Health and People with Usher syndrome
"Being deafblind it’s like being deep under the earth where there is no light or sound, at the beginning I had difficulty breathing, but after I was convinced that there was enough air"

Yolanda de Rodriguez (1945-2002)
Studies from the Swedish Institute for Disability Research 76

MOA WAHLQVIST

Health and People with Usher syndrome
Abstract


The present thesis concerns people with Usher syndrome (USH) and their health. People with USH have a congenital hearing loss of various degrees and an eye disease with a progressive course; for some, the balance is also affected. Three clinical groups have been identified 1, 2 and 3, and 13 genes have currently been identified. USH is the most common cause of deafblindness. Clinical knowledge and the limited research that exists have shown that people with deafblindness can experience difficulties in everyday life. Depression, anxiety and social withdrawal have been described.

The general aim of the present thesis was to describe the health of people with USH. The empirical material employed was based on an extensive survey in which people with USH answered two questionnaires concerning health, anxiety, depression, social trust, work, health-care, financial situation, and alcohol and drug use. The focus of the present thesis is on general health, physical health and psychological health, social trust and finance. Three studies in the present theses focus on USH1, 2 and 3, respectively; finally, the fourth study provides an in-group comparison of people with USH. The results of studies I and III are compared with a cross-section of the Swedish population. The results revealed poor physical and psychological health, a lack of social trust and a strained financial situation regardless of clinical diagnosis. The discussion stresses the importance of taking a biopsychosocial approach when describing the health of people with USH, in which previous research is lacking. Additional research should focus on the mechanisms at different levels that affect people with USH and their health from a life-course perspective. Furthermore, research should include a salutogenic perspective to explore the resources and strengths of people with USH.

Keywords: Usher syndrome, Deafblindness, Health, General Health, Physical Health, Psychological Health, Social Trust, Financial Situation

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## Abbreviations

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<th>Description</th>
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<tbody>
<tr>
<td>ADL</td>
<td>Activity of daily living</td>
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<tr>
<td>CI</td>
<td>Cochlear implant</td>
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<td>dB</td>
<td>Decibel</td>
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<td>ERG</td>
<td>Electroretinography</td>
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<td>GHQ-12</td>
<td>General Health Questionnaire, 12</td>
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<td>HAD-scale</td>
<td>The hospital anxiety and depression scale</td>
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<td>HET</td>
<td>Health on Equal Terms questionnaire</td>
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<tr>
<td>ICD-10</td>
<td>International Classification of Diseases, version 10</td>
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<td>ICF</td>
<td>International Classification of Functioning, Disability and Health (2001)</td>
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<tr>
<td>HI</td>
<td>Hearing impairment</td>
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<td>HL</td>
<td>Hearing loss</td>
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<tr>
<td>OCT</td>
<td>Optical coherence tomography</td>
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<tr>
<td>PTA</td>
<td>Pure tone average</td>
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<tr>
<td>PTA4</td>
<td>Pure tone average for four frequencies (0.5-4kHz)</td>
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<tr>
<td>RP</td>
<td>Retinitis pigmentosa</td>
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<tr>
<td>SOC</td>
<td>Sense of coherence</td>
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<tr>
<td>USH</td>
<td>Usher syndrome</td>
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<tr>
<td>USH1</td>
<td>Usher syndrome type 1</td>
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<tr>
<td>USH2</td>
<td>Usher syndrome type 2</td>
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<tr>
<td>USH3</td>
<td>Usher syndrome type 3</td>
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<tr>
<td>VA</td>
<td>Visual acuity</td>
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<tr>
<td>VF</td>
<td>Visual field</td>
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Included studies

The present thesis is based on the following four studies, which are referred to in the text by their roman numerals:


II. Wahlqvist, M., Möller, C., Möller, K., & Danermark, B. Health among persons with Usher syndrome type 3, Implications of Deafblindness. Submitted for publishing.


IV. Wahlqvist, M., Möller, C., Möller, K., & Danermark, B. Similarities and Differences in Health, Social trust and Financial situation in people with Usher syndrome, a bio- psychosocial perspective. In manuscript.

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Introduction

In the United Nations convention on human rights for people with disabilities, article 25 asserts that State Parties should recognize:

“...that persons with disabilities have the right to the enjoyment of the highest attainable standard of health without discrimination on the basis of disability..”

The present thesis concerns people with Usher syndrome (USH) and their health. People with USH have a congenital hearing loss to varying degrees and progressive eye disease; balance is also affected in some patients. USH is the most common cause of deafblindness. The clinical knowledge and limited research that exists have shown that people with deafblindness experience difficulties in everyday living, including problems with, anxiety, depression, social withdrawal and communication.

The empirical material presented in the present thesis is based on an extensive survey in which people with USH answered a variety of questions concerning health, wellbeing, anxiety, depression, social trust, work, health-care, finances, and the consumption of alcohol and drugs. The present thesis focuses on general health, physical and psychological health, social trust and finances.

The presence of a medical condition or impairment does not automatically implicate poor quality of life or poor health. However, the presence of a disease or disability can, together with other factors, lead to worse health and a poorer quality of life than general population. This result might be because of inequalities in the surrounding environment, restrictions in activity and participation, and unequal positions in the labor market, and the health-care services provided. The present thesis is rooted in an interdisciplinary setting, where competences from different disciplines e.g., medicine, psychology, sociology and social work have contributed to understand the health of people with USH.

Let us begin with the important definitions and concepts of the present thesis. Furthermore, the disability discourse as well as the connection between disability and health will be described. The present research regarding deafblindness, USH and health is introduced. To begin, brief introductions of the anatomy and physiology of the ear, and eye is provided.
The anatomy and physiology of the ear and eye

The ear
The auditory system consists of the ear, the auditory canals and the brain. Functionally and anatomically, the ears consist of the outer ear, the middle ear, the inner ear as well as the central auditory system. The outer and middle ear direct sound to the inner ear. The cochlea consists of three parts, the scala vestibuli, scala tympani and scala media.

From the tympanic membrane to the ossicles in the middle ear, vibrations transmit sound to the inner ear. The sound wave amplified by the ossicles in the middle ear is transmitted from air to liquid and travels through the scala vestibuli and scala tympani. The outer and inner hair cells in the scala media constrict due to the traveling of the perilymphatic wave. The synapses of the hair cells release neurotransmitters that convert the movement of the hair cells into electrical impulses. The electrical impulses are then passed on through the auditory nerve. The auditory nerve ends in the cochlear nucleus in the brainstem where the relocation to new nerves paths is conducted. Approximately 70 percent of the signals travel contralateral to the opposite cerebral hemisphere, whereas 30 percent are projected ipsilaterally. Numerous connections exist where the auditory system sends and receives signals from other systems or senses, e.g., vision. In the auditory cortex, the electrical signals are interpreted as understandable sounds. In the cochlea, every frequency has a specific place for stimulation: High frequencies are in the beginning, whereas low are at the apex.

The inner ear consists of the vestibular i.e., balance organ that features three semicircular canals, the utricle and the saccule. These sensory organs register the movements of the head, changes in position and gravity. The hair cells react to the movements of the endolymph in the semicircular canals because of the movements of the head. The impulses are sent in the same way as auditory impulses through the vestibular nerves to the vestibular nuclei in the reticular formation in the brainstem. From the vestibular nuclei, nerve signals are transmitted to the muscles of the eye to direct the eyes in the reverse direction to the head movements to maintain a stable picture on the retina. This reflex is known as the vestibule ocular reflex.
Definitions of hearing loss

Hearing disorder refers to an umbrella term that encompasses a variety of terms such as hearing loss, hearing impairment, hard of hearing and deafness. Hearing loss (HL) refers to the location in the anatomical system that the damage is located, either in the anatomical or physiological parts of the ear as well as the hearing function. HL can have a variety of causes such as genetics, age, or being exposed to noise or infections. In contrast, hearing impairment (HI) refers to functional hearing loss or the degree of HL. To be deaf is, in some contexts, considered as being part of a cultural and language minority in the hearing community, rather than as an impairment; this view is often referred to as “Deaf”.

A common way to establish the degree of HL is by assessing hearing thresholds with pure tone audiometry (PTA). PTA provides the amplitude of the weakest tones that a person can hear. The frequencies tested are usually 0.25, 0.5, 1, 2, 4, 6 and 8 kHz. An average of four frequencies (0.5, 1, 2, 4 kHz) are often used (PTA4). Normal hearing thresholds are age dependent; a PTA4 of ≤25 dB is considered normal for younger participants. The thresholds used in the present thesis were classified according to the European standard of PTA4 from mild to profound HL: Mild HL >20 dB and <40 dB; Moderate HL, >40 dB and <70 dB; Severe HL, >70 dB and <95 dB; Profound HL, ≥95 dB. The HL of a person is reported as the PTA4 for the best ear.

The eye

The optic system consists of the eye, the optic nerves and the visual nerves fibers in the brain. The function of the eye is to register the light waves transmitted through the cornea, the lens and the vitreous body to be projected on the retina. The anatomy of the eye, briefly described, consists of the cornea, iris, lens, vitreous body, retina, optic nerve and the extraocular muscles. The nerve cells in the eye are specialized photoreceptors known as the rods and cones. The rods are specialized to register brightness and peripheral vision. Three types of cones exist that register the colors red, blue and green as well as control central vision and visual acuity (VA). The central part of the retina is the macula, the primary location of the cones, with VA to see details. The periphery enables us to rapidly adjust to light-darkness and the peripheral vision (i.e., visual field, VF) primarily managed by the rods. The optic nerve transmits impulses via the visual pathways to the visual cortex in the occipital lobes. The movements of the eye are controlled by eye muscles via voluntary and involuntary
movements. The involuntary movements are originate from vestibulo-
ocular reflex where the signals from the vestibular organs in the ear are
directed through the brain stem to move the eye opposite to head move-
ments at the same speed and amplitude to keep a steady image on the
retina.

To have the best possible visual perception, good visual field and visual
acuity are necessary.\cite{77,159}

Definitions of vision loss
Many disorders can lead to the malfunction or impairment of the optic
system. In addition several ways exist to examine vision loss, such as
funduscopy, the visual acuity test, the visual field test, electroretinography
(ERG), optical coherence tomography (OCT), angiography, and so on.

Visual acuity (VA) can be assessed in many ways but the most common
measure is having a patient read the smallest letters on a standardized
chart at a distance of approximately 2 meters. This chart is known as Snell-
en’s chart, and it consists of eleven lines of capital letters with a decreas-
ing font size for every subsequent line. The number of letters included on
each line also increases.\cite{124} European standards for VA range from 1.0
(perfect vision) to 0.00 (blindness). Visual acuity is measured using the
best correction to evaluate visual acuity function.

The Goldman visual field test is one way to measure visual field (VF;
the visual surroundings when the eyes are fixed at a certain point). This
test is constructed to map a person’s visual field by presenting points of
light of various size and intensity. The light might either be at a fixed
point or move toward the center from the perimeter. Goldman perimeter
can be assessed in many ways. One method used in this thesis categorizes
the visual field types into five phenotypes (1-5), where 1 denotes normal
vision, 2 denotes a visual field with partial or complete ring scotoma, 3
denotes a concentric central field loss with a remaining peripheral island, 4
denotes a concentric loss with a visual field of ≤ 10°, and 5 denotes blind-
ness.\cite{64}

The most reliable and objective method of retinal assessment is electro-
retinography (ERG), where the electrical responses of the retina are meas-
ured by creating a flashing light stimulus. If the ERG pattern changes or is
extinguished, then retinal degeneration might be suspected.\cite{66,91}

The definition of visual impairment according to the WHO is included
in the International Classification of Diseases-10 (ICD-10) and describes
four levels of visual function: 1) normal vision, 2) moderate visual im-
pairment (0.3-0.1), 3) severe visual impairment (0.1-0.05), and 4) blindness (0.02-light perception). The definitions of vision impairment have been criticized based on their classifications of vision impairment solely according to visual acuity, which does not cover the whole range of limitations in vision that might cause functional limitations for individuals.

**Deafblindness**

Deafblindness is an umbrella term for conditions in which both hearing and vision are affected. Approximately >200 hereditary syndromes can cause deafblindness (Möller, C., personal communication 2015). In 2003, approximately 50 hereditary syndromes that cause deafblindness in adults were reported. The most common cause of deafblindness is syndromes of genetic origin, but other etiologies can include infections, medication and trauma. Age-related deafblindness is the most common cause of sensory deprivation in the elderly, encompassing age-related hearing loss, macular degeneration, retinitis pigmentosa (RP) and cataracts. The definition of deafblindness has been discussed recently, in research, clinical settings and among people with deafblindness themselves. In 2004, the EU parliament recognized deafblindness as a separate and distinct disability, and in 2007, the Nordic definition of deafblindness was accepted in Reykjavik, by the Nordic Leadership Forum. The definition was revised in 2013 by the Swedish Council of Deafblindness, and defines deafblindness as

“...a distinct disability. Deafblindness is a combined vision and hearing disability. It limits activities of a person and restricts full participation in society to such a degree that society is required to facilitate specific services, environmental alterations and/or technology.”

The definition includes a five-point list of comments that clarifies and highlights different issues in the definition of special interest such as information, variations in disability across different settings, service delivery and environmental adjustments. A variety of research definitions referring to deafblindness exist, but there is no consensus regarding which should be used or whether different definitions denote different concepts. Therefore comparisons and generalizations of results across different studies are limited. Two major streams of definitions can be identified: medical definitions and more functional emphasizing the effect of hearing and vision loss on everyday activities and participation in society. However,
er, neither the medical nor functional definitions of deafblindness refer solely to people who are totally deaf and blind; rather they include people who have remaining hearing and vision and different onsets. In this sense, the term “deafblindness” is misleading with the regard to the use of the terms “deaf” and “blind”. Other terms that are used include “dual sensory loss”, “dual sensory impairment”, and “combined hearing and vision impairment/loss” as well as, “congenital deafblindness” and “acquired deafblindness”. Depending on the context, these terms can be used interchangeably\(^\text{150}\). In the thesis, the term deafblindness is used.

Vision and hearing are complementary senses that enhance each other: they are also the primary senses for communication\(^\text{107}\). Restrictions in both of these senses will have significant consequences for the individual in his or her interactions with the environment, others and society, both with regards to receiving and providing information\(^\text{18, 55, 69, 128}\). The consequences of deafblindness are sometimes expressed as more than the sum of each impairment alone\(^\text{30}\).

**Usher syndrome**

USH is the most common cause of genetic deafblindness. It includes congenital hearing loss (HL), which can vary from profound deafness to moderate HL; an eye condition known as retinitis pigmentosa (RP); and, for some, balance problems due to bilateral vestibular areflexia (i.e., the absence of signals from the balance organs in the inner ears)\(^\text{78, 95}\). The worldwide prevalence of USH has been estimated as 1-4 in 25.000 people\(^\text{95}\), or 3.2- 6.2 in 100,000\(^\text{98}\) depending on the study. In Sweden, the overall prevalence of USH is estimated at 3.3 per 100,000 people\(^\text{131}\). Approximately 800 people in Sweden live with USH, equally distributed across men and women\(^\text{140}\). von Graefe first described USH in 1858, but it was British ophthalmologist Charles Usher who named the syndrome in 1914. USH is an autosomal recessive genetic condition with large heterogeneity; currently, 13 associated genes have been identified\(^\text{95}\). Its autosomal recessive pattern means that both parents must be carriers of the genes that cause USH to pass it on to their children\(^\text{19, 98}\). The USH genes are likely to be integrated in the protein network referred to as “the Usher interactome”. Thus, different mutations might lead to similar hearing and vision phenotypes\(^\text{81}\). The HL in USH is sensorineural which means that it is located somewhere in the inner ear, in the cochlear nerve or somewhere else in the auditory system\(^\text{98}\). RP is a retinal disease that affects the rods and cones in the eye with a progressive degenerative course over the life-
span. Night blindness, light sensitivity, limited visual field and blurred visual acuity are symptoms\textsuperscript{44, 66, 78}. Cataract often accompanies RP and is common among elderly people (85 percent) with USH\textsuperscript{133}. Cataract affects visual acuity, and although people who have cataract can undergo surgery, it is not unusual for problems to reoccur. Three clinical types of USH are known (1-3)\textsuperscript{98, 108, 134}.

People with USH use different communication modalities, e.g., speech, sign language, tactile sign language, lip-reading, braille, and technical devices. It is not uncommon for individuals method of communication to change over the lifespan\textsuperscript{45}.

**USH1**

The estimated prevalence of USH1 in Sweden is 1.6/100.000\textsuperscript{131}. USH1 is characterized by congenital profound deafness, bilateral vestibular areflexia, and RP. For people with USH1, the discovery of RP is usually established during early adolescence. Because of the malfunction of the balance organ in the inner ear, infants with USH1 have problems with motor skills such as crawling and sitting and show as delayed walking age (>18 months), which suggests additional problems and the possibility of an early diagnosis\textsuperscript{78, 131}.

Most adults with USH1 in Sweden today use visual sign language; as their vision deteriorates, they complement this with tactile sign language as their methods of communication. They also use different technical devices to receive information and communicate with the environment. The introduction of cochlear implants in the 1990s slowly changed communication from sign language to oral communication or sign language and oral communication in combination because most children in Sweden who are born with severe-to-profound hearing loss currently receive a cochlear implant.

**USH2**

The prevalence of USH2 is estimated as 1.4/100.000\textsuperscript{131}. People with USH2 are born with a congenital moderate-to-severe HL\textsuperscript{98}. According to the European standard, this diagnosis means an HL over 40 dB and less than 70 dB (moderate) as well as over 70 dB but less than 95 dB (severe)\textsuperscript{145}. This HL remains stable over the years. RP is often discovered in people with USH2 in late adolescence or early adulthood. However, RP might be present before diagnosis, (e.g., for example night blindness or problems
with adaptation between light and dark). People with USH2 do not have problems with their vestibular organs and balance\textsuperscript{78, 95}.

People with USH2 usually communicate via speech and lip-reading. People with USH2 also use different technical devices such as hearing aids, loop system, FM systems, computers, magnifiers, and more to facilitate communication and receive information about the environment. Some also learn sign language to complement oral communication.

**USH3**

USH3 is most rare of the three types of USH in most parts of the western world, and the prevalence of this condition in Sweden is estimated as 0.3/100.000\textsuperscript{131}. The hearing, vision and balance of people with USH3 show a progressive degenerative course over the life-span, with a moderate hearing impairment as children that progresses to deafness and severe vision impairment at approximately 30 years old\textsuperscript{133}. However, the discovery of hearing impairment varies, and in some cases, it is not discovered until 3 or 4 years old\textsuperscript{121}. The balance function deteriorates with increasing problems over the lifespan\textsuperscript{78}. It is not unusual for people with USH3 to have been clinically diagnosed with USH2 as child but to have this decision revised during adulthood as the progression of hearing and vision loss becomes obvious. With the development of genetic testing, the possibility to receive a correct diagnose at an earlier age is improving. The importance of a correct diagnosis concerns being able to provide accurate information about the course of the disease from a lifespan perspective. People with USH3 often use speech as youth but change by using visual or tactile sign language as a complementary communication strategy as adults. People with USH3 use hearing aids and other technical devices such as loop systems, FM systems, magnifiers for computers, and other systems to receive information about activities in the environment.
Health

Health is a comprehensive concept that has been the object of interest and discussed since antiquity. It can be viewed from a multitude of perspectives and is often used as an everyday word without any further explanation. In ancient Greece, Hippocrates spoke of health as the relationship between soul and body, and described how humans should behave to keep their health and body fluids in balance. The definition posed in ancient Greek stated that

“Health is a unity and harmony within the mind, body, and spirit which is unique to each person and is as defined by that person. The level of wellness or health is, in part, determined by the ability to deal with and defend against stress. Health is on a continuum with movements between a state of optimum well-being and illness which is defined as degrees of disharmony. It is determined by physiological, psychological, sociocultural, spiritual and developmental factors.”

The relationship between the soul and body or between the mental and physical has been discussed within different philosophical and religious discourses throughout history. Health has been and is related to numerous other concepts such as wellbeing, quality of life, health related quality of life, happiness and so on. The definition of health provided by the World Health Organization (WHO) is

“A state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.”

Several authors have criticized this definition for possibly confusing health and happiness, for being a utopia, and for meaningless. The problems with operationalizing the different concepts in the WHO definition have also been discussed. These difficulties primarily concerned how to define social well-being and how to adopt the WHO definition to different cultures. Bickenbach stated that if the definition of health as defined by the WHO is used, then no person (regardless of disability), will be able to claim complete physical wellbeing. Another way to discuss health can be found in the normative approach in which, health and disease are used in such a way that they reflect our definitions of physical and psychological states. Desirable states are healthy, and those who are undesirable are labeled as diseased. In some contexts, the triad of disease, illness and sickness (which implies differences in the meaning of these three concepts) is used. Disease refers to a pathological process in which the physical con-
dition is central, and illness is understood as a subjective experience of being unhealthy, which includes a reduced capacity. Sickness is to be understood as a specific social role, while society is obliged to sustain the “sick” person.115

Biological health

Two major theories of health have been present throughout history. Nor-denfelt 118 describes these theories of health and disease as divided into the medical/bio statistical and holistic theories of health. Related theories exist, and theories about health can be positioned on a continuum with endpoints of biological/medical and holistic48. Christopher Boorse argued that the bio statistical theory implies that healthy people are defined as those without disease or illness

“Health is normal functioning, where the normality is statistical and the functions biological.” (p.542)20

This position can be regarded as the normative notion of health: The absence of a medical condition is the definition of health. Hence, the naturalist defines the natural traits of humans48. Arguments exist that determining the correct definitions of health and disease is not of interest; instead, descriptions should focus on physiological or psychological states and what should be valued or disparaged48. What we define as health is related to the concept of disease, changes over time and is relative to environmental and cultural contexts. Boorse describes four criteria that must be fulfilled to describing health and disease. The first considers reference class. Reference class is something smaller than the entire species because of limitations regarding what is normal within a species, related to sex or age21, 48. The second criterion is related to the normal function of the reference class in terms of survival and reproduction. The two last criteria are related to disease. The third is related to disease as either a type of impairment to one’s normal functional ability or a limitation in one’s functional ability caused by the environment48. The last criterion concludes that health is the absence of disease21. A fundamental critique has been levied at the second criteria, given that biology does not exclusively address the survival and reproduction of a species; rather, an organism has many biological functions or states that do not relate to survival or reproduction48.
Holistic Health

Holistic health philosophers such as Nordenfelt define health as something more than the absence of disease; however, the non-presence of disease does not guarantee health. To be healthy is related to the individual as a whole entity in his or her environment and cannot be defined by biology or psychology alone. In this theoretical perspective, ability and individual choice are central; unhealthy people are hindered from taking these actions. In other words, health is connected to the person as a whole, individual ability and intention. On the other hand, diseases are connected to the organs or some part of the body or mind that is not functioning.

Biopsychosocial health

Developments of medical science have led to the emergence of more comprehensible concepts to describe the health of the individual. In a 1977 article, Engel concluded that

“I contend that all medicine is in crisis and, further, that medicine’s crisis derives from the same basic fault as psychiatry’s, namely, adherence to a model of disease no longer adequate for scientific tasks and social responsibilities of either medicine or psychiatry.” (p 129)

The opinion was that within medicine, physicians should only consider the somatic situation of the patient and should not concern themselves with psychosocial issues. This belief led to psychiatrists suggesting that their field of expertise would be excluded from medicine; in fact, it was argued that psychiatry did not conform to the accepted concept of disease. According to Engels, the biomedical model was the predominant way of how disease was understood at the time. He concluded that the way of considering disease as a deviation from the norm leads to thinking of biological variables as measureable. Therefore, it does not have a room within its framework address social, psychological, and behavioral dimensions. The body was considered as a machine where disease is the breakdown of that machine. A discrepancy existed between practice and science within the biomedical field until the 20th century. In practice, emotions are considered as a part of the development and course of a disease.

The transition from “biomedical to biopsychosocial” was present over the last century. According to Engel, the use of biopsychosocial thinking is a more inclusive conceptual framework to guide professionals in their everyday work with patients. Thus, the patient can actively contribute by providing information regarding his or her condition and empha-
size the importance of inter-professional cooperation in the healthcare setting. This viewpoint has not predominated in history, where biomedical and biopsychosocial platforms have been typically pointed in the opposite directions.

Furthermore the biopsychosocial model was developed by the World Health Organization (WHO) and the International Classification of Functioning, Disability and Health (ICF). The ICF is an updated and inclusive classification of the former International Classification of Impairments, Disabilities and Handicaps (ICDH) and complements the WHO International Statistical Classification of Diseases and Related Health Problems (ICD-10). The ICF takes a comprehensive view on health and its effect on different aspects of life, health and wellbeing. Ways to describe non-fatal health outcomes, functioning and disability in all areas of life were missing from the ICD-10. The ICF model integrates the medical and social models and presents a coherent view of different perspectives of health (i.e., biological, psychological and social). The underlying postulation of health in the ICF is that it is not only the absence of disease and injury but also bodily and mental functioning, where the level of functioning is independently determined. The ICF intends to create scientific grounds to understand the concept of health. To make this goal possible, a common language is needed as are common standards for reporting health outcomes to make them comparable across nations and cultures. The ICF defines impairments as problems in bodily function or structure that denote deviations or loss. However, whether the impairments are compatible with health or a representation of disease depends on the individual’s potential to the culture and specific demands put on the individual.
Bickenbach\textsuperscript{12} described the conceptualization in the ICF as “interactional rather than linear in the sense that disabilities are characterized as outcomes of the interaction between underlying health conditions... and physical, human-built, attitudinal, and social environmental barriers.”(p. 825)\textsuperscript{12}

The ICF has two parts. The first is the conceptual model that is built upon a biopsychosocial model to understand health and health-related conditions. The second part includes codes for classification. This part of the ICF is comprehensive and covers approximately 1500 codes at different levels of precision\textsuperscript{153}. This part is not further elaborated in the present text. The structure of the classification makes choosing the level of detail to describe function and disability possible. Human functioning is identified as bodily functions, bodily structures, activities and participation\textsuperscript{160}.

The relationship between health and disease are far from clear and will never be so because their definitions are infused with diverse cultural, social and psychological considerations\textsuperscript{46}. Bircher\textsuperscript{13} describes a dynamic definition of health and disease, combining ideas from both Engel\textsuperscript{46} and Nordenfelt\textsuperscript{117} and stating that essential elements in defining health include both the biopsychosocial nature of human existence, the fact that every person’s health determines his or her future and the relationship between the demands made on the individual’s life and the individual’s abilities to meet them. Living demands change throughout the lifespan and are, according to Bircher\textsuperscript{13}, culture specific and must be met based on one’s personal responsibility. Bircher’s\textsuperscript{13} definition of health is below.

Health is a dynamic state of wellbeing characterized by a physical, mental and social potential, which satisfies the demands of a life commensurate with age, culture, and personal responsibility. If the potential is insufficient to satisfy these demands the state is disease.”(p. 336)\textsuperscript{13}
Furthermore, Bircher and Kuruvilla\textsuperscript{14} developed the theory of health using the Meikirch Model of Health that defines health as

“…a state of wellbeing emergent from conducive interactions between individuals’ potentials, life’s demands, and social and environmental determinants. Health results throughout the life course when individuals’ potentials – and social and environmental determinants – suffice to respond satisfactorily to the demands of life. Life’s demands can be physiological, psychological, or environmental, and vary across individual and context, but in every case unsatisfactory responses lead to disease”\textsuperscript{(p.368)}\textsuperscript{14}

This theory of health includes the social and environmental determinants found within the field of public health. Bircher and Kuruvilla state that this definition of health is limited by being theoretical and conceptual. However, by using the of ICF definition and other tools available to measure health, disability and quality of life, the Meikirch Model can add to the understanding of the factors that contribute to suboptimal health\textsuperscript{14}.

**Quality of Life**

Closely related to theories about health are concepts related to quality of life (QoL) and health-related quality of life (HQoL). Aristotle discusses quality of life using terms such as “the good life”, “doing well” and “happiness”\textsuperscript{50}. Engel\textsuperscript{46} wrote that a need exists for a shift in the thinking of the discourse on quality of life. A vast body of research addresses the definition of quality of life and its related concepts; this discussion will not be fully developed in the present thesis. However, a brief introduction will be provided because of, its close relationships to the concept of health and the measurement of health. The definition of QoL experiences is similarly as jumbled as the concept of health. According to Bergsma and Engel\textsuperscript{10} a need exists to measure what is not defined. Boström and Nyqvist\textsuperscript{22} stated that QoL is a broad concept that refers to many different aspects of life\textsuperscript{22}. However, the different definitions and opinions that exists seem to extensively agree that quality of life is a multidimensional concept\textsuperscript{51}. One definition of quality of life is described as addressing the “goodness of life” in terms of the aspects that are affected by health and health-related quality of life\textsuperscript{23}.
To express quality of life solely in terms of life satisfaction is, according to Felce and Perry\textsuperscript{51}, not sufficient; satisfaction is a personal assessment and grounded in personal experiences without any objective measurements and is, in its construction, not achievable. Felce and Perry\textsuperscript{51} stated that

“life conditions and satisfaction with life will inevitably vary, and neither ideal conditions nor perfect satisfaction can be arranged for or achieved by every member of a society or societal subgroup.” (p.59)\textsuperscript{51}

Moons et al.,\textsuperscript{101} contrasted Felce and Perry\textsuperscript{51} by stating that a growing consensus exists since, the late 1990s and the beginning of the millennium regarding that quality of life is determined by subjective experiences unlikely to be determined by objective life conditions. In this understanding, a personal and subjective appraisal of one’s life conditions is what determines quality of life\textsuperscript{101}. Despite the different existing definitions of quality of life, overlaps exist in domains that are of interest. Felce and Perry\textsuperscript{51} identified five major domains that include the major aspects needed to operationalize quality of life. The first domain addresses physical wellbeing, which includes health, fitness and physical safety. The second includes material wellbeing and sustainable finances or income, the quality of the living environment, privacy, possessions, food, transport, neighborhood, security and stability. Social wellbeing is the third domain, which includes quality and breadth of interpersonal relationships, within the immediate and extended family and one’s general friends and acquaintances. This dimension also includes support from the community, community activity and level of acceptance and involvement. Furthermore, development and activity are identified as a dimension that includes possession and the use of skills in relation to independence, competence, control and choice. Functional activities such as work, leisure, housework, education and productivity are included. The last domain includes emotional wellbeing as affect or mood, satisfaction of fulfillment, self-esteem, status and respect as well as religious faith\textsuperscript{51}.

Felce and Perry\textsuperscript{51} presents a comprehensive overall model of quality of life that defines this term as

“...an overall general wellbeing that comprises objective descriptors and subjective evaluations of physical, material, social and emotional wellbeing together with the extent of personal development and purposeful activity, all weighted by a personal set of values.” (p.60)\textsuperscript{51}

The model is influenced by external factors which might include genetic, social and material inheritance, age and maturation, developmental histo-
ry, employment, and other social, economic and political variables. The overall quality of life model and the influence from the external factors create a flexible model in which the relationships among the different parts are not fixed. Moons et al. described an eight-point list of different conceptualizations of quality of life present in the biomedical and nursing field: normal life, social utility, utility, happiness/affect, life satisfaction, satisfaction with specific domains, personal goal achievement and natural capacity.

A part of the confusion in the use of the concept quality of life concerns the inconsistent use of quality of life, health status and functional status as interchangeable concepts. This confusion is reflected in the fact that instruments (i.e., questionnaires) of health status are being used to measure quality of life or health-related quality of life.

Health-related quality of life
If quality of life is the umbrella term for describing the subjective values of the “good life” for the individual, then health-related quality of life is the operationalization of the concepts of quality of life and health into something objectively measureable. The intention associated with health-related quality of life was to narrow the focus to the effects of health, illness and treatment on quality of life. Health is related to negative aspects of life and death as well as positive aspects such as happiness. According to Guyatt et al. the term “health-related quality of life” is used because widely valued aspects of life exist that are not generally considered “health”; these values include income, freedom, and the quality of the environment. The effect of the other factors described above might adversely affect health. The problems do not have to be related to health or medical concerns. The definition provided by Guyatt et al. differs from that provided by Ferrans et al., who stated that health-related quality of life should exclude aspects of quality of life that are not related to health such as political, cultural or societal attributes. Fayers and Machin provided yet another definition of health-related quality of life, and concluded (as others have) that it is a loose definition. According to Fayers and Machin, the divide between quality of life and health-related quality of life concerns whether it is aspects that are affected by disease or treatments that are of interest (e.g., in clinical trials or clinical medicine). A general agreement exists that aspects can vary across studies, but health-related quality of life often include general health, physical functioning, physical symptoms and toxicity, emotional functioning, cognitive functioning, role
functioning, social well-being and functioning, sexual functioning and existential issues\textsuperscript{50}. In revising a previous conceptual model of health-related quality of life, Ferrans et al.,\textsuperscript{54} focused on five types of measures to be included in health-related quality of life outcomes. These measures are somewhat consistent with those that Fayers and Machin\textsuperscript{50} included in their definition. The five types are

“First, biological function (originally biological and physiological variables) in described as focusing on the function of cells, organs, and organ systems. Biological function would be assessed through such indicators as laboratory tests, physical assessment, and medical diagnoses. Second symptoms (originally symptom status), refers to physical, emotional and cognitive symptoms perceived by a patient. Functional status, the third component, is composed of physical, psychological, social, and role function. Fourth, is general health perceptions, which refers to a subjective rating that includes all of the health concepts that precede it. Fifth, overall quality of life, is described as subjective well-being, which means how happy or satisfied someone is with life as a whole.”(p. 338)\textsuperscript{54}

Ferrans et al.,\textsuperscript{54} argued that this revised model contributes to the understanding of the concept of health-related quality of life in nursing and healthcare because the conceptual confusions that exist do not clearly define what “health-related” includes.
The disability discourse

What has been regarded as a disability has changed throughout history and has often accompanied other changes in society, religion, politics and the view regarding what constitutes a human being. This change has also reflected the terminology used when describing people or groups with impairments. The United Nations stated that

“Disability is an evolving concept and that disability results from the interaction between persons with impairments and attitudinal and environmental barriers that hinders their full effective participation in society on equal basis with others.”

From a historical perspective, the medical/individual model predominated. This model asserts that the disability lies within the individual, and the efforts of society (i.e., medicine) should address, cure or treat of the individual. A deviation from normality was regarded as impairment. During the 1960s and 1970s, a movement that contrasted the medical perspective toward people with impairments began in the United Kingdom. This perspective came to be known as the social model of disability. Here, the emergence of disability concerns how resources in society are distributed. Shakespeare described the social model as a movement that promotes a shift in the thinking of people with disabilities.

“Rather than seeing people with disabilities in terms of their medical condition, rather than having doctors as the experts on disability, rather than thinking in terms of treatment or cure, society should remove barriers and accept that impairment is part of human diversity.”

The critique of the social model of disability has primarily concerned its understanding as something detached from impairment, which is considered a reduction of the understanding of the relationship between disability and impairment.

Another way of understanding of impairment and disability is found within the cultural perspective in which impairment is considered an individual variation in functioning. This perspective is related to discussions regarding normality, gender, sexuality and ethnicity. The relative perspective, has a long tradition within Swedish disability research; it is the view that a variation of functioning in relation to the society can lead to the occurrence of disability, and through efforts targeted both toward society and the individual, this disability can be diminished. The relative perspective has been recently complemented by the WHO’s ICF.
the relative perspective and the ICF are rooted in or were developed under interdisciplinary or multidiscipline settings\textsuperscript{129}. Thus, impairment and disability are possible to discuss from individual and environmental settings as well as across different dimensions both vertically and horizontally. Bhaskar and Danermark described the postulation regarding how to understand a phenomenon or reality as a laminated system that reveals mechanisms that affect people with disabilities in general\textsuperscript{11}. Through the use of a laminated system, a non-reductionist perspective is possible to gain knowledge as well as the epistemological assumption regarding how to explain society and the behaviors of human beings. Within the field of disability sciences, the layers are biological, psychological and social. These are not fixed layers; thus, they can be divided further depending on the empirical question\textsuperscript{111}. 
Disability and Health

Conducting research about people with disabilities and their health is challenging because these people are just as heterogeneous as any other group of humans. Research on comorbidity of people with disabilities has found an increased risk of poor health. Worldwide, 650 million people are estimated to live with disabilities, and in Sweden, between 1.3 and 1.8 million people live with disabilities. Because of differences in definitions and self-report-based statistics, medical diagnoses, work ability and other parameters the number differ across studies. Shakespeare stated that disability cannot simply be equated with impairment and that disability is far more than just a health issue. Impairments often contribute to the disadvantages and difficulties experienced by individuals with disabilities. People with disabilities have healthcare needs that are usually greater than those of the general population; if these needs are neglected, then their quality of life will suffer. Moreover, it will become difficult and sometimes impossible for them to attain their human rights. The health risks among people with disabilities include becoming more vulnerable to age-related conditions, assuming in more risky behaviors, and becoming more at risk for violence or injuries. In some contexts, the term “adjusted disability years” is used. This term implies that disability is defined in terms of a decline in health; thus, it does not capture the complexity of living with a disability.

The WHO’s European Review of Social Determinants of Health revised the gap between health for different groups. The social determinants of health describe the relationship between individual lifestyle factors and interactions at the group level and, in society with regards to living conditions, the environment and socioeconomic factors. Marmot et al. conclude that Europe has made remarkable progress in improving the living conditions for their residents. However, inequalities remain among countries; these concerns include education and healthcare among others, and these must be addressed. In Sweden, research is showing that the health of the general population is improving, although this improvement is unequally distributed. Historically, people with disabilities have not been a focus of the public health discourse. Bickenbach stated that “there is perhaps no pair of concepts more controversial than those of “health” and “disability” (p. 822). However, the status quo is slowly changing, and the health of people with disabilities is an emerging concern within this field. Lollar stressed the importance of viewing people with disabilities, their health and living conditions from a public health
perspective. The field of disability has requested that disability and public health intersect, meaning that both disciplines are motivated to close the divide between public health and disability.

Several studies have reported that people with disabilities have more health-related problems, are more prone to early deaths, have more other chronic conditions, and have more unmet healthcare needs than the general population\(^1,80,126\). Over time disabilities have been considered equivalent to chronic health conditions. This perspective remains present in studies that combine respondents with chronic health conditions (e.g., heart disease) and those with disabilities (e.g., functional limitations) in a single group\(^80\). However, disability can be both a risk factor for chronic health conditions as well as the outcome of living with such conditions\(^80\), this is referred to as a secondary condition\(^93\). The disability can be either from birth or acquired later in life. Furthermore, the secondary condition might be more severe than the primary impairment, and this condition might increase the level of dysfunction. Many secondary conditions are preventable and knowledge about the primary impairment has implications for the development of both prevention and healthcare\(^93\).

The Swedish Public Health Agency described the self-rated health and living conditions of people with impairments in Sweden\(^142,143\). The study of health and living conditions for people with impairments living in Sweden has been conducted once and the identification of person’s with impairments are based on self-reports of different conditions (e.g. hearing impairment, vision impairment, mobility)\(^142\). The health of people with impairments in Sweden was worse than of the general population. The authors concluded that the majority of poor health found concerned unequal living conditions and not the actual impairment\(^142,143\). Danermark and Hanning described the hearing and vision of people living in Sweden using the “Health on Equal Terms” questionnaire\(^38\). They found that people who self-report hearing difficulties (i.e., difficulties hearing what is said in a conversation, regardless of hearing aid use), were more likely to report poor mental health and pain than people without any hearing difficulties\(^38\). When studying the quality of life of people with hearing impairments, Fellinger et al.,\(^52\) found that people with hearing impairment had worse social situations than those who were deaf but using sign language and those with normal hearing.

A cross-sectional study of 70- and 77-year olds who self-reported visual impairments were examined for declines in visual acuity; significant problems with poor health were found compared to persons of same age with
no visual impairment. The participants with vision impairment reported having more problems with both fatigue and performing activities of daily living. They reported to being lonely and had used health services significantly more than those with better vision. The study was longitudinal, and a higher mortality rate was related to poor self-reported health at the seven-year follow-up assessments.

Chia et al., used the Blue Mountains Eye Study to investigate health-related quality of life in 49 to 98-year olds. They found that people with no correctable visual impairment had significantly poorer health related quality of life scores compared to those without visual impairment. Comparisons with other medical conditions were performed, revealing differences with regard to physical and mental health. The effect of poor vision was more severe than that of the other conditions (arthritis, asthma, stroke, diabetes, heart attack and angina). Langelaan et al. compared people with visual impairments with those with other chronic conditions and a healthy reference. They found that the former group (i.e., people with visual impairments) reported poorer quality of life than did a healthy reference group. Compared with other chronic conditions, the overall quality of life was not reported as worse. In that study, people with chronic fatigue syndrome and stroke reported poorer quality of life.

**The disability paradox**

The definition of quality of life as a subjective appraisal of one’s life condition and a purely subjective experience is in line with the understanding of that people with disabilities report high quality of life, despite their impairments or medical conditions. For the outside observer, this finding can be difficult to understand assuming that people with disabilities live incomplete lives. Alternatively as Amundson stated, “disabled and non-disabled people have very different assessments of the quality of disabled persons life” (p. 102). Ereshefsky argued that the terminology “able-bodied” and “disabled” implies an advantage for the former with regard to values and medical descriptions.

The discussion of people with disabilities and high quality of life is usually referred to as “the disability paradox”. The disability paradox describes the disparity between objective conditions and subjective experiences. It stresses the importance of the personal experience with disability in defining the self, one’s view of the world, social context and social relationships. Factors that contribute to the disability paradox concern how the surrounding environment views disabilities, negative attitudes,
judgments by the general public, and unequal positions in society. As Albrecht and Devlieger\textsuperscript{3} described, quality of life refers to the holistic notion of well-being that refers to an experience beyond the activities of daily living or disease, categories that include a more comprehensive understanding of the social, psychological and spiritual being. In this context, disability should be addressed using a salutogenic perspective, avoiding the pathological consequences of a medical condition. Antonovsky developed a salutogenic perspective of health in the 1970s. According to Antonovsky, health is considered as a continuum with total wellness and total disease as its endpoints\textsuperscript{7}. Distinguishing among health, functioning and disability is crucial for how these concepts are viewed, and this act has consequences for how they are measured\textsuperscript{80}. Self-reported quality of life among people with significant disabilities can greatly differ from ratings of others, leading to the disability paradox\textsuperscript{3}. The effect of a disability can be significant for the individual, the family network and society. Disabilities challenge preconceived expectations of what is considered as normal. Disabilities also challenge the values and notions of well-being\textsuperscript{3}.

Constructions of normality and what is regarded as normal and as having a normal body are what constitute the differences between people with impairments and “others”\textsuperscript{148}. People with disabilities are to be regarded as a heterogeneous population. They do not necessarily share common religions, political beliefs and social classes. They can also differ with regard gender, age, ethnicity, sexuality, living region, partnership status and health. This must be kept in mind, also when it comes to people with USH.

Fellinghauser et al.,\textsuperscript{53} explored the “disability paradox” in relation to the general population of Switzerland using the ICF. These authors concluded that environmental and personal factors play significant roles in how people perceive their health and quality of life. Limitations in activity and participation negatively affected how people perceive their health; however, having an impairment was not directly related to how one perceive his or her health\textsuperscript{53}.
Health-related research regarding deafblindness and Usher syndrome

USH is one of the most investigated genetic deafblind syndromes, and a vast body of research describes the genetics of USH\textsuperscript{44, 70, 78, 81, 95}. The importance of creating a public health perspective for deafblindness and Usher syndrome has not been accomplished, although it has been stressed from different points of view\textsuperscript{36, 98, 111}. In his review of the present research on deafblindness, Dammeyer\textsuperscript{36} concluded that it is a condition that can lead to a numerous of health-related difficulties such as, depression, cognitive decline and psychological distress. Möller\textsuperscript{111} addressed deafblindness from public health and health threats perspectives, suggesting that the lack of adjusted information in public health matters among institutions in society such as food recommendations, different warnings (e.g., alcohol/drug use, notes on medicine and more) are problematic and severe for individuals with deafblindness\textsuperscript{111}. Furthermore, Millán et al.,\textsuperscript{98} indicated the importance of early diagnoses in USH as a public health issue, this because of heterogeneity within the group and the social withdrawal that people with USH can experience.

Researchers, even when not specifically discussing USH, have described the psychosocial consequences for people with deafblindness, including anxiety, feelings of isolation, withdrawal and depression\textsuperscript{18, 25, 68, 111}. Saunders and Echt's\textsuperscript{135} findings show that older adults with deafblindness report psychological and social problems, restricted activities of daily living (ADLs), and problems with cognitive functioning and communication. Dalby et al.,\textsuperscript{32} also discussed the risk of experiencing limitations in ADL among people with deafblindness. Bodsworth et al.,\textsuperscript{18} investigated psychological distress among people with deafblindness. The findings of 539 individuals revealed that the percentage of people with deafblindness who met the criteria for psychological distress or were unable to cope with stressful situations was higher than that in the general population and among those who had either hearing or visual impairments\textsuperscript{18}. Numerous studies have attempted to establish the relationship between psychiatric diagnoses and USH; these studies are often based on single cases or members within the same family\textsuperscript{41, 42, 125, 158}. Another example of this limitation is the Dammeyer\textsuperscript{35} study that suggested a higher prevalence of psychiatric illness exists among people with USH\textsuperscript{1}.

Miner\textsuperscript{99, 100} interviewed people with USH\textsuperscript{1} and USH\textsuperscript{2} about their experiences of living with USH from a lifespan perspective and revealed psy-
cholesterol and emotional distress such as depressive episodes, grief, guilt, loneliness and suicidal behavior in the stories told by the participants. The experiences told by the participants were related to time of diagnosis, when life changes occurred because of the progression of RP and participants strived for independence.

The participants’ stories not only concerned themselves but also related their families and friends. Miner wrote that “love, respect, meaningful relationships, and a sense of belonging are universal human needs” (p.13). This conclusion can be understood in relation to Danemark and Möller’s discussion on social recognition and the ontological security of people with deafblindness. To be socially recognized means to be acknowledged as a human being with certain needs and wishes. This recognition is considered a process that contains three types of personal characteristics, namely, self-confidence, self-respect, and self-esteem. Giddens defines ontological security as a sense of order and continuity in an individual’s experiences, arguing that this security relies on people’s ability to provide meaning to their lives. Meaning is found when experiencing positive and stable emotions as well as when avoiding chaos and anxiety. If an event occurs that is not consistent with the meaning of an individual’s life, then it will threaten the individual’s ontological security. Ontological insecurity (i.e., a lack of a deep, inner feeling of security, due to the absence of a sense of continuity in the events of one’s life) might be experienced by people with USH when adapting to changes in vision or hearing impairment is necessary.

An increased risk of mortality was found among people in United States and Australia who self-reported deafblindness.

Several studies have reported that a compromised communication situation can be present for people with deafblindness and USH. Hersh found barriers due to poor knowledge regarding how to communicate. The twenty-eight people with deafblindness who participated in that study also described problems regarding how to live independently and experiences of isolation. Heine and Browning found similar findings, reporting that people with deafblindness felt misunderstood and isolated because of communication breakdowns. Other’s attitudes about people with deafblindness are not only related to the deafblindness per se but also to attitudes regarding what a person with deafblindness is capable of doing (i.e., items related to independence and activities). Miner indicated that the progression of RP in people with USH with regard to communication can
lead to feelings of incompetence when (for example) sign language is no longer possible to see or when lip-reading becomes difficult.

The Usher lifestyle survey\textsuperscript{33} reported challenges maintaining independence with regard to information, communication and mobility when sight and hearing deteriorate. Schneider\textsuperscript{136} found restrictions in participation with regard to the environment often caused by a lack of information on the effects of being deafblind. The participants in her study referred to interacting with the surrounding environment and others while deafblind as “negotiating a place in a hostile world”\textsuperscript{136}.

Henricson et al.,\textsuperscript{69} investigated the cognitive and reading skills of adults with USH2. Their findings indicated that people with USH2 are similar to those with long-term hearing impairment with regard to phonological processing and phonological working memory. Compared with individuals with normal hearing and vision, people with USH2 performed significantly poorer and had longer reaction times. Henricson et al.,\textsuperscript{69} suggested that some of these results might affect the health and wellbeing of people with USH2, and a need exists for more research regarding the communication strategies of this group. In their review on communication and psychosocial consequences Heine and Browning\textsuperscript{68} concluded that the communication strategies of older adults with dual sensory loss must be addressed in a multidisciplinary rehabilitation setting. The communication challenges and psychosocial burden that they revealed were confirmed by later studies\textsuperscript{26, 135, 137}.

Few studies have examined the effects of CIs on people with USH. Damen et al.,\textsuperscript{34} showed that improvements with regard to certain hearing-related tasks (e.g., sound perception and social interaction as well as access to information and activity) were found for those with USH1 who had received CIs compared with the non-implanted. However, no differences in health-related quality of life were identified between people with CIs and those without. Damen et al.,\textsuperscript{34} found improvements in audiological performance among people with USH1. Their findings also showed that implantation at an early age is beneficial for this group. People with USH3 reported their perceived effects after receiving a CI\textsuperscript{120}; they stated that their health had improved after receiving the implant and that they could administer the device by themselves. However, the variation within the group was vast\textsuperscript{120}. Important factors regarding the outcomes of implantation were age at implantation, individual variations in hearing, and other individual factors. A study that addressed people with deafblindness but not specifically people with USH found positive effects associated with
CI, especially among those who had used speech to previously communicate. The benefits associated with CI included environmental sounds and safety\textsuperscript{141}, something that might be important to compensate for visual impairment.

Bodsworth et al.,\textsuperscript{18} asked participants with deafblindness about the support that they received from their families and friends as well as from healthcare or other support providers; most of the respondents replied that they received support from their families and friends. However, they requested more formal support from healthcare and other service providers with regard to diagnosis and assistance with both practical and emotional consequences\textsuperscript{18}. A fragmented healthcare that lacks coordination, thus, resulting in a time-consuming experience, was reported in a biopsychosocial study on deafblindness\textsuperscript{109}. Furthermore, when women with USH1 described their contact with low vision clinics and ophthalmology departments, they reported a healthcare system that was substantially insufficient and uncoordinated\textsuperscript{112}. A study of older adults with deafblindness and their contacts with health care found that support and services were often offered based on a single impairment (i.e., hearing or vision) but not the combination of the two\textsuperscript{137}.

To conclude this section on the previous health-related research on deafblindness and people with USH, much of the research that exists describes the challenges or difficulties (e.g., anxiety, depression, social withdrawal, and barriers to communication and independence) of living with both vision and hearing loss. The consequences of these impairments affect many aspects of being human. However, no study has addressed the biopsychosocial health of people with USH.
Aims

Biological and psychosocial paradigms are part of the discourse and development of both disability and health. Health is viewed as a complex phenomenon, and none of biological, psychological or social variables can be disregarded; the ability and intention of the individual must be addressed in this context. To live with a progressive disease such as USH can have affect one’s life experience at different levels throughout life; to regard life from only one perspective (e.g., biological or psychological or social) would be to reduce complex life situation. Mechanisms at different levels interact and can have consequences for people with Usher syndrome and their health from a lifespan perspective. This thesis takes interdisciplinary (biopsychosocial) approach to describe the health of people with USH. The present thesis investigated variables related to health within general health, physical health, social trust and finances. Hence, the overall aim of the present thesis was to describe health of people with Usher syndrome.

The aims of the following four studies are

1. To describe the physical and psychological health of people with USH2, and to explore if there were any differences in terms of gender.
2. To describe health and social trust in people with USH3 in relation to the degree of hearing, and vision impairment.
3. To describe the physical and psychological health, as well as social trust and financial situation of people with USH1 in comparison with a cross-section of the Swedish population.
4. To describe the similarities and differences in health and social trust among people with USH types 1, 2 and 3.
Methods

Study designs

Four studies are included in the present thesis. All four studies apply a quantitative, cross-sectional approach using two different questionnaires: “Health on Equal Terms” and “the Hospital Anxiety and Depression scale” (table 1). These surveys are described below.

The first cross-sectional study (I) compares people with USH2 with a Swedish reference group using descriptive method to evaluate self-assessed general health, physical and psychological health.

The second study (II) describes self-assessed general health, physical health, psychological health and social trust of people with USH3.

The third comparative cross-sectional study addressed people with USH1 with regard to self-assessed general health, physical health, psychological health, social trust and financial situation (III). A cross-section of the Swedish population was used as reference group.

The fourth study (IV) makes an in-group comparison of self-assessed general health, physical health, psychological health, social trust and financial situation.
Table 1. Design, materials, included data and analysis of the four studies (I-IV).

<table>
<thead>
<tr>
<th>Study</th>
<th>Design</th>
<th>Materials</th>
<th>Included data</th>
<th>Analysis</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Quantitative</td>
<td>Study population: 96 persons with USH2 Age 18-84 Mean age 55 51 women 45 men</td>
<td>“Health on Equal Terms” Questions pertaining to: General health Physical health Psychological health</td>
<td>Frequencies Chi²-test Logistic regression</td>
</tr>
<tr>
<td></td>
<td>Descriptive</td>
<td>Reference group: 5738 persons Age 16-84 Mean age 49 3213 women 2525 men</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Comparative</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cross-sectional</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>II</td>
<td>Quantitative</td>
<td>Study population: 15 persons with USH3 Age 19-71 Mean age 41 11 women 4 men</td>
<td>“Health on Equal Terms” Questions pertaining to: General health Physical health Psychological health Social trust Financial situation</td>
<td>Descriptions of: Number of problems Vision loss Hearing loss Cochlear implant Age Sex</td>
</tr>
<tr>
<td></td>
<td>Descriptive</td>
<td>Reference group: 55738 persons Age 16-84 Mean age 49 3213 women 2525 men</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Comparative</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Cross-sectional</td>
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</tr>
<tr>
<td>III</td>
<td>Quantitative</td>
<td>Study population: 60 persons with USH1 Age 20-79 Mean age 49 36 women 24 men</td>
<td>“Health on Equal Terms” Questions pertaining to: General health Physical health Psychological health Social trust Financial situation</td>
<td>Frequencies Chi²-test Logistic regression</td>
</tr>
<tr>
<td></td>
<td>Descriptive</td>
<td>Reference group: 5738 persons Age 16-84 Mean age 49 3213 women 2525 men</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Comparative</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Cross-sectional</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td>Quantitative</td>
<td>Study population: 162 persons with USH1, 2 and 3 Age 18-84 Mean age 51 91 women 71 men</td>
<td>“Health on Equal Terms” Questions pertaining to: General health Physical health Psychological health Social trust Financial situation “Hospital Anxiety and Depression Scale”</td>
<td>Frequencies Chi²-test Kruskal Wallis one way analysis of variance Logistic regression</td>
</tr>
<tr>
<td></td>
<td>Descriptive</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Comparative</td>
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<tr>
<td></td>
<td>Cross-sectional</td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>
The Swedish Usher database
The Swedish Usher database at the Audiological Research Center at Örebro University hospital has been collecting data for approximately 30 years. Professor Claes Möller created the database. The database includes data from individuals with all three types of USH. The inclusion criterion is to have a clinical diagnosis of USH. The clinical diagnose is based on objective observations of hearing, vision and balance. The database includes data on genetics, hearing loss, vision loss (i.e., visual field, visual acuity and cataract), family history and other medical records. The database is updated on a regular basis and includes data from approximately 400 individuals. Thus, the database most likely includes approximately 60 percent of the known population with USH in Sweden.

Assessment of hearing impairment
Data on hearing loss were retrieved from the database. The Swedish Usher database has been collecting audiograms and other information about the hearing loss of people with USH over the last 30 years. In the present thesis, the pure tone average of four frequencies (PTA4) were used as a measure of the degree of hearing loss, and all of the data on hearing loss were reported as close to the time of the dispatch of the questionnaires as possible. Thresholds were classified from mild to profound hearing loss (Mild over 20 dB and less than 40 dB; Moderate, over 40 dB and less than 70 dB; Severe, over 70 dB and less than 95 dB; Profound, equal to and over 95 dB) according to the European standard [145].

Assessment of vision impairment
Data on vision impairment (i.e., visual field and visual acuity) were retrieved from the database. The data used in the present thesis were chosen to be as close in time to the distribution of the questionnaires. Visual field tests were assessed via Goldman perimetry, and the results were categorized into five phenotypes (1-5), where 1 denoted normal vision, 2 denoted visual field with partial or complete ring scotoma, 3 denoted a concentric central field loss with a remaining peripheral island, 4 denoted a concentric loss and a visual field of ≤ 10°, and 5 denoted blindness [64]. Visual acuity was measured using the Snellen chart-based standard test [124]. The results for the best eye with correction were used to evaluate visual acuity function. Visual acuity ranged from 1.00 (perfect visual acuity) to 0.00 (blindness) [64].
Genetics
USH has been thoroughly genetically investigated; currently, 13 genes have been discovered\(^9\). The development of genetic testing and the possibility to identify genes has been remarkable over the recent years. The genetic data of those who have received the genetic testing procedure are included in the database. The numbers of people who have received genetic testing are reported in the studies (I-IV).

Populations

People with Usher syndrome
In the present thesis, people with USH from all three clinical diagnoses were examined. The clinical diagnose is based on observations of hearing, vision and balance. In total, two questionnaires (the “Health on Equal Terms”\(^5\) and “the Hospital Anxiety and Depression scale”\(^1\)) were sent to 230 individuals, of whom 171 responded (60 USH1, 96 USH2 and 15 USH3). The response rate for each USH group was considered as satisfactory with 69 percent USH1, 79 percent USH2 and 71 percent USH3. As table 2 shows, more women than men answered the questionnaires. The mean age and age range between the three USH groups slightly differed, with people with USH2 having the oldest mean age (55 years) and the widest range 18-84 years. People with USH3 were the youngest, with a mean age of 41 years and an age range of 19-71. People with USH1 had a mean age of 49 and an age range of 20-79. The percent that had a genetic diagnosis at the time of answering the questionnaires ranged from 43-73 (see table 2). All people with USH presented with a mean hearing loss that ranged from severe to profound. They also presented with severe visual field and visual acuity loss when answering the questionnaires (see table 2).
Table 2. People with USH included in the present thesis.

<table>
<thead>
<tr>
<th></th>
<th>USH1</th>
<th>USH2</th>
<th>USH3</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number</td>
<td>60</td>
<td>96</td>
<td>15</td>
</tr>
<tr>
<td>Women</td>
<td>60%</td>
<td>53%</td>
<td>73%</td>
</tr>
<tr>
<td>Mean Age (years)</td>
<td>49</td>
<td>55</td>
<td>41</td>
</tr>
<tr>
<td>Age (min-max)</td>
<td>20-79</td>
<td>18-84</td>
<td>19-71</td>
</tr>
<tr>
<td>Clinical Diagnosis</td>
<td>60</td>
<td>96</td>
<td>15</td>
</tr>
<tr>
<td>Genetic Diagnosis</td>
<td>43%</td>
<td>59%</td>
<td>73%</td>
</tr>
<tr>
<td>Hearing Impairment dB Mean (best ear)</td>
<td>99 dB</td>
<td>73 dB</td>
<td>99 dB</td>
</tr>
<tr>
<td>Visual Field Mean (1-5, best eye)</td>
<td>3</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>Visual Acuity Mean (1.0-0.0, best eye)</td>
<td>0.5</td>
<td>0.4</td>
<td>0.7</td>
</tr>
</tbody>
</table>

Reference population
We had the opportunity to compare the results from the USH population with raw data of a cross-section of the Swedish population who had answered the Health on Equal Terms questionnaire through the Public Health Agency of Sweden\textsuperscript{122, 143, 144}. This reference population includes people living in Sweden in 2007, and hence included individuals with and without impairments. The data of the reference group was taken from an external database administered by the Public Health Agency of Sweden\textsuperscript{144}. This group consisted of a simple random sample of the total population aged 16-84. The sample was identified by the Total Population Register and consisted of 10,000 people living in Sweden whose identification was verified to obtain updated addresses. The respondents answered the questionnaires either by mail or an Internet link \textsuperscript{144}. The reference group consisted of 5738 individuals with a mean age of 49 years (range 16-84 years). The female/male ratio was 56%/44%. Comparisons with the USH population were performed in studies I and III.
Questionnaires

Health on Equal Terms
The “Health on Equal Terms” (HET) questionnaire is a generic instrument aimed at the whole population. The purpose of the HET (created by the Public Health Agency of Sweden) is to describe patterns and changes in health within Swedish population over time. The HET has been administered annually in Sweden since 2004. The questionnaire has not previously been used for people with USH.

The complete questionnaire was not used in the present thesis. The questions included pertain to the general health, physical health, psychological health, social trust and financial situation of the participants.

General health domain
The general health domain includes questions that address “health” as an umbrella term for health and wellbeing. The first question assesses the participant’s current health on a five point scale from very poor to very good. The following questions concern how many days over the previous 30-day period the participant experienced poor physical and mental health and how many days capacity for work and activities of daily living (ADLs) were affected as a result of poor physical health and psychological health; ≥15 days indicates poor physical health, psychological health, and restricted capacities for work and ADL.

Physical health domain
The questions regarding physical health relate to different types of pain (i.e., pain in neck and shoulders, back pain, pain in extremities and headache). Physical health questions inquire about difficulties with tinnitus, bowel trouble, overweight and incontinence. The questions are to be answered on a three-point scale ranging from “no” to “yes, great discomfort”. Questions are asked about if having problems with diabetes, asthma, allergy or high blood pressure. The questions are to be answered on a four-point scale ranging from “no” to “yes, great distress”.

Psychological health domain
The psychological health domain includes questions related to fatigue, sleeping problems, and worry or anguish, and they are answered using a
three-point scale from “no” to “yes, great discomfort”. The psychological domain includes 12 questions about abilities over the past few weeks. The 12 questions were taken from the General Health Questionnaire-12 (GHQ-12), which was constructed in the early 1970s to identify psychiatric illness and the capability to cope with new, stressful situations. The questionnaire has been widely used both in healthy populations and clinical groups. Different versions exist that include different numbers of questions. The questions are summarized in an index, where cut-offs indicate “problem” or “no problem”. The max score is 12, and the cut off is set at ≥3 which indicates poor wellbeing. The questions ask regarding; being able to concentrate, having feelings of worthlessness, inability to appreciate the day and so on. Answers are given on a four-point scale from “not at all”, “no more than usual”, “more than usual” and “much more than usual”. In the present thesis the single questions have been used (study I-IV).

Furthermore, a question about stress is included in the HET, formulated as “Do you feel stressed at present?” this was to be answered on a four point scale from “not at all” to “very much”.

The psychological health domain included two questions concerning suicidal behavior including “Have you at any time found yourself in a situation in which you have seriously considered taking your own life?” and “Have you ever tried to take your own life?” The possible answers were “no”, “yes, once” and “yes, several times”.

Social trust and financial situation domains
Questions related to social relationships and trust are included within the social trust domain. These questions include “Do you ever refrain from going out alone for fear of being attacked, robbed or otherwise molested?”, the question could be answered on a three-point scale from “no” to “yes often”; “Have you been subjected to physical violence over the past 12 months?”, and “Have you been subjected to threats of physical violence so that you became frightened over the past 12 months?”, the answer could be “yes” or “no”. Finally, “Have you been treated or received in such a way that you have felt wronged over the past three months?”, the answerer could be given on a three-point scale from “no” to “yes, several times”.

Furthermore, questions about having anyone to share their innermost feelings and confide in, the possibility of obtaining help if needed and believing that one can trust most people were asked. Answers were given
as “yes” or “no” for the first question but on a four-point scale “yes, always” to “no, never” for the second question. The last question was answered as “yes” or “no”.

The social trust domain includes two questions regarding financial situation. The questions asked were “Should you suddenly find yourself in an unforeseen situation in which you had to get hold of 15,000 Swedish Crowns in a week, could you manage this?” (answered as “yes” or “no”) and “Has it happened during the past 12 months that you have had difficulty in managing your current expenditure for food, rent, bills and so on?” (answered using a three-point scale ranging from “no” to “yes, on several occasions”).

In addition to the questions related to general health, physical health, psychological health, social trust and finances, demographic questions (e.g., sex and age) were asked in the present thesis.

**Development and testing of the HET questionnaire**

Most of the questions in the HET were taken from previous county surveys in Sweden or the Statistic Sweden’s surveys Living Conditions in Sweden (SLC). A method group at Statistic Sweden’s measurement laboratory tested and revised the questions included in the HET. According to Fayers and Machin, the validation process of an instrument includes several steps and contains different aspects of validation. The HET has shown to have a good construct validity and metric capacity to discriminate between different latent variables when used in the Swedish population. To determine the construct validity of a question, each question in the questionnaire was theoretically reviewed regarding its intent to be measured and whether the question generates meaningful correlations to health, gender, age and socioeconomic status. Furthermore, known group validation was of interest. Known group validity addresses the expectation of between-group (e.g., patient groups) differences; the instrument should be sensitive in this aspect. In the present thesis, self-assessed health was expected to differ between people with USH and the reference group, based on clinical experience of the group.

Reliability addresses the repeatability of the measurement and how reproducible and consistent the results are. This study marks the first time that the HET questionnaire was used to describe health of people with USH, and their general health, physical health, psychological health, social trust and financial situation has been sparsely reported previously. To scrutinize the consistency of the questionnaire in this group of people fur-
ther, repeated data collections are required. However, the response rate was satisfactory for all three groups of people with USH who received the questionnaire (table 2).

**The Hospital Anxiety and Depression Scale**

The Hospital Anxiety and Depression Scale (HAD-scale) was developed at the beginning of the 1980s as a screening tool to detect symptoms of anxiety and depression\textsuperscript{161}. The purpose of the questionnaire was to identify anxiety and depression in patients who sought treatment for physical somatic complaints. Patients who reported these complaints were found to have problems with anxiety or depression. The HAD-scale includes 14 questions; seven pertain to anxiety, and seven consider depression. Questions about the emotions of anxiety and depression were formulated as “I feel tense or wound up” and “I still enjoy the things that I used to enjoy”\textsuperscript{161}. In the process of psychometric testing of the instrument, the psychiatric rating was evaluated to determine the scores that were sufficient to make assumptions of the presence of anxiety or depression. Seven or fewer points was considered as non-cases, 8-10 points was considered as probable cases and 11 or more was considered as cases that needed treatment\textsuperscript{161}.

**Development and testing of the HAD-scale**

The HAD-scale was constructed so that somatic symptoms were not included, only those related to the emotional aspects of anxiety and depression\textsuperscript{50, 161}. Numerous of premises were established when the questionnaire was constructed. These premises were that the instrument should be brief, clearly distinguish between anxiety and depression, and not include any somatic symptoms such as headache or dizziness. The initial scale included two subscales related to either anxiety or depression with eight questions each. Responses were graded on a scale ranging from 0-4\textsuperscript{161}. Internal consistency of the subscales was controlled and one item in each subscale was excluded. These items were excluded because a weak item was found in the depression subscale and, to keep a balanced instrument, one item was therefore excluded from the anxiety subscale\textsuperscript{161}.

This questionnaire has been widely used in both clinical settings and research among, the general populations and clinical groups\textsuperscript{5, 15, 16, 105, 106}. In Sweden, the reference material was taken from Lisspers, Nygren and Söderman\textsuperscript{89}. In Sweden the prevalence of possible cases of anxiety and depression are 12\% and 9\%, respectively; the prevalence of definite cases
of anxiety or depression are 8% and 6%, respectively. These figures are based on 624 thirty to 59-year olds living in the county of Jämtland. The HAD-scale has also been used to generate norm data concerning anxiety and depression for the general population in UK.

Procedure

Data collection
The data of the questionnaires were collected on two separate occasions. In 2008 people with USH2 and USH3 answered the questionnaires. At this point, the HET was not adjusted to increase visibility; however, some adjustments were made to the HAD-scale, (e.g., a larger font was used). In 2012, people with USH1 answered the questionnaires. The reason for not including them in the first dispatch was that the questionnaires were adjusted to be accessible to people who do not use spoken or written Swedish as their first language. This was to improve the response rate. Most adults with USH1 in Sweden use sign language as their first language. To our knowledge, no adjustments were previously conducted to improve accessibility for people with vision losses and sign language. Therefore, in practice this group of potential respondents has been excluded from surveys to a large extent. The procedure to adjust the questionnaires is described below. At the dispatch of the questionnaires people with USH1 received the questionnaires in written form, on a memory stick and on a DVD; they could answer by filling out the questionnaires on paper or the memory stick provided. To return the questionnaires people with USH1 could send it by post or by attached in an email to one of the researchers (i.e., Moa Wahlqvist). Most participants with USH1 returned the questionnaires by post, either on paper or by returning the memory stick; a few participants choose to send their answers attached in an email.

Adjustments of the questionnaires*

Accessibility for those who use Swedish sign language
The translation of the text into sign language presents with many challenges, not only because of the translation per se but also because of nec-

* The procedure to adjust the questionnaires has been derived from the study “Physical and Psychological health, social trust and financial situation in persons with Usher syndrome type 1” (Wahlqvist et al., 2015, accepted).
ecessary linguistic and cultural adjustments\textsuperscript{102}. The questionnaires were translated from written Swedish to Swedish Sign Language (SSL) with the help of a professional SSL interpreter who had expertise with people with deafblindness. The researchers and the interpreter discussed the translation process (i.e., the choices of signs for different concepts and words). One of the researchers (M. Wahlqvist) is bilingual (Swedish and SSL). Following the translation process a professional filmmaker filmed the signed questionnaires in a studio. In addition to the questionnaires, a short presentation providing information in SSL about the research project, previous studies, the Swedish Usher Database and informed consent was filmed. In this presentation, two of the researchers (C. Möller and M. Wahlqvist) provided the information in spoken Swedish with a simultaneous translation into SSL. Additional information about how to return the questionnaires was provided in SSL by one of the researchers (M. Wahlqvist). Adjustments were made to the background as well as the clothing and make-up of the researchers and interpreter in to improve contrast. The filmed material was then produced on a DVD. A screen with a yellow background and black text appeared between each question on the DVD to indicate the following question, and the interpreter repeated the same information in SSL.

**Visible accessibility including Braille**

The written material was also adjusted to make the questionnaires accessible to those who use Braille script readers via a computer. Thus, all text was ordered in rows, and no checkboxes or columns were used. The questionnaires and information about the study were saved on a memory stick that, allowed respondents who used a magnifier on their computer to access the material. Most people with deafblindness in Sweden have access to a computer or other technical aids to read text. Adjustments of the material were made in hard copy to improve the contrast, such as font type, color, and size. A one-sided print was used.

**Pilot study**

A pilot study of six people with USH1 was conducted to obtain feedback on the material. The six people received the material on DVD, memory stick and on paper. Overall, their comments were positive. Their negative comments concerned the number of questions and the time needed to answer the questionnaires. Nevertheless, the length of the questionnaires was
not adjusted. The people who participated in the pilot study were included in the III.

**Spokesperson**

The people who participated in the pilot study were asked whether they would be willing to act as an information spokesperson in the study. If they consented, then their name and contact details were added to the information provided in the material. As a spokesperson they had to be accessible and answer the questions of other participants about the questionnaire. The intention was to get help to spread information about the study and to make information about the study accessible in different ways.

**Analysis**

The process of coding of variables we have followed the recommendations in the objective and background of the questions in the HET\(^2\) and the technical description of the HET\(^1\). The present thesis and articles describe different aspects of health in people with USH. To do so, descriptive statistics as frequencies, the Chi\(^2\)-test results, and cross tabs were used. To further explore aspects of health, logistic regression was used. In studies I, III and IV, logistic regression were used to evaluate the risk (i.e., the odds ratio(OR)) of a poor outcome based on group (people with USH or reference; studies I and III) and within people with USH exploring independent variables (i.e., sex, age, visual field, visual acuity, degree of hearing loss, and clinical diagnose) (study IV). Logistic regression can be used to predict the probability that an event will occur\(^1\). For group-comparison in study IV, Kruskal Wallis test of one-way variance was used, which is a non-parametric test\(^2\),\(^8\). All analyses were conducted using the Statistical Package for Social Sciences (SPSS) versions 20 and 21.

**Ethical approval and considerations**

In the present thesis including the studies I-IV, the guidelines of the WMA Declaration of Helsinki, Ethical principles for medical research involving humans\(^1\) and the Ethics of research involving humans\(^2\) has been considered. The Ethics Committee of Linköping University Hospital and the Institutional Review Board of the Boys Town National Research Hospital Omaha USA, in 1990 and 1997 approved the use of material in the Usher database for research. All participants with USH have signed informed consent forms to participate in this clinical and genetic research on Usher syndrome. In 2012, the Ethics Committee of Uppsala University approved the translation and the sending of the HET and HAD-scale to people with USH\(^1\) to collect data regarding their health (Dnr 2012/515). In connection of the collection of data (i.e., sending the questionnaires) all people with USH have been informed about that the participation in the study is confidential and voluntary.

People with USH comprise an exclusive and heterogeneous group, and conducting research with this group can be challenging for several reasons. People with USH likely represent approximately 50% of all adults with deafblindness; however, this group can still be relatively small, with an estimated prevalence of 3.3/100 000 in Sweden\(^1\). Thus, only approximately 800 people in Sweden have USH\(^1\). The material that constitutes the empirical data in the present thesis is comprehensive; therefore, it is important to consider the response burden placed on a single individual. The experience of the individual can be vast and might conflict with the need for more knowledge about the life circumstances of people living with USH. This supposition implies that a careful decision-making process must be undertaken because of what is possible with regard to the limitations of the empirical material, methodological challenges, and in relation to what is lacking in the current research and knowledge.

Considerations also include the possibility of being recognized by others or recognizing one's self in the results. This effect might especially be concerning for the only 15 people with USH\(^3\) included. This effect is addressed in the interpretation and reporting of the results. There was also choices made of the researchers not to include detailed data of family circumstances for each individual, and several people have received a CI after answering the questionnaires.

Answering questions about health can be sensitive and evoke additional questions as well as feelings related to what it is like to live with USH. These effects were addressed to stress the importance of the participants’ voluntary participation, and that the decision to participate in the research would in no way affect the support provided by healthcare or other services.

Another ethical consideration concerns how the results are reported to the participants with USH. Throughout conducting the present thesis, it has been important to provide feedback to the participants themselves. The results of this thesis have been presented at different conferences,
consent forms to participate in this clinical and genetic research on Usher syndrome. In 2012, the Ethics Committee of Uppsala University approved the translation and the sending of the HET and HAD-scale to people with USH1 to collect data regarding their health (Dnr 2012/515). In connection of the collection of data (i.e., sending the questionnaires) all people with USH have been informed about that the participation in the study is confidential and voluntary.

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Considerations also include the possibility of being recognized by others or recognizing one’s self in the results. This effect might especially be concerning for the only 15 people with USH\textsuperscript{3} included. This effect is addressed in the interpretation and reporting of the results. There was also choices made of the researchers not to include detailed data of family circumstances for each individual, and several people have received a CI after answering the questionnaires.

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workshops and meetings. This fact represents an ethical concern such that participant feedback is important to gain more knowledge, reveal that others are in the same situation, and show that research is not conducted solely for the sake of the research.

Summary of the studies

Study I

Title: Physical and Psychological health in persons with deafblindness that is due to Usher syndrome type II.

This study sought to describe the physical and psychological health of people with USH2 and explored differences in terms of gender (sex). Only a small body of research has focused on comorbidity and deafblindness. USH has so far not been associated with disorders other than hearing loss, vision loss and (for some) balance problems. No previous study has focused on the health differences between men and women with USH2. A total of 122 adults with USH2 from the Swedish USH database at the Audiological Research Center received the questionnaire HET, and 96 answered and were included in the study. The results of people with USH2 were compared with those of a reference group composed of 5738 people. The reference group was taken from an extant database administered by the Swedish Institute of Public Health and Statistic Sweden.

The current focus was on questions pertaining to general health, physical health and psychological health. Significant differences in both physical and psychological health were found between people with USH2 and the reference group (diagram 1). People with USH2 expressed problems with tinnitus, headache, shoulders and neck pain, and had significantly worse problems with eczema and skin rashes. Psychological health (including fatigue, inability to concentrate, being unable to accomplish things, feelings of worthlessness, and a feeling of being constantly under strain) was significantly worse among those with USH2. Those people with USH2 reported unhappiness and depression as well as anxiety significantly more than those in the reference group. Suicidal behaviors (both thoughts and attempts) were more evident among those with USH2 than the reference group (diagram 1).

Summary of the studies

Study I
Title: Physical and Psychological health in persons with deafblindness that is due to Usher syndrome type II.

This study sought to describe the physical and psychological health of people with USH2 and explored differences in terms of gender (sex). Only a small body of research has focused on comorbidity and deafblindness. USH has so far not been associated with disorders other than hearing loss, vision loss and (for some) balance problems. No previous study has focused on the health differences between men and women with USH2. A total of 122 adults with USH2 from the Swedish USH database at the Audiological Research Center received the questionnaire HET, and 96 answered and were included in the study. The results of people with USH2 were compared with those of a reference group composed of 5738 people. The reference group was taken from an extant database administered by the Swedish Institute of Public Health* and Statistic Sweden.

The current focus was on questions pertaining to general health, physical health and psychological health. Significant differences in both physical and psychological health were found between people with USH2 and the reference group (diagram 1). People with USH2 expressed problems with tinnitus, headache, shoulders and neck pain, and had significantly worse problems with eczema and skin rashes. Psychological health (including fatigue, inability to concentrate, being unable to accomplish things, feelings of worthlessness, and a feeling of being constantly under strain) was significantly worse among those with USH2. Those people with USH2 reported unhappiness and depression as well as anxiety significantly more than those in the reference group. Suicidal behaviors (both thoughts and attempts) were more evident among those with USH2 than the reference group (diagram 1).

Men and women in the reference group significant differed with regard to physical and psychological health; women reported having more problems with regard to 30 of 36 variables controlled. Comparisons between men and women with USH2 presented minor differences regarding shoulder and neck pain and incontinence; women expressed to have significant more problems with regard to both variables.
Furthermore, women with USH2 were compared with their reference counterparts, revealing significant differences on five questions pertaining to physical health and two questions concerning psychological health; in which women with USH2 exhibited poorer health in all cases (diagram 2).

*Diagram 2. Significant differences in physical and psychological health among women with USH2 compared women in the reference group

*Significance set at ≤0.05.
Significant differences were found between men with USH2 and their reference counterparts on 15 of 36 questions (six pertaining to physical health and nine concerning psychological health). Men with USH2 reported more severe situations for all significant differences found (diagram 3).

*Diagram 3. Significant differences in physical and psychological health among men with USH2 compared with men in the reference group.*

The result finding describes poor physical and psychological health for people with USH2 compared with the reference group. Special attention must be devoted to the high frequency of suicidal behavior revealed as well as the physical and psychological health situations that men with USH2 reported.
Study II
Title: Health in persons with Usher syndrome type 3, Implications of deafblindness

This study described the health and social trust of people with USH3 in relation to the degree of hearing and vision loss. This study included 15 people with USH3 who answered the HET questionnaire; questions pertaining to general health, physical health, psychological health as well as social trust and financial situation were included. They also answered HAD-scale. In table 3, the demographics, degree of hearing loss, presence of a cochlear implant, visual field and visual acuity are presented with the general health, physical health, psychological health and social trust domains. The table also presents the total number of problems for each person.

Within the general health domain, self-assessed health was reported as good or fair; one person (table 3, person 8) reported poor health and that she had experienced a restricted capacity to work and activities of daily living (ADLs) over the previous 30 days; poor psychological health days were also frequent for a large part of the month. Person 11 reported that poor physical health days had restricted her 15 of the last 30 days, and she experienced five days of poor mental health. This person reported a lowered capacity for work and reduced ADLs for 20 of the last 30 days. Person 15 answered that she was completely restricted from work and ADLs during the last month. Her poor physical and psychological health could be interpreted but not whether her physical and psychological days were equally distributed (table 3).

Participants reported between 0-9 physical health problems, which included symptoms such as headache and shoulder and neck pain. Further, back pain, pain in the extremities, bowel problems, tinnitus and eczema were reported. Two people reported no physical health problems, and seven people reported fewer than five problems (table 3).

The most frequently reported psychological health problem was fatigue, which was reported by all participants except person 1, followed by stress which was reported by thirteen people. Other psychological health problems included worry, sleeping problems and poor wellbeing. The results from the HAD-scale showed that, three people met the diagnostic criteria for depression and four people had anxiety disorders. Eight people reported suicidal thoughts, and three people reported suicide attempts.

The most frequent problem within the social trust domain was refraining from going out alone. Person 1 was the only one not reporting any
problems in this domain (table 3). Not obtaining help when needed and a general mistrust in others were also demonstrated. Two people reported not having someone with whom to share one’s innermost feelings and confide in. Eight people reported being violated, and two people had experienced violence. A severely strained financial situation was apparent for four people who reported problems with being unable to pay for expenditures such as food, rent and other bills at least once over the last twelve months. These people also reported being unable to obtain 15,000 Swedish crowns within a week in the event of an unforeseen situation. Five people reported a difficult financial situation (i.e., they had problems with either paying for expenditures over the previous twelve months or obtaining 15,000 Swedish crowns within a week).

When the demographics and medical data were added to the matrix, some patterns emerged (table 3). Women reported both the fewest and most problems. The four men who participated in the study reported between 9-15 problems. No differences in term of age and the number of problems reported were identified.

Three people with USH3 had a unilateral cochlear implant at the time they answered the questionnaires, and they reported far fewer problems than the others, had good self-assessed health, few poor physical health days, few poor mental health days and few days of restricted capacity for work and ADL (table 3). For the other participants, a greater diversity existed in poor days and restrictions in work and ADL. Major differences were found with regard to the number of psychological health and social trust problems reported; those with cochlear implants reported fewer problems. They also reported fewer physical problems; however, the other participants’ problems overlapped with those with cochlear implants (table 3).

The total number of problems reported was affected when severe hearing loss were combined with both poor visual field and visual acuity scores (e.g., participants 14 and 15). Both participants 14 and 15 had profound hearing loss, visual fields of approximately 10° and a visual acuity score that met the criteria for legal blindness (i.e., 0.3). Participants 4, 10 and 11 did not report as many problems; however, they also presented with either better visual field or visual acuity scores (table 3).

This study marks the first time that health and social trust were described for this group, and more research is needed to explore the health and social trust of people with USH3.
Table 3. The demographics, degree of hearing impairment, the presence of a cochlear implant, and visual field and visual acuity scores as well as general health, physical health, psychological health and social trust of people with USH3

<table>
<thead>
<tr>
<th>Person</th>
<th>Sex</th>
<th>Age</th>
<th>Degree Hearing Impairment, dB(PTA)</th>
<th>Cochlear Implant</th>
<th>Visual Field</th>
<th>Visual Acuity</th>
<th>Self-assessed Health</th>
<th>Physical Days&lt;sup&gt;a&lt;/sup&gt;</th>
<th>Mental Days&lt;sup&gt;a&lt;/sup&gt;</th>
<th>Capacity for Work and ADL Days&lt;sup&gt;a&lt;/sup&gt;</th>
<th>Physical Health</th>
<th>Mental Health&lt;sup&gt;b&lt;/sup&gt;</th>
<th>Social Trust&lt;sup&gt;c&lt;/sup&gt;</th>
<th>Total Problems Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>F</td>
<td>36</td>
<td>101</td>
<td>X</td>
<td>4</td>
<td>1,00</td>
<td>Good</td>
<td>3</td>
<td>3</td>
<td>3</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>F</td>
<td>20</td>
<td>96</td>
<td>X</td>
<td>2</td>
<td>1,00</td>
<td>Good</td>
<td>4</td>
<td>2</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td>1</td>
<td>2</td>
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<td>105</td>
<td>X</td>
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<td>0</td>
<td>0</td>
<td>0</td>
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<td>1</td>
<td>1</td>
<td>6</td>
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<td>4</td>
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<td>Fair</td>
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<td>9</td>
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<sup>a</sup>In the article, "Health and Social Trust in persons with USH3, Implications of Deafblindness", the visual acuity score is reported in American equivalents (foot scale). However, the decimal equivalents used in Sweden are given here. <sup>b</sup>The general health, physical health, psychological health and social trust domains include questions pertaining to the HET. <sup>c</sup>The number of bad days over the previous 30 days; ≥15 days indicates poor physical health, psychological health, a lowered capacity for work and restricted activities of daily living (ADLs). <sup>d</sup>Psychological health includes questions from the HAD-scale. <sup>e</sup>Social trust includes financial situation. n.a. no answer.
Study III
Title: Physical and Psychological Health, Social Trust and Financial situation for people with Usher syndrome type 1.

This study described the physical health, psychological health, social trust and financial situations of people with USH1 compared a cross-section of the Swedish population.

Two adjusted questionnaires, the HET and the HAD-scale, were sent to 87 adults with USH1. The questionnaires were translated into Swedish sign language. The written material was adjusted for better contrast (i.e., yellow paper with black text) and larger font, and a structure compatible with the Braille script reader was provided. The questionnaires adjustment procedure is included in this article.

Sixty people with USH1 answered the questionnaire. People with USH1 were compared with a reference group that included 5738 people living in Sweden in 2007. This article presents the results from the HET questionnaire.

The findings revealed significant differences in psychological health, social trust and finances such that people with USH1 presented with a worse situation. People with USH1 had fewer physical health problems than those in the reference group. Headache was the only symptom in which people with USH1 presented with significantly more problems than the reference group. People in the reference group presented with significantly more hand, elbows, knees and leg pain as well as more tinnitus (diagram 4).
Diagram 4. Significant differences in physical health among people with USH1 compared with the reference group.

People with USH1 reported significantly more psychological distress than the reference group with regard to fatigue, lost confidence, constantly under strain, feelings of worthlessness, being unable to face problems and being unhappy. People with USH1 also reported significantly suicidal thoughts and attempts (diagram 5).

Diagram 5. Significant differences in psychological health among people with USH1 compared with the reference group

*Significance set at p≤0,05.
The most prevalent social trust domain problem was refraining from going out alone (diagram 6). Other problems included a general mistrust of others and not having anyone with whom to share one’s innermost feelings and in whom to confide. The largest difference between the two groups was “Not receiving help when needed” (diagram 6). A difficult financial situation, defined as not having the opportunity to obtain 15,000 Swedish crowns within a week of an unforeseen situation, was also more common for the USH1 group than the reference group (diagram 6).

Diagram 6. Significant differences in social trust in people with USH1 compared to the reference group.

*Significance set at p≤0.05

The results in the study revealed a more severe psychological health situation and lack of social trust including financial situation for people with USH1 compare to the reference group. It is possible to assume that some of the problems reported could be related to environmental factors, social recognition, ontological insecurity as well as self-esteem and identity. The strained financial situation reported has not been described previously and needs to be further scrutinized. Furthermore special attention needs to be devoted to the high frequency of suicide thoughts and attempts.
Study IV
Title: Similarities and differences in the health, social trust and finances of people with Usher syndrome: a biopsychosocial perspective

This study described similarities and differences in general health, physical health, psychological health, social trust and finances among people with USH, types 1, 2 and 3. This study also sought to examine whether numerous independent variables (i.e., sex, age, clinical diagnose, visual field, visual acuity and degree of hearing impairment) were associated with poor health, social trust problems and strained finance outcomes among people with USH.

Of the 46 outcomes scrutinized, 11 significant differences in health and social trust were found with regard to people with USH1, 2 or 3. Most differences were found within the social trust and financial situation domains.

Sixteen outcomes differed by more than 10% between the clinical USH group who reported the least number of problems and the group who reported most problems. However, people with USH1, 2 and 3 were more similar than different for almost half (19/46) of the outcomes.

No significant differences were observed in the self-assessed poor health of people with USH1, 2 and 3. No significant differences in the number of reported physical poor health days, psychological poor health days or days in which capacity for work and activities of daily living had been lowered were found. However, people with USH3 reported fewer days affected by poor physical health (diagram 7). People with USH2 reported fewer problems with a lowered capacity for work and activities of daily living due to physical and psychological poor health than people with USH1 and USH3 (although this result was not significant). People with USH3 were more likely to report problems with capacity for work and restricted activities of daily living (ADLs).
Diagram 7. Self-assessed general health, physical health and psychological health days, capacity for work and activities of daily living over the past 30 days for people with USH1, 2 and 3

*A report of ≥15 days over the last 30 days was considered as poor health.*

The regression model regarding how the independent variables were associated with the general health dependent outcomes, revealed that age had a significant high odds ratio (OR) with regard to poor self-assessed health (table 4).
In three of the thirteen physical health outcome measures, differences were found among people with USH1, 2 and 3; in ten outcomes, no differences were revealed (diagram 8). However, the between-group comparison showed that the significant difference in tinnitus was between people with USH1 and those with USH2. People with USH3 reported far more (although not significant) problems with tinnitus than those with USH1 (diagram 8).

*Significance set at $p \leq 0.05$

The logistic regression model regarding how different independent variables contributed to the poor physical health outcomes did not reveal a clear contributor (table 4). For six of the physical outcomes, USH category (i.e., clinical diagnosis of USH2 or 3) or sex (i.e., being a woman) was associated with a higher risk for poor outcomes (table 4). For two outcomes an older age was represented with significant higher risk of a poor outcome, and a poor visual field was significantly related to one poor outcome (table 4).
Psychological health included 20 different outcomes, of which two were significant: fatigue and suicide thoughts (diagram 9). The group comparison for fatigue revealed that the difference between people with USH1 and those with USH3 was huge, in which the latter reported far more problems than the former (93% compared to 62%), however, this result was not significant. The same pattern of significance was revealed for suicide thoughts with regard to the same participants.

Diagram 9. Group comparisons of psychological health among people with USH1, 2 or 3

The logistic regression model of how different independent variables contributed to poor psychological health outcomes did not reveal a clear tendency regarding which independent variable that was most associated with poor psychological health. USH category was significantly associated with risk of fatigue. This finding indicates that people who have USH2 or USH3 were at greater risk for problems with fatigue. Both age and USH category were associated with sleeping problems. Those with a more severe hearing loss were at risk of having suicide thoughts. Interestingly, association was negative for some of the variables, such as visual field and to managing problem meaning those with a better visual field had a higher
risk of problems. A negative association was also found between visual acuity and being incapable of making decisions (table 4).
Six of the nine social trust and finance outcomes showed significant differences. Regardless of significance, people with USH3 reported the most problems, closely followed by those with USH1. People with USH2 reported either the least or second most social trust and financial situation problems (diagram 10 and 11).

Diagram 10. Group comparisons of social trust among people with USH1, 2 or 3

*Significance set at p≤0.05.
Diagram 11. Group comparisons of financial situation among people with USH1, 2 or 3

Significance set at $p \leq 0.05$.

Different independent variables contributed to the social trust and financial outcomes (table 4). Women were more almost six times more likely to refrain from going out alone. Both sex and visual acuity contributed to poor outcomes in general trust in others. Specifically, women and those with better visual acuity were more at risk for a poor outcome. Age and visual acuity contributed to poor outcomes regarding no one to share innermost feelings and confide in, respectively. Specifically, older participants and those with better visual acuity were at significantly higher risk. Younger participants had a higher risk of being violated. People with USH3 were most likely to have a difficult financial situation (table 4).
Table 4. Logistic regression model for independent variables USH category, sex, age, visual field, visual acuity, and degree of HI and poor dependent outcomes regarding health, social trust and financial situation

<table>
<thead>
<tr>
<th>Dependent Outcome Measures</th>
<th>Independent Variables</th>
</tr>
</thead>
<tbody>
<tr>
<td>USH Category</td>
<td>Sex</td>
</tr>
<tr>
<td>Poor health</td>
<td>4.07 (1.04-15.97)</td>
</tr>
<tr>
<td>Headache</td>
<td>2.65 (1.39-5.99)</td>
</tr>
<tr>
<td>Tinnitus</td>
<td>3.43 (1.59-7.42)</td>
</tr>
<tr>
<td>Pain shoulders, neck</td>
<td>3.06 (1.46-6.45)</td>
</tr>
<tr>
<td>Pain hand, elbow, knee, legs</td>
<td>2.16 (1.10-4.27)</td>
</tr>
<tr>
<td>Incontinence</td>
<td>3.98 (1.126-14.060)</td>
</tr>
<tr>
<td>Bowel trouble</td>
<td>2.66 (1.28-5.51)</td>
</tr>
<tr>
<td>Allergy</td>
<td>3.22 (1.09-9.46)</td>
</tr>
<tr>
<td>High blood pressure</td>
<td>2.90 (1.10-7.63)</td>
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<td>Fatigue</td>
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<tr>
<td>Sleeping problems</td>
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<td>Lost sleep over worry</td>
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<td>Manage problems</td>
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<tr>
<td>Incapable of making decision</td>
<td>0.10 (0.01-0.75)</td>
</tr>
<tr>
<td>Suicide thoughts</td>
<td>1.77 (1.01-3.11)</td>
</tr>
<tr>
<td>Suicide attempts</td>
<td>2.31 (1.06-5.05)</td>
</tr>
<tr>
<td>Refrain from going out alone</td>
<td>6.70 (2.95-15.24)</td>
</tr>
<tr>
<td>No general trust in most people</td>
<td>1.84 (1.03-3.32)</td>
</tr>
<tr>
<td>No one to share innermost feelings with and confide in</td>
<td>2.14 (1.14-4.02)</td>
</tr>
<tr>
<td>Violated</td>
<td>0.57 (0.33-0.96)</td>
</tr>
<tr>
<td>Difficult financial situation</td>
<td>2.26 (1.07-4.76)</td>
</tr>
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</table>
Discussion

This section discusses the following themes about people with USH and their health:

- Brief introductions regarding the previous research of people with USH and their health and what is missing from this research are provided, to underpin the rationale for this thesis.
- This section is followed by a discussion of the major findings of the four studies (I, II, III and IV) included in the present thesis in the context of previous research. Each domain (i.e., general health, physical health, psychological health, and social trust including finances will be discussed separately.
- After discussing the main findings, the need for a biopsychosocial approach for the health of people with USH1, 2 or 3 is addressed.
- The challenges present during this thesis that concern conducting research with people with USH1, 2 or 3 are then scrutinized.
- This section is followed by a section on people with USH1, 2 or 3 and their health when framing the disability paradox.
- The last section addresses the need for a salutogenic health perspective when trying to understand the health of people with USH1, 2 or 3.

A brief introduction

The present state of knowledge regarding people with USH and their health is fragmented. This thesis provides knowledge on the general health, physical health, psychological health, social trust and financial situations of this group. The extensive empirical material used in the thesis provides detailed descriptions of the present health state for this group who, because of their combined impairments, might be at risk for poor health. The present thesis does not explain the underlying mechanisms that affect people with USH and their health; rather, it thoroughly describes their general health, physical health, psychological health, social trust and financial situations.

Möller\textsuperscript{111} described deafblindness \textit{“as a bio-psycho-social and contextual matter”}(p.64). This perspective is important for healthcare, other support services and research. Deafblindness, such as that observed in people
with USH, is a complex phenomenon. Therefore, describing health, social trust and financial outcomes in terms of causality is challenging.

Some studies that have addressed the importance of taking a health perspective with regard to USH\textsuperscript{36, 98, 111}. However, Millán et al.,\textsuperscript{98} did not develop this idea other than to conclude that people with USH are clinically and genetically heterogeneous and at risk for social isolation. Therefore, they need attention from a public health perspective. Möller\textsuperscript{111} addressed the public health perspective that concerns safety, such as the informational text on medicine and other information provided by society. Studies have described people with USH\textsuperscript{99, 100}, deafblindness\textsuperscript{18, 71, 136}, or both as “exposed”. Ellis and Hodges\textsuperscript{45} described diversity in all aspects of life and addressed how to cope with changes in life situations.

**People with USH1, 2 and 3 and their health**

The adult people with USH1, 2 or 3 included in the Swedish Usher database self-assessed their health using a comprehensive questionnaire the (HET) and a 14-item questionnaire regarding to anxiety and depression (the HAD-scale). Hearing and vision loss and genetic background information were retrieved from the Usher database when the questionnaires were dispatched. The results of studies I and III were compared with a cross-section of the Swedish population who completed the HET. Study II presents in-group comparisons of people with USH3, and the results in study IV present in-group comparisons among people with USH1, 2 or 3. The results show that poor physical health, psychological health, a lack of social trust and a strained financial situation are present for people with USH, regardless of their clinical diagnosis.

**General health**

The general health state described in the HET did not reveal significant differences between people with USH1, 2 or 3 and the cross-section of the Swedish population that was, compared in studies I and III with regard to poor health. The global question in the HET, “How do you rate your general health state?”\textsuperscript{57}, might be regarded as vague and non-specific to be used by itself\textsuperscript{50}. However, self-assessed are a reliable measure of health\textsuperscript{76, 97}. Several studies have reported that self-reported health predict mortality\textsuperscript{9, 40, 58, 72}.

Questions were asked about how many days over the last 30-day period had physical and psychological health been affected as well as whether restrictions of capacity on work and activities of daily living were present.
due to these poor health days. People with USH1, 2 or 3 reported that they experienced poor physical and psychological health days and that their capacity for work and activities of daily living had been restricted. The question about capacity for work and activities of daily living does not define what “activities” includes. Activities of daily living can include a variety of activities such as housework, picking up the children at kindergarten, personal hygiene, dressing, leisure activities more. A person with deafblindness who experiences limitations in these activities is included in the Nordic definition of deafblindness, with the addition that the effort to limit the restrictions in activity and participation is a responsibility of society. Saunders and Echt\textsuperscript{135} studied older people with age related deafblindness and found that restrictions in activities of daily living occur in a vast majority of life areas such as shopping, leisure activities, housework and so on. However, restrictions in work and activities of daily living have not been previously reported as a consequence of poor physical and psychological health days among people with USH1, 2 or 3.

**Physical Health**

Different pain-related problems were frequently reported for people with USH, which somewhat differed with regard to where the pain was located among the three USH types (e.g., shoulder and neck pain, headache, back pain, or pain in extremities). People with USH3 most frequently reported having headaches (75%); people with USH2 reported having the most problems with shoulder and neck pain (61%); and people with USH1 reported having the most problems with back pain (45%; see diagrams 1, 4, and 8). Great efforts are needed to understand the issue of those living with USH1, 2 or 3 because of the limited possibility to compensate for either vision or hearing when both senses are affected. This effect might lead to bodily tension that can explain the pain reported by people with USH. Headache can be a consequence of the struggle to see and hear for people with deafblindness\textsuperscript{109}. Physical problems are seldom reported in the USH research; however, if the physical symptoms are a consequence of living with sensory loss these must be addressed and not ignored. Pain-related problems might contribute to a lower quality of life for the individual. Albrecht and Devlieger\textsuperscript{3} reported that pain might be a key factor when people with disabilities report that their quality of life is reduced.

Study I compared men and women with USH2 with those in the reference group. This comparison revealed small differences in physical health for men and women with USH2. Significant differences in reported physi-
cal health were found with regard to tinnitus and shoulder and neck pain. This finding was not the case for men and women in the reference group; here, women were more likely to express poor physical health. The differences in physical health between men and women with USH have not previously been the focus of research. The differences that have been addressed include visual prognosis. Specifically, Sadeghi et al., studied people with USH1 or 2 and found that the disease progression was faster for men with USH1 than women with USH1 or 2. Differences between men and women have not been the focus of the physical health investigated the other studies of the present thesis (II, III, IV); rather, these were differences controlled for in the study that compared people with USH1 and the reference group (study III). Here, differences were not revealed in terms of sex.

**Psychological Health**

The most frequently reported problem, regardless of USH type, was fatigue. Fatigue refers to a state of long-term tiredness that can affect people with both acute and chronic conditions. Fatigue can have long-term effects for an individual’s health. Fatigue was previously reported among people with deafblindness as a consequence of the efforts to compensate for sensory loss. This compensation is related to the consequences of their hearing and vision loss and the efforts needed to keep up with their surroundings when their sensations are restricted. Self-esteem, identity and how others perceive us are important with regard to how people cope with new life situations.

The psychological consequences of deafblindness have been reported primarily in relation to anxiety and depression. These problems have also been reported by the people with USH1, 2 or 3, and anguish or worry was the third most reported problem (see studies I, III and IV). Furthermore, some people met the criteria for having a depression or anxiety disorder as defined by the HAD-scale (see studies II and IV). With the comprehensive questionnaire used in the present thesis (i.e., HET), together with the HAD-scale a more detailed description of psychological health is possible than has been reported in previous research.

Within the psychological domain people with USH reported several problems including those with concentration, sleeping, feelings of worthlessness, anxiety, and depression and more (for details see studies I-IV). Previous research has attempted to establish causal links for psychiatric illness in people with USH, based on cases or family members.
However, no large epidemiological study has established that person with USH are more prone toward psychiatric diseases than others. Our studies showed that people with USH, regardless of type, are at risk for psychological health problems. This finding does not imply a causal relationship such that people with USH have a higher prevalence of psychiatric illness because they have USH. The psychological problems reported (e.g., fatigue, problems with concentration, anxiety, being unable to solve problems and feelings of worthlessness) might be consequences of living with a condition that has a progressive cause and leads to lifelong challenges and constant adaptations to new life situations. These consequences might be similar to those faced by other groups of individuals who are live with chronic, progressive conditions; however, this topic is not the objective of the present thesis to study. Furthermore, the reported problems with psychological health might be the consequences of a situation where communication, the exchange of information and orientation in the social world are compromised due to deafblindness. The struggle to maintain independence can be difficult for people with USH because as vision loss (as in USH1 and USH2) and hearing loss (as in people with USH3) might be risk factors for poor psychological health. Damen et al.,\textsuperscript{33} did not include people with USH3 but found that people with USH1 were more restricted in their independence than people with USH2.

The most extreme consequence of not being recognized as a human being, experiencing ontological insecurity and poor psychological health is suicide. Suicidal behavior is also connected to different life events when challenges become overwhelming and people can see no other way out. Suicidal behaviors such as suicidal thoughts and attempts have been reported by a small number of previous studies (e.g., Miner\textsuperscript{99,100} and Schneider\textsuperscript{136}). No large studies have described suicidal thoughts and attempts among people with deafblindness or USH to the extent found in studies I-III. The frequencies of suicidal thoughts and attempts among people with USH1, 2 or 3 ranged between 23-53\% and 9-20\% respectively. Both variables were least likely in people with USH2 and most likely in people with USH3 (study IV). People with USH1 and those with USH2 reported a risk of suicide attempts that was five times (study III) and, almost three times higher (study I) than the reference group, respectively. The risk for men with USH2 was almost six times higher than the reference group (study I). People with USH3 were not compared with the reference group because of the low number of participants.
At this point, we do not know how many people actually succeed in their attempts to commit suicide, but these data are something important to scrutinize. A higher risk of mortality has been reported for people with deafblindness\textsuperscript{83, 88}. The risk of suicidal behaviors has been described after receiving a diagnosis of USH1 or 2\textsuperscript{99, 100}. Recent research, unrelated to people with USH or deafblindness, showed that people in Sweden who attended psychiatric clinics after having attempted violent suicide i.e., via hanging or shooting were at risk for successfully reattempting suicide\textsuperscript{127}.

**Social trust and financial situation**

Fear of the future and of how life will be can arise when living with deafblindness. Social trust is a mental state that has different orientations. Social trust implies trust in other people and environments. Social trust has both cognitive and emotional dimensions, and it is related to relationships with others as well as how we find reality to be reliable, predictable and consistent\textsuperscript{39}. As vision deteriorates people with deafblindness have reported that their relationships with others and the environment came under attack, and they began to struggle to find a place in the seeing and hearing world\textsuperscript{99, 100, 136}. Some of the questions within the social trust domain refer to having the possibility obtain help when needed and having someone with whom to share their innermost thoughts and feelings. The results revealed that people with USH1 reported that these qualities were lacking significantly more than the reference group (study III) and people with USH2 or 3 (study IV). In previous studies\textsuperscript{18, 45}, people with deafblindness or USH reported that they received support from family and friends in the way they wanted. However, people were not satisfied with the formal support that they received\textsuperscript{18, 45}; here, they wanted more support with both practical and emotional issues. However, people with USH have also reported feeling uncertainty or fear regarding being rejected or treated differently when telling a partner or friend about their condition. Negative consequences of being diminished or perceived in a different way have been reported by people with USH1\textsuperscript{99} or USH2\textsuperscript{100} as their vision decreased and they could no longer do the same tasks as before.

Refraining from going out and having a general mistrust in others were reported in studies II, III and IV. Most people with USH1 and those with USH3 reported these variables as problems, but people with USH2 also did not feel that they could trust others in general or felt that they did not want to go out by themselves for fear of being attacked, robbed or otherwise molested (study IV). Withdrawal from social activities has been re-
ported in other studies (e.g., Brennan and Bally\textsuperscript{25} and Hersh\textsuperscript{71}), but the mistrust and fear of going out has not been reported previously.

In studies II, III, and IV, the social trust problems were discussed in relation to being socially recognized and experiencing ontological security. These concepts concern how we are being recognized as human beings and whether we find our existence to be meaningful and understandable. The challenges of living with USH and the repeated changes of everyday life connected to the progressive course of the condition can, as mentioned previously, lead to fear and anxiety for the future. This fear also includes the attitudes and understanding of others. The lack of knowledge about living with USH from professionals, which is demonstrated in the fragmented healthcare\textsuperscript{25, 109, 112} described, that focuses on a single impairment\textsuperscript{137} is problematic in this situation.

How people with USH cope with the challenges of living with their diagnosis and how it can affect their lives in different ways are related to ontological security. Ellis and Hodges\textsuperscript{45} had people describe their different ways of coping with USH. Some internalized it as a part of themselves, whereas others did not. Both people with USH1 and those with USH2 reported that as their vision deteriorated meaningful relationships, respect and a sense of belonging became more important than before. These people described situations where they were no longer regarded as participating on equal terms as others, were excluded and not viewed as people who could contribute in the same way as previously\textsuperscript{99, 100}.

Barriers of communication (e.g., misunderstandings, a lack of knowledge and understanding regarding what adjustments are needed, and reluctance from others) are frequently described in the stories told by people with deafblindness or USH\textsuperscript{45, 71, 99, 100, 136}. Most adults with USH1 currently living in Sweden use sign language as their primary language. Fear of losing vision in combination with deafness has been described\textsuperscript{99} and not merely in relation to the people themselves. It might also raise fear in others in the Deaf community. The Deaf community is regarded as a separate minority group within the hearing community, with its own cultural and social structure\textsuperscript{75}. The fear that is being experienced within this community is related to communication, perception and how one will be accepted in the Deaf community if one becomes deafblind. The fear of losing one’s vision and what the reactions might be for a deaf person who defines him or herself as a part of the Deaf community might make the person reluctant to tell others he or she has a vision problem\textsuperscript{110}.
Financial situation can be measured in various ways, but one way to understand the financial situation of people is to scrutinize the questions pertaining a strained situation over the last 12 months, where the people answered whether they had problems with paying expenditures such as rent, food, or other bills. The other question was related to the likelihood that a person could obtain 15,000 Swedish Crowns within a week in the event of an unforeseen situation. This amount is known as “cash margin” a measure based on a normal Swedish salary. The results of the studies II, III and IV revealed that people with USH1, 2 or 3, had strained financial situations. People with USH3 reported significantly more problems with regard to having difficulties over the 12 last months in paying for expenditures (diagram 11). Regarding the occurrence of an unforeseen situation, both person with USH1 and those with USH3 reported significantly more problems than did those with USH2 (study IV).

The financial situations of people with USH have not been reported previously, and a need for additional studies exists. Strained financial situations might be related to poor health outcomes and insecurity in everyday living. Strained financial situations are related to unequal access to education and an unequal position in the labor market. In the report from the Public Health Agency of Sweden, the risk for poor psychological health (e.g., anguish and worry) were higher among those who had a strained financial situation and had an impairment.

**People with USH and health: A biopsychosocial perspective**

The health of people with USH can be understood at different hierarchies through which mechanisms affect the outcomes at the different levels. At the biological level (or a lower molecular level), USH affects genes that cause hearing and vision loss and functional sensations in the ear and eye. At a psychological level, the experience of living with USH and how to cope with such a condition are affected by mechanisms. At the social level, mechanisms are in play regarding the attitudes and norms about people with deafblindness such as that in USH. The interplays of these mechanisms at different levels produce reality for people with USH. No previous research has attempted to describe the health of people with USH from a biopsychosocial perspective. This has been the overarching goal of the present thesis.

The biopsychosocial perspective applied and discussed here was inspired by Bircher’s merging of Engel and Nordenfelt. The essential elements for defining health include the biopsychosocial nature of human
existence and the relationship between the demands made on the individual and his or her abilities to meet them when changes in life occur, including social determinants. Events in life can happen as chains, where one event leads to a consequence that affects new events that occur. In other words, an event that affects a person’s life is not unrelated to other events or people. However, how one copes with these events are connected to their abilities to cope and meet challenges. This effect could not be captured by applying a strict biological theory of health, which, in practice, would focus on malfunctions in the body or organs. For people with USH, this effect includes impairments in hearing, vision and (for some) balance. The holistic perspective described by Nordenfelt considers the person as whole. However, the social determinant included in the Bircher definition of health adds another dimension to the biopsychosocial definition of Engel as well as the abilities and intention of Nordenfelt, which to a greater extent address the involvement of the environment. The people with USH who participated in the Ellis and Hodges study did not report that it was the actual level of hearing and vision losses that affected how life changes were experienced and managed. Research on people with hearing loss has shown that how people perceive their hearing loss is more related to how they describe their health than what the actual audiogram implicates. Functional limitations, abilities and intentions, rather than medical diagnoses or level of impairment, might be what affects everyday living for people with USH. Recourses, or in ICF terms, “personal factors” might affect how the life challenges due to a progressive disorder such as USH affect adjusting or coping with events that occur. However, if hearing and vision loss are not reported or defined, then no conclusion can be made regarding the above statement about people with hearing loss.

The objective of study IV was to scrutinize whether similarities and differences were present between the three clinical diagnoses of USH. A model was constructed to investigate how sex, age, visual field, visual acuity, hearing loss and the clinical diagnosis of USH1, 2 or 3 affected poor outcomes in health, social trust or financial situation. Some of the poor health outcomes that the people with USH1, 2 or 3 reported might be considered as a secondary condition, which in itself might have a greater effect on the individual’s health than the restrictions of hearing and vision loss. Marge addressed this discussion by concluding that introducing the ICF model would expand the thinking of secondary conditions; however, it is not enough to cover the complexity of the interactions between the primary disability and secondary conditions.
The consequences of combined vision and hearing loss go beyond the actual degree of impairment and must be understood with regard to its functional dimensions. This understanding has been expressed such that the sum of the loss in hearing and vision is more than each part alone. Functional limitations in vision and hearing might be a better indicator of how the individual perceives his or her deafblindness in everyday life and from a life course perspective. The results of study IV indicate the complexities of living with deafblindness such as that in USH1, 2 or 3. No previous studies have attempted to scrutinize variables such as age, sex, visual field, visual acuity or degree of hearing loss in relation to health outcome. Participants in smaller qualitative studies have reported that they have experienced psychological distress, or in other ways, a compromised situation, at critical points (e.g., Miner and Schneider). Critical points can include time of diagnosis, changes in life due to progressive vision/hearing loss, and being perceived by others as more different than before.

According to Möller, it is of great importance to take a biopsychosocial perspective in healthcare regarding people with deafblindness to provide support and care. A lack of knowledge among professional healthcare providers or assumptions made due to poor knowledge of what living with deafblindness means might have far reaching consequences for the individual. For example, the risk of suicidal behaviors among people with USH was significantly higher than that among the reference population (see studies I and III as well as study IV for in-group comparison). Using the question in the HET, participants were able to report whether they had attempted taking their own life once or several times. A fragmented healthcare where knowledge and coordination of support or care are lacking might miss the complexity of living with USH, which could have fatal consequences for the individual.

The UN convention, article 25, states that human health is a right, regardless of impairment or other circumstances. It also states that the healthcare offered should be equivalent for all people. None of our studies (I-IV) scrutinized what type of healthcare people with USH received or wanted or healthcare contact was experienced. Thus, we described that people with USH experience problems with their health at different levels. The overrepresentation of poor physical health, psychological health, problems with social trust and financial situations must be understood from an interdisciplinary biopsychosocial perspective together with specific knowledge about deafblindness. This idea is stressed in the Nordic definition of deafblindness.
Challenges for research on people with deafblindness or USH

Challenges with definitions of deafblindness
Numerous definitions of deafblindness exist, and some research does not clearly define the group being studied. Hence, comparisons of results become difficult. Schneider et al. stated that future research should use mixed methods when describing hearing and vision; otherwise, a risk of simplifying the consequences of vision and hearing function and their combined affects exists. The absence of clinical and functional definitions for vision loss, hearing loss, and type of deafblindness as well as to understand the consequences of living with deafblindness has been addressed by other studies. The people with USH in the present thesis were well defined, and two established questionnaires were used. The present thesis adds an inclusive approach that has not been used by previous studies on the health of people with USH. Previous research focused on the psychological and social consequences of deafblindness such as depression, anxiety, independence and social withdrawal, but it lacks the dimensions of general health, physical health and financial situation.

Challenges with generalization
This research is constrained in terms of generalization to other the functional dimensions and disabling conditions that characterize differences in people with deafblindness. Generalizations in qualitative research are possible from different standpoints that might be applicable for the present thesis. Different arguments are possible, and one of them is “generalization through recognition of patterns” (p. 28). Studies are often performed with a limited number of people in specific contexts. According to Larsson, pattern recognition is an accepted way to gain knowledge about unexplored groups. The knowledge gained in the present thesis, might be transferred to the greater context of the health of people living with deafblindness. This expansion is possible based on the argument that no reason exist that the patterns in health revealed for the USH group should differ from other groups with deafblindness. People with USH also constitute a large part of the population of people with deafblindness. The second line of argument is that the knowledge about the health of people with deafblindness is sparse.
**Challenges with the questionnaires**

Many instruments (i.e., questionnaires) to measure health exist. Some of the instruments that have been developed are generic, meaning that they are suitable for any group of people. These types of questionnaires are often used in surveys aimed at healthy people. The HET is an example of a generic instrument\(^{22}\). A generic instrument covers a wide range of conditions; however, they fail to focus on specific issues regarding people with diseases or impairments. On the other hand, a disease-specific instrument aims at the special concerns of a specific group with disabilities or health condition. Instruments also exist that focus certain aspects of health or quality of life\(^{50}\). One such instrument is the Hospital Anxiety and Depression Scale\(^{161}\). Measuring of health and quality of life is challenging\(^{51}\). The questionnaires used must be relatively short and should not contain a high level of abstraction in complex domains such as social life, home life or work. This presupposes knowledge from those who construct the questionnaire and the respondents in terms of their language abilities and capabilities to respond to abstract terms such as the extent of a problem\(^{51}\). The measurement of quality of life has become an important way to evaluate quality and outcomes within healthcare, especially for patients with chronic conditions\(^{101}\).

Both the HET and the HAD-scale are widely used questionnaires. The HET has been administered annually in Sweden since 2004, and is evaluated on a regular basis. The use of the HAD-scale has been both in clinical groups and in general populations, and the HAD-scale is widely psychometrically tested. These facts are strengths of both the questionnaires. Crawford et.al.,\(^{31}\) concluded that the HAD-scale was applicable in general populations across different background variables such as age, education level, and socioeconomic status; However, sex (gender) cannot be excluded from the analysis\(^{31}\).

Neither the HET nor the HAD-scale have previously been used for people with USH. These facts, together with a good response rate of people with USH included in the present studies, implies a satisfactory and reliable start to gain more knowledge regarding how people with USH perceive their health. The empirical material presented in the current thesis has not been presented in research previously. The Swedish Usher database enables the retrieval of a well-defined group together with data on vision, hearing, and genetics, providing gives opportunities to thoroughly describe the participants.
Studies I and III compared people with USH1 or 2 with a cross section of the Swedish population. The HET was chosen based on the premise people with USH could be compared with a reference group. Two reports discuss the health of people with impairments living in Sweden. However, the HET has been criticized for how it discriminates among different impairments and regarding who is included. It is possible to identify impairments in the population based on numerous questions. These questions have been criticized for being blunt and missing certain impairments (e.g., psychological illnesses and cognitive difficulties). The impairments that can be identified by the HET include hearing and vision loss based on whether the respondent can hear a conversation between two or more people, answered on a three-point scale from “yes, without hearing aid”, “yes, with hearing aid” or “no”; whether the respondent can see written text in a newspaper, answered in a three-point scale from “yes, without spectacles”, “yes, with spectacles” or “no”. Questions are also asked regarding whether the respondent has difficulties with walking over a short distance, walking up steps, whether help is needed either by another person or a technical aid move around outside. Questions concerning whether the respondent has one, two, three or more diseases or impairments are also included. In the present thesis, the questionnaire was used with regard to a specific group, namely people with USH, and describes different health indicators for this group. When comparing people with USH with the cross-section of the Swedish population, no people with impairments were excluded from the reference group. The reason for not comparing with the reference group after separating those who self-reported impairments or had medical conditions and those who did not is that it is difficult to identify who to include based on unclear definition of “healthy”.

Furthermore, no question in the HET addressed communication or communication strategies. Communication is a dimension of health that needs attention because it plays a central role in human life and human relationships. Research has shown a better self-reported mental health and quality of life among people who are deaf than those who had hearing loss. suggests that inclusion in a cultural community with a common language (i.e., sign language) is a protective factor for mental distress and poor quality of life. Deafblindness, such as that in USH, is largely an impairment that has implications for communication and interactions as well as giving and receiving information. For some people with USH who have been brought up using one way to communi-
cate (i.e., speaking, lip-reading or visual sign language), the progress of vision and hearing loss observed in USH3 or the progression of vision loss observed in USH1 and USH2 might lead to a need to change one’s communication style.

**Challenges in adjustment of the questionnaires**

A further challenge of the current research concerns the layout of the questionnaires. Little research exists on the process of adjusting a questionnaire for people who use sign language as their primary way to communicate. One study described the challenges of adjusting a psychological health questionnaire for people who use sign language\textsuperscript{102}. They described the process of adjustment as not merely translation from one language to another but also as a cultural adjustment\textsuperscript{102}.

The questionnaire adjustments made in the present thesis were not solely aimed at people with vision loss or those who use sign language as their primary way of communication. The intention was to provide people with USH1 with different ways to access the questionnaires: in written text with an adjusted layout, on a DVD and on a memory stick. Therefore, this survey could also be used by people with a hearing and vision loss or both. Some of the adjustments primarily concerned the visual accessibility of the material, and some of the adjustments concerned accessibility for people with hearing losses who used sign language as their primary means of communication. Adjusting the accessibility of a questionnaire for the target group is complicated; vision, hearing and language are important in the adjustment process. The adjustments made were a first attempt to provide people with deafblindness with different opportunities to access the material. Further study design should consider the possibility to respond in different ways. In study III, participants had the possibility to give their answers either by filling out the questionnaires on paper or by computer on the memory stick provided. They could choose to return the filled out questionnaires by post or attached in an email. However, the response rate of people with USH1 was considered as satisfactory (study III).

**People with USH and the disability paradox**

Reporting good self-assessed health and while having numerous physical and psychological poor outcomes, problems with social trust, difficult financial situations and a medical diagnosis might seem as a paradox. This conundrum is the disability paradox\textsuperscript{3}. People with USH described their
general health to be good, and no significant differences were found in comparison with a cross-section of the Swedish population (Study I, III and IV). At the same time, people with USH reported more psychological days as well as restricted capacity for work and reduced activities of daily living than the reference group. Certain problems within the different health domains (physical, psychological, social and financial) were significantly more common among people with USH than the reference group.

Krahn et al. stated that the essence of the problem with measuring self-reports of health is that confounding effects exists between what is actually measured and what is intended to measure. Thus, health status is often confused with functional abilities. In practice, a person with USH might self-assess lower health score that reflects not a poorer health but a lower functional capacity. Krahn et al. stated that distinctions in health, function and disability have important implications for how they are measured. Function is not invariably related to health; rather, it interacts with health. Fellinghauer et al. showed how general health and impairment were not related; instead, it was limitations in activity and participation that affected perceived general health and not the impairments per se. The experience of a poor quality of life is a result of a complex system through which mechanisms at different levels are in play.

Engel addressed the “disability paradox” when discussing that the use of the bio-psychosocial model enables explanations for why some certain individuals experience “illness” conditions while others merely have “problems of living”, regardless of emotional reactions to circumstances or physical symptoms. Engels discussion did not concern people with disabilities; however, the notion of what constitutes an illness might be a subjective experience regardless of disability and something related to what it is to be human in a social context. Ellis and Hodges reported that people had perceived their USH in different ways. Some considered it as a natural part of who they were and had internalized it as a part of themselves. For the outside observer, this might be a challenge because USH can cause restrictions that greatly affect everyday life. Because deafblindness and USH are rare, knowledge of the experience of living with it is sparse in the general population and among professionals. A common misunderstanding is that people with deafblindness experience a totally dark and silent world; this idea can be frightening for those who have never been in contact with a person who is deafblind.

There is no doubt that the disability paradox exists. This paradox is framed by what people with disabilities themselves say influences their
quality of life and how maintain balance in life. Developing the disability paradox further is beyond the scope of the present thesis, as are the assumptions made by the people with USH regarding how they perceive this discussion. However, the preconceived assumptions of others in society or the network of family and friends regarding what constitutes health and quality of life most likely effects how people with USH are approached. This consequence might affect how a person with USH is perceived professionals, family members or friends. This effect might predict how and what questions about health that are asked. People with deafblindness or USH have reported a lack of knowledge, misunderstandings, and not being regarded as a competent person45, 71, 136.

Being socially recognized and having a feeling of inner consistency and meaningfulness are related to social recognition and ontological security, concepts that can be addressed within this context. If people are not recognized as human or they experience ontological insecurity described by Möller and Danermark110 as well as Danermark and Möller39, then the consequences for the individual might include a compromised self-image and self-identity. These effects would imply that the balance among the body, mind and spirit that Albrecht and Devlieger3 describe is important for people with disabilities to report high quality of life.

**Salutogenic health**

Another perspective that is poorly represented relates to what Danermark and Möller39 addressed in their article on deafblindness, ontological security and social recognition stress: the importance of research with a salutogenic perspective on deafblindness. The salutogenic perspective would enable a shift in orientation within the research on deafblindness from problem oriented to solution oriented to show the potential of people with deafblindness. This idea might lead to highlighting efforts that promote health in a life course perspective.

Aron Antonovsky, professor of the sociology of medicine in Israel, began his work on what later was known as the salutogenic paradigm in 1970. Antonovsky described the sources of the salutogenic orientation as all of the living organisms that are exposed to the irregularities that create disorder instead of order. This assumption contrasts with the pathogenic orientation, which suggests that the purpose of the organism is to create order out of chaos7. His conclusion is that “disease, however defined, is very far from an unusual occurrence” (p49)7. Health, according to Antonovsky, is considered as a continuum with total wellness and total dis-
ease as its endpoints. The central concept of Antonovsky’s understanding is sense of coherence (SOC). Antonovsky suggested that the individual’s degree of sense of coherence in life effects their position on the continuum. Three dimensions were introduced as determinants of coping with the stressors that each individual exposed to each day: comprehensibility, manageability and meaningfulness. Antonovsky stated that SOC from the beginning is a cognitive concept; comprehensibility addresses the belief that things happen in an orderly and predictable fashion, and a sense they are understandable events in life and can reasonably predict what will happen in the future. Manageability concerns the skills or abilities that a person possesses, the support or resources necessary to address issues, and the belief that things are manageable and control. Meaningfulness concerns the things in life that are interesting and a source of satisfaction. A strong sense of coherence does not necessarily imply that life as a whole is ordered with high degrees of comprehensibility, manageability and meaningfulness, but areas of subjective importance in a person’s life must fulfill these criteria.

As stressed in 2008, the salutogenic perspective is missing from the research on people with deafblindness. Much of this research has a problem-oriented focus, although the resources that people with this condition possess are briefly described. For example, Ellis and Hodges described how people with USH relate to their diagnose as something that they have internalized or something separate from themselves. A salutogenic perspective might reallocate resources to benefit people with USH from a health perspective.

The inclusion of a salutogenic perspective would complement the picture described in the present thesis of the problems with poor physical, psychological health, social trust and finances among people with USH as well as previous research about people with deafblindness. A salutogenic perspective would enable a more inclusive description of people with USH as well as their health, social trust and individual resources, which are vital for maintaining a high quality of life.
Conclusions

The present thesis describes the poor physical health, psychological health, social trust and financial situation of people with USH compared with a cross-section of the Swedish population. Major problems with fatigue, different types of pain, inabilities to manage problems and feelings of anguish and worry were found. Suicidal behaviors were overrepresented in the USH group, which is an important finding that should be investigated further. A lack of social trust and strained finances were found. People with USH experience problems with going out alone, and they did not feel that they could trust people in general. Some also reported that they did not have anyone with whom to share their innermost feelings and in whom to confide. A strained financial situation was found both with regard to not being able to pay for expenditures such as rent, bills and food but also not having the ability to obtain 15,000 Swedish crowns within a week.

The present thesis argues that an interdisciplinary biopsychosocial perspective is important to describe the health of people with USH, something that has been lacking in previous research. To describe the health of people with USH from one perspective only reduces the complexity of living with USH from a health perspective. Using the Bircher definition of health and disease (which combines both biopsychosocial aspects according to Engel, 1977 and the holistic health perspective represented by the Nordenfelt theory of health (2001)), enables the examination and discussion of the complexity of health for people with USH. This pathway is commensurate with the discussion about quality of life within the disability paradox. Incorporating the salutogenic health perspective fits both with the Bircher definition and the disability paradox as well as makes it possible to expand the knowledge about resources and mechanisms at different levels that construct health and quality of life for people with USH.

The postulation of health, quality of life, and health-related quality of life is, as Bergsma and Engel, stated a jungle of different concepts and overlaps. The intent of the present thesis was to further explore health in a life course perspective using biopsychosocial approach; otherwise, a risk of oversimplification exists. Mechanisms at different levels affect on the health outcome of people with USH, regardless of their diagnosis. In this sense, it is a disability that has functional consequences for everyday life. In this context, the Bircher definition of health provides a theoretical...
framework that includes biological, psychological, social, intentional and cultural dimensions as well as others.
Future Research

The present thesis described the general health, physical health, psychological health, social trust and financial situations of people with USH1, 2 or 3. The need for a biopsychosocial approach was addressed, and this element is what has been lacking in research about the health of people with USH. The current results of the empirical results add more pieces to the puzzle to understand the health of people with USH from a life course perspective. Future research should be both quantitative and qualitative. Future research should attempt to understand the mechanisms in play at different levels that affect the health of people with USH. A need also exists for a salutogenic perspective to identify the variables that promote health and the resources that people with USH possess. Furthermore, the quality of life of people with USH should be addressed and, the strategies that are used when changes occur. Future research should also follow up with the previous surveys that have been conducted, to obtain longitudinal perspective on the health of people with USH. The findings of the included studies that must be scrutinized further, for example, for the possible health benefits among people who have received a cochlear implant (as indicated by study II). The suicidal behavior found in the current thesis has not been previously reported and must be addressed. No current reports exist regarding of how many people with USH successfully commit suicide. In addition, it would be of interest to examine people who have attempted suicide: What help did they receive, and what would they have needed earlier from healthcare or other support service providers?
Sammanfattning på Svenska/Swedish summary

Titel: Hälsa hos Personer med Ushers syndrom.

Introduktion


Det empiriska materialet i denna avhandling utgörs av ett omfattande enkätmateriel, där personer med USH har svarat på en mängd frågor om olika aspekter på hälsa, ångest, depression, social tillit, arbete/sysselsättning, vårdkontakter, ekonomisk situation, alkohol och tobaks användning. Fokus i avhandlingen ligger på generell hälsa, fysisk hälsa, psykologisk hälsa, social tillit och ekonomisk situation.

Att leva en funktionsländersättning behöver inte innebära att livskvaliteten eller hälsan är försämrad. Men tillsammans med andra faktorer så kan ett funktionsländersättning innebära att livskvalitet och hälsa är sämre än i övriga befolkningen. Ett antal studier både nationellt och internationellt har visat att personer med funktionsländersättning har en sämre hälsa än övriga befolkningen, detta beror troligen på sämre levnadsvillkor snarare än funktionsländersättningen i sig. Begränsningar i möjligheten till aktivitet och delaktighet, begränsningar i den omgivande miljön, en ojämlik position på arbetsmarknaden och tillgång på vård och stöd har beskrivits.

Föreliggande avhandling har sina rötter i en tvärvetenskaplig kontext där olika perspektiv; medicin, psykologi, sociologi, pedagogik och socialt arbete, har bidragit till förståelsen av hälsa för personer med USH.
Dövblindhet


"Dövblindhet är ett specifikt funktionshinder.

Dövblindhet är en kombinerad syn- och hörselnedsättning.

Dövblindhet begränsar en persons möjlighet att delta i aktiviteter och inskränker full delaktighet i samhället i sådan grad, att samhället måste underlätta genom att tillhandahålla specifika insatser, anpassa omgivningen och/eller erbjuda tekniska lösningar." 114

Till definitionen finns fem kommentarer som har till syfte att förtydliga definitionen, dessa punkter behandlar syn och hörsel som centrala för informationsutbyte, behovet av specifika insatser och anpassning av omgivningen beroende på när dövblindheten uppstått och dess omfattning, delaktighet och aktivitet, anpassning på jämliga villkor samt vikten av ett tvärvetenskapligt synsätt som även innefattar specifika kunskaper om dövblindhet 114.

Usher syndrom

Kliniskt kan USH delas in i tre olika typer som benämns USH1, USH2 och USH3.78

USH1 innebär en medfödd dövhet samt att balanssinnet i örat inte fungerar, vilket leder till bland annat svårigheter att stabilisera huvudet som spådbarn och försenad gångålder. På grund av den kliniska bilden så upptäcks också RP ganska tidigt hos personer med USH178.

Hos personer med USH2 är den medfödda hörselnedsättningen måttlig till grav. Balanssinnet är inte påverkat98. Hos dessa personer diagnostiseras RP senare, ofta i anslutning till att personen skall ta körkort. Personer med USH2 kan ha haft problem med till exempel mörkerseendet sedan tidigare, men det är inget som föranlett vidare undersökningar.


Personer med USH använder sig av olika sätt för att kommunicera och interagera med omgivningen. Några exempel är läppavläsning, visuellt teckenspråk, taktillt teckenspråk, olika hörsetekniska hjälpmedel (hörapparat, CI, teleslinga, FM-system), men även tekniska lösningar på dator och telefon till exempel förstoring, talsyntes, punktdisplay med mera. Det är inte ovanligt att personer med USH behöver komplettera eller byta strategier för kommunikation vartefter försämring av framförallt synen sker (USH1 och USH2) men även när hörseln försämras (USH3).

Hälsa

Hälsa har intresserat människan sedan urminnes tider, redan i antikens Grekland hade man tankar om vad som var hälsa och hur den skulle upperrätthållas96. Två huvudspår kan identifieras i hur man har sett på hälsa; dels ett biologiskt perspektiv där ohälsa innebär avvikelse i kropp eller organ och god hälsa innebär att kroppens organ fungerar ”felfritt”. Det andra perspektivet brukar beskrivas som det holistiska perspektivet. En av företrädarna är Nordenfelt som menar att hälsa inte kan beskrivas i endast biologiska termer utan även inkluderar psykologiska faktorer samt en persons intention och möjligheter att göra det hon vill118. Världshälsoorganisationen WHO definierar hälsa som ett ”ställande av fysiskt, psykiskt
och socialt välbefinnande, och inte endast frånvaro av sjukdom eller skada”\textsuperscript{139}. Denna definition har fått kritik för att vara normativ och för att det är problematiskt att definiera vad välbefinnande är\textsuperscript{86, 103, 104}. Under 1970-talet växte det fram kritik, framförallt inom psykiatrin, mot det reduktionistiska perspektivet som ansågs vara förknippat med det biologiska synsättet\textsuperscript{46}. Detta ledde till att ett bio-pyskosocialt perspektiv utvecklades, där biologiska, psykologiska och sociala aspekter av hälsa beaktas. Inom detta perspektiv tillskrevs patienten själv ha betydelse genom att berätta om sitt tillstånd och konsekvenser som det medförde. Internationella klassifikationen av funktionsstillstånd, funktionshinder och hälsa, som lanserades av WHO, 2001 kan ses som en vidare utveckling av detta perspektiv\textsuperscript{153}. Genom klassifikationen har man försökt att skapa ett gemensamt sätt att beskriva olika funktionsstillstånd och funktionshinder i relation till hälsa. Klassifikationen består av en tankemodell samt 1500 klassificeringskoder. Med hjälp av ICF skall biologiska företeelser som kropps-funktion och kroppsstruktur, aktivitet, delaktighet och omgivningsfaktorer vara möjliga att beskriva utifrån vad som är underlättande och hindrande för en individ.

**Funktionshinder och hälsa**

Synsättet på funktionsnedsättning respektive funktionshinder har förändrats över tid. Liknande strömningar som finns och har funnits inom hälsoområdet kan återfinnas även här. I ett historiskt perspektiv har personer med funktionsnedsättningar beskrivits i ett biologiskt/medicinskt ramverk, där funktionsnedsättning hos individen är en avvikelse som skall åtgärdas genom medicinska insatser\textsuperscript{129}. En motreaktion till detta perspektiv startade inom det som beskrivs som den sociala modellen, där fokus istället är på samhället och samhälleliga insatser\textsuperscript{138}. Ett samhälle som inte är anpassat leder till att funktionshinder uppstår. Här är inte funktionsnedsättningen i sig intressant utan ansvaret för att funktionshinder inte uppstår ligger hos samhället. Ytterligare perspektiv har utvecklats, där intentionen har varit att kombinera det medicinska med det sociala och i tillägg införliva, kultur, normer, omgivande miljö med mera\textsuperscript{129}.

Inom folkhälsoområdet har personer med funktionsnedsättningar länge varit en exkluderad grupp. Detta beror till stor del på att utgångspunkten inom folkhälsoområdet har varit att förebygga skada, sjukdom och för tidig död\textsuperscript{90}. Under de senaste tio åren har detta börjat förändras och intresse finns inom folkhälsoområdet och inom funktionshindersområdet att överbygga klyftan. Att hälso för personer med olika funktionsnedsätt-
ningar är sämre än i övriga befolkningen har visats i en mängd forskning och rapporter. I Folkhälsomyndighetens två rapporter som kom 2008, beskrivs ohälsa för personer som självskattat sina funktionsnedsättningar inom en rad områden. I rapporterna slås fast att mycket av den ohälsa som beskrivs inte kan härledas till funktionsnedsättningen i sig utan till ojämlika levnadsvillkor inom områden som berör möjligheter till utbildning och arbete, sjukvård på lika villkor\textsuperscript{142,143}.

**Hälsorelaterad forskning om dövblindhet och Usher syndrom**

Inom den genetiska forskningen om dövblindhet är USH ett av de mest kartlagda. I nuläget har 13 gener identifierats som kan ge USH\textsuperscript{95}. Vad gäller hälsorelaterad forskning kring dövblindhet och USH så har forskningen till stor del varit inriktad på att beskriva symptom som ångest och depression, isolering och svårigheter med att behålla sitt oberoende\textsuperscript{18,24,33,99,100,110} liksom begränsningar i delaktighet och aktiviteter\textsuperscript{111}. Vidare beskriver svårigheter som relaterar till möjligheter för kommunikation och interaktion med andra\textsuperscript{71}. Tidigare forskning som studerat hälsa i ett biopsykosocialt perspektiv hos personer med USH saknas.

**Syfte**

Det övergripande syftet i denna avhandling var att beskriva hälsa hos personer med USH. För att uppnå detta syfte genomfördes fyra delstudier, med nedanstående syften:

1. Syftet var att beskriva fysisk och psykisk hälsa hos personer med dövblindhet, orsakad av Usher syndrom typ 2, samt att undersöka skillnader beroende på kön.
2. Syftet var att beskriva fysisk och psykisk hälsa, liksom social tillit hos personer med Usher syndrom typ 3, i relation till grad av syn- och hörselnedsättning.
3. Syftet var att beskriva fysisk och psykisk hälsa, social tillit och ekonomisk situation hos personer med Usher syndrom typ 1, i jämförelse med ett tvärsnitt av Sveriges befolkning.
4. Syftet var att beskriva likheter och skillnader i generell hälsa, fysisk hälsa, psykisk hälsa, social tillit och ekonomisk situation hos personer med Usher syndrom 1, 2 och 3.
Material och metod

De fyra delstudierna bygger på ett omfattande empiriskt material baserat på två olika frågeformulär; den nationella folkhälsoenkäten ”Hälsa på Lika villkor” (HLV) och ”The hospital anxiety and depression scale” (HAD-skalan). De fyra delstudierna är kvantitativa tvärnittsstudier. I studie I och III har studiepopulationen jämförts med en referenspopulation bestående av ett slumpmässigt urval av personer ur Sveriges befolkning som har svarat på HLV. Det empiriska studiematerialet är hämtat från den Svenska Usher databasen vid Audiologiskt forskningscentrum i Örebro. Databasen har funnits i cirka 30 år och inkluderar, förutom enkätdata också syn- och hörseldata, kopior av medicinska journaler, uppgifter om genetisk diagnos för cirka 400 personer med USH. Samtliga av dessa personer har samtyckt till att deras uppgifter används i forskning om USH. Uppdatering av databasen sker regelbundet.

Resultat

Sammantaget visar resultaten i de fyra delstudierna att personer med USH har problem med såväl fysisk som psykisk hälsa. De beskriver bristande social tillit och svårigheter med sin ekonomiska situation. I två av studierna har svaren från personer med USH1 och 2 jämförts med ett urval av Sveriges befolkning (studie I och III). Denna jämförelse gjordes inte vad gäller personer med USH3 var och ett urval av Sveriges befolkning (studie I och III). Denna jämförelse gjordes inte vad gäller personer med USH3 beroende på det låga antalet respondenter (studie II). Den fjärde studien är en inom grupps jämförelse mellan personer med USH, med syftet att om möjligt identifiera variabler (klinisk diagnos, ålder, kön, synfält, synskärpa och grad av hörselnedsättning) som påverkar hälsa hos personer med USH (studie IV).

Det vanligaste problemen som personer med USH1, 2 och 3 rapporterar är trötthet (fatigue) och värk i nacke/axlar. Många rapporterar även sömnsvårigheter, känslor av oro/ängest och huvudvärk. Självmorstankar och självmordsförsök var kraftigt överrepresenterat hos personer med USH, jämfört med referens gruppen. Förekomsten av självmordsförsök varierade mellan 23-53 % och mellan 9 och 20 % i studiepopulationen rapporterade tidigare självmordsförsök. Motsvarande siffror i referensgruppen var 12 % för självmordstankar och 4 % för ett eller flera självmordsförsök. För både självmordstankar och självmordsförsök var det personer med USH2 som stod för den lägsta frekvensen och personer med USH3 den högsta, se studie I, III och IV.
Fler likheter mellan personer med USH1, 2 och 3, än skillnader vad gäller generell hälsa, fysisk, psykisk och social hälsa identifierades. Av 46 variabler som undersöktes identifierades signifikanta skillnader mellan grupperna för elva av variablerna. De flesta skillnaderna som identifierades återfanns inom de variabler som relaterade till social tillit och ekonomisk situation (studie IV).

**Slutsatser**

I föreliggande avhandling har fysisk och psykisk ohälsa samt problem med social tillit och en ansträngd ekonomisk situation beskrivits för personer med USH. Förståelsen av hälsa hos personer med USH bör ske på flera nivåer där biologiska, psykologiska och sociala faktorer på olika sätt påverkar utfallet i hälsa för dessa personer. I avhandlingen argumenterats för att ett tvärvetenskapligt bio-psykosocialt angreppssätt är viktigt för att beskriva hälsa hos personer med USH. Att beskriva hälsa utifrån ett enstaka perspektiv, oavsett vilket, blir begränsande och visar inte den komplexitet som det innebär att leva med USH och hur det kan påverka utfall i hälsa. Vidare saknas ett hälsorömnande perspektiv för att lyfta fram resurser och friskfaktorer för personer med USH i ett hälsperspektiv.
Acknowledgments

It began with a phone call: “-Moa, shouldn’t you take a look at this job listing as a doctoral student at the University of Örebro? They are looking for someone with a background in social science and knowledge of deafblindness.” Thank you, Camilla W, for drawing my attention to this possibility! I also want to thank my dear friend Kicka for believing in me from the very beginning and ending our discussion about what a doctoral education and writing thesis would bring with it, with “I will make the canapés for your dissertation”, not for a second doubting that I would make it.

I am very grateful to the people with Usher syndrome who participated in my research; without you, there would not be any articles or thesis, and my work would have been impossible. I also want to thank those of you who contributed with your knowledge and input in the process of adjusting the questionnaires and those who participated as spokespersons in the process of collecting data. I sincerely hope that the present thesis help to ease the burden put on you by spreading knowledge about living with deafblindness and Usher syndrome.

I want to express my grateful and sincere thanks to my supervisors, Berth Danermark, Claes Möller and Kerstin Möller for patiently guiding me through my journey as a doctoral student. Berth thank you for your astonishing ability to understand what I mean. Claes, your interest in and dedication to the life of people with Usher syndrome and the founding of the Swedish Usher database have significantly improved the possibility of performing research within the field of deafblindness. Kerstin, thanks for your hands-on supervision in the process of becoming a researcher (even if I am not there just yet) and for friendship over the years. Your different perspectives and knowledge have enriched my work and the present thesis.

Many have contributed to and facilitated the process of this work. My most grateful and sincere thanks to all of you at the Swedish Institute of Disability Research (SIDR), School of Health and Medical Sciences, Örebro University as well as those at the Audiological Research Center, University Hospital of Örebro. Colleagues in scientific discussions and friends at coffee breaks and lunches, including Johanna, Lotta, Sif, Julia, Stephen, Emelie, Antonia, Thomas, Åsa, Sara B, Helena, Peter, Susanne, Parivash, Marianne, Frida, Jonas K and Jenny, to quote myself: “It’s ok to talk about anything from science and research to the curtain set and lace.”
Thanks to Ann-Marie Helgstedt for your wonderful work with all of the administrative details that I don’t have a single clue about and for reading the manuscript.

Thanks to Tobias and Jonas B for having a solution to all the technical problems that occurred during this work. Additional thanks goes to you, Jonas, for your help with the diagrams, tables and the picture on the front page. Maybe your hair has become a bit greyer in the process, but mine is not.

Thanks to Camilla S and Hanna for your patient and thorough work with the Usher Database, data collection, help with retrieving data from the database and much more. Margareta, no doctoral student should be without your knowledge of scientific databases, search strategies and not least of all EndNote®.

To my fellow doctoral students and senior researchers engaged in the research of people with deafblindness, Berit, Cecilia, Hans-Erik, Mattias, Agneta (a special thanks for valuable comments of the manuscript!), Berth, Björn, Claes and Kerstin: our discussions and meetings increased my knowledge, and I think together we have been striving toward interdisciplinarity.

I thank Susanna Larsson-Tholén and Björn Lyxell for their constructive critiques and excellent reviews of my thesis work at the 60%-seminar and at the final seminar.

Thanks to the HEAD Graduate School, for providing me with the opportunity to participate in workshops, conferences and meetings, expanding my knowledge of what it is to be a researcher.

I would also like to express my gratitude for the financial support that I received from the Swedish science council, the Stinger foundation, the Foundation of MoGård, the Research Foundation of the Hearing Impaired (HÖrselForskningsfonden), the Silent School Foundation (Tysta skolan), the Association for Retinitis Pigmentosa, and the Örebro Region County.

Over the years, some of you have become close friends; special thanks goes to Ulrika (“...det jag inte kan förändra det kan jag acceptera, och det jag kan acceptera det är redan förändrat” (från ”Tommy och hans mamma”, TAW)) and Sarah G (“Ge mig frid nog att kunna ta ett helvete i taget” (från ”Ett helvete i taget”, TAW)) for always supporting, caring and taking your time, through the laughs and sometimes tears.

A deep and grateful thanks go to all of you who I consider to be part of my big family, in Grönbo and elsewhere, always supporting and encourag-
ing, allowing me to be a “nerd”. To my dear friend Thérèse and her family, consider the paths that life takes.

To my mother and father, deep and grateful thanks for the support and help with practical things during this process, especially during the last intense period. To my sisters and brother and their families, no matter what happens, family is always family, and thanks for being a part of mine. Thanks to my mother in law, Ireen, for help with practical things and taking care of Elis during the summer.

To my animal family, Skrudur, Dalheim and Julius the horses: When I am with you, I do not have to impress anyone. Thanks goes to Atlas (“Scooby Doo”) the dog, unconditionally loving me no matter what, and to Mordor Morr the cat, for doing whatever cats do, and not caring less.

Lastly and most importantly to my small family, Andreas: “I love you!” Elis: “Thanks for keeping me present in the small important things of life” and to the baby to come: Thank you for pushing me to finish this work in time. We long to meet you!

Moa
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