F Clinical care and management

F.1 ORAL CARE IN HUNTINGTON'S DISEASE

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Background: The Mun-H-Centre is a Swedish national orofacial resource centre for rare disorders and a national resource centre for orofacial aids and assistive devices. Since 1999 the authors have been members of the Huntington's disease (HD) team at Sahlgrenska University Hospital/Östra in Göteborg and because of that have had the unique opportunity of treating a number of patients with the diagnosis of HD.
Aim: To collect, document and develop information relating to rare diagnoses and to circulate this knowledge as a contribution to ensure that patients with rare disorders receive better treatment and can have an improved quality of life.
Methods/Techniques: Dentists and dental hygienists at Mun-H-Centre have followed 20 patients with HD and observed oral problems over time. At Mun-H-Centre there is a multiprofessional team for trying out orofacial assistive devices to patients with disabilities. The team members represent occupational therapy, physiotherapy, speech–language pathology and dentistry.
Result/Outcome: Our experiences are documented and illustrated as follows: orofacial symptoms, risk factors for deterioration of oral health, preventive dentistry and cooperation with the patient's personal assistants, treatment in the dental chair, examples of orofacial aids and assistive devices for oral care, eating and drinking.
Conclusions: It is important for a person with HD to see a dental team regularly in order to maintain a good quality of life through a healthy mouth, for the purpose of speaking and communicating as well as eating and enjoying meals. Preventive dentistry from an early stage of the disease and orofacial assistive devices that can compensate for lack of function are recommended.

F.2 NEW TOPICS FOR HUNTINGTON'S DISEASE IN EUROPE?

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Background: During our project we have found that there are topics concerning the care of persons with Huntington's disease (HD) that are necessary to agree at an international level. One of these topics is to find a good model for taking care of and informing children in families with HD about the disease. Some other important issues are orofacial care and speech as well as cognitive/linguistic communication problems.
Aims: To search for models and to create new models in order to facilitate life for persons with HD and for their families.
Methods: Cooperation with professionals, caregivers and associations in Europe. We will plan workshops together and we will arrange meetings over the internet. We will participate in the working group Standard of Care and Quality of Life.
Result: To create good models and a networking team in these fields.
Presentation: We will present these ideas in a poster presentation.

F.3 IMPACT OF HUNTINGTON'S DISEASE ON QUALITY OF LIFE: A QUALITATIVE STUDY

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Background: Even though Huntington's disease (HD) is an autosomal dominant neurodegenerative disease characterised by motor, cognitive and and behavioural disturbances, there has been little research reflecting the first-hand experience of the health-related quality of life of people living with HD.
Aims: This qualitative study examines patients' personal experience of the impact of HD as a part of a larger EHDN project to develop a disease-specific patient-centred health-related quality of life instrument for HD. The development of such an instrument would benefit from examining patients' direct experience and how they themselves perceive the impact of HD on their health-related quality of life.
Methods: Thirty-nine semistructured interviews were conducted primarily with people living with different stages of HD, with some input from their companions, carers and health professionals familiar with HD. The computer-assisted qualitative data analysis software package NVivo 7 was used for sequential analysis of the data using grounded theory.
Results: Six dimensions of quality of life emerged: (1) physical and functional dimension; (2) cognitive dimension; (3) emotional dimension; (4) social dimension; (5) legal and financial dimension and (6) self dimension.
Conclusions: A model of quality of life for people living with HD is conceptualised to capture the interaction of these dimensions in mediating the subjective construct of health-related quality of life of people living with HD.

F.4 IMPROVING CARE AT END OF LIFE: HOW DOCUMENTING WISHES CAN ENABLE INFORMED DECISION-MAKING

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End-of-life care for individuals with Huntington's disease is an important but difficult area for both patients and clinicians. One of the key components of support at late-stage disease is knowledge about what the preferences and wishes of the affected person are in relation to feeding and medical treatment.

These informed decisions ought to be made and recorded when a patient is known to be able to make such choices. For this to be demonstrated, individuals should undergo regular neuropsychological assessment. Patients should be encouraged to appoint someone who can manage their financial and personal affairs when they can no longer do so, and these contact details should be recorded in the medical notes. This information must be easily accessible to clinicians so that they can act appropriately, following the patient's wishes, if for example, the patient has an emergency hospital admission. This should then prevent any additional distress to the patient and their families or carers. This poster describes a document in which the clinician can record affected individuals' wishes and their recommendations for end-of-life care, which we feel should be part of a recognised standard of care in Huntington's disease.

F.5 WHO CARES ABOUT MY FEELINGS? CHILDREN'S SITUATION IN FAMILIES WITH HUNTINGTON'S DISEASE

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Background: Living with a parent with Huntington's disease (HD) can be difficult when the parent's personality is gradually changing. It can be hard for a child to understand what is going on. Children can show signs of anxiety, grief and being burdened with a sense of guilt. Another pressure for the children is that they themselves are at risk.
Aims: The primary aim is to answer the questions: How should we inform children about HD? When? Who should do it? In what way should we talk to children? A secondary aim is to develop a model of management for Sweden and we searched for models in other countries.

Methods: Questionnaires were sent out to 29 international Huntington associations in 22 different countries and the number of answers was 15. Three interviews with children were performed and three visits, two of them in Sweden and one in Scotland.
Results: The results showed that there is a definite need to give attention to the children at an early stage. In our study, 33% of the associations answered that they do not have any particular strategy for coping with the children's situation. The other associations answered that they attend to the children's needs in different ways. The best model was found in Scotland.
Conclusion: There is an obvious and urgent need for guidelines in this area and also specifically formulated, well structured, good information for children in families with HD. A majority of associations were eager to collaborate and the EURO–HD might be a very suitable forum to create a unified model to improve and facilitate the children's situation.

F.6 CURRENT PHARMACOLOGICAL MANAGEMENT IN JUVENILE HUNTINGTON'S DISEASE

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Background: Huntington's disease (HD) is a progressive neurodegenerative disorder. Onset under 20 years is termed ''juvenile Huntington's disease'' (JHD). The clinical presentation of JHD can be strikingly different from adult-onset HD. The management of JHD is aimed at symptom relief; there is little published evidence to guide this.
Aim: To survey the current pharmacological management of JHD in the United Kingdom.
Methods: Patients were identified through the Huntington's Disease Association and hospital practitioners. A questionnaire was sent to each family. Information gathered included symptoms, medications, side effects and perceived effectiveness.
Results: Seven families responded. The mean time since diagnosis was 7 years. Common symptoms included speech difficulties (7), swallowing difficulty (6), stiffness (6), sleeping difficulty (5), pain (4) and behavioural problems (4). The mean number of medications was six (range two to 15). The most commonly prescribed agents were atypical antipsychotics (4). The indications for these included behavioural problems and agitation. Treatments for stiffness included: antimuscarinics (2), levodopa with dopa-decarboxylase inhibitors (2), baclofen (2) and tizanidine (1). The following medications were also prescribed: benzodiazepines (3) and opiate analgesia (3).
Conclusions: Identifying the current practice is a first step towards establishing an evidence base for possible intervention. We hope to extend this survey to Europe and the United States. We are still recruiting in the United Kingdom. In this survey of patients with JHD most were on an anti-parkinsonian medication and/or muscle relaxants and sedation/analgesia. Atypical antipsychotics were commonly prescribed for agitation and behavioural problems in this group of patients. Pain is a feature of JHD that must not be overlooked.

F.7 BEST CARE IN HUNTINGTON'S DISEASE: A FIRST CONSENSUS DOCUMENT IN ITALY

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The Italian lay association for Huntington's disease (HD) (AICH-Roma Onlus and AICH-Milano) provided the initial input in July 2007 asking our research group to create a multidisciplinary panel of experts with the main task of providing recommendations for the best care of HD patients.

The consensus document the panel has produced (the first in our country) answers some key questions, such as the real characteristics of patients involved, the alternative strategies for management, the clinical problems, and outcomes and offers suggestions about the best approach to patients and families.

The document is now in press (10 000 copies) and will then be disseminated to the relevant audience (neurologists, geriatrists, medical professionals involved with movement disorders). The lay association will also be active in distributing the handbook to family doctors involved with HD patients.

As levels of clinical expertise in HD vary considerably among different regions, this consensus document is aimed at promoting a common and better approach to the care of these patients. It can also constitute an important tool of continuous medical education and of appraisal of practice standards.

F.8 EXPLORING SUPPORTIVE CARE FOR INDIVIDUALS AFFECTED BY HUNTINGTON'S DISEASE AND THEIR FAMILY CAREGIVERS IN A COMMUNITY SETTING

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Background: The Heathleigh Unit provides a specialist service for adults affected with Huntington's disease (HD). Two years ago the configuration of the service changed from residential care to a community and day service. This study focussed on the provision of day and community care from the perspectives of service users, caregivers and professionals.
Objectives: To identify the support needs of affected persons and their caregivers. To ascertain the extent to which the specialised unit meets the needs of users and to identify any unmet needs of affected persons and caregivers.
Method: Research participants were invited to take part in one of five focus groups or individual interviews. Audio-taped discussions were transcribed and analysed using an inductive coding technique.
Sample: Participants in the focus groups were (1) affected men; (2) affected women; (3) caregivers; (4) staff of the unit and (5) individuals using the community service. A total of 33 people were involved.
Results/Outcome: Three main themes emerged: (1) transitions and the journey; (2) facing challenges and (3) the role of the service. It was important to affected persons to be known as the person they were, despite the changes associated with the disease. The service provided was considered very appropriate, but flexibility in using the service, especially at the onset of the condition, was also important. Conflicts arose for staff between encouraging affected persons to take part in activities that might be therapeutic and enabling choice. Stigma due to lack of understanding was a barrier to undertaking social activities.
Conclusion: Provision of service in a dedicated unit is an appropriate way to support those affected with HD. Enhancing understanding of the disease in the community may enable patients to undertake a broader range of activities. Allowing the individual to make choices, even when risk is involved, can assist those who are affected and their carers to cope.

F.9 HUNTINGTON'S DISEASE: A PRESENTATION OF COURSES OFFERED TO PATIENTS, RELATIVES, INDIVIDUALS AT RISK AND PROFESSIONALS

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Background: The Centre for Rare Disorders, Rikshospitalet University Hospital, Oslo, Norway, is an interdisciplinary, national

competence centre, which offers information, counselling and courses on a selected range of rare disorders. The centre has responsibility for over 50 rare diagnoses and focuses on both medical and psychological problems. The Centre's service is aimed at the appropriate patients and their families and at professionals working with rare disorders.
Aims: Huntington's disease (HD) is an inheritable, chronic disease. It is therefore necessary to provide ongoing information regarding the individual patient over a period of several years, both to professionals, family members and those genetically at risk so they can plan for today but also for the future. The presentation will illustrate the variety of courses we offer to those affected by HD, both for patients, their families and professionals.
Methods: We arrange three types of course: for professionals; for patients and their families; for family members genetically at risk.
Results: We arrange: information meetings for professionals in the patient's local environment; regional courses when there are several patients in the same district, with up to 60 participants; regular courses for professionals, held in each county; courses for the newly diagnosed and their families; courses for persons at risk. This year we have held a course for professionals from seven counties at Rikshospitalet.
Conclusions: Our courses provide comprehensive information on HD to professionals, patients, their families and persons at risk. They meet others who are in the same situation and are encouraged to establish a network.