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LIVING WITH OSTEOGENESIS IMPERFECTA

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LIVING WITH OSTEOGENESIS IMPERFECTA

THESIS FOR LICENTIATE DEGREE

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“Jag har uppfattat det (OI) som en,
någon slags medresenär eller kompis”
(intervju 12, sidan 1)

ABSTRACT

Osteogenesis Imperfecta (OI) is a hereditary connective tissue disorder with fragility of the bones as the dominant symptom. There is no cure for OI, but symptoms are treated with medical, orthopaedic and physiotherapy methods. The disease group is very heterogeneous and knowledge about adults living with the milder forms, type I or IV is limited.

This thesis is based on two studies with the overall aim to describe health conditions in adults with OI, type I or IV. The aim of Study I was to describe physical activity, pain, joint mobility, muscle function in lower extremities, quality of life, and satisfaction of life in adults with mild-to-moderate OI. The aim of Study II was to explore experiences of living with OI and how this affects functioning in daily life.

The participants were recruited through patient records from the national resource centre at Karolinska University Hospital, by the Swedish OI association, and among family members of already recruited participants. Inclusion criteria were being over 18 years of age, diagnosed with OI, type I or IV, and ability to walk, with or without walking aid. Forty men and women between 21-71 years were identified during 2002-2009, and 30 out of these consented to participate in both Study I and II. Assessment methods were self-reported questionnaires, a clinical examination, and in-depth interviews.

The results showed that all domains in International Classification and Functioning, disability and health (ICF) were affected by the disease. Pain, balance impairment, scoliosis, kyphoscoliosis, contractures, hyper-mobility of joints, decreased muscle function of lower extremities, hearing deficit and dental problems were found in the domain body function and structure. In the domain activity/participation a decreased health related quality of life, an overall high satisfaction of life, trouble with running, lifting heavy objects and conducting heavy work were found. Despite this, 19 out of 27 reported adherence to the general recommendations for daily physical activity. From the interviews several important environmental factors and personal factors were identified.

The heterogeneity of the disease group makes it difficult to give a general view of an adult person living with OI, type I or IV. Even though the identified impairments, activity limitations, participation restrictions, environmental and personal factors found in this thesis do not apply for all adults with OI, it is important to remember that they may be present when caring and treating an adult with OI.

SAMMANFATTNING

Osteogenesis Imperfecta (OI) är en ovanlig ärftlig bindvävssjukdom med ett skört skelett som det dominerande symptomet. Det finns ingen bot för OI, men det finns symptomatiska behandlingsmetoder som innefattar medicinska, ortopediska och fysioterapeutiska metoder. Sjukdomsgruppen är mycket heterogen och kunskapen om vuxna med de mildare formerna, typ I eller IV, är bristfällig.

Den här avhandlingen baseras på två studier, där det övergripande syftet var att beskriva hälsotillståndet för vuxna med OI, typ I eller IV. Syftet med studie I var att beskriva daglig fysisk aktivitet, förekomst av smärta, ledrörlighet, muskelfunktion i nedre extremiteterna, livskvalitet och livstillfredsställelse hos vuxna med mild-måttlig form av OI. Syftet med studie II var att utforska hur vuxna med OI typ I eller IV upplever att leva med sjukdomen och hur den påverkar den dagliga funktionen.

Rekryteringen av deltagarna skedde dels via nationella resurscentrets patientregister på Karolinska sjukhuset, via den svenska OI-föreningen samt genom redan identifierade familjemedlemmar. Inklusionskriterier var en ålder över 18 år, diagnostiserad med OI, typ I eller IV, och gångare med eller utan gånghjälpmedel. Fyrtio män och kvinnor identifierades under 2002-2009 varav 30 personer, 21-71 år, samtyckte att delta i Studie I och Studie II. Utvärderingen av daglig fysisk aktivitet, smärta, ledrörlighet, muskelfunktion i nedre extremiteterna, livskvalitet och livstillfredsställelse, samt upplevelsen av att leva med OI skedde dels genom självskattningsformulär, genom en klinisk undersökning och djupintervjuer.

Resultaten visade att deltagarna var påverkade inom alla domäner enligt WHO's klassifikation av funktionstillstånd, funktionshinder och hälsa (ICF). Smärta, skolios, kyfos, kontrakturer, överrörliga leder, nedsatt muskel funktion i nedre extremitet, hörsel och tandproblem förekom i domänen kroppsfunction och anatomisk struktur. I domänen aktivitet/delaktighet förekom en nedsatt hälsorelaterad livskvalité, en hög livstillfredsställelse, svårigheter att springa, lyfta tungt och utföra hårt/ansträngande arbete. Trots detta rapporterade 19 av 27 att de uppfyllde de generella rekommendationerna för daglig fysisk aktivitet. Från intervjuerna identifierades flera viktiga omgivnings- och personliga faktorer.

Heterogeniteten i sjukdomsgruppen gör det svårt att ge en generaliserad bild av hur det är att som vuxen leva med OI, typ I eller IV. Även om de i denna avhandling identifierade funktionsnedsättningar, aktivitetsbegränsningar, delaktighetsinskränkningar, omgivnings- och personliga faktorer inte gäller alla vuxna med sjukdomen, är det viktigt att komma ihåg att dessa kan förekomma när man vårdar och behandlar vuxna personer med OI.

LIST OF SCIENTIFIC PAPERS

This thesis is based on the following original papers, which are referred to in the text with their Roman numerals:

- I. Balkefors V, Mattsson E, Pernow Y, Sääf M. Functioning and Quality of Life in Adults with Mild-to-Moderate Osteogenesis Imperfecta.
Physiother Res Int 2013;18:203-11.
- II. Balkefors V, Ståhle A, Biguet G. Being an expert on my rare condition – for better or worse. Adults’ experience of their Osteogenesis imperfecta.
In manuscript.

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LIST OF ABBREVIATIONS

AROM	Active Range Of Motion
ADL	Activities of Daily Living
BMD	Bone Mass Density
DI	Dentinogenesis Imperfecta
DRI	Disability Rating Index
HRQL	Health Related Quality of Life
ICF	International Classification of Functioning, disability and health
IMF	Index of Muscle Function
IPAQ	International Physical Activity Questionnaire
Li-Sat-11	Life Satisfaction -11
MET	Metabolic equivalent
OI	Osteogenesis Imperfecta
PA	Physical activity
PBM	Peak bone mass
SF-36	The Short form 36-item Health Survey
SRH	Self-Rated Health
WCPT	World Confederation of Physical Therapy
WHO	World Health Organization
VAS	Visual analogue scale

1 INTRODUCTION

My interest in the lives of adults with Osteogenesis imperfecta (OI) started when I did my Master of Science education, and had a clinical post at a children's hospital team caring for children with OI. I had the privilege to be educated by the interprofessional OI-team and I had the chance to follow the child and its family through the continuum of care. During this time I met children with different types of OI, but most importantly I met parents who also had OI as the disease is hereditary. I noticed that these adults had little or no experience of or access to the same interprofessional team care as that of the child. During this time I had a chance to informally interview the parents and also to read the existing literature concerning OI. I noticed that the literature was limited when it comes to adults with OI, type I or IV. Often these types are described as mild to moderate, almost "normal" whereas the parents reported difficulties that the health care might be able to support further. My curiosity made me want to explore how adults with OI managed to live with their chronic disease and how they, in spite of the difficulties that clearly exists, seemed to be able to live a relatively normal life.

1.1 OSTEOGENESIS IMPERFECTA

Osteogenesis Imperfecta, (OI) is a hereditary connective tissue disorder (1). The most common kind is autosomal dominant, i.e. one of the parents have one mutant gene and might pass OI on to their child. The chance to pass it on is 50 % in each pregnancy. Autosomal recessive OI also exists, but is rare and not seen in OI type I or IV. Osteogenesis Imperfecta can also occur as a new mutation (2).

The prevalence of OI is uncertain due to the fact that the very mild forms might be undetected, but approximately 1:10 000 live births have been mentioned recently (2). According to the National Board of Health and Welfare in Sweden, OI occurs in between six to 20 individuals per 100 000 inhabitants and equally in males and females. In most cases the underlying pathology is believed to be a defect in collagen, both quality and quantity (1). Collagen is the protein that makes/gives structure to bone but also occurs in ligaments, the white of the eye and in the teeth (3). Osteogenesis imperfecta affects the person on both an organic and functional level. Fragility of the bones is the dominant symptom. Other symptoms are short stature, bowing of the long bones, increased joint flexibility and fragile vessels (1,4).

Osteogenesis Imperfecta affects both heart and lung function, especially type III and partly type IV (4,6). As clinical features the following may occur: blue/black sclera (the white of the eye), brittle teeth (Dentinogenesis imperfecta), hearing impairment, and thin soft skin. The disease also results in secondary bone deformities following fractures and vertebra compressions. Functional ability depends on the form of OI (1,4). In the OI literature, type I has been described as normal or with little clinical attributes and type IV as mild deformity of bones and mild to moderate short height (7,3). It has also been pointed out that fracture rates in OI type I decreases after puberty, but that the fracture frequency might increase again with

ageing, in both men and women (3). Further, persons with OI type I have a normal life expectancy (8).

Some literature suggest an addition of descriptive words and classifications such as mild, lethal, severe progressive and moderate severe form when describing OI. Lindahl et al. (9) presents an overview of new OI types discovered after the first classification by Silience et al. consisting of 17 different types. However, the classification by Silience et al. (table 1) is still the base and widely used due to difficulties in how to best define and classify this disorder.

As a disease group the symptoms are very heterogeneous and clinical features differ between as well as within individuals and over time (1,4). There is no cure for OI. Symptoms are treated with physiotherapy, orthopaedic and medical methods (3). The goal of rehabilitation is improvement of physical function (10,11).

Table 1. Classification according to Silience et al. 1979 (7).

Type IA	Normal height and little or no skeletal features, blue white of the eye (sclera), 50% loss hearing. Autosomal dominant.
Type IB	Type IA with addition of Dentinogenesis imperfecta.
Type II	The most severe form. The child dies at birth or during new-born. Autosomal dominant.
Type III	Substantial skeletal changes at birth. Sclera varying nuances, common with Dentinogenesis imperfecta and hearing loss. Short stature. Autosomal dominant or recessive.
Type IVA	Normal sclera, moderate skeletal changes, and varying length. Sometimes hearing loss. Autosomal dominant.
Type IVB	Type IVA with addition of Dentinogenesis imperfecta.

1.2 TREATMENT OF OSTEOGENESIS IMPERFECTA

Treatment should be multidisciplinary with involvement of an endocrinologist, rehabilitation specialist, orthopaedic surgeon, dentist, geneticist, social workers/psychologist, physiotherapist and occupational therapist to be able to address important issues for a person living with OI (11). It is essential with a specialist team for providing the support, information and care the person with OI and their families ask for (12,13). Already in the beginning of 1990s Byers & Steiner (3) pointed out that the care for adults with OI is fragmented and there would be benefits in developing specialised assembled care such as that provided for children with OI.

1.2.1 Medical treatment

Bisphosphonates are the most commonly used medical treatment of OI (14). It is not curative but a supplement to other symptomatic treatments. Bisphosphonates works as osteoclast inhibitors, i.e. the osteoclasts do not resorb the bone to the usual extent (15) leading to increased bone density. There is an agreement that bisphosphonate increases bone mineral density (BMD) (9,16,17), but it is not yet established if the treatment affects fracture incidence and improves function. Also the optimal method (oral or intravenous), dosage, length of therapy and optimal therapeutic window are still not known (9). The treatment has been evaluated in several studies (18-24), where the participants are mostly children. However, two studies (23,24) concluded both that bisphosphonate given to adults with OI increases bone density and the first (23) also associated it with lower risk for fractures, but the second (24) points out a need for further studies to evaluate the effect on fracture incidence.

1.2.2 Orthopaedic treatment

Indications for orthopaedic/surgical treatment are fractures and recurrent fractures and severe bowing of bones. Treatment consists of fracture management with external fixation (casting/splinting and braces), aiming for immobilisation time to be as short as possible to prevent further decline in bone strength and secondary deformities due to this fixation. Further, internal fixation and correction of long bone bowing with osteotomy and intra medullar rodding, i.e. rods inserted into the marrow of the long bones are methods used (3,16,25,26). One study indicated reduced pain and increased walking capacity when hip and knee replacement surgery was performed in six patients aged 46-62 years (27).

If there is scoliosis present and the curve is progressive and/or affects the person negatively in terms of respiratory function and/or back pain, spinal fusion might be the option. However, the treatment and the decision must be well thought through, both if the surgery is appropriate and when it should be performed to get the best result for the individual (25). Another alternative is external fixation with a brace, but with caution, because of the risk of deformities of the rib cage, thereby affecting lung function.

1.2.3 Physiotherapy

The world confederation for physical therapy (WCPT) describe physiotherapy as:

“Providing services to individuals and populations in order to develop, maintain and restore maximum movement and functional ability throughout the lifespan. This includes providing services in circumstances where movement and function are threatened by ageing, injury, disease or environmental factors. Functional movement is central to what it means to be healthy. Physical therapy is also important for identifying and maximizing quality of life and

ability for movement in the scopes of promotion, prevention, treatment/intervention, habilitation and rehabilitation. This includes physical, psychological, emotional, and social well-being. Further, physical therapy involves the interaction between physical therapist, patients/clients, other health professionals, families, care givers, and communities in a process where movement potential is assessed and goals are agreed upon, using knowledge and skills unique to physical therapists” (28).

Countries have their own definition of physiotherapy and the Swedish association of physiotherapy describe it as follows:

“A knowledge and professional area, where these two are interacting with each other. In the area of knowledge, the science of physiotherapy, characterized by the view of humans as physical, social and existential whole in a health perspective. In centre of knowledge stands the understanding of, that inside the human are forces that heals and resources to change and these can be activated with movement in a therapeutic interaction. Through awareness of the body and adequate movement the human can influence his/her health. Movement are considered a ground to the whole human function and as a tool to reach individual goals in relation to the environment” (29).

Physical activity (PA) is an important part of physiotherapy treatment and is defined as: *“any bodily movement produced by skeletal muscles that results in energy expenditure”* (30). Physical activity in daily life can be categorised into occupational, sports, conditioning, household, or other activities. Exercise is a subset of PA that is planned, structured, and repetitive resulting in improved or maintained physical fitness. Physical fitness is a set of attributes that are either health- or skill-related (30).

It is known that bone mass and trabecular orientations are adapted to the intensity and directionality of external forces and that physical activity increases bone mass (31). The design of exercise toward maintaining or improving bone density, movements that cause high loads at high rates and load from variable directions should be used (32). Studies support the idea that high intensity exercise can increase or maintain bone mass in premenopausal women (33,34).

Most studies in this area are focused on children, and have shown that physiotherapy is important for the child with OI when developing motor skills, but also to make sure that weight bearing activities are possible to perform, both to minimise future fractures but also when recovering from fractures (16). Recommendations for children with OI type I and IV are to promote an active life style and moderate aerobic training in combination with strengthening exercises without weights (35). Further, exercise should be supervised and performed with safe activities chosen to reduce the risk of injury. Contraindications for exercise for children with OI are participation in contact sports, sports with sudden rotation moments and any form of intense interval training (35). Children and adolescents with OI have been shown to have lower aerobic capacity and muscle strength than peers (36). However, it has also been shown a significant improvement in these parameters in

comparison with healthy peers after participating in a 12 weeks (two times per week) exercise program (37).

There is a limited amount of studies concerning exercise recommendations and exercise programs for adults with OI, type I or IV. One article has described a rehabilitation program for children (38), but specific exercise recommendations and rehabilitation programs for adults with OI, type I or IV, are lacking. The heterogeneity of the condition makes it hard to present a specific exercise program that can be used for all patients with OI. However, physical activity is important in preventing chronic diseases, for example cardiovascular diseases and diabetes type 2 (39). International recommendations state that adults need at least 30 minutes of moderate-intensity activity preferably every day of the week, and at least ten minutes at a time (39). Very little has been written about physical activity and adults with OI and whether individuals with OI type I or IV are able to follow the recommendations or not.

1.3 THE INTERNATIONAL CLASSIFICATION OF FUNCTIONING, DISABILITY AND HEALTH (ICF)

In the present thesis the International Classification of Functioning, disability and health (ICF) is used as a theoretical and conceptual framework. It was developed by the World Health Organization, WHO (40). The International Classification of Functioning, disability and health describes all aspects of human health and health related conditions. The overall aim is to provide a unified and standardised language for describing and classifying health domains and health related states.

Health and health related states that are connected to different health conditions can be described by using the ICF, giving a description of situations according to human functioning conditions and restrictions. It serves as a frame to organise the information, gives it a structure so it can be presented in a meaningful, all together easy manner. The ICF consists of two parts. The first part is functioning and disability, with the components body functions and structures, and activity and participation. The other part consists of contextual factors with the components environmental and personal factors (40), figure 1.

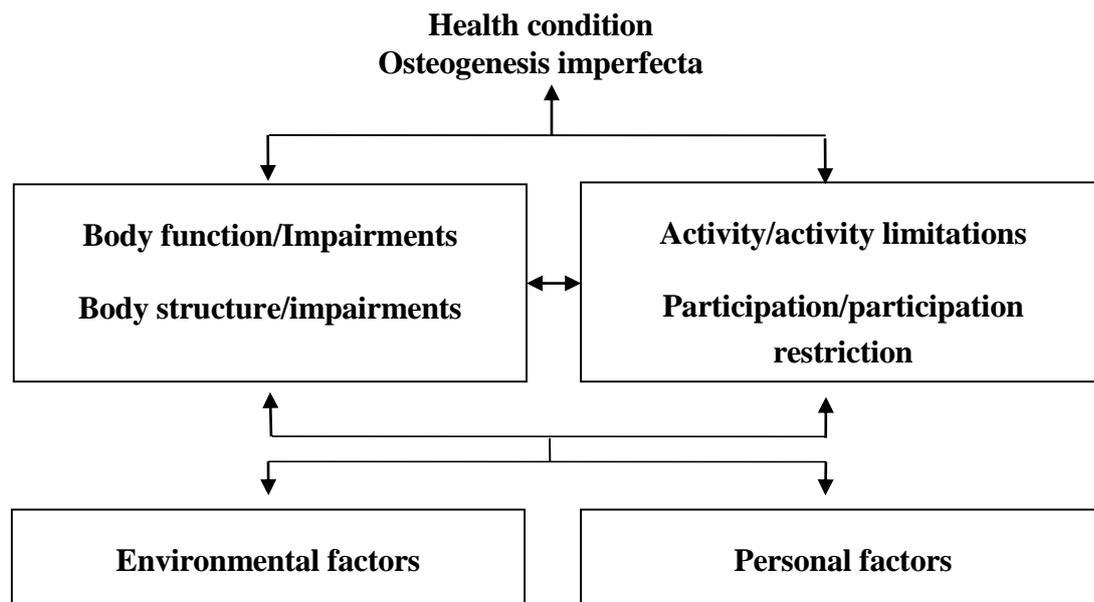


Figure 1. Diagram of the ICF domains and their interaction with each other. Reproduced with permission from WHO permission team.

Personal factors are not classified in ICF due to social and cultural variance. However, personal factors contribute to a more comprehensive description of a health condition because they deal with the individual's life and living (e.g. other health conditions, lifestyle, habits, coping styles, social background, education, profession) (40). From evidence and experts opinions the definition of ICF Core Sets have been developed to make ICF more useful for clinical practice. The ICF Core sets is a list of categories relevant to most persons with a specific health condition and for the health professionals giving the care for this condition (41). The ICF Core Sets for Osteoporosis (42) have been defined and also contextual factors concerning physical disability due to four different musculoskeletal conditions: Rheumatoid arthritis, Low back pain, Osteoarthritis and Osteoporosis are described (43). These are listed in table 2. Existing core sets might be useful in clinical practice to promote teamwork communication and when describing functioning and health in persons with OI, since the dominant symptom in OI is fragility of bones and may therefore be considered as a musculoskeletal condition. The ICF is recommended by the WHO to be used when developing equal health care where the care is patient centred and inter-professional. A health care that makes sure that the person is central in decision making in their life and understands the personal needs for a reasonable good living. In a study where students and supervisors were using ICF during assessments of patients, the conclusion was that ICF made it easier to achieve patient-centred care and strengthen the importance of contextual factors as well as the promotion of teamwork and in addition the patients felt listened to and cared for (44).

Further, contextual factors should be addressed in the prevention of musculoskeletal conditions and in prevention of disability when a condition is present such as OI (43). Most of the literature concerns the symptomatic treatments such as bisphosphonates and surgical treatments with aim to increase physical functioning outcomes. Continuous physiotherapy and muscle training is highlighted in one study as probably the most important part when describing interprofessional care and symptomatic treatments for OI (45).

Table 2. Contextual factors relevant to disability in musculoskeletal conditions.

Environmental factors	Personal factors
Obesity	Coping strategies
Drugs	Self-efficacy
Walking aids	Beliefs and attitudes of the person towards disease and disability
Cars	Stress
Work environment	Depression
Functional access to public buildings	Physical activity/level of fitness
Modification of the home	Comorbidity
Weather and climate	Socioeconomic status
Immediate family	Educational status
Health professionals	Profession/working conditions
Attitudes	Job satisfaction
Public transportations	
Mobility service	
Social security services	
Waiting lists for care	

1.4 HEALTH RELATED QUALITY OF LIFE

Quality of life is defined by WHO as “an individual’s perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards and concerns” (46). Health Related Quality of Life (HRQL) is important when living with rare and/or chronic conditions. The main purpose is to assess and illuminate the burden of the disease (47). Self-Rated-Health (SRH) is a formalised measure of subjective health and has been found to be an independent predictor of mortality, but also useful for measuring clinical outcome (48). These instruments make it possible to compare across health conditions, populations and interventions concerning functioning and health (49).

In this thesis the Short form 36-item Health Survey (SF-36) was used for assessing health related quality of life (HRQL) and Life Satisfaction -11 (LiSat-11) for life satisfaction. The SF-36 is the most widely used general health status instrument (49). The SF-36 does not cover environmental factors, but provides useful information on functioning and disability. The SF-36 has been used in a study concerning adults with OI in the US (50) which concluded that adults with OI had significantly lower outcomes on the physical function

parts compared to adult norm data but the mental parts were equal when compared with adult norm data. Assessing life satisfaction with LiSat-11 gives an added dimension to the health related quality outcomes measured with SF-36. When describing HRQL, ICF has been demonstrated to be useful and may serve as a common framework (49).

1.5 RATIONAL FOR THE PRESENT THESIS

Osteogenesis Imperfecta is a rare condition and health professionals in general have limited experience of the disease. Specialist teams are needed to provide a good support to children with OI and their families, with a broader focus than only on medical issues. In Sweden specialised assembled care is provided for children and adolescents with OI \leq 18 years of age, but not for adults. There is a lack of knowledge about adults with OI, type I or IV, concerning physical activity level, pain incidence, joint mobility, muscle function, quality and satisfaction of life. There is also a need for a deeper understanding of the complex relationship between disability, environmental factors, personal factors and health in everyday living with OI.

2 AIM

The overall aim of this thesis was to describe the health conditions in adults with Osteogenesis Imperfecta, type I or IV.

The aim of **study I** was to describe physical activity, pain incidence, joint mobility, muscle function, quality and satisfaction of life in adults with mild-to-moderate Osteogenesis Imperfecta, to form the basis of improved clinical care and physiotherapy treatment.

The aim of **study II** was to explore the experiences of adults living with Osteogenesis Imperfecta, type I or IV, and how it might affect functioning in daily life.

3 METHODS

3.1 DESIGN

The present thesis is descriptive and ICF is used as a theoretical and conceptual framework to describe and discuss the health condition of adults living with OI, type I or IV. Study I is a cross sectional study involving a clinical assessment and self-assessed questionnaires. Study II is an explorative qualitative interview study where the participants' individual perceptions about living with the condition are recognized.

3.2 PARTICIPANTS

Inclusion criteria in Study I and II were being over 18 years of age, diagnosed with OI, type I or IV and ability to walk, with or without a walking aid. Forty persons with OI type I or IV were during 2002-2009 identified mainly through patient records at the national resource centre at Karolinska University Hospital, but also by the Swedish OI association or by already identified family members. Everyone identified was informed and asked to participate by an information letter (some also by their physician). Thirty individuals consented to participate in both Study I and II. Ten persons chose not to participate mostly because of lack of time and acute illness not related to OI.

One participant was excluded from Study I due to uncompleted questionnaires. Thus, 29 participants were included in Study I and 30 in Study II.

Median age of the participants was 41 (range: 21-71) years. Median length of the female participants' was 158 cm (range: 130-170) and of the male participants 171 cm (range: 160-178). Median body weight of the female participants was 61 kg (range: 44-77) and of the male participants 79 kg (range: 64-116). Median Body Mass Index (BMI) of the female participants was 24 kg/m² (range: 19-35) and of the male participants 26 (range: 23-37). Twenty-four participants reported having an elderly relative also having OI.

Four participants were not born in Sweden, but spoke Swedish well. Table 3 describes the participants' demographic data further.

Table 3. Number of participants and demographics for Study I and II.

Demographic variable	Number of participants
Participants Study I (Female/male)	18/11
Participants Study II (Female/male)	18/12
20-29 years old	6
30-39 years old	8
40-49 years old	7
50-59 years old	5
60 + years old	4
≥ 5 fractures	21
< 5 fractures	9
Upper extremity fractures	22
Spinal fractures/compression	11
Lower extremity fractures	27
Worked full time	23
Active work with physical demands	8
Sedentary work situation	15
Studying in higher education	3
Pensioners	3
Disability pension	1
In a relationship	21
Having children	18
Child/children with OI	9
Diagnose investigation at children specialist centre	4
Walking aid	3
Wheel chair transporting longer distance	1
Shoe insertions or specially made shoes	11
Length difference between their legs	14
Lived in Stockholm and suburbs	21
Lived in smaller towns and rural areas in the middle part of Sweden	9

3.3 DATACOLLECTION

An information letter was sent to the identified adults with OI and included both Study I and Study II, i.e. the clinical assessment and answering questionnaires (Study I) and the interview (Study II). All data was collected on one single occasion. The individuals needed to respond to the information letter if they decided to participate and then, prior to the interview and clinical assessment, the self-assessed questionnaires and pain drawing were sent by mail for completion at home and brought to the test occasion. The interview and clinical assessment were conducted at three different hospital sites in Stockholm or in the participants own home, and the total time for the meeting was 75-180 minutes. The assessment started with questions about demographic data and then continued with the interviews that were taped and transcribed partly by the author and partly by a secretary not involved in the study in any other way. The interviews lasted between 15-120 minutes. All data collection were performed by the same physiotherapist (VB). Data collection methods were selected so it would be easy to bring equipment required if the participants wanted the interview and clinical assessment to be conducted in their own home.

Some of the participants were excluded from different parts of Study I due to uncompleted questionnaires or their own decision not to participate in a clinical assessment they considered as a risk for injury. Table 4 gives an overview of number of participants included in the particular data collection method.

Table 4. Overview of number of participants in the different parts of this thesis.

Data collection method	Number of included participants
Pain, pain drawing	29/29
Joint mobility, goniometer	28/29
Spine, kyphometer	25/29
Lower extremity muscle function, IMF	29/29
Daily physical activity, DRI	29/29
Physical activity, IPAQ	27/29
Health related quality of life, SF-36	28/29
Life satisfaction, LiSat-11	29/29
Interview	30/30

3.4 MEASUREMENT METHODS

3.4.1 Clinical assessment - Study I

All methods used in the clinical assessment are mainly described in ICF domain body function/impairment except for the Index of Muscle Function (IMF), which had one item concerning the domain activity/activity limitations (general indoors walking ability).

Curvature of the spine was measured with a Kyphometer (Debrunner's Kyphometer, original design, Protek AG, Bern, Switzerland). The instrument is placed on palpated referent points between Th11-12 and S1-2 and between Th 2-3 and Th11-12 (51).

Visible scoliosis was noted in standing and forward bending.

Range of motion (ROM) was evaluated with a goniometer in degrees (52). Measurements were performed with the person in a supine position. Hip flexion, knee extension/flexion and elbow extension/flexion were also measured. The method has been shown to have good intra-reliability with repeated measures in a standardised environment, and with standardised reference points and body positions (53).

Muscle function in lower extremity was assessed with IMF (54). An observer scores the performance using a three-point scale (0-2). Areas evaluated are general movement and muscle function, muscle strength, endurance and balance/co-ordination. The IMF has been found to be reliable and valid for persons with rheumatoid arthritis or osteoarthritis of the hip or knee (55).

3.4.2 Questionnaires – Study I

Pain is described within the ICF domain body function/impairment. Pain was assessed using a pain-drawing instrument where the person describes the pain with a word, for example dull or shooting. The pain drawing is tested for reliability for patients with chronic pain (56,57).

Daily physical activity was assessed with the Disability Rating Index (DRI) (58). The DRI's items are described in the ICF domains activity/activity limitations, participation/participation restrictions and partly in environmental factors. The DRI is a questionnaire covering 12 items concerning common daily activities. The person marks on a 100-mm visual analogue scale (VAS) in accordance to his/her presumed ability to perform the daily physical activities in question. Zero stands for performance without any problem and 100 stands for impossible to perform. The DRI has shown good reliability, validity and sensitivity to clinical changes (58).

Physical activity (PA) was assessed using the International Physical Activity Questionnaire (IPAQ) (59). The IPAQ questionnaire is described within the ICF domain activity/activity limitations. The questions concern time spent walking, moderate-intensity and vigorous-intensity activities during one day of a normal week. Exercise intensity is based on the Metabolic Equivalent (MET) system. The amount of oxygen the body consumes is directly proportional to the energy expended during PA. At rest, the body uses approximately 3.5 ml of oxygen per kilogram of body weight per minute ($3.5 \text{ ml} \cdot \text{kg}^{-1} \cdot \text{min}^{-1}$). This resting metabolic rate is referred to as 1.0 MET (60). A classification of the relationship between the intensity of an activity and the required oxygen levels and a list of MET intensities for various PA has been developed (61). The IPAQ guidelines recommend that data is presented as median MET-minutes/week. This is calculated as number of minutes during a

normal day multiplied by number of days and intensity level of the activity (IPAQ, 2005). Intensity levels were decided as follows: vigorous intensity = 8 MET, moderate intensity = 4 MET and walking = 3.3 MET. The results of three levels of PA are presented as low, moderate and high depending on the frequency (number of days) and time spent performing at the three activity intensities mentioned. The result is compared to a Swedish adult population data (62). The IPAQ version used has been tested for reliability (59,63). Time spent sitting is presented as minutes/week.

Health related quality of life was assessed with the self-administrated questionnaire SF-36 (64). The SF-36 is described within the ICF domains activity/activity limitations and participation/participation restrictions. The questionnaire comprises of 36 questions with fixed answers on different raw scales. The questions are divided into eight domains: physical functioning, role-physical, bodily pain, general health, vitality, social functioning, role-emotional and mental health. Scores vary between 0-100. High score indicates high quality of life. The result are compared with norm data for the Swedish population (65).

Life satisfaction was assessed with the questionnaire Li Sat-11 (66); a questionnaire with 11 questions concerning life satisfaction. The person estimates each question on a six-point scale (1-6). One is not at all satisfied and six is completely satisfied (66). The items included in the Li-Sat's questionnaire are mainly described within the ICF domain participation/participation restrictions and partly environmental factors.

3.4.3 Interview - Study II

The results from the interviews are described in all ICF domains, but most important in contextual and personal factors. Individual in-depth interviews were performed in order to add information that the clinical assessment and questionnaires failed to capture and to assess the persons own experiences of living with the disease. An interview guide was created together with a physiotherapist experienced in interview methodology. The initial question was: "Could you describe what living with OI means for you?" Then four areas were covered: limitations and consequences in daily life, problem solving strategies, suggestions to others living with OI and positive experiences. The interview ended with a question where the person could add information important for them concerning OI. The interviews were taped and transcribed word by word and then analysed with inductive qualitative content analysis (67,68).

Figure 2 shows an overview of the methods used to collect data and in which ICF domains the result from the methods are described.

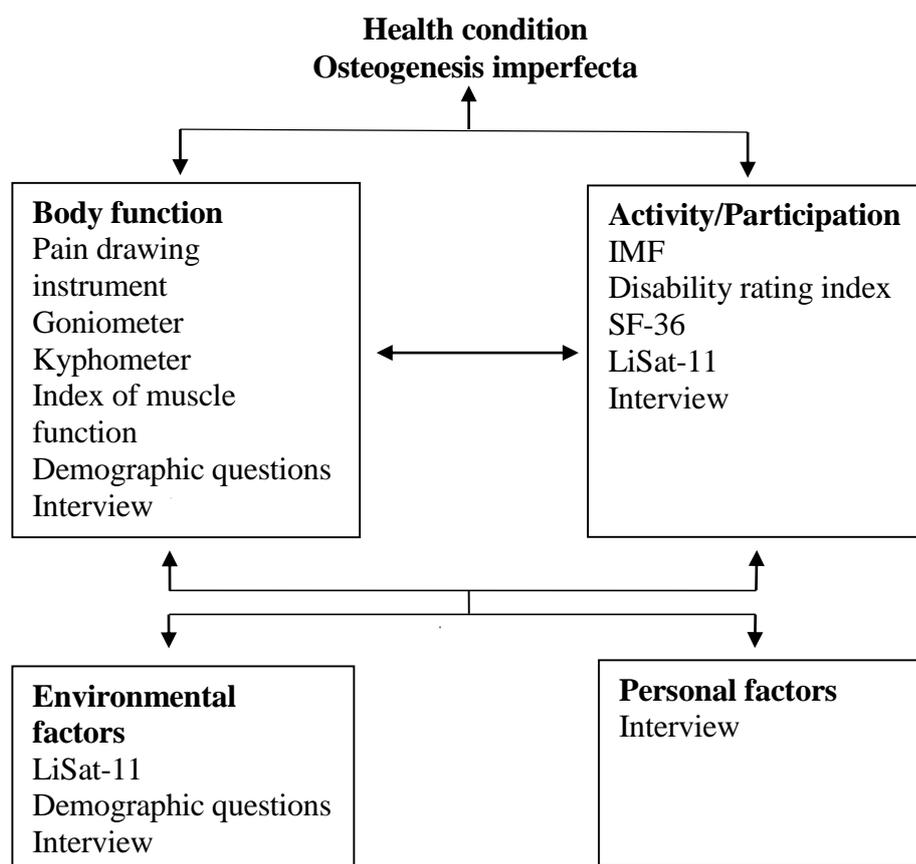


Figure 2. Overview of methods and in which domain the information is described in the ICF.

3.5 DATA ANALYSIS

In Study I descriptive statistics were used. The results are presented with mean, median and range. Mann Whitney U-test for analysis of differences between groups and t-test for single samples were used to test differences between results of participants and norm data. A significance level was set to $p > 0.05$.

In Study II qualitative content analysis (67,68) was used to analyse the transcripts, consisting of 359 pages. The transcripts were read several times and reduced to meaning units which after that were condensed and labelled with codes. Preliminary sub-categories and categories were abstracted. During the whole process the emerging result was discussed with a person with great experience of qualitative methods. After thorough discussions concerning alternative interpretations and underlying meanings in the texts an overall theme, categories with several sub-categories could be described.

3.6 ETHIC APPROVAL AND CONSIDERATIONS

This thesis is a part of a multi-centre study that evaluates the effects of bisphosphonates. The multi-centre study was approved by the Karolinska Institutets Regional Ethics Committee for research, Solna, Dnr: 02-261. The two studies in this thesis, were considered included in the ethical application at the time when it was submitted.

However, this thesis does not take into consideration whether the participants were treated with bisphosphonates or not. Thus, this thesis does not include interventions.

All participants were informed that they could withdraw from the study at any time and it would not affect any future care and participants who did not have former contact with an endocrinologist were offered an appointment if they so wished. To ensure the participants identities were not revealed the presentation has been carefully thought through and the results are presented as a group, although OI is a very heterogeneous disorder. Therefore, it is important to remember that the description does not apply for every person with OI, type I or IV, but it is possible when caring and treating a person with OI that these described disabilities and contextual factors may be present.

4 RESULTS

Health in adults with OI, type I or IV, may be influenced by their disease in all ICF domains. In these participants' participation and contextual factors are important to recognise, for optimising functioning and minimising disabilities. From the interviews in Study II environmental and personal factors were identified from the overall theme, categories and sub-categories shown in table 5.

Table 5. Overview of the overall theme, categories and sub-categories regarding being an adult living with OI, type I and IV found in Study II.

Overall theme	Being an expert on OI - for better or worse	
Categories	To confront a wide range of challenges	To develop creative strategies and change attitudes – an ongoing process
Sub-categories	<i>Being restricted</i> <i>Being different, marginalised and excluded</i> <i>Being forced to represent your own interests</i> <i>Feeling guilt</i> <i>Being concerned about an uncertain future</i>	<i>Striving for normality</i> <i>Striving to adjust and adapt</i> <i>Striving to be foresighted and proactive</i>

4.1 BODY FUNCTION AND STRUCTURE

Osteogenesis imperfecta affects the person with OI on the domain impairments of body function and structure in the ICF.

The clinical assessment revealed that 13 of 25 participants had a kyphosis 40 degrees or more, measured with a kyphometer. Twenty-three had a clinical visible scoliosis. The participants were very heterogeneous according to joint active range of motion. Both hyper-mobility and hypo-mobility were shown between the participants, but also a single participant could differ in joint range of motion in their legs and arms. Hypo-mobility was almost always due to earlier injuries and secondary deformation of the joint or bone.

Lower extremity muscle function showed that some had difficulties with muscle strength, balance and endurance and some had no problems at all. Regarding muscle strength 10

participants had no problem performing the tasks and 19 had problems. Median for the whole group was 3 (range 0-14). Regarding balance 8 participants had no difficulties performing the tasks but 21 had. Median for the whole group was 3 (range 0-10). On endurance measurements 15 had no problem performing the tasks and 11 had some problems and 3 participants had difficulties performing the tasks. Median for the whole group 0 (range 0-10). Two participants were having extensive difficulties with the whole test.

Twenty-five of the participants reported pain. Fifteen had neck and shoulder pain, 11 had chest and back pain, 8 participants reported pain in their feet and the clinical assessment revealed that several of the participants had fallen arches, probably due to loose ligaments between bones in the feet. Table 6 shows what kind of sensation they perceived, and how it was marked according to the pain drawing instrument. An example of other described types of pain is “it is like you are pressed together from the top but also from the sides” (back pain).

Table 6. Words for description of pain sensations in the pain drawing instrument.

 Dull	 Numb
 Cutting (“soda feeling”)	 Muscle spasm
 Burning	 Other, describe the type of pain

Figure 3 shows an overview of the pain drawing, the localisation where the participants reported pain. Nine participants reported having pain in 5 or more localisations in their body at the time of filling in the Pain drawing instrument. During the interviews the acute pain when injured was also mentioned as well as a sense of discomfort. Further, on the SF-36 the body pain component was lower in comparison with Swedish norm data (figure 4, on next page). Fourteen perceived a length difference in their legs. Some had hearing deficits and teeth problems. They described that their teeth were worn down and cracked easily.

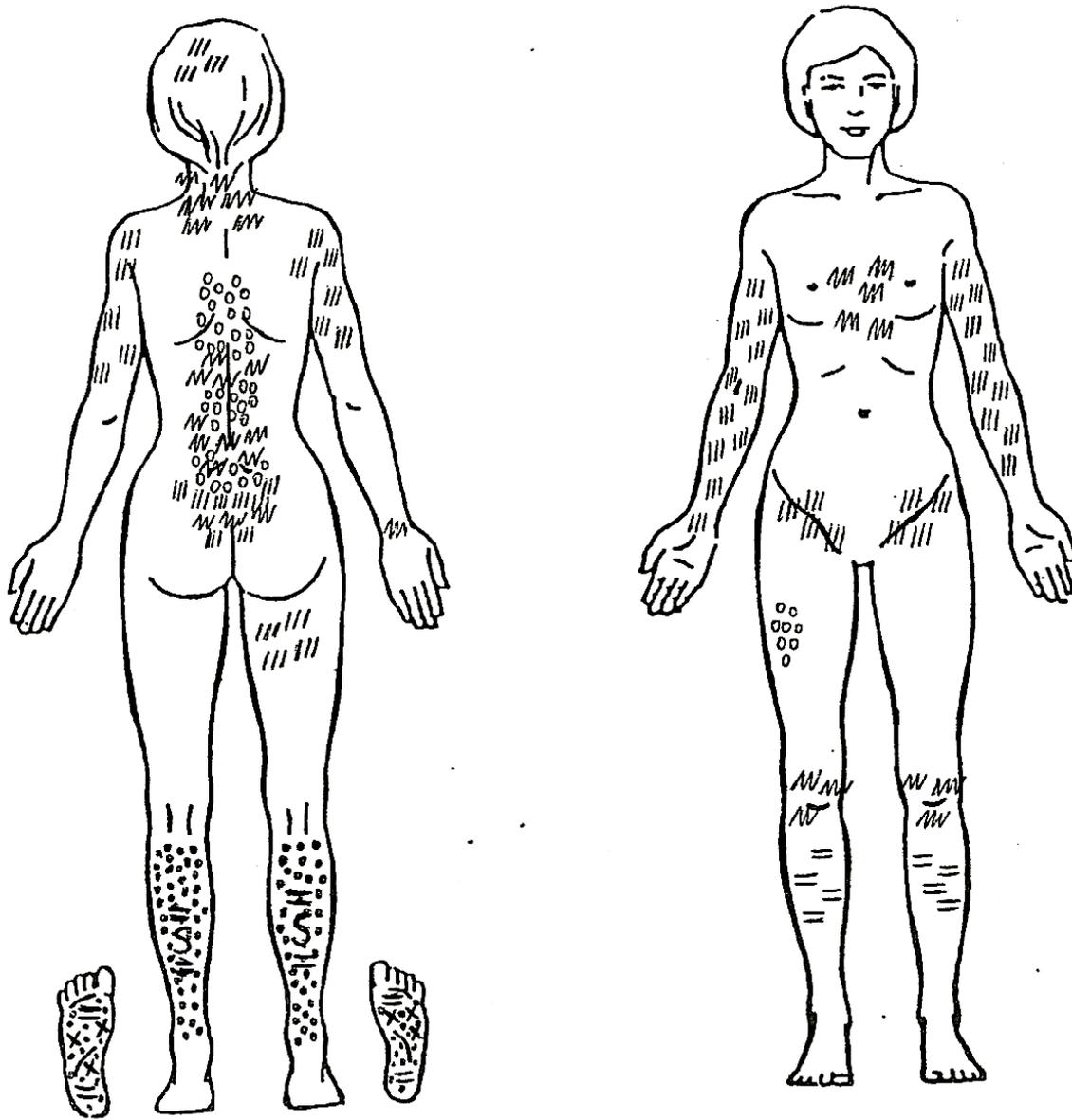


Figure 3. Illustration of the participants reported pain sensations and localisations, assembled in one pain drawing by the author.

4.2 ACTIVITY AND PARTICIPATION

Daily physical activities such as running, lifting of heavy objects, heavy work and participation in sports were reported as hardest to perform on the DRI questionnaire (table 7).

Table 7. Physical activity assessed with the Disability Rating Index (DRI) presented by median and range on a Visual Analogue Scale, 0-100 millimetre, n=29. 0=without difficulty. 100=Not at all. Number of participants marking ≥ 50 on the scale presented separately. Reproduced from Study I.

	Median	Range	n ≥ 50
Running	91	0-100	18
Lifting heavy objects	74	0-100	15
Heavy work	40	0-100	13
Participating in exercise/sports	28	0-98	9
Climbing stairs	12	0-94	6
Carrying a bag	12	0-96	6
Standing bent over a sink	9	0-93	4
Outdoor walks	4	0-87	5
Light work	3	0-82	4
Sitting longer time	2	0-93	3
Making a bed	2	0-92	3
Dressing (without help)	1	0-46	0

Participants estimated a median of total physical activity of 1 608 MET-min/week. In comparison with a normal Swedish population (n=1 000) (62) that estimated 1 119 Met min/week. Nineteen participants estimated that they adhered to the recommended 30 minutes of moderate-intensity activity preferably every day of the week. Two participants reported no physical activity during a normal week and six participant reported low level. Thus, 8 did not reach recommended physical activity during a normal week. The time the participants estimated spent in sitting was median 3 600 minutes/week, i.e. 60 hour/week.

Health related quality of life assessed with the SF-36 was lower when compared to Swedish norm data (65) (figure 4). The domain Physical Function comprises of activities performed daily, e.g. running, lifting heavy objects, climbing stairs, walking, and the domain Role Physical comprises difficulties in performing work related activities or other daily activities due to the persons health condition. Vitality relates to how strong, full of energy and how tired and exhausted the participants felt during the past four weeks. The domain Social Functioning covers how the bodily health condition disturb the ability to consort together with family, friends, neighbours and others. The domain Role Emotional covers the ability to perform work and other activities concerning time spent doing it, the quantity and quality of performed work. Mental health relates to how unhappy, low and depressed or happy, calm

and harmonious the participant feels. The four latter domains are described as mental components and seems somewhat less negatively influenced in adults with OI.

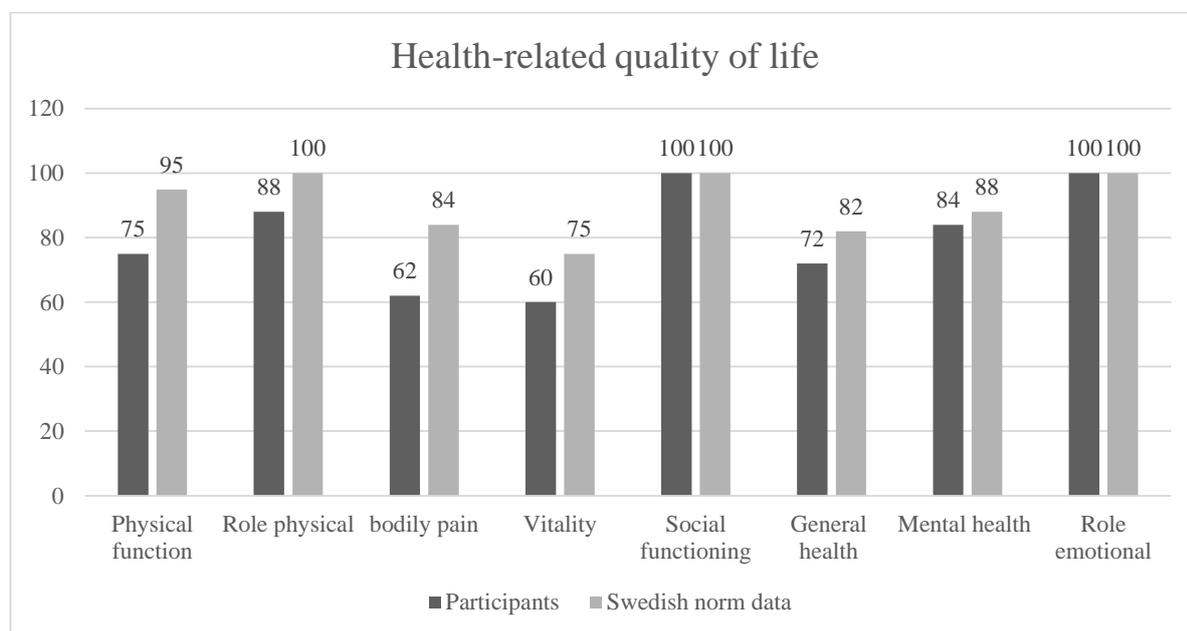


Figure 4. SF-36 components presented as mean for participants and Swedish norm data (65). High score meaning better health related quality of life. For physical function, role physical, bodily pain and vitality are $P < 0.001$. For Social functioning are $P = 0.005$, General health $P = 0.006$, Mental health $P = 0.006$ and Role emotional $P = 0.007$.

Estimated life satisfaction on the LiSat-11 was over all high when asking for vocational situation, financial situation, leisure time, contacts with friends, sexual life, family life, partnership relation and mental health. All of these items were rated with a median of 5 (range 1-6). The last sentence/statement about the ability to manage self-care was rated as median 6 (range 2-6), participants felt very satisfied. However, the sentence/statement “My physical health is” was rated with a median 4 (range 1-6) and 18 participants rated 4 or less, meaning the participant felt in this matter quite satisfied to very dissatisfied.

4.3 ENVIRONMENTAL FACTORS

From Study II and self-reports during the assessment the participants mentioned environmental factors that may influence the management of functioning in daily life when living with OI, type I or IV.

Participants mentioned they were restricted during school years by close family, health professionals and teachers, i.e. they were not allowed to attend ordinary gym classes, ride bicycles, ride horses or perform winter sports. However, the importance of having supportive family and friends that can help when the prerequisite for the day abruptly change was also

mentioned, i.e. OI is unpredictable and an injury may be just around the corner if one is not observant at all times. Further, friends can also be a wall of protection when in crowded places, at a concert or night clubs. One participant said “sometimes I need a body guard to feel safe”. However, often the choice is to not attend these activities because of the risk for injury. Older relatives with OI might also be role-models and might be the expert on OI such that the participant can lean on them when the constant responsibility is overwhelming.

Access to different health care and aids may provide a facilitator on the participation domain. For example the use of walking aids (pain relief) to minimise risk of losing balance and for protection, i.e. a walker may serve as an obstruction for preventing other people bumping in to you, use of hearing aids or access to surgery for the ability to participate in group conversations, ability to get reduced dental costs when extensive dental treatment is needed. The possibility, financial and practical, for rebuilding the kitchen (for example lower the sink and cupboards) when body height is reduced may help to prevent wear of shoulders, neck and back in the future or when pain is already present. Door sills can be taken out to prevent stumbling and falls.

The attitude from health professionals was highlighted as positive when recognised as unique and when accepted as an expert on the own personal type of OI. Attitudes of health professionals are perceived as negative when the individual does not feel listened to and feels abandoned, with important health issues. There might be a feeling of loneliness when making important life decisions. Environmental factors and attitudes from society are also mentioned, i.e. participants perceived sometimes being different and excluded from others and from activities. The preconceived beliefs of others were perceived as barriers. Beliefs that assume that the person with OI cannot attend an activity without first asking if they can. Stigma is also an aspect of being different and being treated differently. It was mentioned that the participants may have a choice between disclosure or non-disclosure of the diagnose OI, for example to employers. The participants perceived being treated with overprotection and scepticism about the seriousness of the health condition, due to the fact that the disabilities are not always visible.

In the interviews a fear of getting worse was mentioned and not having had the ability to do as much as possible to prevent it. Very few participants had an ongoing physiotherapy treatment with preventive/preserving physical activity/exercise in mind and in the interview it was mentioned that even after fractures they did not have a contact with either an occupational therapist or physiotherapist. Participants felt an uncertainty about the future concerning body changes and a need for role-models to be able to prevent getting worse.

The fragmented health care the participants perceived was pointed out as a negative aspect of living with OI and also the absence of regular check-ups by an interprofessional team to get support to be able to, if possible, prevent decline in health. Having access to assembled health care, someone that recognises you and can keep the care together were wished for. It was pointed out that it is a struggle to find what is needed when it comes to health care.

4.4 PERSONAL FACTORS

Identified personal factors from Study II, from questionnaires and self-report during the assessment are described in this section.

Personal factors that most likely influenced functioning and disability were the developing of creative strategies and changes of attitudes which were a way to confront the wide range of challenges that are present, i.e. the role of being an expert on OI. An ongoing process of learning to become and to maintain the role, for better or worse. The expert role leads to mixed feelings. The participants both wanted and needed to be an expert on OI, due to lack of knowledge in health care but also in society in general. Because of the rarity of the disorder they needed to develop strategies as described in Study II: striving for normality, ability to adjust and adapt and being foresighted and proactive.

The process of striving for normality includes generating a positive outlook and having independence to live a normal life despite the OI. It also includes not being labelled as sick or disabled. One strategy was to compare functioning with others with worse conditions for example persons with OI, type III. However, this might lead to minimising of symptoms and trivialisation of the health condition.

The participant's attitudes are important when it comes to disease and disability. Adults with OI most likely does not let the disease hold them back and have a remarkable way of adjusting to challenges that are coming their way during life. Further, the participants have the ability to adapt to new situations and with their self-reliance and control they are good advocates for their own interest. Adults with OI are mostly able to speak for themselves, concerning their own health condition.

Being foresighted and proactive means that the participants perceive that they constantly needed to be observant and alert to avoid obstacles and situations that could be a possible risk of injury. Further, it includes a constant need for being prepared to solve problems and find solutions to changes of the planned agenda for the day. It is hard being spontaneous and the feeling of being forced to have control at all times was pointed out.

Personal factors also influencing the participants' life were life-style factors such as physical activity (IPAQ). The participants report a span from sedentary to a very active life-style. The group was heterogeneous, but still 19 persons reported they did adhere to general physical activity recommendations (39).

Several of the participants have chosen to educate themselves and are managing it well. However, there was also a feeling of a need to be more educated to compensate for an unavoidably higher absence from work due to their health condition. Twentythree worked full time, at the time of the interview and clinical assessment. One participant was on disability pension mostly due to pain problems. The participants that were pensioners had worked until retirement age and one still worked part-time.

4.5 SUMMARY OF FUNCTIONING AND CONTEXTUAL FACTORS

Figure 5 shows a summary of possible functioning and disabilities and contextual factors influencing an adult living with OI, type I or IV.

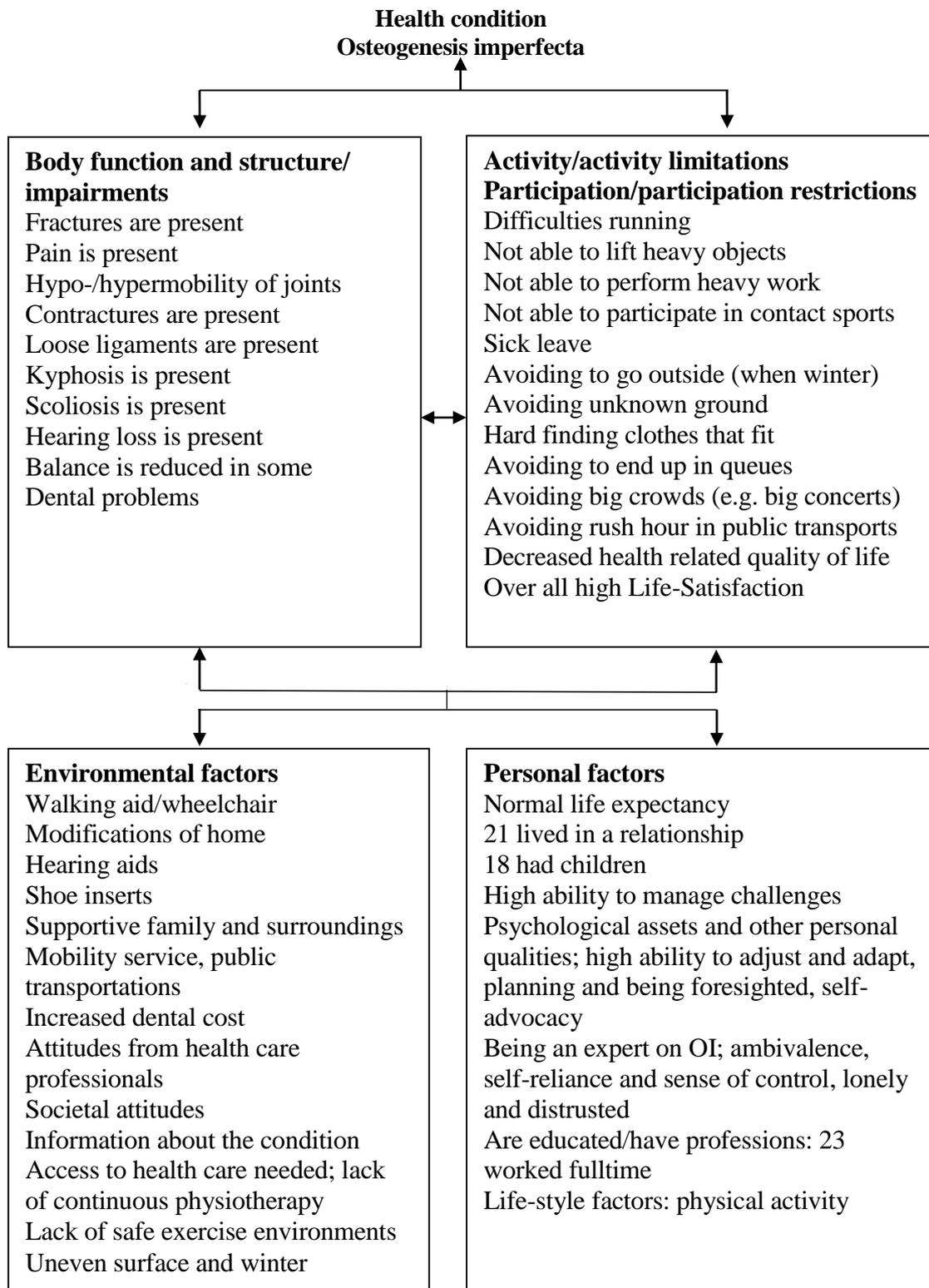


Figure 5. Overview of possible functioning and disabilities and contextual factors influencing when living with OI, type I or IV.

5 DISCUSSION

The description of adults living with OI in this thesis was done within the framework of the ICF, since ICF's standardised language is useful in understanding functioning and disability, as well as the impact of environmental and personal factors as facilitators or barriers to health in chronic disease states. By combining clinical testing with questionnaires and in depth interview, a broad understanding was reached.

Osteogenesis Imperfecta, (OI) affects all domains according to the ICF for those afflicted. The present thesis describes some of them. An essential part is the positive personal factors that were defined. Participants in Study I and II were incredible when it comes to managing their disease, very independent and strived to live as normal a life as possible. Nevertheless, information from the interviews revealed that the participants had a wish for support, above all, from health care. It is hard work being forced to be an expert in their own illness, OI, and responsible for the care of the same. Most likely there are several additional disabilities and contextual factors involved when living with OI as an adult, the health condition is multifaceted and differs between individuals, but also from time to time in the same individual. As most studies are conducted with children and adolescents type III or IV, there is a lack of studies concerning adults with OI type I or IV, and especially how environmental and personal factors affect their health condition. Thus, this thesis might help to identify important parts that may facilitate and improve multidisciplinary care and support for adults with OI type I or IV. In almost every study describing treatment strategies of OI it is concluded that this group would benefit from a specialised team care (11,12,13). The conclusion from the results of the present thesis confirms this. Persons with rheumatoid arthritis, one of the musculoskeletal conditions described before (43), is evidently having benefits from multiprofessional team care (69).

Due to heterogeneity, diagnose and classification of OI is hard. It has been suggested to use descriptions of the disorder as mild, moderate and severe form. The type I is thus considered as a mild form, but this might not be an adequate description as a person with type I (a mild form) may perceive his/her form of OI as not being so, depending on how it affects their life. It is only the adult self that can express the severity of the disease. Every person perceives things differently.

5.1 BODY FUNCTION AND STRUCTURE

Spine and joint range of motion showed a wide range, with both decreased and increased mobility, confirming large heterogeneity within the group. MacKiernan (70) reported that joint hyper-mobility and functional impairment are common in OI, type I, and 47 % of the participants had kyphoscoliosis or scoliosis. In Study I in present thesis a clinical examination revealed that nearly 80 % of the participants had kyphoscoliosis or scoliosis, i.e. an increased curvature of the spine.

Pain, both acute and chronic, is central in OI, and persons with OI probably have to learn to live with pain in early age to manage life. Twenty-five participants (86 %) reported pain at the time of the clinical assessment. Fifteen had neck and shoulder pain, 11 had chest and back pain. Two studies (50,70) confirmed that back pain is a common symptom in adults with OI, type I or IV. It is reported (71) that persons with pain are developing functional limitations that are associated with aging earlier in life, i.e. difficulties with running/walking, stair climbing, upper extremity tasks such as lifting objects and activities in daily living. The participants in Study I and II perceived those activities mentioned above as hard to perform and ageing did not seem to be the main reason for their difficulties. It is most likely that pain is one reason for functional limitations, such as walking longer distance, but it could also be the impairments developed from earlier fractures/injury. However, pain in feet and legs, were described to be worse when walking longer distance or when standing for longer times. One other study indicates that hip and knee replacement operations reduced pain and improved walking capacity, and this may be of importance since the OI type I or IV have a normal life span. Pain reduction and increased or preserved walking capacity would most likely affect HRQL outcomes.

5.2 ACTIVITY AND PARTICIPATION

Physical activity (PA) is important when it comes to how functioning and disabilities influences living with OI. Nineteen out of 30 participants reported adhering to PA recommendations (WHO), i.e. 30 minutes of moderate-intensity PA five days or more per week. In contrast to one study by Pouliot-Laforte et al. (72) that reported that children with OI and a control group did not reach the recommended daily PA. The groups in that study were almost equally active, but the OI group had a decreased muscular function compared to the control group. Thus, the conclusion from that study was that an inactive life-style could not alone be the reason, and that the OI group would need more PA/exercise to be able to reach the same muscular function as the control group. An increased amount of PA could prevent adverse effects of ageing on muscular function observed in OI patients type I (72). It is well known, that PA is an important part of the prevention of cardio-vascular diseases, type 2 diabetes, osteoporosis etc. (39). The participants in Study I and II might have had a wish that they were as active as the recommendations says, and thus reporting of PA was overestimated (73). On the other hand, it might also be so that the perception of the intensity differs from persons without OI, because of a possibility of decondition due to the disease and a feeling that the PA was performed with a higher intensity than it is for a physical fit person (74). There is, therefore, a need for further studies measuring PA with objective methods, such as pedometer or accelerometer, to be sure that the recommendations of PA are reached (75). It is also important to find out if muscle weakness is present, and if so to study if increased PA may positively influence muscle strength in adults with OI.

An inactive life-style is leading to negative consequences like high blood pressure, cardio-vascular diseases and higher loss of bone mass density, which is important to prevent for

persons with OI. In one study (35), children with OI are recommended to avoid PA such as contact sports, sudden rotation moments and intense interval training, and this is in the literature described as a PA that is beneficial for restoring/building strong bones (32). Due to the risk of injury, the participants in Study I and II reported that they had not attended such activities from early age. However, physiotherapists might be able to find ways to replace these not recommended exercises to make it possible for persons with OI to prevent comorbidities and strengthening bone in a safe environment during the whole life span.

Health related quality of life, for the participants in Study I and II was lower than norm data in Sweden, when measured with SF-36 (65), and DRI items running, lifting heavy objects were reported being hardest to perform (58). The physical parts in SF-36 are also comprising questions of running and lifting heavy objects. These perceived disabilities might be due to the risk or fear of falling, which may lead to an injury when having OI. The participants had experiences of falling or taking a wrong step leading to fractures. It might also be back, hip, knee, feet problems with muscle weakness, changed alignment due to bone deformity secondary to injuries and loose ligaments not functioning as they are supposed to, i.e. connect bones to each other and assist in the control of body movements. However, it has been reported that disability alone is not decreasing HRQL. Important parts in increasing HRQL are achieving meaningful occupational activities, social adaptation and the ability to cope with challenges in life (76). There are values that the participants seemed to have the capability to achieve, but still the HRQL is decreased compared to Swedish norm data and the reported physical health is lower than other items on LiSat-11, which might indicate that OI is affecting mostly the physical health and not so much mental health. A study from the US supports that the Physical Health domain in SF-36 is decreased in adults with OI when compared to US norm data. The overall life satisfaction was high in this group when it comes to participation in different activities and in this thesis 23 of 30 of the participants worked full time, and 21 lived in a relationship, supporting the satisfaction-with-life data. The high life satisfaction might be due to the fact that they have adapted and adjusted so well to life that the participation restriction is not considered a restriction for the individual. The participant is the judge of participation restrictions. Can it be that adults with OI, type I or IV, are having an ability to accept the situation and adapt to the new, so the restrictions are reduced according to this?

5.3 ENVIRONMENTAL FACTORS

Environmental factors relevant to functioning and disability for the participants were somewhat similar to the ones relevant to other musculoskeletal conditions: for example RA, Osteoarthritis and Osteoporosis. Examples of similar relevant factors are; health professionals, attitudes modification of the home, weather and climate, immediate family, and public transportations (43).

It was reported of the importance for access to needed health care. The care for an adult with OI is still fragmented even though it was pointed out already in the 1990s (3) and also that it would be beneficial with development of specialized assembled care. Only a few participants reported having contact with a physiotherapist and an ongoing treatment. However, none of these physiotherapists were a part of an interprofessional team and with special knowledge of OI. It was not even common to have met a physiotherapist after a fracture, not more than general information on how to handle the situation was reported. This may be due to the lack of knowledge and/or the heterogeneity of OI.

Movements are considered a base for the whole human function, and physiotherapists provides knowledge and expertise how to develop, maintain and restore maximum movement and functional ability throughout the life span, including circumstances where movements and functions are threatened, according to the WCPT's definition of physiotherapy. When it comes to promoting PA and providing a safe exercise environment throughout life for the participants in Study I and II, the physiotherapy profession does not live up to what this definition says. This may be due to the general health care structure and the small number of persons with OI. However, as a part of an interprofessional care, physiotherapy is important to ensure that individuals with OI have at least the possibility for PA to prevent other diseases due to an inactive life-style (45).

If there was a competence center for adults with OI, comprising knowledge of an assembled care, it would be easier to find information about the disease and health care professionals caring and treating these persons would easier find answers to their questions. Furthermore, persons with OI would feel recognized and the rarity of the disease would be less of a problem (77,45).

5.4 PERSONAL FACTORS

Personal factors that might be relevant to an adult living with OI, and are similar to the ones relevant to other musculoskeletal conditions are; coping strategies, beliefs and attitudes of the person towards disease and disability, physical activity/level of fitness, educational status and profession/working conditions (43).

Coping style with an active approach seem to be present in participants in this thesis. Most of the time they seem to have a positive outlook for their health condition. It has been described before that adults with OI have resilience (78), i.e. they meet challenges over and over through life and seem to manage to dust it off and get going every time. They know how to take care of themselves. An optimistic view of life and a stronger perception of pain control are associated with less disability and better physical function (79). It is easier to ignore for example pain sensations and easier to reinterpret sensation when optimistic. The participants are good at self-advocacy and are fantastic in adjusting and adaptation. Planning is a part of life with OI and it is perceived as forced to be able to avoid injury.

Participants seem to have a high ability and believe in them self to be capable of managing future situations. Their whole life they have been practicing to do so and they manage to adjust their life due to the challenge and are able to adapt to the new course. There are similarities with other chronic diseases (Scleroderma) such as the need to take control over the disease, a function of develop self-advocacy (80). Further, because of the rareness of OI, the adult are forced to tell their medical story over and over again, sometimes perceived negative. However, this might also be positive, since telling the story may give the person a chance to identify resources and managing strategies (80) that they did not know they had. This might be one reason why the participants are good at self-advocacy.

There is a need for future studies about PA in adults with OI with more specific measures, such as accelerometers or pedometers and PA diaries to ensure that the recommendations about PA are reached and if not be able to support the participants to increase their activity. But it is also important to explore how the exercises are performed, what kind of physical activity are performed. Is there any risk of disability problems later in life due to how the PA is performed today?

The participants mentioned a feeling that there is a risk of not being selected in a job-interview due to OI. That it is necessary to have higher education than peers, due to the fact that the health condition will occasionally cause higher amount of sick days due to fractures and pain. The participants in this thesis were also describing the possibility not to reveal their OI, which are mostly invisible. Not revealing was a choice and was giving the person a chance to be normal and not be discriminated against (81).

5.5 RELATIONSHIP BETWEEN FUNCTIONING, DISABILITY, ENVIRONMENTAL AND PERSONAL FACTORS IN OI

An essential part of the participants identified disabilities was the relationship between the individuals functioning, environmental and personal factors. You might say it is the person's health characteristics in relationship with their environmental and personal factors that define their disabilities (40).

Adults with OI are forced to live with the ambivalence between being an expert of their own health condition and the fact that they need support to achieve best possible outcome when it comes to living with their disabilities. If an adult with OI has the possibility of a continuous contact with a physiotherapist that has knowledge about OI, disabilities might be prevented or decreased.

The resilience (78) many of the participants showed may have developed from the constant confrontation with challenges that forces them to develop creative strategies and changes of attitudes. The expert role promotes several positive personal factors such as self-advocacy and a sense of control of the health condition, which might help to decrease the negative

environmental factors such as the attitudes from health professionals that the interviews revealed (being distrusted and not listened to).

5.6 METHODOLOGICAL CONSIDERATIONS – STRENGTHS AND LIMITATIONS

ICF as a framework to describe health conditions proved to be useful and able to give a holistic view of the participants. The importance when describing health conditions is to include contextual factors. Personal factors are relevant to disability and to achieve person centered care (41). The ICF is useful for persons with all forms of disability for identifying their health care and rehabilitation needs. This thesis provides information about adults with OI asking for support from health care, but we do not know exactly what this stands for. Future studies should address this issue and might thereby empower the development of a health care that is supportive and patient centered.

Disability is always an interaction between a person's properties and the properties in the overall context where the person lives (40). Without the interview part in this thesis the description of an adult living with OI would have been incomplete. The interview provided additional data for functioning and disability along with contextual factors that broaden the description of these persons. The description given here is not complete, but might provide useful knowledge for future care for adults with OI.

When using questionnaires there is always a recall bias (82). Many of the participants taking part in this thesis seem to have excellent recall of their medical history, it seems close in their mind. This might be due to them having to tell their medical history in every contact with health care. There is also a risk of overestimation of the intensity and amount of PA when using questionnaires (73). When a person has a low level of fitness, vigorous activity might be perceived as more intensive than it really is (74).

All data were collected at the same time. It was easy for the participants to have one appointment to relate to. Had the interviews instead taken place after the analyses of the questionnaires, the interview guide might have been different, or the interviewer would have had a different pre-understanding and follow-up questions may have been different. That is something we do not know, but the approach with a novice interviewer seemed to provide a safe situation where the participants seemed to be able to share their experiences from health care and society. Furthermore, one single appointment is easier for the participant to relate to, especially the ones not living in Stockholm area, and the equipment used were chosen to have the possibility to move from place to place. The ICF core sets for chronic conditions would have been useful when collecting data with interview, a help to create the question guide (42). However, the ICF Core sets did not exist when the studies in this thesis started.

6 CONCLUSION

The conclusion from the present thesis is that adults with OI, type I or IV, have bodily impairments, activity limitations and participation restrictions such as pain, scoliosis, contractures and trouble with running, heavy lifting, heavy work and sports. They also perceive a reduced HRQL, despite an estimated high life satisfaction.

Furthermore, adults with OI are forced to become experts on their condition, for better or worse, but the participants have the ability to adapt to new situations and with their self-reliance and control they are good advocates for their own interest.

The heterogeneity of the disease group makes it difficult to give a general view of an adult person living with OI, type I or IV. Even though the identified impairments, activity limitations, participation restrictions, environmental and personal factors found in this thesis do not apply for all adults with OI, it is important to remember that they may be present when caring and treating an adult with OI.

7 CLINICAL RELEVANCE

With an increased knowledge of adults' experiences of living with OI, type I or IV, health professionals can better understand, educate and care for these individuals, so that they are not forced to be experts on their own disease, at all time.

Participants with experience of elderly relatives as role models mentioned the importance of having someone to lean on when the constant responsibility of their own care becomes overwhelming. Therefore, it would be of importance to develop a mentorship program.

8 FUTURE

One way of compensating for the lack of role models might be the development of a mentorship program or a social forum on the internet (83). The development of an internet based self-management program (84) together with a specialised interprofessional health care team providing group delivered courses in self-management (85,86) might be other ways of compensating when a role-model is missing and in the end achieve satisfying assembled health care with support in self-management and self-care.

Another potential future development would be a center with a specialized interprofessional team where the adult person with OI can get support and answers to questions raised. The center might also provide support when it comes to coaching for a physically active lifestyle to prevent non-communicable diseases.

For future research, it would be of interest to explore the physical activity pattern in adults with OI with the aim to develop suitable exercise programs for this heterogeneous group.

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