

3rd World Congress on Huntington's Disease

by Diana Raffelsbauer

The 3rd World Congress on Huntington's Disease (HD), held in Dresden, Germany, from September 8th to 11th, 2007, was a joint event of the World Federation of Neurology (WFN) Research Group on Huntington's Disease and the International Huntington Association (IHA). Under the motto "Joining forces for HD", the Congress offered more than 500 scientists, clinicians, representatives of lay organisations and members of HD affected families from all continents the opportunity to share results, ideas and experiences. **Elizabeth McCusker** (WFN), **Christiane Lohkamp** (IHA) and **Bernhard Landwehrmeyer** (EHDN) opened the Congress, which was organised in joint plenary sessions on topics of general interest and more specialised parallel sessions targeted either to scientists, clinicians or lay people.



Meeting of the International Huntington Association

The Meeting of the IHA within the scope of the 3rd World Congress on Huntington's Disease was held in Dresden, Germany, on September 10th and 11th, 2007. Different aspects of HD were presented by outstanding HD specialists and lay organisation representatives. The topics, which were divided into six sessions, included support systems for HD families, quality of life of HD patients and their partners, and challenges of genetic information in HD. One of the main focuses of the Meeting was on better understanding the symptoms and developing more efficient strategies to care for HD patients and to cope with the disease in daily life.

IHA sessions I and II: How people with HD think, how to accommodate these changes and how to more efficiently interact with those whom you love

The Meeting of the IHA was opened by **James Pollard** from the Tewksbury Hospital, Massachusetts (USA). Huntington's disease (HD) is characterised by a clinical triad of cognitive, psychiatric and motor disorders. Based on neuropsychological studies, personal testimonials and observations, Pollard used examples to simulate the challenges resulting from some cognitive symptoms of HD. He described how HD patients think and how these impairments affect daily life. A better understanding of the disease may help family members and caregivers to accommodate the changes, to more efficiently interact with affected people and to explain the disease to others in a new way.

According to Pollard, HD patients typically show the following cognitive patterns: slower thinking, recognition is easier than recall, topic changes are difficult, difficulty organising and planning, and impulsivity: "can't wait". These features result in three conditions: narrow focus on what is coming next, apathy and irritability.



HD patients have less healthy brain cells available to process information efficiently. This slows thinking, delays response and changes behaviour. Every movement and every thought which was unconscious once becomes conscious during the disease, explained Pollard. Thinking becomes an exhausting, annoying process which requires more effort. HD affected people need more time to process information and give answers. The best way to help is giving them more time to respond. Pollard postulated that to interrupt this slow thinking is like resetting the timer and puts the whole process back to the beginning. Therefore, it is important to wait for the person's response and tolerate the often long silences.

The universal principle that recognition is easier than recall applies particularly for HD patients. It is more difficult to answer an open question than to choose an item of a multiple choice question or to answer with yes or no. Pollard recommended making things as easy as possible, for example by asking yes or no questions.

Using the Word Color Identification Test (Naming Colored Words Test) developed by John Ridley Stroop, Pollard explained the principle of cognitive interference. HD patients are more sensitive to distractions. It is more difficult for them to divide attention and follow topic changes. The maxim here is "one thing at one time".

Pollard used the Serial Sevens Test, which is part of the Mini-Mental State Examination, to illustrate the difficulty of thinking and concentrating that HD patients face in daily life. He said that the ability to organise and plan is affected to such an extent that it resembles wearing a blindfold: an exhausting process.

Another cognitive feature of HD is reflected by the inability to wait. This phenotype is unrelated to personality, but results rather from neurological (organic) problems. Bearing in mind that HD patients can't wait would help to minimise unpleasant reactions. Therefore, avoid "no" and "wait". "Do not reason", said Pollard, "it is easier to do it now".

Pollard described the three main conditions which are caused by the cognitive features mentioned above. The narrow focus on what is coming next seems to result from an overload of information. Indeed, HD patients are only concerned about now and what is happening next. Apathy (the absence of emotion, passion or excitement) is a common symptom of HD and probably related to frontal lobe dysfunction. Certain changes in the brain, which impair controlling impulses and expressions of emotion (often anger), result in irritability. Both apathy and irritability are stress responses: the first one is flight, the second fight.

In the second session, Pollard presented strategies of how to better accommodate the cognitive changes and how to more efficiently interact with people suffering from HD. These accommodations should avoid confusion, anger and apathy, help in daily activities (such as bathe and eat) and maintain a warm relationship throughout the course of the disease. Examples include, for instance: "no surprises", keep the same sequence and order, go slower, give a preview and a review, do one thing at a time, give continuous reassurance (what is going on?, how much longer?) with milestones and play-by-play descriptions (like in a baseball game), and establish a clear



beginning and end. Our accommodations can be seen as “terms (conditions) of endearment”.

Pollard also discussed which accommodations are more helpful at which stage of the disease (advanced, middle and early). In the advanced stage, HD patients encounter more physical disabilities which involve cognition and movements including speech. Family members might experience disconnection, loss and despair. “But still there is hope that endearing moments can endure”, said Pollard. “If we accommodate the cognitive symptoms of HD and use simple strategies to deal with them, we will be able to see both, the person we love and HD with its cognitive features”, said Pollard. The maxim KISS (keep it short and sweet) should be present in all our activities. No matter what the topic is, slow down, concentrate on one thing and enjoy the moment. “With them we find wonder in everyday things”, said a family member of an advanced HD patient.

In the middle HD stage, the same accommodations as in the advanced stage are helpful. HD patients typically show an obsessive desire to maintain sameness. Therefore, it is important to prepare any activity well, to give the same help in the same order and way (i.e. with system) and always to give reassurance.

Next, Pollard explained the so-called ‘Huntington’s disguise’, which is seen already in the early stage of HD. It comprises both motor and cognitive features. HD patients frequently suffer from dystonia, a disorder in which muscle contractions cause twisting and repetitive movements or abnormal postures. When affecting the facial muscles, this gives a peculiar expressionless. The difficulty maintaining a muscle contraction (motor impersistence) may affect social interaction, for instance sustaining eye contact or smiling. Abnormal postures include hanging shoulders and head. Other factors may be uncontrolled movements including muscle jerks, altered voice, impaired balance and altered depth perception, explained Pollard. Slow thinking and delayed responding make conversations difficult. In addition, people suffering from HD can’t recognise emotions well, particularly the negative ones. They are to some extent unable to interpret facial expressions of anger and sadness, for example. This inability to react may resemble emotionless.

The Huntington’s disguise might make people appear different than usual, sometimes even unrecognisable. It might make us believe things that are not true about HD affected people. “If we reveal this mask, we can see through it and prevent false beliefs”, said Pollard.

IHA session IIIa: Illness perceptions, coping mechanisms and well-being of HD patients and their partners

The IHA session III was devoted to support systems for HD patients and their families. The topic was introduced by **Lucienne van der Meer**, who works as a psychologist at the Department of Clinical Genetics of the Leiden University Medical Center (Leiden, The Netherlands).



Beyond any doubt, HD impacts the quality of life (QoL) of both patients and their partners. However, psychosocial aspects of suffering from HD have not received much scientific attention to date. Van der Meer started her presentation with a summary of the available literature about how HD impacts QoL of patients and their partners/caregivers. HD patients suffer from psychological disorders such as depression, anxiety, alcoholism and increased risk of suicide. They fear the physical, mental, financial and social consequences of the disease. A disintegration of social networks is often observed. Especially in the early stages, HD patients feel ashamed and tend to deny the disease as a defence mechanism.

The partner/caregiver perceives an increased responsibility (patient, children, home, work, etc) and constantly feels overburdened. 82% of them show stress symptoms, which depend on the behavioural traits of the patient, severity and duration of the disease, increased parental responsibilities and feelings of loss (relationship with spouse, career, social activities, personal freedom, etc). An increased risk of marital breakdown is observed. Whereas patients suffer as well from the physical impacts of the disease, partners are mostly disturbed by mental and personality changes.

Van der Meer presented data from a QoL study performed with 77 early to middle stage HD patients at the Leiden University Medical Center (The Netherlands). The study describes how HD affects quality of life, how patients and their partners perceive HD and how they cope with the problems they encounter in daily life. According to this study, the largest impacts of HD on patients' QoL are seen in the parameters of work, alertness, home management, recreation, communication, psychosocial aspects and emotions.

The question "How do patients perceive their illness?" was assessed along five dimensions: identity (symptoms), cause, control/cure, time-line and consequences. In general, HD patients think that they have many severe symptoms, that they will be ill for a long time, that HD influences their whole life and that there is nothing that can be done.

Subjective illness perception influences quality of life as follows: If patients perceive many impairing symptoms, there is a decrease in vitality, physical, mental and general health. If partners perceive the disease to be long, there is rather an increase in vitality and physical functioning. If they perceive the disease to have fewer consequences, an increase in vitality and mental health can be observed, said van der Meer. Taken together, well-being of patients is better when they perceive fewer negative consequences of HD and their partner perceives the disease as less severe. Similarly, well-being of partners is better when they perceive less negative consequences of HD and the patient feels more in control of the disease.

HD patients use different strategies to cope with the disease: acceptance (frequent); seeking social support, focusing on emotions and mental disengagement (infrequent); and denial (seldom). Acceptance of the disease seems to improve mental health, noted van der Meer. Similar strategies are also used by partners of HD patients, as well as active coping (frequent) and suppression of competing activities (infrequent).



In the future, more research using HD specific QoL instruments is needed. Intervention programmes should be developed in which illness perceptions and coping mechanisms of HD patients and their partners are influenced in the hope of enhancing their well-being, concluded van der Meer.

IHA session IIIb: Young people's view of growing up in a family with HD

Continuing on the topic support systems for HD patients and their families, **Karen Keenan** and **Catherine Martin** reported on their work at the National Youth Service of the Scottish Huntington's Association (SHA). Founded in 1989 by families living with HD, the SHA now has 35 staff members throughout Scotland providing expert care to families living primarily in their own homes. Support is offered by families and professionals via a network of local family support groups and paid HD specialists.

The main focus of the presentation was on young people's views of what it is like for them to grow up in a family affected by HD. Young people's experiences can be quite varied: While some cope successfully, others experience considerable problems and may even be at risk of physical and/or emotional harm. Keenan began with an impressive DVD presentation showing the story of Simon, a 15-year old Scottish boy who is alone caring for his HD affected mother at home.

Keenan then gave an overview of the social situation of children and young people in Scotland in general. About 25% of the Scottish population is under 19 years of age, and nearly one third of Scottish children live in poverty. One in seven young people (age 16-19) have "disappeared" from the system, i.e. they are not in education, employment or training. 100,000 children live with domestic abuse. Between 40-60,000 children live with at least one drug using parent.

Regarding health, Scottish teenagers show high rates of alcohol, tobacco and drug abuses. High pregnancy rates are observed (42.4 cases per 1,000 between 13-19 years of age). Males aged 11-24 are at highest risk of committing suicide. 10% of young people have mental health problems which interfere with daily life. In Scotland the number of young carers is estimated between 17,000-100,000, said Keenan. The United Kingdom has been identified as the worst country for child well-being across 21 industrialised countries (UNICEF Report 2007).

Keenan mentioned the difficulties which young people growing up in a family with HD may encounter in daily life. These include issues around caring activities, coping strategies, communication, family relationships, understandings of inheritance, risk perceptions, attitudes to genetic testing and having children. HD can affect parenting capacity, e.g. other parent's availability and attachment problems. Children may experience multiple losses, e.g. of parent, quality of life prior to the disease, income, etc. The burden of risk and possible lack of information may lead to prolonged anxiety. Young people may also experience stigma and taboo associated with HD, all of which can lead to some children feeling considerably isolated.

Next, Keenan presented the results of the research study that she conducted by interviewing 33 young people (aged 9-28). Among these, 26 were at risk, one was a



tested gene carrier, one had juvenile HD and five were not at risk. 12 from the 33 interviewees were/had been young carers of a parent with HD, assuming tasks and responsibilities which in usual circumstances an adult would do. In certain cases, the other parent worked long hours or was also ill, or they lived with a single parent. These young carers were particularly at risk for physical and/or emotional impairments.

Young people also experienced the negative impact of their own risk, e.g. physical (tiredness), emotional (lack of self-esteem, avoid relationships) and psychological (panic attacks, excessive symptom searching) aspects. They need social support and effective coping strategies. For example, some used problem solving, planning, having a positive attitude to disability and gaining certainty to cope. Growing up with information can help some young people better cope with HD, whereas others do not want an overload of information, said Keenan.

In the second part of the presentation, Martin summarised the history of the SHA, which currently offers 9 specialist HD services, palliative care training and 10 family branch support groups. The need to support children and young people living in families with HD emerged in the SHA through the 1990s, said Martin. Hence, the Youth Service was founded in 2001 as a "South West Pilot Project" with financial support from Children in Need. Based on further evidence of need, the Service was extended to cover the national area with funding until November 2007.

The National Youth Service strives for the following goals: to increase children and young people's knowledge and awareness of HD; to provide one-to-one, group and interactive support, helping to reduce isolation; to support parents to communicate with their children about HD; to improve professional awareness of the impact of HD on children and young people; and to provide young people with a safe and confidential platform to discuss HD and their personal circumstances. The role of the Youth Service is divided into six main groups: advice and information, advocacy, peer mentoring, personal development, legislation, encouragement and support, noted Martin.

Within its mission to educate and disseminate information about HD and the impact of living with HD on children and young people, the SHA provides families with different informational materials, such as age-oriented booklets, books, websites and newsletters, which regularly report on group activities, research updates, forthcoming events, new projects, etc. The SHA also conducts its own research.

The Youth Service is committed to providing support and advice on an individual and group basis for children and young people living with HD in their transition from childhood to adulthood. Many group activities are offered, such as daily trips and summer camps (the focus is on having fun and outdoor activities, e.g. climbing, canoeing, rafting, zip wire, trapeze, high ropes and abseiling). Contact between young people is also mediated through the Youth Service website (which has a chat room and discussion forum) and Bebo page.

At the moment, the Service is providing direct support to over 100 young people and their families. "It makes you remember you aren't alone, and there are others your



age you can talk to with the same problems”, said one of the children. However, the SHA still has to face problems of funding, transport and logistics, reaching all young people of affected families and legislation (child protection, confidentiality, etc), concluded Martin.

IHA session IIIc: Rehabilitation in Huntington's Disease – an experience from Italy

Rehabilitation therapy has been shown to improve quality of life in patients with neurodegenerative disorders. However, most studies on the use of physiotherapy in HD have not been scientifically robust enough in order to strongly recommend it.

Paola Zinzi and **Stefano Maceroni** reported on a pilot project they conducted at the home care “Nova Salus” (Italy) with the aim of providing a quantitative and qualitative assessment of the effect of rehabilitation therapy on HD patients.

The intensive rehabilitation programme, provided in both single and group formats, comprised physical, respiratory, speech, cognitive and occupational therapies, explained Maceroni. In the context of occupational therapy, the programme included playful activities and artistic techniques such as decoupage and patchwork, offered by the recently established creativity laboratory.

Each treatment took three weeks and could be repeated three times a year. A standard quantitative assessment was performed at the beginning of each admission using the following tests: Zung Scale (depression), Mini-Mental State Examination (cognition), Barthel Index (activities of daily living), Tinetti Scale (gait and balance) and Physical Performance Test (PPT; functional timed tasks). Tinetti and PPT were also measured at the end of each 3-week treatment to evaluate motor and functional outcomes. A qualitative evaluation of the results was given by a questionnaire mailed to the patient's family.

40 HD patients were enrolled in the project between 2000 and 2002. Baseline functional characteristics of these patients fulfilled the selection criteria: No patient was beyond stage III of the Shoulson Rating Scale; they were on average moderately depressed (Zung test mean score 34.3, i.e. below the critical value of 40), and had a mental performance at the lower border of normality (MMSE 24.9). Total functional capacity (7.8/13) and activities of daily living (86.3/100) scores were on average moderately reduced.

Considering the 11 patients who completed six admissions over a period of two years, both the Tinetti Scale and the Physical Performance Test showed a marked improvement in scores at the end of each admission (+4.7 on Tinetti Scale and +5.21 on PPT). No carry-over effect between subsequent admissions was apparent, but at the same time, no deterioration with respect to baseline over the 2-year period was detected.

Regarding the tests for depression, cognition and daily living functionality, no significant changes were found, indicating that the patients maintained the initial level



and did not deteriorate beyond the baseline scores over the time, added Zinzi. The qualitative data from the questionnaires confirmed the above results and showed positive effects also on psychosocial aspects. An overall positive effect of the rehabilitation experience was reported by 100% of the respondents. Main improvements were reported for motor control, speech, balance, swallowing and gait. Positive effects were also observed for mood state, family and social relationships.

In summary the results indicate that the rehabilitation protocol developed by Zinzi, Maceroni and colleagues is an effective tool to improve both physical and psychosocial symptoms caused by HD. This confirms that the rehabilitative approach allows HD patients, as well as their families, to live the course of the disease in an overall better physical and psychological well-being.

IHA session IVa: Building up support systems for families and people with HD

Continuing on the topic of support systems for HD patients and their families, **Don Lamont** from the Huntington Society of Canada (HSC) presented the Canadian Model as an example of how lay associations can serve people with HD.

Since its foundation in 1973, the HSC has been committed to supporting individuals and families affected by HD. The HSC strives to maximise the quality of life of people living with HD by delivering services, enabling others to understand the disease by offering educational programmes and furthering HD research. "HSC is the HD specialist: It acts as an independent advocate - integrated within 'the system' - to get 'the system' to support families with a continuum of care and to link families to services", said Lamont. The HSC believes that family support and research must be tightly linked. For instance, an active supply of scientific information to the families should be part of the multidisciplinary care just as the option to participate in clinical trials.

The geographic and demographic features of Canada represent a challenge: 33 million people spread over a vast area, resulting in a population density of 3.2 people/km². Accessibility to services can be restricted, especially in remote areas. The number of HD specialists (neurologists, psychiatrists, geneticists, etc) in the country is limited, and other causes compete for their attention.

This required a unique solution: hence the Canadian Model to support individuals and families. This model is based upon circumstances unique to Canada. The core service network consists of 10 staffed HSC family resource centres in larger communities, as well as another 12 part time individual and family service workers essentially in areas with lower populations. HSC staff in the network is equipped to provide affected individuals and those at risk, families and professionals with information about HD and support, as well as to link them to community-based services, explained Lamont. Independent of HSC, there are eight multidisciplinary HD clinics and six Huntington Study Group sites in Canada, with whom HSC staff collaborates.



In summary, HSC's Individual and Family Service goals are: to supply information to the families, to help people to manage the disease, to identify the families' need for services and to make sure that these needs are met, to support and educate service providers and to create integrative systems where individuals and families can exchange their experience. These goals are met through two complementary services: support service (individual and group counselling focused on individuals and families) and HD service development (for local, regional and national services), as well as lay support groups and social interaction through local Chapter groups.

Lamont also emphasised the activities of the HSC as an information facilitator, through regional family day information sessions, national and local websites, informative e-mail bulletins and newsletters and other educational materials (e.g. books and pamphlets).

The HD Clinic in North Bay (Ontario) and the HD Clinic in Vancouver (British Columbia) are two distinct (rural and urban) examples of multidisciplinary care. The North Bay clinic connects the patient with the clinician by way of video conference, said Lamont.

IHA session IVb: Building up support systems for families and people with HD

Next, **Michael Orth** reported on the HD Clinic in Hamburg (Germany), where a multidisciplinary team of specialists treat and advise patients with HD and their families. The catchment area of the clinic comprises four other German federal states with a total population of around 4.5 million people. Among them 400-500 people are HD patients with many more at risk.

The goals of the clinic are: predictive genetic counselling and testing, management of neurological and psychiatric symptoms, social support including legal matters, therapeutic clinical trials and research, explained Orth.

The core clinic team consists of one neurologist/neuropsychiatrist and one neurologist (both specialised in movement disorders), two neurology registrars, one human geneticist and one study and research nurse. To provide HD patients with a continuum of care services, the clinic works closely with other health care professionals, such as psychologists, specialist nurses, social workers, HD lay organisations, dentists, speech and language therapists, occupational therapists, physiotherapists, as well as nursing homes and research institutes. The clinic is a member of the European HD Network (EHDN) and is currently participating in the Registry Project.

Since 2006 the clinic has seen 40 patients with a diagnosis of HD or for predictive testing. In total, about 100 patient contacts have been established. For the future, Orth mentioned the need for more manpower in his clinic and the importance of further research.

The HD Clinic in Hamburg works in close cooperation with the Northern German HD Support Network, represented by **Gabriele Ritter**. She is a social worker at the



specialist HD ward based in Heiligenhafen (Germany). This facility offers not only symptomatic neurological-psychiatric treatment, but also a series of other services for HD patients and their families. These include therapies (psychotherapy, occupational therapy, physiotherapy, speech and language therapy), dietary advice and treatment, psychological and social support, and legal advice.

The ward accommodates about 140 HD patients per year. 40% of them need permanent nursing home care. The remaining 60% continue receiving assistance through the Support Network after discharge. The Northern German HD Network covers a vast area. "Out-patient support requires commitment and dedication", said Ritter.

Ritter raised issues, some of them ethical, concerning the responsibility for patients' well-being. The solution may require efforts from all parts. The Network brings together HD affected families, self-help groups and professionals of different institutions and care facilities to a well-functioning support system. One successful example is a care facility in Itzehoe (Germany) with 120 HD patients at different stages of the disease.

IHA session IVc: Development of a disease-specific quality of life instrument for people with Huntington's disease – hearing what patients say

On behalf of the EHDN Quality of Life Working Group, **Aileen Ho** and **Mevhibe Hocaoglu** presented the interim results of the Huntington's Disease Quality of Life Project. The aim of the project is to determine and measure HD patients' health-related quality of life (QoL) by developing an HD-specific instrument.

Quality of life is a major issue over the course of the disease. This raises the need for an accurate and objective assessment of QoL in HD which is valid, reliable and sensitive, said Ho. Since the features of HD are unique, existing generic quality of life instruments might not be adequate.

Ho first reported on QoL studies that she and colleagues performed with HD patients using two generic quality of life questionnaires: the sickness impact profile (SIP) and the short form 36 health survey questionnaire (SF-36). In the SIP testing, the impact of HD on well-being was measured in 14 different dimensions. Most impairments were seen in the dimensions of work, alertness, recreation and pastimes, home management, communication and psychosocial dimension. In general, SIP is poorly tolerated because the questionnaire is too long, said Ho. The SF-36 questionnaire comprised eight dimensions, from which the physical ones were markedly affected. On a pragmatic level, the SF-36 is shorter than the SIP and is therefore easier for patients at various stages of the disease to complete.

A close scrutiny of the data revealed that there were weaknesses in the SIP and SF-36 questionnaires in evaluating quality of life in HD. For some items, the questionnaires were not specific enough, reported Ho. Also, some items were missing or underrepresented, e.g. behaviour (disinhibition, problem solving) and



mood. Furthermore, motor symptoms appeared to influence some strictly non-motor dimensions of the tests (item contamination).

Hence, Ho, Hocaoglu and the EHDN Quality of Life Working Group are working on the development of a questionnaire which addresses the specific ways in which patients' ability to live their lives has been affected by HD. It is important to ascertain patients' perspective of their well-being using an instrument that is valid and reliable. "This is why this project is so important", said Ho.

The development of this HD-specific QoL instrument will be conducted in three stages: Stage 1 is aimed at the generation and selection of items by interviewing HD patients and their carers. In Stage 2 suitable items will be derived to conceive a prototype instrument which will be tested. Stage 3 will evaluate the psychometric properties of the instrument.

Ho stressed the importance of understanding how HD affects HD patients' perceived quality of life and capturing the reality of the living with HD reflected through the subjective experience of the patients. A priori, items were derived from the literature. Secondly, indigenous items were generated from the interviews that capture patient specific expressions.

The development of the interview questions in Stage 1 of the project was based on a review of the literature regarding motor disorders, cognitive impairment, behavioural and psychiatric disturbances seen in HD. This yielded semi-structured interview questions reflecting different social, cognitive, emotional, physical and functional aspects of health-related quality of life. The pilot testing for relevance, acceptability and comprehensibility of the interview was carried out by three HD patients, one care advisor and six HD specialists.

The interview comprised open-ended questions and specific probes to elicit reflection and response. To date, 20 interviews have been conducted with 10 HD patients from early to advanced stages, seven carers, two health care professionals and one juvenile HD patient. The interviews were transcribed and analysed with a computer-assisted qualitative data analysis software (NVivo 7). Preliminary qualitative analysis of these interviews can be summarised under the following dimensions: physical and functional, cognitive, emotional, social, legal and financial, as well as the dimension "self", which was created specifically for HD and includes items such as attitudes towards life, self-esteem, personal goals, safety, body image, sexuality, health, fitness and treatment satisfaction, explained Hocaoglu.

Preliminary data show that, in the cognitive dimension, concentration and memory are particularly affected in HD, said Hocaoglu. In the emotional dimension, depression and anxiety were reported as the main problems. Regarding the physical and functional dimension, HD patients suffer mostly from impairments in the ability to articulate speech, sleep, walk and drive. In the social dimension, HD patients markedly perceive a deterioration of their relationships, especially in their role in the family. The issue "How to cope with the disease?" assumes a central part in daily life. HD undoubtedly also impacts independence, interest and hobbies.



By exploring associations among the emergent dimensions, Ho and Hocaoglu proposed a working model on how HD affects patients' quality of life. According to this model, the emotional, cognitive, social, physical and functional dimensions interact with each other and affect the patient self.

Future aims are the further development of the proposed model, further exploration of associations between the different dimensions (also involving participant attributes such as gender and stage of HD), the completion of the targeted number of interviews, and item reduction by eliciting responses from patients. Thereafter, an initial prototype will be generated, and subsequently refined and validated in the following stages of the project.

IHA session Va: Identifying, responding to and living with the challenges of genetic risk for HD

The IHA session V was dedicated to the challenge of genetic information in HD. The topic was introduced by **Elizabeth McCusker** from the World Federation of Neurology. McCusker presented the challenges that patients encounter by deciding whether to undergo genetic testing or not. The advantages and disadvantages of genetic tests for predicting HD were outlined. She reported on certain cases of patients with HD-like symptoms who were afterwards tested negative for HD. Genetic testing is therefore a valuable diagnosis instrument, concluded McCusker.

Next, **Claudia Downing** from the Centre for Family Research at the University of Cambridge (UK) shared with us the experience she gained when researching families facing HD. Downing presented the challenges that arise for those aware of their or their partner's risk for HD when considering becoming and being parents at a time when genetic testing and new reproductive techniques have become available. Challenges comprise identifying the risks, responding to genetic information, deciding whether to have children, living with uncertainty or clarifying it, and parenting with the knowledge of a gene positive status, which also includes the uncertainty about the at-risk parent's ability to sustain a parenting role.

Becoming aware of HD is a process which involves reconciling family experiences with factual information, noted Downing. Possible barriers include misdiagnosis, lack of communication and denial. According to Downing, the reproductive decision-making process involves negotiating risks that are predetermined, those which are a matter of choice, concerns about values (especially responsibility), children's future, being a parent and eugenic legacy, at a time when new options for clarifying and avoiding risk are becoming available. These comprise genetic testing and *in vitro* fertilisation (IVF) including preimplantation genetic diagnosis (PGD). These options, however, can result in new challenges, outlined below.

Downing presented a model of responsibility which provides a framework in which to describe how people face these challenges. According to the model, awareness (either factual or lived) of risk in two dimensions (child-centred or parenting risk) defines reproduction as problematic. This leads people to redefine elements of their situation, such as their risks, fertility and relationships. The form that redefining takes



reflects modifying factors, such as their values, concepts of the future and perceptions of social support. Redefining enables them to tell a different story when subsequently accepting, modifying or avoiding risks. Concerns about one value, responsibility, dominate participants' stories, identifying responsibility as the core concept for the model. Establishing themselves as responsible decision-makers helps restore feelings of well-being.

Responsibility is not enacted from a single script. For example, modifying the risk can be sought by restricting family size: "I had only one child - with a hope that he may just be the one that's good", said an HD patient. Others tried to modify the risk by having more children than they might otherwise have had. Their hope was that the chances of having non-affected children would increase and HD negative children could "compensate" for the parent's possible compromised ability to care for any affected ones.

Downing then focused on challenges that specifically arise from genetic testing and new reproductive technology. These include undergoing the procedures, justifying rejecting the new options, making decisions about disclosing the parent's status and having a biological or non-biological child. She described how a positive predictive test result creates many challenges for parents. These comprise living with uncertainty about when they will develop HD, how best to use this time to meet the responsibilities towards their dependent children and to each other, and how to plan for the future.

Parents report rethinking priorities, such as having holidays now. It may also involve delegation of parental responsibilities (e.g. around who disciplines the children), role reversals and making children independent sooner. Competing needs have to be reconciled. The biggest problem parents report surrounds telling their children that they are at risk. Their instinct to protect the child conflicts with the child's need to know. A parent's needs for medication may also conflict with children's needs. For example, one single mother only took sleeping medication during the school holidays, as she had to be awake to take the child to school when school was in session, observed Downing.

Planning for the future involves passing on values as well as genes, finding ways of "being there" for children's milestones, negotiating appropriate housing, ensuring that the other parent is aware of the implications for the children, making care arrangements that don't involve one's children and, for some, contemplating the possibility of euthanasia, said Downing. She drew on the example of trying to move into suitable housing. Although parents have good reasons to move into suitable accommodations before they become affected (such as making this less traumatic for their children), they face problems. For instance, testing positive makes arranging mortgages and insurance difficult. Those in council accommodation find that authorities focus on the parent rather than the family. The children's needs are not taken into account, as they are not council tenants until they are older or start paying council tax. Downing concluded her presentation with an optimistic emphasis, stressing the need to have fun and retain hope, despite HD.



IHA session Vb: Open discussion on genetic testing in HD – PGD, PND, predictive and diagnostic testing

To round off the topic, **Asunción Martínez Descals** conducted a discussion on the use of genetic diagnostic techniques in HD. Recommendations concerning the use of a predictive test for the detection of HD were drawn up by a committee consisting of representatives of the International Huntington Association (IHA) and the World Federation of Neurology (WFN) Research Group on Huntington's Disease.

The establishment of a committee with the specific task of preparing such guidelines was agreed upon at the WFN and IHA conferences in Lille (France) in 1985. The first recommendations were adopted by each of the organisations at their respective meetings in Vancouver (Canada) in 1989 and published in the *Journal of the Neurological Sciences* (1989; 94:327-332) and the *Journal of Medical Genetics* (1990; 27:34-380). Revision of these guidelines was necessary in view of the report, published in March 1993, on the detection of the gene defect. These revised guidelines were published in the *Journal of Medical Genetics* (1994; 31:555-559) and in *Neurology* (1994; 44:1533-1536).

In fact, the international guidelines developed by the IHA and WFN for predictive and prenatal testing have been applied successfully since 1994. Based on the experience of HD affected families, guidelines for diagnostic testing have been developed and adopted by the IHA in 2005. However, ethical concerns emerge about predictive testing in children and prenatal diagnosis (PND). Particular issues arise in connection with modern diagnostic procedures using assisted reproductive technology (i.e. *in vitro* fertilisation) such as the preimplantation genetic diagnosis (PGD).

Martínez addressed the question "Do we need guidelines for predictive testing in children and for PGD?" In particular, the aspects "which criteria must be fulfilled to recommend the tests" and "when (at which age) testing in children should be performed" were discussed. Genetic testing brings patients and parents the challenge of how to cope with the results. Therefore, caution should be taken especially when considering performing the test in children. A minimum age of 18 is recommended, as it is hoped that at this age a person has the maturity needed to deal with the awareness of carrying the HD gene. However, in reality we face situations in which pregnant women under the age of 18 want to undergo the test.

Legal and financial issues of predicting HD were also outlined. Results may affect health and life insurances and lead to discrimination (e.g. in employment). This may occur in a pre-symptomatic stage and may also affect healthy individuals with a family history of HD, said Martínez. Furthermore, it also influences the decision of having children.

In some countries PGD is forbidden by laws aimed at embryo protection (e.g. in Germany). The discussion pointed out the need for a worldwide standardisation of guidelines for genetic testing based on international rules for hereditary diseases under the involvement of ethics committees. In the meantime, all present representatives from different countries agreed on the continued use of the existing guidelines for predictive testing.



IHA session VI: Open discussion of the future direction of IHA – expectations and ideas

As the president of the IHA, **Christiane Lohkamp** concluded the sessions with an evaluation of the meeting and an outlook on future activities. Participants praised the meeting as very successful and congratulated the organisers. In summary, the speakers were carefully selected, the topics were well-defined and the discussions were very interesting. The programme was flexible, enabling participants to choose among different sessions. Participants approved the running of the two parts of the Congress (IHA and WFN) for 1 ½ days separately. This model should be maintained in the future.

The meeting gave the opportunity to hear members of the lay community and share experiences. Moreover, it was a platform to meet researchers from different countries (including the home country), as well as the language coordinators from the European Huntington's Disease Network (EHDN), and establish relationships with them, said one of the participants.

The site of the IHA Meeting in 2011 was discussed, taking into account travelling costs. Among the candidates Mexico, Japan and Australia, the IHA favoured the last one (Melbourne). Also, it was suggested that sessions be divided according to the target groups' families, members of lay organisations and nurses was suggested.

Electronic issues of the IHA website were discussed, such as the need for changing the internet provider. Furthermore, the establishment of a chat room or discussion forum with frequently asked questions about HD was examined. Examples of such services in Australia, the Netherlands and Italy were mentioned. The responsibility of transferring wrong information could be circumvented by disclaimers. Nonetheless, to assure that the information on the site is medically correct would require the engagement of a medical doctor 24 hours a day, which would be too expensive. Furthermore, the need of translating contents into different languages would also be an issue, since English might not reach affected families all over Europe.

The platform for a chat room could be provided by the EHDN. However, the need for translation would again be an issue. Security problems (viruses, misuse of contents, etc) were also mentioned. Taken together, the IHA decided not to establish a chat room and to further provide support and advice to HD affected families directly by e-mail and phone contact.

Future activities of the IHA will include developing strategies to better communicate outcomes from scientific and clinical research to the lay community worldwide, and to help setting up nursing homes and educating nursing home personnel, concluded Lohkamp.