals. Furthermore, the Hosmer-Lemeshow goodness-of-fit statistics were computed. Prior to conducting the logistic regressions we investigated multicollinearity. The variables with correlation coefficients >.70 were not entered in the regression analyses. All analyses were conducted using SPSS version 21.0; SPSS Inc. Chicago IL.

Results

Socio-demographic and clinical description of the patients

The socio-demographic characteristics for the total sample and across disease stages are summarized in Table 1. The median age was 57.5 years, and 54.7 % of the patients were men. The majority of patients lived at home (62.8 %). Of the complete sample of 86 patients, 12 (14 %) were in stage I, 23 (26 %) in stage II, 19 (22 %) in stage III, 15
At stage IV, and 17 (20 %) in stage V. Overall, significant group differences across disease stages were found for variables Occupational situation, Housing situation and Informant.

Clinical characteristics for the total sample of patients across disease stages are presented in Table 2. The median values for disease duration, total FAS score and FAS independence scores were respectively 6.1 (IQR 6.8) years, 15 (IQR 17), 70 (IQR 35). Of the complete sample, 36 (42 %) patients had comorbid conditions, 5 (42 %) in disease stage I, 13 (56 %) in disease stage II, 10 (53 %) in disease stage III, 4 (27 %) in disease stage IV, and 4 (23 %) in disease stage V.

**Table 1** Socio-demographic statistics for complete sample and divided across V disease stages

<table>
<thead>
<tr>
<th>Variables</th>
<th>Categories</th>
<th>Complete sample (N = 86)</th>
<th>Stage I (n = 12)</th>
<th>Stage II (n = 23)</th>
<th>Stage III (n = 19)</th>
<th>Stage IV (n = 15)</th>
<th>Stage V (n = 17)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age</td>
<td></td>
<td>58 (15)</td>
<td>49 (20)</td>
<td>54 (21)</td>
<td>58 (11)</td>
<td>58 (8)</td>
<td>59 (16)</td>
</tr>
<tr>
<td>Education (years)*</td>
<td></td>
<td>12 (5)</td>
<td>13 (5)</td>
<td>12 (7)</td>
<td>11 (2)</td>
<td>12 (6)</td>
<td>11 (6)</td>
</tr>
<tr>
<td>Gender</td>
<td>Female</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td>47 (55)</td>
<td>7 (58)</td>
<td>14 (61)</td>
<td>12 (63)</td>
<td>7 (47)</td>
<td>7 (41)</td>
</tr>
<tr>
<td>Education</td>
<td>Lower</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
</tr>
<tr>
<td></td>
<td>Higher</td>
<td>34 (39.5)</td>
<td>7 (58)</td>
<td>11 (48)</td>
<td>6 (21)</td>
<td>6 (40)</td>
<td>6 (35)</td>
</tr>
<tr>
<td>Sivil status</td>
<td>Single</td>
<td>36 (42)</td>
<td>4 (33)</td>
<td>7 (30)</td>
<td>9 (47)</td>
<td>8 (53)</td>
<td>8 (47)</td>
</tr>
<tr>
<td></td>
<td>Married</td>
<td>50 (58)</td>
<td>8 (67)</td>
<td>16 (70)</td>
<td>10 (53)</td>
<td>7 (47)</td>
<td>9 (53)</td>
</tr>
<tr>
<td>Occupation</td>
<td>Manual</td>
<td>41 (48)</td>
<td>5 (42)</td>
<td>10 (43.5)</td>
<td>12 (63)</td>
<td>6 (40)</td>
<td>8 (47)</td>
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<tr>
<td></td>
<td>Non-manual</td>
<td>42 (49)</td>
<td>7 (58)</td>
<td>13 (56.5)</td>
<td>7 (37)</td>
<td>8 (53)</td>
<td>7 (41)</td>
</tr>
<tr>
<td>Occupational status</td>
<td>Employed</td>
<td>14 (16)</td>
<td>11 (92)</td>
<td>3 (13)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>0 (0)</td>
</tr>
<tr>
<td></td>
<td>Unemployed</td>
<td>72 (84)</td>
<td>1 (8)</td>
<td>20 (87)</td>
<td>17 (100)</td>
<td>15 (100)</td>
<td>17 (100)</td>
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<tr>
<td>Informant</td>
<td>Patient</td>
<td>27 (31)</td>
<td>9 (75)</td>
<td>14 (61)</td>
<td>4 (21)</td>
<td>0 (0)</td>
<td>0 (0)</td>
</tr>
<tr>
<td></td>
<td>Patient &amp; informant/informant only</td>
<td>59 (69)</td>
<td>3 (25)</td>
<td>9 (39)</td>
<td>15 (79)</td>
<td>15 (100)</td>
<td>17 (100)</td>
</tr>
<tr>
<td>Housing situation</td>
<td>Living at home</td>
<td>54 (63)</td>
<td>12 (100)</td>
<td>23 (100)</td>
<td>13 (68)</td>
<td>6 (40)</td>
<td>0 (0)</td>
</tr>
<tr>
<td></td>
<td>Not living at home</td>
<td>32 (37)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>6 (32)</td>
<td>9 (60)</td>
<td>17 (100)</td>
</tr>
<tr>
<td>Residence</td>
<td>Rural</td>
<td>13 (15)</td>
<td>1 (8)</td>
<td>4 (17)</td>
<td>2 (10.5)</td>
<td>3 (20)</td>
<td>3 (18)</td>
</tr>
<tr>
<td></td>
<td>Urban</td>
<td>73 (85)</td>
<td>11 (92)</td>
<td>19 (83)</td>
<td>17 (89.5)</td>
<td>12 (80)</td>
<td>14 (82)</td>
</tr>
</tbody>
</table>

IQR Interquartile range; Group comparison across the five disease stages performed using Chi-square tests for independent samples (categorical values). *normally distributed and therefore reported result from ANOVA. **not normally distributed therefore performed and reported Kruskal-Wallis test. ***responses missing (1 in stage IV and 2 in stage V). Remaining proportions and comparisons are crosstabs / Chi-square. IQR: Interquartile range.

Description of healthcare and social support needs, provision and unmet needs
Bargraphs with median values for disease stages I-V for the NPCS total, domain and subscale scores for Needs, Gets and Unmet needs are presented Fig. 2. In general, the median values for NPCS total score and domain scores for Needs and Gets increase from disease stage I – IV, and remain stable from stage IV to V. NPCS Needs and Gets median values for subscales Personal care and Accommodation follow a similar pattern.

Proportions of patients with unmet needs in the total sample and across disease stages are presented in Table 3. Results show high proportions of patients with unmet needs for NPCS total score (92 %), domains Health and personal care (83 %), and Social care and support (79 %), and subscales Rehabilitation (74 %) and Social and family support (66 %). The highest proportion of patients with unmet needs for the overall and domain scores was found in disease stage III (95 % each). Comparing the proportions of patients with unmet needs between stages, significant group differences were found on subscales Personal Care ($p = 0.00$) and Accommodation ($p = 0.00$).

Factors associated with level of unmet needs across the disease phases (early, middle and advanced)
The results of modelling total level of unmet needs (NPCS total score unmet needs) and level of unmet
needs for the two NPCS domains Health and personal care and Social care and support respectively are displayed in Table 4. Being in the middle and advanced phase of HD increased the odds of having a high level of total unmet needs by 3.5 times (OR = 3.5) and 1.4 times (OR = 1.4) respectively, whereas the patient reporting their needs without help from an informant tended to decrease the odds of having high level of unmet needs (OR = 0.52). Similar results were found for level of unmet needs in the domain Health and personal care: the middle and advanced phase of HD tended to increase the odds of having a high level of unmet needs (OR = 2.77 and OR = 2.20 respectively). Additionally, higher education and patients reporting their needs without help from an informant tended to decrease the odds of reporting a high level of unmet needs in this domain (OR = 0.48 and OR = 0.57 respectively). Furthermore, higher education tended to increase the odds of reporting a high level of unmet needs in the domain of Social care and support (OR = 1.3). Having comorbid conditions tended to decrease the odds of reporting a high level of unmet needs for Social care and support (OR = 0.65). Results of residual analyses identified two extreme cases for the model for the Total level of unmet needs. Removing the cases from the analyses did not change the results. No outliers were identified for the level of unmet needs on either of the NPCS domains. Hosmer and Lemeshow tests for goodness-of-fit were satisfactory for all three models.

**Discussion**

This is the first study to systematically investigate to which extent healthcare and social needs are met in a

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**Table 2 Sample clinical characteristics**

<table>
<thead>
<tr>
<th>Variables</th>
<th>Complete sample (N = 86)</th>
<th>Stage I (n = 12)</th>
<th>Stage II (n = 23)</th>
<th>Stage III (n = 19)</th>
<th>Stage IV (n = 15)</th>
<th>Stage V (n = 17)</th>
<th>Sign</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disease duration</td>
<td>Median (IQR)</td>
<td>6 (7)</td>
<td>2 (2)</td>
<td>5 (6)</td>
<td>7 (5)</td>
<td>10 (8)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Total FAS score</td>
<td>Median (IQR)</td>
<td>15 (17)</td>
<td>24 (2)</td>
<td>20 (2)</td>
<td>15 (4)</td>
<td>5 (2)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Independence score</td>
<td>Median (IQR)</td>
<td>70 (35)</td>
<td>97 (9)</td>
<td>80 (5)</td>
<td>65 (10)</td>
<td>45 (20)</td>
<td>P &lt; 0.001</td>
</tr>
</tbody>
</table>

FAS Functional Assessment Scale, IQR Interquartile range; Group comparisons are completed using Chi-square tests for categorical variables and Kruskall-Wallis for continuous/interval variables, as none of the continuous variables were normally distributed.

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**Fig. 2** Bargraphs illustrating level of needs, provision and unmet needs for NPCS total, domain and subscale scores.
representative sample of HD patients by using the newly developed Needs and Provision Complexity Scale (NPCS). As expected, the results indicate a high frequency of unmet needs and gaps in provision of healthcare and social support services at the overall level and in the domains health and personal care and social support services across all five disease stages. More than half of the patients had unmet needs in the NPCS subscales, including Rehabilitation and Social and family support. The results are, in general, in agreement with a previously mentioned survey in the UK [28], and a recent study of HD patients in Europe [29], and thus, may represent the real needs of the HD population.

Contrary to our hypothesis, the results suggest that the most substantial gaps between healthcare and social support service needs and provision are in the middle phase (disease stage III) of HD, in terms of both proportion of patients and level of unmet needs. Patients in this phase represent a heterogenic group, due to higher variation in symptom presentation and progression [31]. A prerequisite for offering adequate help in the middle phase of HD is that healthcare providers understand the needs of these patients and collaborate with family caregivers [32]. Thus, a stronger focus on monitoring patients’ symptoms and functioning is warranted, and healthcare services need to be targeted to this specific group of HD patients. Even though the level of needs for social and healthcare services as a total are greater in disease stages IV and V, these patients receive a higher amount of services, resulting in a smaller amount of unmet needs. This may be due to symptoms being more overt in these stages causing patients to no longer being able to carry out daily activities independently, which may lead to greater awareness of the needs of these patients. Indeed, observations from clinical practice indicate that HD patients in later stages tend to have a higher caregiver frequency and are more often cared for outside the home. Our findings are also in line with research on patient with long-term neurological conditions reporting that patients whose rehabilitation needs were met were more dependent at 12 months after discharge from hospital than those with unmet needs [33]. Furthermore, studies identifying unmet needs after traumatic brain injury indicate that patients with more visible needs have a higher degree of met needs, which may reflect that health professionals are working actively and responsibly for the patient; thus, the patients are more satisfied and perceive their needs as met [34].

<table>
<thead>
<tr>
<th>Table 3 Proportions of unmet needs among participants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Variable</td>
</tr>
<tr>
<td>-------------------------------------------------------</td>
</tr>
<tr>
<td>NPCS\textsuperscript{a} total score</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td>Domain score Health and Personal care</td>
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<td></td>
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<tr>
<td>Subscale</td>
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<tr>
<td>Healthcare</td>
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<tr>
<td>Personal Care</td>
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<tr>
<td>Rehabilitation</td>
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<tr>
<td>Domain\textsuperscript{a} score Social Care and Support</td>
</tr>
<tr>
<td>Social and family support\textsuperscript{a}</td>
</tr>
<tr>
<td>Specialist equipment</td>
</tr>
<tr>
<td>Unmet needs</td>
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<tr>
<td>Accommodation</td>
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<tr>
<td>Unmet needs</td>
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<tr>
<td>Accommodation</td>
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<tr>
<td>Unmet needs</td>
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</tbody>
</table>

\textsuperscript{a}NPCS Needs and Provision Complexity Scale; Chi-square test for categorical variables was used; \textsuperscript{*}Five or less cells have expected count less than five. \textsuperscript{a}missing data (1 missing NPCS total score, disease stage III; 1 missing domain score social care and support, disease stage III; 1 missing subscale social and family support, disease stage III)
Factors associated with level of unmet needs across the disease phases

Furthermore, we aimed to assess which socio-demographic and clinical factors were associated with the level of unmet needs for healthcare and social support services. Modelling unmet needs further illustrated the association between disease phase and level of unmet needs for the total level of unmet needs and in the domain Health and personal care. A lower level of education tended to decrease the odds of having a high level of unmet needs in health and personal care domain. One possible explanation for this finding can be related to trends in the general population; people with higher education are more resourceful and have a better understanding and awareness of what healthcare and social support services are available or that they are entitled to, as well as more resources to follow up on receiving the services they need [35]. As a consequence they may report a greater amount of unmet needs. Regardless, we have to interpret this result with caution, as we did not address patients cognitive or behavioral states in this study.

Furthermore, if the survey interview was conducted with the patient only, results showed trends toward decreased odds of having high levels of unmet needs. This may reflect patients’ having reduced awareness of their symptoms causing them to underreport and deny needs for health and personal care services. On the other hand, research has shown that in some cases, proxies tend to overestimate patient disability [36] and hence, may overestimate the patients’ needs for healthcare and social services. This often appears to be the case in clinical practice and the “truth” often seems to be somewhere in the middle.

None of the socio-demographic and clinical factors included were significantly associated with level of unmet needs in the social care and support domain. However, education level tended towards an impact on this domain as higher education increased the chance of having higher levels of unmet needs. A contradiction seemed to emerge with this result as a higher educational attainment, in general, has been associated with higher levels of social support. Future studies are needed to tease this apart, as our data are limited in their ability to provide more insight.

Comorbidity tended to decrease the chance of having higher levels of unmet needs for social care and support in this study. The presence of comorbid conditions has been associated with poorer social functioning and further research is needed for a fuller understanding of this finding.

Modelling the gaps in healthcare and social support services provide additional support to the overall level of

Table 4 Factors associated with level of unmet needs using binary multiple logistic regression models for total level of unmet needs (NPCS total score), level of unmet needs for health and personal care services (NPCS health and personal care score) and for level of unmet needs for social care and support services (NPCS social care and support score)

<table>
<thead>
<tr>
<th>Socio-demographic and clinical Variables</th>
<th>NPCS unmet needs: total (N = 79)</th>
<th>NPCS unmet needs: domain health and personal care (N = 71)</th>
<th>NPCS unmet needs: domain social care and support (N = 68)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age</td>
<td>1.03 (0.98 – 1.08)</td>
<td>1.02 (0.97 – 1.07)</td>
<td>1.00 (0.96 – 1.05)</td>
</tr>
<tr>
<td>Education Level</td>
<td>0.78 (0.28 – 2.22)</td>
<td>0.48 (0.16 – 1.45)</td>
<td>1.40 (0.467 – 3.95)</td>
</tr>
<tr>
<td>High education vs. lower education*</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Disease duration</td>
<td>0.97 (0.86 – 1.10)</td>
<td>0.98 (0.86 – 1.13)</td>
<td>1.00 (0.89 – 1.13)</td>
</tr>
<tr>
<td>Comorbidity</td>
<td>0.91 (0.31 – 2.70)</td>
<td>0.83 (0.25 – 2.74)</td>
<td>0.65 (0.22 – 1.90)</td>
</tr>
<tr>
<td>Comorbidity vs. no comorbidity*</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>HD Phase</td>
<td>3.57 (0.89 – 14.4)****</td>
<td>2.77 (0.62 – 12.36)</td>
<td>1.06 (0.27 – 4.18)</td>
</tr>
<tr>
<td>Middle phase vs. early phase*</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>HD Phase</td>
<td>1.38 (0.32 – 6.0)</td>
<td>2.20 (0.42 – 11.67)</td>
<td>1.06 (0.24 – 4.75)</td>
</tr>
<tr>
<td>Advanced phase vs. early*</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Informant</td>
<td>0.52 (0.13 – 2.04)</td>
<td>0.57 (0.14 – 2.30)</td>
<td>1.03 (0.24 – 3.6)</td>
</tr>
<tr>
<td>Patient vs. patient with informant*</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*Reference group
* OR > 1 increase the Odds of having a high level of unmet needs; OR < 1 decrease the odds of having high level of unmet needs
** Hosmer and Lemeshow Goodness-of-fit test χ² 0.12; df 8; P = 0.332
*** Hosmer and Lemeshow Goodness-of-fit test χ² 0.63; df 8; P = 0.612
**** Hosmer and Lemeshow Goodness-of-fit test χ² 6.65; df 8; P = 0.575
***** Approached significance (p = 0.074)
unmet needs and level of unmet needs for health and personal care increasing considerably for patients in the middle phase of HD. As mentioned before, the middle phase of HD represents a largely heterogenic group of patients, and this phase may present the most challenges and unmet needs as the family starts to see an increase of symptoms and the person with HD may have cognitive deficits and struggle to see their own problems. It can be considered a transitional phase, where patients progress from being relatively independent to becoming increasingly dependent in various areas of daily life. The transitional character of this phase may cause additional challenges in providing the tailored multidisciplinary healthcare these patients need. Providing adequate care to patients with HD should include regular monitoring and evaluation of symptoms and functioning from the moment of diagnosis, but may be particularly important in the middle phase. One of the important messages to clinicians is to not only evaluate clinically but also follow up patients and assess needs for healthcare and social services in a systematic manner, in order to provide adequate comprehensive care during the course of disease.

Limitations and strengths

The present study has some methodological limitations. Firstly, a cross-sectional study design prevents us from discussing any causal relationship between independent variables and NPCS scores representing levels of unmet needs. The results need to be further studied with a longitudinal study design and, ideally, in a larger population of HD patients.

Secondly, the response rate of 54.4% cases reduced power of our analyses, and some form of population bias cannot be excluded. Patients with reduced self-awareness may not be in contact with healthcare institutions and therefore may have fallen out of reach. If they did receive an invitation they may have declined or chose not to reply because they do not perceive themselves as being ill. Additionally, patients in late stages of the disease are highly dependent on their primary (family) carers who may not have had the time or energy to reply. Yet, considering the clinical picture of HD, the population in this study may be considered representative and our response rate can be considered satisfactory. A strength in the present study is that contrary to many studies (i.e., [29], patients in the advanced phase (stages IV and V) were not underrepresented. However, one of the most important limitations is that behavioral and cognitive domains were not addressed in this study due to the fact that the data were collected by a survey and not by clinical evaluation. Thus, the interpretation of the study results should be made with caution.

The NPCS was not validated for Norwegian language and circumstances. Yet, the instrument has shown good psychometric properties [37] and is originally developed for this type of patient populations. There are currently no other instruments available to assess needs for healthcare and social support services in a similar normative and systematic manner. In order to ensure obtaining best possible reliability of results obtained using the NPCS Scale we discussed each of the NPCS items regarding the interpretation for rating in the context of the Norwegian healthcare system and this particular patient group, in addition to a carefully executed translation process.

Implications and future directions

The results of the present study suggest that particular focus should be warranted for patients entering the middle phase of HD. Investigating factors associated with the level of unmet needs for healthcare and social support services further suggest that the patients’ education level may be of importance when surveying the needs for healthcare services in HD patients. Also, when discussing needs and provision of services, an informant closely related to the patient should ideally be present. Further studies including a larger population and longitudinal study design should be performed in order to verify the results of the present study and to shed further light on the predictive factors for level of unmet needs. Of particular interest are the needs for social and family support [20]. Moreover, closer investigating the potential influence of cognitive and psychiatric symptoms and self-awareness on levels of unmet needs for healthcare and social support services deserves further research.

Conclusions

This study indicates unmet needs for health and personal care and social support services among HD patients and across all five disease stages. However, the most substantial gaps in healthcare and social support services were identified in the middle phase (disease stage III) of HD (in terms of both proportion of patients and level of unmet needs). One important message to clinicians is to not only evaluate the patients clinically, but also follow up patients and assess needs for healthcare and social services in a systematic manner, in order to provide adequate comprehensive care during the course of disease.

Additional file

Additional file 1: The Needs and Provision Complexity Scale. (PDF 102 kb)

Competing interests

The authors declare that they have no competing interests.
Authors’ contributions
MRvW, EIH, RJ, JCF and NA were involved in study concept and design and acquisition of data. MRvW, EIH, JCF and NA analyzed and interpreted the data, in addition to drafting the manuscript. All authors have read and approved the final version of the manuscript.

Acknowledgements
We thank all patients, their family members and healthcare professionals participating in the study. We further thank Nancy Borgerød, Gunvor Ruud and Ragnhild Wehus for their assistance with the recruitment procedure.

Author details
1Centre for Habilitation and Rehabilitation Models and Services (CHARM), Institute for Health and Society, University of Oslo, P.O. Box 1130, Blindern 0318 Oslo, Norway. 2Department of Neurohabilitation, Oslo University Hospital, P.O. Box 4950, Nydalen 0424 Oslo, Norway. 3Department of Physical Medicine and Rehabilitation, Oslo University Hospital, P.O. Box 4950, Nydalen 0424 Oslo, Norway. 4Centre for Rare Disorders, Oslo University Hospital, Rikshospitalet, P.O. Box 4950, Nydalen 0424 Oslo, Norway. 5Department of Health Management and Health Economics, University of Oslo, P.O. Box 1130, Blindern 0318Oslo, Norway. *Department of Neurology, Oslo University Hospital, P.O. Box 4950, Nydalen 0424Oslo, Norway.

Received: 27 May 2015 Accepted: 18 August 2015
Published online: 28 September 2015

References
PAPER II
Health-related quality of life and unmet healthcare needs in Huntington’s disease

Authors: Marleen R. van Walsem, Emilie I. Howe, Gunvor A. Ruud, Jan C. Frich, Nada Andelic.

Authors’ email addresses:
r.m.v.walsem@medisin.uio.no
emilie.howe@medisin.uio.no
guruud@ous-hf.no
jan.frich@medisin.uio.no
nadand@ous-hf.no

(*Corresponding author)

1 Centre for Habilitation and Rehabilitation Models and Services (CHARM), Institute for Health and Society, University of Oslo, P.O. Box 1130 Blindern, 0318 Oslo, Norway

2 Department of Neurohabilitation, Oslo University Hospital, P.O. Box 4950 Nydalen, 0424 Oslo, Norway

3 Department of Physical Medicine and Rehabilitation, Oslo University Hospital, P.O. Box 4950 Nydalen, 0424 Oslo, Norway

4Centre for Rare Disorders, Oslo University Hospital, Rikshospitalet, P.O. Box 4950 Nydalen, 0424 Oslo, Norway

5 Institute of Health and Society, University of Oslo, P.O. Box 1130 Blindern, 0318 Oslo, Norway

6Department of Neurology, Oslo University Hospital, P.O. Box 4950 Nydalen, 0424 Oslo, Norway
Abstract

**Background:** Huntington’s disease (HD) is a rare neurodegenerative disorder with a prevalence of 6 per 100,000. Despite increasing research activity on HD, evidence on healthcare utilization, patients’ needs for healthcare services and Health-Related Quality of Life (HRQoL) is still sparse. The present study describes HRQoL in a Norwegian cohort of HD patients, and assesses associations between unmet healthcare and social support service needs and HRQoL.

**Methods:** In this cross-sectional population-based study, 84 patients with a clinical diagnosis of HD living in the South-East of Norway completed the HRQoL questionnaire EuroQol, EQ-5D-3L. Unmet needs for healthcare and social support services were assessed by the Needs and Provision Complexity Scale (NPCS). Furthermore, functional ability was determined using the Unified Huntington’s Disease Rating Scale (UHDRS) Functional assessment scales. Socio-demographics (age, gender, marital status, occupation, residence, housing situation) and clinical characteristics (disease duration, total functional capacity, comorbidity) were also recorded. Descriptive statistics were used to describe the patients’ HRQoL. Regression analyses were conducted in order to investigate the relationship between unmet healthcare needs and self-reported HRQoL.

**Results:** The patients were divided across five disease stages as follows: Stage I: n = 12 (14 %), Stage II: n = 22 (27 %), Stage III: n = 19 (23 %), Stage IV: n = 14 (16 %), and Stage V: n = 17 (20 %). Overall HRQoL was lowest in patients with advanced disease (Stages IV and V), while patients in the middle phase (Stage III) showed the most varied health profile for the five EQ-5D-3L dimensions. The regression model including level of unmet needs, clinical characteristics and demographics (age and education) accounted for 42 % of variance in HRQoL. A higher level of unmet needs was associated with lower HRQoL (β value - 0.228; p = 0.018) whereas a better total functional capacity corresponded to higher HRQoL (β value 0.564; p < 0.001).

**Conclusions:** The study findings suggest that patients with HD do not receive healthcare services that could have a positive impact on their HRQoL.
Key words: Huntington’s disease, Health-related Quality of Life, Healthcare needs, Healthcare services, EQ-5D, NPCS
Background

Huntington’s disease (HD) is a rare neurodegenerative disorder with a prevalence of 6 per 100,000 in European, North American and Australian populations [1]. This chronic and complex disease is characterized by a triad of symptoms, including motor impairment, decline in cognitive function, and psychiatric disturbances. Symptoms develop gradually and result in progressive functional decline, and a complex and continuously changing clinical picture [2]. Although a clinical diagnosis of HD is based on the presence of undisputable motor symptoms, psychiatric symptoms and changes in cognitive function may precede clinical diagnosis by several years [3-6]. Most patients receive clinical diagnosis in mid adult life (between 30 – 50 years of age), with an estimated disease duration of 17 – 20 years [7].

Clinical care of patients with HD is focused on disease management, alleviating symptoms and maintaining functional ability and health-related quality of life (HRQoL) [7]. Treatment requires multidisciplinary, comprehensive care from several groups of healthcare professionals, and may include both pharmacological and non-pharmacological interventions, as curative treatment currently does not exist [8-11]. HRQoL has emerged as an increasingly important patient and clinician reported outcome measure alongside other endpoints of symptom ratings of HD [12, 13]. Furthermore, the establishment of several large observational studies during the last 5 to 10 years, such as REGISTRY and PREDICT-HD [14, 15] has led to rapidly increasing research activity on HRQoL in HD. HRQoL is a multidimensional concept reflecting impact of disease and/or its treatment on an individuals’ physical, emotional and social well-being [16]. Research has found lower HRQoL in patients with clinical HD compared to HD premotor manifest patients, persons at risk for HD and their partners [17]. Furthermore, studies have aimed at identifying which disease related factors that most strongly correlate with HRQoL in patients with HD [10, 11, 17, 18]. Two studies found that functional ability and depression were strongly associated with a decline in HRQoL in HD patients [11,
17, 18]. Read et al. 2013 found that neuropsychiatric symptoms and cognitive impairment had the strongest negative relationship with HRQoL [17]. One study identified depression and cognitive impairment as the strongest determinants of HRQoL [10]. A reason for the slight differences in findings may be due to the studies comprising of different populations of HD (i.e. only HD patients with early HD).

In recent years, there has been an increased focus on the association between unmet health care needs and HRQoL in health services research [19]. Despite general knowledge of the need for multidisciplinary and comprehensive healthcare for patients with HD, there is a lack of research investigating the relationship between healthcare service delivery (or lack thereof) and HRQoL. One previous study has shown that HD patients have a considerable level of unmet needs for healthcare and social support, indicating that many patients do not receive the comprehensive care they need [20]. To the authors’ knowledge, only one study that included patients with rare long-term neurological conditions, including HD, has investigated both HRQoL and healthcare services, more specifically access to supportive health and social care. In addition to providing support to previous studies, indicating reduced HRQoL in HD patients compared to the general population and other diseases, the study suggested that patients with rare complex neurological disorders do not use health and social care services that could have a positive effect on their HRQoL [21]. No studies specifically investigating the potential association between gaps in healthcare service needs and provision and HRQoL in HD have been performed.

Thus, the aims of this study are:

- to describe the health status (HRQoL) in a Norwegian cohort of patients with HD.

- to assess the association between unmet needs for healthcare and social support services and HRQoL.
We expected to find a higher level of unmet needs for health care services to be associated with lower HRQoL.

**Methods**

*Participants and participant recruitment*

Patients with a clinical diagnosis of HD living in the South-Eastern region of Norway (population 2.7 million inhabitants) were invited to participate in a survey. Eligible patients were identified and recruited through the regional academic medical center, Oslo University Hospital, through the Department of Neurology, Department of Neurohabilitation, Department of Medical Genetics and through the national advisory service for HD, the Centre for Rare Disorders. Additionally, Vikersund Rehabilitation Centre, offering a rehabilitation program for patients with HD, provided invitations to additional patients. Furthermore, we collaborated with a Norwegian professional network for community care in HD (Fagnettverk Huntington) and the Norwegian HD lay association (Landsforeningen for Huntingtons sykdom), in order to attempt reaching as many eligible patients as possible. We identified 158 eligible patients (corresponding to a prevalence of 5.9/100.000). A written invitation enclosing study information and an informed consent form was sent to these patients. Informed consent was obtained from 88 of the invited patients. Two patients were excluded after careful review of the patients’ medical records by a medical expert (JF), as we questioned if the patients had sufficient clinical symptoms to formally have a clinical diagnosis at the time of inclusion.

*Ethics*
The study was approved by the Regional Ethical Committee (ref. 2013/2089). Informed consent was obtained for all patients prior to inclusion in the study. Consent was obtained from the primary caregiver or legal representative for patients who were unable to give informed consent themselves.

Data collection

Data were collected during study visits either as outpatient study visits (39 %) or in patients’ homes (61%). Appointments for study visits were made by contacting the patient/carer upon receiving the informed consent form. Socio-demographic, clinical and disease specific data were collected and a clinical rating and needs assessment was performed as part of a survey interview with the patient and/or primary carer. Data collection and assessments were conducted by the same two experienced clinical raters (MRvW & EIH). Additional information from the patients’ medical records was used to estimate years of education and level of education (lower vs. higher) for four patients and occupational type (manual vs. non-manual) for three patients. We estimated disease duration (number of years with clinical diagnosis of HD) using clinical information that was available through the patients’ medical records for three patients, as we were unable to collect this information at the study visit. We were unable to obtain information concerning the number of CAG repeats in the HTT-gene for three patients. Next, patients were rated regarding their functional ability and their needs for healthcare and social services. At the end of the visit patients were asked to report their HRQoL by completing a generic questionnaire for HRQoL. If the patients were unable to independently fill in the questionnaire (i.e. due to motor impairment), their primary carer assisted them. They were explicitly informed that the questionnaire was a self-report measure aiming to reflect the patients rating of their health status. The carer assisted the patient in indicating their choice on the form in case of motor impairment, or aided the patient by reading or explaining the questions. For eight patients with advanced disease their primary carer completed the questionnaire on behalf of the patient. These primary carers were instructed to reflect the patients’ experienced health status and
HRQoL as well as possible. If they were unable to do so the questions were kept open and became missing values. All carers were either family members or health care personnel involved with patients on a daily basis. When questionnaires were not completed during the study visit, they were returned using a prepaid reply envelope. Patients, who had not returned the questionnaire by the end of the inclusion period, were followed up by telephone.

Measurements

The patients’ functional ability was evaluated with the three scales of the Unified Huntington’s Disease Rating Scale (UHDRS) - Functional assessment including the: a) Total Functional Capacity Scale (TFC) with a scoring range of 0-13, b) the Functional Assessment scale (FAS), a checklist for daily living, range 0-25 and c) the Independence Scale (IS), indicating the level of independence in %, scoring range 0-100. The TFC is used to classify HD patients into five functional disease stages: Stage I corresponds to a TFC score of 11-13, Stage II to a TFC score of 7-10, Stage III to a TFC score of 3-6, Stage IV to a TFC score of 1-2 and Stage V to a TFC score of 0. Higher scores on these scales indicate higher functioning, corresponding to higher independence [22].

In order to rate the level of unmet needs for healthcare and social services, we used the Needs and Provision Complexity Scale (NPCS), clinician version [23]. This tool is recently developed in the UK in order to identify healthcare and social support needs among patients with long term neurological conditions. It measures the patients’ needs for healthcare and support services, Part A (NPCS-Needs) and to which extend these needs are met through service provision Part B (NPCS-GETS). In the clinician version, needs (Part A) are assessed in a systematic and normative way by the clinician, and part B is systematically recorded by the clinician based on information provided by the patient and/or carer. The measure includes 15 items with a total scoring range of 0-50 covering low and high levels of needs. The items are divided over 6 sub-scales representing two domains: a) Health and Personal care needs and b) Social and support needs, both having a score range of 0-25. The NPCS
can be used at the population level to identify gaps in health service provision. At the individual level the NPCS can be used to monitor the changing needs and provisions of patients along the care pathway over time. The scale has shown good psychometric qualities and has been translated to Norwegian [20, 24].

We used the EQ-5D-3L questionnaire to measure HRQoL. This is a generic self-report measure developed by the EuroQol Group [25]. The measure is used in several health conditions, including HD [26-29]. The scale consists of two parts. The first part comprises five single item dimensions of health: mobility, self-care, usual activities, pain/discomfort and anxiety/depression, which can be rated on three levels of severity representing no problem (1), slight problem (2), major problem (3). The level scores for these five dimensions can be presented in health profiles as well as global health indices with a weighted total value for HRQoL. Part two is a Visual Analogue Scale (VAS) ranging from 0 (worst health-state) to 100 (best health state), and is often used as a general measure for HRQoL. The EQ-5D 3L has been found valid to use in the Norwegian population [30]. For the purpose of our study we use the level scores in order to describe health profiles for the five disease stages and the VAS scores as an overall measure of perceived HRQoL.

Statistical analyses

Descriptive statistics of mean values and standard deviation (SD) (normally distributed variables) and median values with interquartile range (IQR) (non-normally distributed variables) were calculated for socio-demographic and clinical sample characteristics and the total level of unmet needs and VAS-score (HRQoL) for the complete sample and each of the five disease stages. Overall group differences between disease stages were computed using one-way ANOVA (normally distributed continuous variables) and Kruskal-Wallis for K-samples (non-normally distributed variables). Group differences for nominal variables were calculated using cross-tabulation Chi-square tests. NPCS total levels of unmet needs were calculated as the discrepancy between the total level of Needs and Gets: NPCS
Needs score – NPCS Gets score = NPCS Unmet needs score. In order to describe the health-status for the complete sample and each of the five disease stages, n and % with level scores of 1, 2 and 3 for each of the five dimensions were calculated, resulting in a health-status profile.

Regression analyses were performed in order to investigate the relationship between the total level of unmet needs and the patients’ overall self-reported HRQoL. Data were inspected for violation of assumptions, which resulted in logarithmical transformation of the scores for total level of unmet needs and disease duration. The relationship between each of the independent variables (level of unmet needs and socio-demographic and clinical variables) and the dependent variable HRQoL was investigated using simple linear regression. The independent variable Informant was collapsed to a dichotomous variable grouping patient alone (N = 27) vs. patient with informant or informant only (N = 57). Four disease-related variables (disease duration, TFC, informant and housing situation) and the independent variable of interest, total level of unmet needs reached levels of significance. All variables, with exception of housing situation due to the high correlation with TFC score, were entered into the regression analyses as control variables. Additionally, despite lacking significance in simple regression, comorbidity and education as these were shown to be associated with levels of unmet needs [20], and age known to influence HRQoL in the normal population, were entered in regression analyses as control variables. Multiple regression analyses were performed using a hierarchical approach (a block-wise analysis). Independent variables were entered in three blocks divided according to the variable of interest, total level of unmet needs for healthcare and social support services (block 1), clinical and disease related variables, including disease duration, TFC score, informant, and comorbidity (block 2) and socio-demographic variables, age and education (block 3).

Results are presented in Adjusted \( R^2 \) and \( R^2 \) Change, and standardized Beta (\( \beta \)) values with confidence intervals. The direction of Beta value was expected negative for total level of unmet needs indicating lower levels of unmet needs corresponding to higher HRQoL. Prior to carrying out the multiple regression analyses, possible multicollinearity between independent variables was investigated using variance inflation factor (VIF). We examined influential data points using Cook’s distance. Variables
with correlation coefficients > .70 were not included in the analyses. Residual analyses were performed and no outliers on any of the variables included in the analyses were identified. Levels of significance were set at \( p = 0.05 \) and all statistical tests were two sided. Statistical analyses were performed using SPSS version 21.0; SPSS Inc. Chicago IL.

Results

Participants’ socio-demographic and clinical characteristics

84 out of the 86 participants (97.7%) (53.2% of the 158 eligible patients) included in the survey study filled out the EQ-5D-3L questionnaires, and were included in the data-analyses. The mean age was 56.7 (SD 11.4) years. The patients were divided across five disease stages as follows: Stage I: \( n = 12 \) (14 %), Stage II: \( n = 22 \) (27 %), Stage III: \( n = 19 \) (23 %), Stage IV: \( n = 14 \) (16 %), and Stage V: \( n = 17 \) (20 %). Socio-demographic characteristics for the complete sample and for each disease stage are presented in Table 1.
### Table 1. Socio-demographic statistics for total sample and divided across disease stages

<table>
<thead>
<tr>
<th>Variables</th>
<th>Categories</th>
<th>Complete sample (N=84)</th>
<th>Stage I (n=12)</th>
<th>Stage II (n=22)</th>
<th>Stage III (n=19)</th>
<th>Stage IV (n=14)</th>
<th>Stage V (n=17)</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age*</td>
<td></td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>0.084</td>
</tr>
<tr>
<td>Education (years)*</td>
<td></td>
<td>56.7 (11.4)</td>
<td>49.8 (9.5)</td>
<td>54.6 (12.9)</td>
<td>58.9 (11.1)</td>
<td>61.1 (11.5)</td>
<td>57.8 (9.0)</td>
<td></td>
</tr>
</tbody>
</table>

| Gender                         | Female                            | 37 (44)                | 5 (42)         | 8 (36.4)       | 7 (37)         | 7 (50)         | 10 (59)        | 0.616   |
|                                | Male                              | 47 (56)                | 7 (58)         | 14 (63.6)      | 12 (63)        | 7 (50)         | 7 (41)         |         |
| Education                      | Lower (≤ 12 years)                | 51 (60.7)              | 5 (42)         | 11 (50)        | 15 (79)        | 9 (64.3)       | 11 (65)        | 0.221   |
|                                | Higher (>12 years)                | 33 (39.3)              | 7 (58)         | 11 (50)        | 4 (21)         | 5 (45.7)       | 6 (35)         |         |
| Civil status                   | Single                            | 36 (42.9)              | 4 (33)         | 7 (31.8)       | 9 (47)         | 8 (57.1)       | 8 (47)         | 0.560   |
|                                | Married                           | 48 (57.1)              | 8 (67)         | 15 (68.2)      | 10 (53)        | 6 (42.9)       | 9 (53)         |         |
| Occupation*                    | Manual                            | 40 (47.6)              | 5 (42)         | 9 (40.1)       | 12 (63)        | 6 (46.1)       | 8 (47)         | 0.643   |
|                                | Non-manual                        | 41 (48.8)              | 7 (58)         | 13 (59.1)      | 7 (37)         | 7 (53.8)       | 7 (41)         |         |
| Occupational status            | Employed                          | 14 (16.7)              | 11 (92)        | 3 (13.6)       | 0 (0)          | 0 (0)          | 0 (0)          | 0.000   |
|                                | Unemployed                        | 70 (83.3)              | 1 (8)          | 19 (86.4)      | 19 (100)       | 14 (100)       | 17 (100)       |         |
| Housing situation              | Living at home                    | 52 (61.9)              | 12 (100)       | 22 (100)       | 13 (68)        | 5 (35.7)       | 0 (0)          | 0.000   |
|                                | Not living at home                | 32 (38.1)              | 0 (0)          | 0 (0)          | 6 (32)         | 9 (64.3)       | 17 (100)       |         |
| Residence                      | Rural                             | 12 (14.3)              | 1 (8)          | 3 (13.6)       | 2 (10.5)       | 3 (21.4)       | 3 (18)         | 0.859   |
|                                | Urban                             | 72 (85.7)              | 11 (92)        | 19 (86.4)      | 17 (89.5)      | 11 (78.6)      | 14 (82)        |         |
| Informant                      | Patient                           | 27 (32.1)              | 9 (75)         | 14 (63.6)      | 4 (21)         | 0 (0)          | 0 (0)          | 0.000   |
|                                | Patient & informant               | 49 (58.3)              | 3 (25)         | 8 (36.4)       | 15 (79)        | 14 (100)       | 17 (100)       |         |
|                                | Informant only                    | 8 (9.6)                | 0 (0)          | 0 (0)          | 0 (0)          | 0 (0)          | 0 (0)          |         |

SD: Standard deviation; # 3 responses missing (1 in Stage IV and 2 in stage V); * using ANOVA, all other variables Chi-square.
Overall group differences across disease stages were significant for all clinical characteristics (p < 0.001) except for comorbid conditions (p = 0.143) (having no comorbid conditions vs. having comorbid conditions) (see Table 2). As expected, patients in advanced disease stages have longer disease duration compared to patients in the early disease stages, while FAS total scores and IS scores declined from disease stage I to V, with scores in stage I being close to normal (FAS median (IQR) = 24 (2) & IS mean (SD) = 95.8 (± 5.1)) and very affected in stage V (FAS median (IQR) = 0 (2) & IS mean (SD) = 20.9 (± 5.7)).

A significant group difference was found for total level of unmet needs for healthcare and social support services (p = 0.013). The level of unmet needs peaked in stage III (median= 8 (IQR = 7)). The levels of unmet needs for patients in stages I and II were lowest (median values and IQR were 3 (4) and 5 (4) respectively), while levels of unmet needs in stage IV and V were constant and higher than stages I and II, but lower than stage III, with median values and IQR of 6.5 (10) and 6 (9), respectively. Median scores and interquartile ranges for Needs (Part A), Gets (Part B) and Unmet needs for total unmet needs, and for the two domain scores “health and personal care”, and “social care and support” are also reported in table 2.
Table 2. Clinical characteristics and NPCS scores for the total sample and all disease stages

<table>
<thead>
<tr>
<th>Variables</th>
<th>Complete sample (N =84)</th>
<th>Stage I (n=12)</th>
<th>Stage II (n=22)</th>
<th>Stage III (n=19)</th>
<th>Stage IV (n=14)</th>
<th>Stage V (n=17)</th>
<th>Sign.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disease duration</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>6 (7)</td>
<td>2 (2)</td>
<td>5 (6)</td>
<td>7 (5)</td>
<td>8 (7)</td>
<td>11 (7)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Total FAS score</td>
<td>15 (17)</td>
<td>24 (2)</td>
<td>20 (2)</td>
<td>15 (4)</td>
<td>5 (3)</td>
<td>0 (2)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td>Independence score*</td>
<td>60 (26.5)</td>
<td>95.8 (5.1)</td>
<td>79.1 (2.9)</td>
<td>64.7 (6.3)</td>
<td>40.4 (10.8)</td>
<td>20.9 (5.7)</td>
<td>P &lt; 0.001</td>
</tr>
<tr>
<td></td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>P-value (2-sided)</td>
</tr>
<tr>
<td>Comorbid conditions</td>
<td>No(ne)</td>
<td>48 (57.1)</td>
<td>7 (58)</td>
<td>9 (41)</td>
<td>10 (71)</td>
<td>13 (76)</td>
<td>0.143</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
<td>36 (42.9)</td>
<td>5 (42)</td>
<td>13 (59)</td>
<td>4 (29)</td>
<td>4 (24)</td>
<td></td>
</tr>
<tr>
<td>NPCS variables</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td>Median (IQR)</td>
<td></td>
</tr>
<tr>
<td>Total score*</td>
<td>Needs</td>
<td>21 (17)</td>
<td>7.5 (6)</td>
<td>11 (6)</td>
<td>23 (12)</td>
<td>29 (7)</td>
<td>28 (10)</td>
</tr>
<tr>
<td></td>
<td>Gets</td>
<td>13 (16)</td>
<td>3 (3)</td>
<td>6.5 (8)</td>
<td>13.5 (12)</td>
<td>21 (7)</td>
<td>21 (10)</td>
</tr>
<tr>
<td></td>
<td>Unmet needs</td>
<td>6 (6)</td>
<td>3 (4)</td>
<td>5 (4)</td>
<td>8 (7)</td>
<td>6.5 (10)</td>
<td>6 (9)</td>
</tr>
<tr>
<td>Domain score Health and personal care</td>
<td>Needs</td>
<td>11 (7)</td>
<td>6 (5)</td>
<td>7.5 (5)</td>
<td>11.5 (5)</td>
<td>13.5 (5)</td>
<td>14 (3)</td>
</tr>
<tr>
<td></td>
<td>Gets</td>
<td>8 (6)</td>
<td>2.5 (2)</td>
<td>5 (6)</td>
<td>6.5 (6)</td>
<td>9 (4)</td>
<td>9 (7)</td>
</tr>
<tr>
<td></td>
<td>Unmet Needs</td>
<td>3 (4)</td>
<td>2 (3)</td>
<td>2.5 (3)</td>
<td>5 (4)</td>
<td>4 (5)</td>
<td>4 (6)</td>
</tr>
<tr>
<td>Domain score Sosial care and support**</td>
<td>Needs</td>
<td>10 (10)</td>
<td>2 (3)</td>
<td>4 (2)</td>
<td>11 (7)</td>
<td>13.5 (8)</td>
<td>14 (2)</td>
</tr>
<tr>
<td></td>
<td>Gets</td>
<td>5 (10)</td>
<td>0 (1)</td>
<td>1.5 (2)</td>
<td>5.5 (6)</td>
<td>11.5 (4)</td>
<td>11 (3)</td>
</tr>
<tr>
<td></td>
<td>Unmet needs</td>
<td>2 (3)</td>
<td>1 (3)</td>
<td>2 (2)</td>
<td>2.5 (5)</td>
<td>2 (6)</td>
<td>2 (4)</td>
</tr>
</tbody>
</table>

FAS: Functional Assessment Scale; IQR: Interquartile range; *normally distributed: reported mean (sd) and Anova; * one missing from stage III; ** one missing from stage III; Needs = NPCS Part A; Gets = NPCS Part B
HRQoL of the total sample and across disease stages

Figure 1 shows bar graphs of the health profiles for the five disease stages, illustrated by the level scores for each of the five EQ-5D dimensions. In general, the health profile is most wide-ranging for patients in stage III, showing level scores across the whole range (1 (no problems) to 3 (major problems)) for each of the five dimensions except for mobility. Approximately two thirds of the total sample report slight or major problems (n = 53 (65.4%)) for mobility. Major problems are all reported in advanced disease (n = 1 (7%) in Stage IV and n= 8 (47%) in Stage V). For the dimension self-care, more than half of all patients report slight or major problems (n = 45 (55.6%)). Major problems are mainly reported in advanced disease (Stage IV: n = 11 (78.6%), Stage V: n = 16 (94.1%), except for 1 patient in stage III. Three quarters of the patients have slight or major problems for usual activity (n = 62 (74.6%)). Problems with usual activity increase across disease stages with a peak in stage III (n = 15 (79%)), and are reported by all patients in stage IV and V. Approximately half of all patients (n = 41 (51.2%)) experience pain/discomfort, but only three patients, one in each disease stage III, IV and V, report major pain/discomfort. A total of 56 (68.2%) patients report slight or major problems with depression/anxiety. In stage I most patients report no problems (n = 9 (75%), while half or more report slight problems in all other stages (Stage II: n = 16 (73%), Stage III: n = 10 (53%), Stage IV (n = 7 (47%) and Stage V (n=11 (65%)).

Insert Figure 1. Bar-graphs showing health profiles of HD patients in Stage I to Stage V.

Overall self-reported HRQoL measured by EQ-5D VAS (N = 82), shows an average score of 52.1 (SD = 26.1), and decreases across disease stages with mean and SD values of 83 and 16.4 for stage I, 57.9 and 20.3 for stage II and 49.3 and 23.5 for stage III, and stable scores for Stage IV and V (N = 15) (mean (SD) values 35 (25.5) & 38.3 (20.9) respectively).
The relationship between unmet needs and HRQoL

Figure 2 shows the decline in HRQoL measured by EQ-5D VAS score for the five disease stages for patients with unmet needs for healthcare and social support services (N = 73), divided into patient with a low level (median score for total unmet needs 1 – 6) and a high level (median score for total unmet needs > 6) of unmet needs. Overall, patients with a high level of unmet needs have lower HRQoL.

Insert Figure 2. Bar-graph of average HRQoL scores across disease-stage for low vs. high levels of unmet needs.

Results of the hierarchical regression analysis investigating the association between level of unmet needs for healthcare and social support services and HRQoL are presented in table 3. The collinearity diagnostics suggested an acceptable degree of collinearity (VIF 1.1 – 2.6, and < 5). Cooks’ distance (D max = 0.205) indicated that no single case in the data induced undue influence on the model. The level of unmet needs was entered at the first step with statistically significant effect, explaining 9% of the total variance in HRQL. When the clinical disease characteristics (disease duration, TFC score, comorbidity and informant variable) were entered into the second step, the amount of explained variance improved for 30%. Only TFC showed significant effect. The model was controlled for demographics (age and education) which explained only 2% of variance. The β value for the level of unmet needs was negative (β = 0.228; p = 0.018) indicating that a higher level of unmet needs was associated with lower HRQoL whereas β value of TFC score (β = 0.564; p < 0.001), was positive indicating that a better functional ability corresponded to higher HRQoL. The model was validated by removing the eight patients for whom only a rating performed by informant was available from the analysis,
revealing no significant changes to the reported regression model, with level of unmet needs remaining significantly associated with HRQoL and no significant change in explained variance of the model.

Table 3. Hierarchical multiple regressions of the total level of unmet needs on self-reported HRQoL (N = 81).

<table>
<thead>
<tr>
<th>Step variables</th>
<th>Variables</th>
<th>$R^2$ Change</th>
<th>$\beta$</th>
<th>$\beta$ (95% CI)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Level of unmet needs†</td>
<td></td>
<td>0.092 (p = 0.006)</td>
<td>-0.228 (p = 0.018)</td>
<td>-9.711 - -0.933</td>
</tr>
<tr>
<td>2. Clinical characteristics</td>
<td>Disease duration</td>
<td>0.302 (p &lt; 0.001)</td>
<td>-0.196 (p = 0.105)</td>
<td>-12.634 – 1.222</td>
</tr>
<tr>
<td>Informant</td>
<td></td>
<td></td>
<td>0.116 (p = 0.352)</td>
<td>-7.320 – 20.3</td>
</tr>
<tr>
<td>Comorbidity</td>
<td></td>
<td></td>
<td>-0.152 (p = 0.114)</td>
<td>-17.99 – -1.959</td>
</tr>
<tr>
<td>TFC</td>
<td></td>
<td>0.564 (p &lt; 0.001)</td>
<td>1.677</td>
<td>5.133</td>
</tr>
<tr>
<td>3. Socio-demographic characteristics</td>
<td>Age</td>
<td>0.026 (p = 0.205)</td>
<td>0.116 (p=0.253)</td>
<td>-0.194 – 0.725</td>
</tr>
<tr>
<td>Education</td>
<td></td>
<td></td>
<td>-0.138 (p = 0.144)</td>
<td>-2.450 – 0.363</td>
</tr>
</tbody>
</table>

$R^2 = 0.42$; Adjusted $R^2 = 0.364$; †1 is missing NPCS total score

Discussion

This is the first study describing self-reported health status (HRQoL) of HD patients in all five disease stages. The lowest rated health status was found in moderate to advanced phases of HD (stages III-V), while patients in stage III showed the most wide-ranging health profile. The study also investigated the association between level of unmet needs for healthcare and social support services and HRQoL. The main results support the view that higher level of unmet needs is related to lower HRQoL. The findings are discussed in detail below.

The study results highlight the significant burden of HD on HRQoL. These findings are consistent with previous studies on HD [11, 17, 21, 26, 31]. Patients in advanced disease stages reported the most extensive reduction in HRQoL. However, already in the middle phase (stage III) patients rated their HRQoL below the average of the present sample and of the sample in Hocaoglu et al., 2012 [26].
Furthermore, the most wide-ranged health profile was found for these patients. Patients in this phase represent a largely heterogenic group, due to higher variation in symptom presentation and disease progression [20]. The phase can be considered a transitional phase, where patients transition from being relatively independent to becoming increasingly dependent in various areas of daily life; thus resulting in these patients experiencing considerable difficulties. However, some patients may report experiencing no problems at all, while other experience major problems with mobility, usual activities, self-care, pain and discomfort and anxiety and depression.

HRQoL is a multidimensional phenomenon influenced by several factors. The HRQoL regression model in this study covered data on the level of unmet health service needs, disease duration, comorbidity and functional ability, informant in the study, patient age and education, explaining 42% of the variance in HRQoL. A possible explanation why a large proportion of the variance remained unexplained is that we did not evaluate in depth the clinical status of patients, such as presence of cognitive impairment, psychiatric symptoms and motor impairment.

Some of the remaining explanatory factors may also be related to environmental conditions such as local health care organization, costs and quality of delivered care and socio-economic inequities among patients. In general, Norwegian universal public health care system is based on a principle of equal access for all citizens according to needs, and not according to wealth. However, a recently published Norwegian study on self-reported health care utilization in the general population found social inequalities in utilization of specialized health care services [32].

Evidence on health care utilization, patients’ needs for health care services and HRQoL in the HD population is sparse [20]. In a qualitative study of healthcare experiences of families affected by HD from Canada, complex needs for healthcare services and emotional support were found. Participants expressed frustration at the lack of knowledge about HD displayed by their family physicians [33]. Several suggestions to improve the quality of care to their families, including better education of
healthcare professionals regarding the complex nature of HD and the provision of regular follow-up support, were offered.

The strong association between functional capacity and HRQoL found in this study is consistent with previous studies on HD and HRQoL [11, 18]. Furthermore, this study found that higher level of unmet needs for healthcare and social services were related to lower self-rated health status (HRQoL).

Information about such needs in the general Norwegian population or in individuals with disabilities does not exist. However, it is worth mentioning that a study from Canada, where there is a health care system with a similar accessibility to the Norwegian system, found that adults (aged 20-64 years) with physical, sensory and cognitive disabilities reported more than three times as many unmet health care needs as their non-disabled counterparts [34].

The study results are also supported by studies on unmet healthcare and social services needs and HRQoL in other patient groups [35-40]. Investigating unmet health needs in patients with coronary heart disease results showed associations between physical and social needs and HRQoL [35].

Research within the field of cancer, revealed that in adolescents and young adults with cancer unmet support service needs were associated with lower overall HRQoL [36]. Also, cancer survivors with unmet supportive care needs in the physical, psychological and patient care domains were shown to have poorer HRQoL [37].

Trying to identify factors related to quality of life in people with severe mental illness, researchers found the strongest predictors to be unmet basic, social and functioning needs [38]. A study on patients with dementia in residential care uncovered that sensory and physical disability needs, mental health and social needs were often unmet [39].

Taken together, the studies highlight that identifying healthcare needs is a vital part of providing comprehensive healthcare due to impact on HRQoL.

This study has limitations that should be addressed. A cross-sectional study design prevents us from discussing any causal relationship between independent variables and HRQoL. Future studies with a larger sample and longitudinal design need to be carried out in order to further tease apart the
associations and predictors of HRQoL including disease specific aspects and level of HRQoL. Several studies on HD have used the SF-36 when investigating associations between disease specific symptoms and HRQoL. The SF-36 has demonstrated good validity in the context of HD [13]. Our study specifically focused on including HD patients in moderate to advanced disease stages, often underrepresented in HD research. The EQ-5D VAS score for HRQoL includes less complicated questions and applying a VAS Scale can be considered easier to administrate to advanced patients, compared to SF-36. While being a self-reported measure of HRQoL, eight EQ-5D questionnaires for advanced HD patients were completed on behalf of the patient by the primary family caregiver or professional carer, as the patients were not able to fill out the questionnaires themselves. Hocaoglu et al. 2012 found good correlations between proxy and patient ratings of HRQoL in HD, but was lower for middle stages of HD compared to early and advanced stages on their HD-HRQoL measure [26]. If a patient needed assistance from their primary carer to fill out the form, the carer was specifically instructed to assist in order to ensure best reflection of the patient’s rating. Further, our model did not change when we analyzed the data without the eight patients for whom only a rating by informant was obtained. We used a generic measure, which is less complex and well correlated with HD-HRQoL measures in a validation study [29]. Although we acknowledge our study’s limitations, we included a relatively large group of patients in the middle to advanced stages of HD: thus comprising a relatively representative sample of the HD population, covering the whole spectrum of disease.

Conclusions

The present findings of the association between level of unmet needs for healthcare and social support services and overall HRQoL might have potential benefits for clinical practice where
comprehensive care is targeted. In order to improve functioning and HRQoL of patients with HD, it is important that clinicians assess, record and monitor healthcare and social support service needs, as well as follow up that needs are met. Building partnerships with family caregivers may improve exchange of information and facilitate tailored health care delivery [41]. Our findings underline the importance of continuity of care through the whole disease spectrum (early to advanced HD) acknowledging the complex and changing nature of this disease [8, 9, 42].

**Abbreviations**

**HD:** Huntington’s Disease; **HRQoL:** Health-related Quality of Life; **UHDRS:** Unified Huntington’s Disease Rating Scale; **TFC:** Total Functional Capacity; **FAS:** Functional Assessment Scale; **IS:** Independence Scale; **NPCS:** Needs and Provision Complexity Scale; **VAS:** Visual Analogue Scale; **SD:** Standard deviation; **IQR:** Interquartile Range; **CI:** Confidence Interval

**Declarations**

**Ethics approval and consent to participate**

The study was approved by the Regional Ethical Committee (ref. 2013/2089). Informed consent was obtained for all patients prior to inclusion in the study.

**Consent for publication**

Not applicable.
Availability of Data and Materials

Requests for data access may be sent to r.m.v.walsem@medisin.uio.no. Requests will be individually evaluated by the Oslo University Hospital and in accordance with the Norwegian personal data legislation act.

Competing interests

The authors declare that they have no competing interests.

Funding

The study was funded by the Norwegian Research Council, project number 209748.

Authors’ contributions

MRvW, EIH, GAR, JCF and NA were involved in study concept and design, as well as in the acquisition of data. MRvW, EIH, JCF and NA analyzed and interpreted the data, in addition to drafting the manuscript. All authors have read and approved the final version of the manuscript.

Acknowledgements

We thank the patients, their family members and their healthcare professionals for participating in the study. Additionally, we thank Nancy Borgerød, Ragnhild Wehus and Kristin Iversen for their assistance with the recruitment of participants.
References


Figure 1. Bar-graphs showing health profiles of HD patients in Stage I to Stage V.
Figure 2. Bar-graph of average HRQoL scores across disease-stage for low vs. high levels of unmet needs.
Research Report

Assistive Technology for Cognition and Health-related Quality of Life in Huntington’s Disease

Marleen R. van Walsema\textsuperscript{a,b,1,*}, Emilie I. Howe\textsuperscript{a,c,1}, Jan C. Frich\textsuperscript{d,e} and Nada Andelic\textsuperscript{a,c}

\textsuperscript{a}Centre for Habilitation and Rehabilitation Models and Services (CHARM), Institute of Health and Society, University of Oslo, Blindern, Oslo, Norway
\textsuperscript{b}Department of Neurohabilitation, Oslo University Hospital, Nydalen, Oslo, Norway
\textsuperscript{c}Department of Physical Medicine and Rehabilitation, Oslo University Hospital, Nydalen, Oslo, Norway
\textsuperscript{d}Institute of Health and Society, University of Oslo, Blindern, Oslo, Norway
\textsuperscript{e}Department of Neurology, Oslo University Hospital, Nydalen, Oslo, Norway

Abstract.

\textbf{Background:} Assistive technology for cognition (ATC) can be defined as external devices aimed at supporting cognitive function. Studies in neurological populations suggest that use of ATC is a promising strategy to ameliorate negative effects of cognitive impairment and improve Health-related Quality of Life (HRQoL). There is a lack of studies on the effects of ATC in HD.

\textbf{Objective:} This study aimed to describe the use of ATC in patients with HD, and to investigate the association between ATC and HRQoL.

\textbf{Methods:} A cross-sectional population-based study, including eighty-four patients with a clinical HD diagnosis (stages I–V). Socio-demographic and clinical data were collected, including information regarding various aspects of ATC use and an evaluation of cognitive impairment was performed. The Unified Huntington’s Disease Rating Scale (UHDRS) Total Functional Capacity scale (TFC) and the EQ-5D Visual Analogue Scale were used to evaluate functional ability and HRQoL. Descriptive analyses were conducted to describe ATC use and regression analyses to investigate associations between ATC and HRQoL.

\textbf{Results:} Thirty-seven percent of the patients had ATC, and ATC was used most frequently in stages I–III. Information about ATC, needs evaluation and training was provided to 44%, 32.1% and 20.2% respectively. The regression analysis showed a significant association between TFC and HRQoL ($\beta$ value = –0.564, $p$ = 0.001), but there was no association between ATC and HRQoL.

\textbf{Conclusions:} One-third of all patients used ATC, mainly those with mild to moderate cognitive impairment (stage I – III). No association between ATC and HRQoL was found. More research is needed to investigate effects of ATC in HD.

Keywords: Neurodegenerative diseases, huntington disease, self-help devices, cognition, quality of life

\textsuperscript{1}Authors contributed equally.
*Correspondence to: Marleen R. van Walsem, Centre for Habilitation and Rehabilitation Models and Services (CHARM), Institute of Health and Society, University of Oslo, P.O. Box 1130 Blindern, 0318 Oslo, Norway. Tel.: +47 22859237; Fax: +47 22850570; E-mail: r.m.v.walsem@medisin.uio.no.

INTRODUCTION

Cognitive impairment is one of the hallmark symptoms of Huntington’s disease (HD), an autosomal dominant hereditary neurodegenerative disease.
Subtle signs of cognitive decline appear more than a decade prior to clinical diagnosis and develop progressively [1–5]. Neuro-imaging studies have revealed alterations in brain function, structure and connectivity in individuals with pre-manifest and manifest HD, and has found relationships between neuro-imaging measures and poorer performance on cognitive tasks [3, 4, 6, 7]. While changes in cognition vary from individual to individual, cognitive impairments are usually most pronounced in the cognitive domains of psychomotor speed, executive functions and memory (specifically visuo-spatial memory), progressing until developing global cognitive impairment and dementia in advanced stages of HD [4, 8, 9]. Cognitive impairment in HD can have a detrimental effect on health-related quality of life (HRQoL) through affecting ability to work and partake in leisure activities, interpersonal relationships, and ability to maintain self-care [10–13]. Two studies on HD have identified impaired cognitive function as the strongest negative determinants of HRQoL [12, 14].

Despite extensive efforts, no known cure exists for HD at present and no pharmacological interventions have been shown to improve cognitive function in HD [15]. Patients suffering from HD are dependent on the provision of individually tailored multidisciplinary comprehensive healthcare across the disease spectrum [14, 16–18]. The broad established knowledge about cognitive impairment in relation to brain function and mechanisms that has emerged during the last decades, has contributed to an increased interest in non-invasive, non-pharmacological interventions that may have a positive effect on cognitive function [4, 19]. Studies conducted in patients with traumatic brain injury (TBI), Alzheimer’s disease and elderly with cognitive deficits, have shown that assistive technology for cognition (ATC) has the potential to support and maintain cognitive function and thereby improve functional ability and HRQoL [20–25]. Assistive technology for cognition (ATC) can be defined as an item that increases, maintains or improves functional capabilities for individuals whose cognitive changes limit their effective participation in daily activities [26]. ATC are external aids that can address disabilities in memory, executive functions such as planning, organization and attention, in addition to reduced psychomotor speed [26, 27]. Wilson et al. assessed a personalized electronic paging system as a method of reducing everyday problems in individuals with cognitive impairment (memory, attention, planning, and organizational problems) following TBI, stroke and other acquired progressive and non-progressive brain injuries. They found use of the paging system to be significantly associated with greater ability to carry out daily activities, such as self-care and keeping appointments [23]. Two studies conducted in the HD population investigated the use of talking mats as a way to support communication in nine patients. Ferm et al. examined the effect of talking mats in one-to-one communication in five patients and found that they were able to have more structured conversations compared to a control group with no communication aids [28]. Hallberg et al. investigated the use of talking mats in group conversations and found that patients had more effective conversations and asked more questions with the help of talking mats [29].

Despite broad knowledge regarding cognitive impairment in HD, and literature investigating ATC as a beneficial intervention to compensate negative effects of cognitive dysfunction in patients with other neurological conditions, the two previously mentioned studies are the only studies that have been conducted on ATC in HD [28, 29]. No studies systematically describing ATC have been conducted in HD. Moreover, to the authors’ knowledge, ATC as a way of alleviating cognitive impairment in HD has not been proposed as a potential cognitive intervention or as a part of comprehensive multidisciplinary care in literature on cognition in HD. Thus, there is a need for studies systematically describing the use of ATC and to investigate its potential as a non-pharmacological intervention in HD, by exploring the association between ATC and HRQoL. Otherwise we may discount an important non-pharmacological intervention for patients with HD.

The aims of the present study are to:

- Describe the use of ATC across the disease stages in a Norwegian cohort of HD patients.
- Investigate the association between ATC and HRQoL.

MATERIALS AND METHODS

Participants and recruitment procedure

A total of 158 patients with a clinical diagnosis of HD residing in the South-Eastern region of Norway (population of 2.7 million), equal to a prevalence of 5.9/100,000 inhabitants, were identified. These patients were invited to participate in a survey study on healthcare needs and utilization and quality of
life. Eighty eight patients gave their consent to participate and were included in the survey. Of those, two patients were excluded due to lack of clinical diagnosis of HD. Of the 86 patients, 2 did not return the EQ-5D-3L for Health-related Quality of Life (HRQoL) questionnaire, resulting in 84 patients included in the data-analyses (53.2% of the total number of patients invited to study participation) (see flow chart in Fig. 1 illustrating patient recruitment).

The identification of eligible patients was completed through the regional academic medical center, Oslo University Hospital, the Department of Neurohabilitation, Department of Neurology and the Department of Medical Genetics, and through the national advisory service for HD, the Centre for Rare Disorders. Furthermore, Vikersund Rehabilitation Centre, offering a rehabilitation program for HD patients, distributed invitations to additional patients. We also collaborated with the Norwegian Professional Network for Community Care in HD (Huntington fagnettverk) and the Norwegian HD lay association (Landsforeningen for Huntington sykdom), in a further attempt to reach as many patients as possible.

Approval for the study was obtained from the Regional Ethical Committee (ref. 2013/2089). All patients included in the study provided their informed consent. For patients unable to give informed consent themselves, consent was given by the primary caregiver or legal representative.

Procedures for data collection

Data was collected from January to August 2014 either as outpatient study visits (39%) or as study visits at the patients’ home (61%). The survey interviews and patient ratings were performed by two experienced clinical psychologists (MRvW and EIH). At the beginning of the visit information on socio-demographic and clinical characteristics was collected. For three patients we were unable to obtain information about the number of CAG repeats in the HTT-gene. Furthermore, for four patients we used supplementary information from the patients’ medical records in order to estimate educational level (low vs. higher) and for three patients we determined the occupational type (manual vs. non-manual). For three patients the disease duration (number of years with clinical diagnosis of HD) was estimated from information in patients’ medical records. As part of the clinical characteristics a clinical evaluation of cognitive impairment was conducted. Furthermore, patients were rated regarding their functional ability and asked to report their self-experienced health-related quality of life (HRQoL) by filling out a generic questionnaire. Primary carers, either family members or healthcare personnel involved with the patient on a daily basis assisted patients who were unable to complete the questionnaire themselves. The primary carer completed the questionnaire on behalf of eight patients with advanced disease. They were explicitly instructed to reflect the patients experienced health status and HRQoL to the best of their ability. When unable to do so the questions were kept open and became missing values. A prepaid reply envelope was used to return questionnaires, which were not filled out during the study visit.

Collecting information on assistive technologies for cognition (ATC)

For the specific purpose of the present study, information concerning ATC was collected. The existing literature on ATC (29, 31) suggests that individuals with cognitive impairment may start using external aids of their own in order to support impaired cognitive function, and include mainstream products such as cell phones, calendars, planner books, alarm clocks etc. We wanted to document the use of such devices and chose to define such items as informal ATC. Formal ATC was defined as items or software specifically designed to support patients with cognitive impairment acquired through a formal process of implementation. The information regarding ATC that was collected included the following: a) whether patients had ATC and whether these were formal or informal b) whether they used ATC, c) whether they
had been informed about the possibility of receiving ATC, d) whether they had undergone a formal needs assessment for ATC, and e) whether they had received training in the use of the ATC. This information was recorded as part of the survey interview at the beginning of the study visits.

Clinical evaluation of cognitive impairment

A clinical evaluation of the patients’ cognitive status was performed. This evaluation was based on information concerning cognitive function and symptoms obtained from the patient and information from their relative and/or professional caregiver, in addition to the clinicians’ observations during the evaluation. The clinical evaluation of cognitive impairment included the following categories: 1) mild cognitive impairment, defined as a slight reduction in one or more cognitive domains causing changes in or impaired ability to perform daily activities and the need for minor adjustments in order to be able to perform everyday activities, with next-of-kin starting to notice changes in the patient, 2) moderate cognitive impairment, defined as overt cognitive impairment in more than one cognitive domain as compared to premorbid function, with clear need for support/adjustments in order to continue carrying out daily activities and no longer being able to perform complex tasks, evident to next of kin, 3) severe cognitive impairment, defined as severe cognitive dysfunction in all domains, impaired communication, no longer being able to carry out daily activities or maintain self-care and in need of around the clock care, 4) Unable to evaluate, defined as cases where the raters were in doubt of the patients cognitive function due to lack of comparative information from next-of-kin or primary healthcare professional.

Measurements

As a measure of functional ability, we used the Unified Huntington’s Disease Rating Scale (UHDRS) – Functional Assessment, including the Total Functional Capacity scale (TFC) (scoring range of 0 – 13), the Functional Assessment scale (FAS) rating ability to perform activities of daily living (scoring range 0–25), and the Independence Scale (IS), indicating the level of independence (scoring range 0–100). The TFC was used to classify the patients in five functional stages of HD: Stage I corresponds to a TFC score of 11–13, Stage II to a TFC score of 7–10, Stage III to a TFC score of 3–6, Stage IV to a TFC score of 1-2 and Stage V to a TFC score of 0. Higher scores on these scales indicate better functioning.

In order to measure self-perceived overall HRQoL, we used the Visual Analogue Scale (VAS) of the EQ-5D-3L. The EQ-5D-3L is a generic measure developed by the Euro-Qol Group [30]. The VAS has a scoring range from 0 (worst health-state) to 100 (best health state) and is often used as a general measure for HRQoL. The EQ-5D-3L has been used in various health conditions including HD, and it has been found valid to use in the Norwegian population [31–34].

Statistical analysis

Descriptive statistics of mean values and standard deviation (SD) were calculated for normally distributed variables and median and interquartile range (IQR) for non-normally distributed socio-demographic and clinical variables. Frequencies and percentages were calculated in order to describe the nominal socio-demographic and clinical variables, as well as to describe ATC use as recorded by the five information items regarding ATC. Group differences between disease stages for the socio-demographic and clinical data were calculated using one-way ANOVA for normally distributed variables and Kruskal-Wallis tests for K-samples for non-normally distributed variables. Chi-square tests were performed in order to calculate overall group differences on ATC use between stages I – V.

In order to investigate associations between the independent variables including formal and informal ATC and the dependent variable representing HRQL (EQ-5D VAS score) simple regression analyses were performed. These revealed significant associations for all disease-related variables (TFC, cognitive impairment, disease duration, informant, housing situation), while none of the socio-demographic variables reached significance. Further, we performed multivariate linear regression analyses in order to assess the contribution of the variables of interest and other potentially confounding variables (control variables) on the dependent variable. Based on results of the simple regression analyses and clinical importance, all disease-related variables, except for living situation due to high correlation with TFC of >0.7, were entered into multiple regression model as control variables. Additionally, the model was controlled for variables age and education, both known to influence HRQoL [35, 36].
and cognitive impairment were entered as disease-specific control variables in separate models as they correlated highly with each other and are considered important relating to both variables of interest and HRQoL. Findings are presented in Adjusted $R^2$ and in standardized Beta ($\beta$) values with confidence intervals and partial $r^2$. Inspection of violation of assumptions resulted in logarithmical transformation of the values of disease duration. Multicollinearity between independent variables was investigated using inflation factor (VIF). Influential data points were examined using Cook’s distance and residual analyses were conducted revealing no outliers among any of the variables included in the analyses. Significance levels were set at $p = 0.05$ and all statistical tests were two sided. SPSS version 21.0; SPSS Inc. Chicago IL was used to perform all statistical analyses.

**RESULTS**

**Description of participants**

Socio-demographic and clinical characteristics of all 84 included patients are presented in Table 1. Disease Stage I included 12 patients (14%), Stage II 22 patients (26%), Stage III 19 patients (23%), Stage IV 14 patients (17%) and Stage V 17 patients (20%). The mean age of the patients was 56.7 (SD 11.4) years. Significant overall group differences ($p < 0.001$) were

### Table 1

Socio-demographic and clinical characteristics for total sample and divided across disease stages

<table>
<thead>
<tr>
<th>Variables</th>
<th>Categories</th>
<th>Complete sample (N = 84)</th>
<th>Stage I (n = 12)</th>
<th>Stage II (n = 22)</th>
<th>Stage III (n = 19)</th>
<th>Stage IV (n = 14)</th>
<th>Stage V (n = 17)</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
<td>Mean (SD)</td>
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<tr>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Age</td>
<td></td>
<td>56.7 (11.4)</td>
<td>49.8 (9.5)</td>
<td>54.6 (12.9)</td>
<td>58.9 (11.1)</td>
<td>61.1 (11.5)</td>
<td>57.8 (9.0)</td>
<td>0.084</td>
</tr>
<tr>
<td>Education (years)*</td>
<td></td>
<td>12.9 (3.5)</td>
<td>14.3 (3.3)</td>
<td>13.8 (3.8)</td>
<td>11.7 (3.2)</td>
<td>12.5 (3.7)</td>
<td>12.4 (3.3)</td>
<td>0.179</td>
</tr>
<tr>
<td>Gender</td>
<td>Female</td>
<td>37 (44)</td>
<td>5 (42)</td>
<td>8 (36.4)</td>
<td>7 (37)</td>
<td>7 (50)</td>
<td>10 (59)</td>
<td>0.616</td>
</tr>
<tr>
<td></td>
<td>Male</td>
<td>47 (56)</td>
<td>7 (58)</td>
<td>14 (63.6)</td>
<td>12 (63)</td>
<td>7 (50)</td>
<td>7 (41)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Education</td>
<td>Lower (≤12 years)</td>
<td>51 (60.7)</td>
<td>5 (42)</td>
<td>11 (50)</td>
<td>15 (79)</td>
<td>9 (64.3)</td>
<td>11 (65)</td>
<td>0.221</td>
</tr>
<tr>
<td></td>
<td>Higher (&gt;12 years)</td>
<td>33 (39.3)</td>
<td>7 (58)</td>
<td>11 (50)</td>
<td>4 (21)</td>
<td>5 (45.7)</td>
<td>6 (35)</td>
<td>0.560</td>
</tr>
<tr>
<td>Marital status</td>
<td>Single</td>
<td>36 (42.9)</td>
<td>4 (33)</td>
<td>7 (31.8)</td>
<td>9 (47)</td>
<td>8 (57.1)</td>
<td>8 (47)</td>
<td>0.643</td>
</tr>
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<td></td>
<td>Married</td>
<td>48 (57.1)</td>
<td>8 (67)</td>
<td>15 (68.2)</td>
<td>10 (53)</td>
<td>6 (42.9)</td>
<td>9 (53)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Occupation*</td>
<td>Manual</td>
<td>40 (47.6)</td>
<td>5 (42)</td>
<td>9 (40.1)</td>
<td>12 (63)</td>
<td>6 (46.1)</td>
<td>8 (47)</td>
<td>0.643</td>
</tr>
<tr>
<td></td>
<td>Non-manual</td>
<td>41 (48.8)</td>
<td>7 (58)</td>
<td>13 (59.1)</td>
<td>7 (37)</td>
<td>7 (53.8)</td>
<td>7 (41)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Employment</td>
<td>Employed</td>
<td>14 (16.7)</td>
<td>11 (92)</td>
<td>3 (13.6)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>Unemployed</td>
<td>70 (83.3)</td>
<td>1 (8)</td>
<td>19 (86.4)</td>
<td>19 (100)</td>
<td>14 (100)</td>
<td>17 (100)</td>
<td></td>
</tr>
<tr>
<td>Housing situation</td>
<td>Living at home</td>
<td>52 (61.9)</td>
<td>12 (100)</td>
<td>22 (100)</td>
<td>13 (68)</td>
<td>5 (35.7)</td>
<td>0 (0)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>Not living at home</td>
<td>32 (38.1)</td>
<td>0 (0)</td>
<td>6 (32)</td>
<td>9 (64.3)</td>
<td>6 (35)</td>
<td>17 (100)</td>
<td></td>
</tr>
<tr>
<td>Residence</td>
<td>Rural</td>
<td>12 (14.3)</td>
<td>1 (8)</td>
<td>3 (13.6)</td>
<td>2 (10.5)</td>
<td>3 (21.4)</td>
<td>3 (18)</td>
<td>0.859</td>
</tr>
<tr>
<td></td>
<td>Urban</td>
<td>72 (85.7)</td>
<td>11 (92)</td>
<td>19 (86.4)</td>
<td>17 (89.5)</td>
<td>11 (78.6)</td>
<td>14 (82)</td>
<td></td>
</tr>
<tr>
<td>Informant</td>
<td>Patient Patient &amp; informant only</td>
<td>27 (32.1)</td>
<td>9 (75)</td>
<td>14 (63.6)</td>
<td>4 (21)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>informant only</td>
<td>57 (67.9)</td>
<td>3 (25)</td>
<td>8 (36.4)</td>
<td>15 (79)</td>
<td>14 (100)</td>
<td>17 (100)</td>
<td></td>
</tr>
<tr>
<td>Clinical characteristics</td>
<td>Disease duration**</td>
<td>6 (7)</td>
<td>2 (2)</td>
<td>5 (6)</td>
<td>5 (6)</td>
<td>8 (7)</td>
<td>11 (7)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>Total FAS score**</td>
<td>15 (17)</td>
<td>24 (2)</td>
<td>20 (2)</td>
<td>15 (4)</td>
<td>5 (3)</td>
<td>0 (2)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>Independence score***</td>
<td>60 (26.5)</td>
<td>95.8 (5.1)</td>
<td>79.1 (2.9)</td>
<td>64.7 (6.3)</td>
<td>40.4 (10.8)</td>
<td>20.9 (5.7)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>n (%)</td>
<td>P-value</td>
</tr>
<tr>
<td>Overall cognitive impairment**</td>
<td>Mild to moderate</td>
<td>46 (54.8)</td>
<td>12 (100)</td>
<td>22 (100)</td>
<td>12 (63.2)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td></td>
<td>Severe</td>
<td>34 (40.5)</td>
<td>0 (0)</td>
<td>0 (0)</td>
<td>6 (31.6)</td>
<td>12 (85.7)</td>
<td>16 (94.1)</td>
<td></td>
</tr>
<tr>
<td>Comorbid conditions No/yes</td>
<td>No(No)</td>
<td>48 (57.1)</td>
<td>7 (58)</td>
<td>9 (41)</td>
<td>9 (47)</td>
<td>10 (71)</td>
<td>13 (76)</td>
<td>0.143</td>
</tr>
<tr>
<td></td>
<td>Yes</td>
<td>36 (42.9)</td>
<td>5 (42)</td>
<td>13 (59)</td>
<td>10 (53)</td>
<td>4 (29)</td>
<td>4 (24)</td>
<td></td>
</tr>
</tbody>
</table>

FAS: Functional Assessment Scale; SD: Standard deviation; IQR: Interquartile range; *using ANOVA; **Kruskall-Wallis for k samples ***normally distributed: reported mean (sd) and Anova; all other variables Chi-square; #3 responses missing (1 in Stage IV and 2 in stage V); ##4 patients unable to evaluate (1 in Stage III, 2 in Stage IV, and 1 in Stage IV).
found for employment, housing situation and informant. Furthermore, overall group differences for disease-specific clinical characteristics were significant \((p < 0.001)\). As expected, patients with advanced disease had longer disease duration compared to patients in the early phase of disease, while total scores for scales indicating functional disability (FAS and IS) showed a decline from stage I to stage V. The number of patients with severe cognitive impairment increased in advanced disease (all patients in stage IV and V had severe cognitive impairment, while all patients in stage I had mild to moderate cognitive impairment). There were no significant group differences for comorbid conditions \((p = 0.143)\).

**Description of distribution of informal and formal ATC**

Table 2 presents frequencies and percentages of ATC. Overall, approximately one third (36.9\%, \(n = 31\)) of all patients had ATC (either formal or informal) with the majority of patients in stages I to III (Stage I: 75\%, \(n = 9\), Stage II: 63.7\%, \(n = 14\), Stage III: 36\%, \(n = 7\), respectively). Formal ATC was most frequent in stage III (36.8\%, \(n = 7\)), while informal ATC was most frequent for patients in stages I and II (75\%, \(n = 9\) and 45.5\%, \(n = 10\), respectively). Overall significant group differences across the five disease stages were found for having and using ATC \((p < 0.001)\). Forty four percent of all patients \((n = 37)\) had received information about the possibility of receiving ATC. Only 32.1\%, \(n = 27\) had undergone a formal needs assessment for ATC, while even fewer patients (20.2\%, \(n = 17\)) had received training for use of the provided ATC. Information regarding ATC provision was predominantly given to patients in stage II (45.4\%, \(n = 10\)) and III (73.7\%, \(n = 14\)). Information regarding a formal needs assessments and training provided for ATC showed similar patterns. In stage I, four patients (33.3\%) had received information about ATC and none of the patients in stage I had yet received an evaluation regarding provision of formal ATC or received training. A similar number of patients in the advanced disease phase (stage IV and V) had received information, evaluation and training for the use of ATC. Overall group differences across disease stages were significant for the provision of information about ATC and performance of ATC evaluations \((p = 0.045 & p = 0.006\) respectively), while no general group difference was found for ATC training \((p = 0.150)\).

**Health-related quality of life**

An average score of 52.1 (SD 26.1) \((n = 82)\) was found for overall self-reported HRQoL measured by EQ-5D VAS. Average scores declined across disease stages I to III (stage I: mean 83 (SD = 16.4); stage II: mean 57.9 (SD = 20.3); stage III: mean 49.3 (SD 23.5)) and remained stable in advanced disease (stage IV: mean 35 (25.5); stage V: mean 38.3 (20.9) \((n = 15)\)).

**The relationship between ATC and health-related quality of life**

When investigating the relationship between formal and informal ATC on HRQoL \((n = 82)\) using...
simple regression analysis, we found a positive β value that reached significance for informal ATC, indicating that informal ATC was associated with higher HRQoL (β value = 0.356, β 95% CI: 8.45 – 34.16, p = 0.001). The formal ATC was not significantly associated with HRQoL (β value = –0.045, β 95% CI –19.04 – 12.58, p = 0.685) in the simple regression analysis.

Results of the multivariate linear regression analysis investigating the associations between having formal and informal ATC and HRQoL (n = 82), are displayed in Table 3. The final model controlled for TFC score, disease duration, informant, age and education explained one-third of the variance in HRQL scores (adjusted R² = 30%, p < 0.001). The model produced only one significant predictor, the TFC. TFC had a negative Beta value (β value = –0.564, β 95% CI 1.47 – 5.34, r² = 0.142, p = 0.001) showing that a higher TFC score corresponded to higher HRQoL, and explained almost all variance in the model. Formal ATC, informal ATC or other control variables were not associated with HRQoL in the final model.

Collinearity statistics were found to be acceptable (VIF: 1.15 – 2.99). No outliers were identified. No single case had undue influence on the model as indicated by Cook’s distance (D = max 0.145). The external validation of the model performed using the variable cognitive impairment instead of TFC showed the same value of the explained variance and other results.

**DISCUSSION**

The current study is the first to describe ATC and its association with HRQoL in HD. We found that 31 of 84 patients had ATC. Use of ATC was most frequent in disease stages I-III, in patients with mild to moderate cognitive impairment, with informal ATC most frequently used in disease stages I-II and formal ATC in disease stages II-III. Information about ATC, needs evaluation and training was provided most frequently in stages II-III. Multiple regression analysis showed that neither formal nor informal ATC were significantly associated with HRQoL. Of the assessed variables, functional capacity as assessed by TFC was the only variable significantly associated with HRQoL.

We found that relatively few patients in this study used formal ATC. The fact that a minority of the patients had formal ATC may be explained by differences between formal and informal ATC: informal ATC is introduced by the patients themselves in order to compensate for self-experienced reduced cognitive function and maintain their daily functioning. These items are well-known to the patients and easily accessible. Formal ATC, on the other hand, requires an implementation process and training. Additionally, patients in earlier disease stages have less pronounced cognitive impairment and higher functional capacity. They may be able to recognize and compensate for early symptoms of cognitive impairment by using such familiar objects. Needs for more complex aids may arise as the disease progresses and cognitive impairments increase in severity. More than 50% of the assessed patients had not received information or undergone a needs assessment for ATC, and few had received training. This finding is in line with other studies on patients with cognitive disabilities, showing that patients often do not receive information concerning ATC, and pointing to lack of knowledge regarding available ATC and a lack of trained personnel to implement them [37, 38]. In this study formal ATC was most frequently used in disease stages II-III, while informal ATC was used in stages I and II. This could be a result of the needs for formal ATC arising later in the course of disease. An additional explanation could be lack of awareness for and knowledge about the provision of formal ATC among professionals working with HD patients resulting in late performance of ATC.

**Table 3**

<table>
<thead>
<tr>
<th>Independent Variables</th>
<th>β</th>
<th>β (95% CI)</th>
<th>Partial r²</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Formal ATC</td>
<td>–0.067</td>
<td>–19.05–9.5</td>
<td>0.006</td>
<td>0.511</td>
</tr>
<tr>
<td>Informal ATC</td>
<td>0.018</td>
<td>–14.8–17.03</td>
<td>0.002</td>
<td>0.889</td>
</tr>
<tr>
<td>TFC</td>
<td>0.564</td>
<td>1.47–5.34</td>
<td>0.142</td>
<td>0.001</td>
</tr>
<tr>
<td>Disease duration</td>
<td>–0.156</td>
<td>–11.63–2.56</td>
<td>0.021</td>
<td>0.207</td>
</tr>
<tr>
<td>Informant</td>
<td>0.097</td>
<td>–8.81–19.51</td>
<td>0.008</td>
<td>0.454</td>
</tr>
<tr>
<td>Age</td>
<td>–0.108</td>
<td>–2.29–0.67</td>
<td>0.016</td>
<td>0.280</td>
</tr>
<tr>
<td>Education</td>
<td>0.037</td>
<td>–0.37–0.54</td>
<td>0.002</td>
<td>0.715</td>
</tr>
</tbody>
</table>

Note: β = standardized coefficients, partial r² = squared partial correlation coefficients. R-square = 0.36. Adjusted R² = 0.30, p < 0.001.
needs assessment and implementation. The latter may negatively influence patients’ ability to benefit from ATC as the progressive nature of cognitive impairment may reduce ability to learn to use ATC through training and thereby reduce the period of time for effectively being able to use ATC. Overall use of ATC was least frequent in stages IV-V, including the patients with more severe cognitive impairment. This may be expected as patients in advanced disease stages have global cognitive impairment (dementia) and motor symptoms causing increasing difficulty applying external aids.

In this study, we did not find an association between ATC and HRQoL. The TFC was the only factor significantly associated with HRQoL. This finding is in line with previous studies [12, 39, 40]. One possible explanation may be that improving HRQoL through the use of ATC requires more than access to devices. One may have to take into account the interactions between the device and everyday life environments in order to make ATC an effective intervention [41]. Prior studies have identified critical factors for successful use of ATC underlining the importance of establishing a match between the person with impaired cognition and the ATC through comprehensive assessment of individual characteristics [20, 26, 37]. These characteristics include personal characteristics of the individual, the environment and the ATC [37, 41]. Our results may indicate that ATC has not been successfully implemented. Several reviews and clinical studies show promising results for ATC as a mean to support and maintain cognitive function and to improve functional ability in patients with impaired cognition [22, 23, 42–46]. An improvement in the quality of life (QoL) and in the ability to perform daily activities was also found in a recently published study on home-based electronic assistive technology for memory in individuals with memory deficits [43]. ATC is thought to positively affect HRQoL through improving functional ability. Therefore, it may be that the effect of ATC is reflected in the TFC score in the present study.

**Limitations and strengths**

A number of limitations of the present study should be addressed. Firstly, this is a cross-sectional study that prevents us from describing the process of ATC implementation over time and discussing causal relationships. Secondly, our evaluation of cognitive impairment was based on clinical evaluation during the interview with the patient without using a standardized instrument. However, we pre-defined three categories of cognitive impairment (mild, moderate and severe) based on our clinical knowledge of cognitive functioning and work experience with patients suffering HD. The high correlation (>0.9) between cognitive impairment and TFC indicates that our evaluation can be considered sufficiently reliable. Thirdly, the present study does not include other disease-specific clinical assessments, which may provide more information about the characteristics of the patients who use ATC, and may help to tease apart associations between ATC and HRQoL. It should further be noted that a disease specific measure of HRQoL could potentially have been more sensitive to an association between ATC and HRQoL. Yet we consciously chose a generic measure in the form of the EQ-5D VAS as it is less complicated and therefore, easier to administer for advanced patients, which we specifically aimed to recruit. Some form of population bias cannot be excluded as patients with reduced self-awareness may not be in contact with healthcare institutions. They may also be more likely to decline participating in the study as they perceive themselves symptom free. Yet, despite reducing the statistical power, the response rate of 53.2% included in the analyses of this study can be considered satisfactory, given the clinical picture of HD. The present study also comes with strengths. Patients in stages IV and V (advanced stages) are relatively equally represented. Moreover, this is the only study to describe use of ATC in HD, a potential additional resource to existing comprehensive healthcare services.

**Clinical recommendations and recommendations for future research**

Previous studies on ATC and cognitive impairment propose that ATC can be effective and that a comprehensive process of identifying needs, personal and environmental factors, implementation through training (matching the person with the device) increases the likeliness of patients being able to benefit from ATC. The results of the present study suggest the need for increased awareness about ATC as an intervention to support and compensate cognitive abilities in HD in Norway. Our findings further suggest that professionals need to be aware of the requirements for successful implementation and awareness of the process towards successfully providing ATC to patients with HD. It is important to bear in mind the complex and changing character of HD, including motor impairments, calling for regular monitoring of ATC.
needs and use. The relatively low number of patients using formal ATC and the absence of an association between ATC and HRQoL may indicate an insufficient ATC implementation process, lacking comprehensive and individually tailored assessment. The results call for further research on ATC as a treatment intervention, and for further investigation of the effectiveness of ATC on cognition and ADL in HD. Future studies should also seek to further tease apart associations between ATC and other disease specific factors, including cognitive function and functional abilities, and HRQoL.

Conclusion

ATC was used mainly in stages I-III, in patients with mild to moderate cognitive impairment. Use of informal ATC was most frequent in stages I – II, and formal ATC in stages II-III. Information about ATC, needs evaluation and training was predominantly provided to patients in stages II-III. No association between ATC and HRQoL was found. Results may reflect lack of awareness and knowledge about the availability of ATC among healthcare professionals. Results further suggest the importance of a thorough assessment and implementation process matching device with the individual, requiring healthcare professionals to see the patient and ATC in a social context.

CONFLICTS OF INTEREST

The authors have no conflicts of interest to report.

ACKNOWLEDGMENTS

The present study was funded by the Norwegian Research Council (209748). We thank the patients, their family members and their healthcare professionals for their time and effort as part of participating in the study. Additionally, we thank Kristin Iversen, Gunvor Ruud, Nancy Borgerød and Ragnhild Wehus, for their assistance with the recruitment of participants.

REFERENCES


[18] Andrews SC, Dominguez JF, Mercieca EC, Georgiou-Karistianis N, Stout JC. Cognitive interventions to enhance...


ERRATA

Page 3: the second sentence: added the word ‘and’: before: ‘…located on chromosome 4, inheritance…’. Now: ‘…located on chromosome 4, and inheritance…’

Page 24: first aim of Paper II: removed the repeated words ‘look like’ at the end of the sentence of the first aim.

Page 36: 4th sentence from the top removed the word various: before: ‘…has been applied to various several conditions…’. Now: ‘…has been applied to several conditions…’

Page 44: next to last sentence: corrected from: ‘One such issue is population the bias as a result of…’ to ‘One such issue is the population bias as a result of…’

Page 63: 7th sentence under chapter 6.2.3.2. corrected the sentence by removing words ‘…been…were… show a …’: Before: ‘The present results may thus reflect that ATCs were not been successfully implemented and were did not show a favorably impact on HRQoL’. Now: ‘The present results may thus reflect that ATCs were not successfully implemented and did not favorably impact on HRQoL’

Correction of references due to incorrect conversion from Pubmed to End note:

Page 68, reference 23: Added: pii: ecurrents.hd.9504af71e0d1f87830c25c394be47027.

Page 70, reference 61: Added and corrected name: Health Soc Care Community. 2015 Sep;23(5):569-76.

Page 71, reference 82: Added: pii: ecurrents.hd.2c56ceef7f9f8e239a59ecf2d94cddac.


Page 73, reference 119: Title of the journal was written without capital letters. Corrected to Health and Quality of Life Outcomes.

Page 73, referance 133: Last word of the Journal name was written with small instead of capital letter: corrected to: BMJ Open.