

FOREWORD

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A standard of care for Huntington's disease: who, what and why



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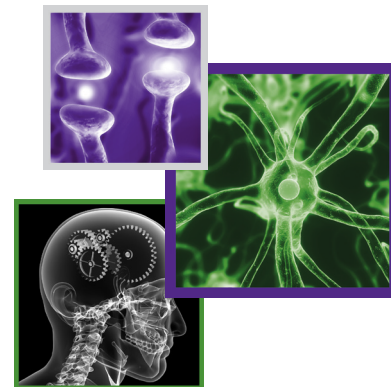
“This paper is an introduction to seven papers that accompany it, all of which are from the Standards of Care working group from within the European Huntington's Disease Network. It gives the background to the group and its work, and presents a summary of the Managed Care Network which is advocated by the group.”

Huntington's disease (HD) is a relentlessly progressive and incurable neurodegenerative disease that affects men and women, usually presenting late in their 4th decade. Each offspring of an affected person has a 50% risk of inheriting this autosomal dominant disorder. It bears the name of George Huntington who described the disease in his talk, then paper, in 1872 [1] although others had written in earlier years about what is clearly the same disease [2].

The cardinal signs of HD include the movement disorder that is usually but not always choreiform in nature, as well as cognitive impairment, personality change and neurological features, which include swallowing disorder and speech impairment. Some of these outcomes of the disease can be managed with some success, but there is no cure. ‘This man has Huntington's disease; there is nothing more I can do’ has been seen in patients' hospital notes on a regular basis, but there is none the less much that can be done to improve the quality of life of the HD patient and their families.

There is no internationally recognized standard of care for Huntington's disease

and there are very few peer-reviewed papers [3–6] on this topic, despite clear evidence of the need for such work [7]. The evidence for best practice is lacking. In part, this reflects the fact that in many countries there are no specialist clinics for this patient group, and statistically significant data about care from large patient groups are lacking. Throughout the world, the care provided for HD families varies widely. Some clinics are led by clinicians with an interest who have developed their service on an *ad hoc* basis. Some are part of general psychiatric or neurology clinics. There are differing approaches to such clinics, and differing opinions about what is necessary at such a clinic. Data presented by Simpson were collected from 28 clinicians involved in specialist care of HD in nine European countries [8]. In total, 68% of the respondents were neurologists, 54% of them shared clinical responsibility for the patients with another clinician. These other specialities included neurology, genetics, psychiatry, neuropsychiatry, neuropsychology and specialists in



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movement disorders. All but two clinicians worked within a hospital setting. A total of 57% offered outreach to nursing homes. Despite the lack of use of guidelines, other critical staff were either present at most clinics, or referrals were made, with 57% having a neuropsychologist present. The most common referral was to a family group representative (79%).

Methods used for clinical assessment of the signs and symptoms of HD were not uniform, although most clinics used the Unified Huntington's Disease Rating Scale (UHDRS) [9]. For cognitive testing, a variety of tools were used to supplement the UHDRS. To assess behavior, the Problem Behaviours Assessment [10], psychiatric interviews and other methods were used in addition to UHDRS. General neurological examination, as well as or instead of the UHDRS, and other tests (such as hand tapping and timed walk) were also reported. Audit of these practices is rare, with publication of evidence even more so.

The role of the European Huntington's Disease Network

The European Huntington's Disease Network (EHDN) [101] has allowed the creation of a network within Europe and beyond of like-minded clinicians and scientists involved in the care of HD, to stimulate and support research into the pathology and treatment, and ultimately cure, of this devastating disease. The Network provides a platform for professionals and families affected by HD to work together. It is already proving to be an invaluable tool and resource for families, clinicians and scientists [102]. Working groups exist to encourage new collaborative work, and the Standards of Care (SoC) working group is one of these to use the expertise of families and clinicians involved in HD. It has the aim of producing an internationally approved guideline for care for this disease.

A recognized standard of care can be expected to produce benefits for patients, but in addition carefully managed clinics will allow for the collection of invaluable data. Knowledge of the pathophysiology of HD is an essential component of research into its cause and prevention. Without detailed and accurate clinical information, such research cannot proceed. Regular assessment and documentation of stage of disease is essential for this process. Trials of therapy will depend on the ability to properly compare progress before and after intervention.

The Standard of Care Working Group

We are aware of the need for peer-reviewed evidence of best practice to be available to those who create guidelines. In the UK, the National Institute for Health and Clinical Excellence (NICE) [103] provides guidance, sets quality standards and manages a national database to improve people's health and prevent and treat ill health. The Scottish Intercollegiate Guidelines Network (SIGN) [104] was formed in 1993. Their objective is to improve the quality of healthcare for patients in Scotland by reducing variation in practice and outcome, through the development and dissemination of national clinical guidelines containing recommendations for effective practice based on current evidence. Both of these UK-based bodies evaluate published, peer-reviewed evidence to produce their guidance, and this can be seen as best practice. A first step in gathering information about the different approaches to management of HD around Europe was to systematically review the existing literature on care of HD in the many disciplines involved using SIGN methodology [105]. But we are also aware that a consensus of experts about what is regarded as best practice can be used to guide clinicians in their care of patients. Within the context of a largely evidence-free environment, the groups have chosen to follow the latter approach, and have suggested care pathways encompassed within a managed care network (MCN), based on that published on the EHDN website (Figure 1). It has its origins from a draft produced by John Eden from the Scottish Huntington's Association [106], and has been accepted at an international level as describing the complex needs of the HD family and as a proposed pathway for meeting these needs.

The definition of best practice is controversial, but using groups of individuals with considerable clinical experience of the management of HD within their field has been the approach of this group. HD is a family disease. Indeed, the impact of HD goes beyond the immediate symptoms experienced by the person who is ill. It affects the whole family: the carer and the person living at risk, the person in receipt of an unfavorable test result as well as the symptomatic patient. HD is a complex disease and requires a multidisciplinary approach, involving a range of services that are required at each, differing stage of a person's life with the disease.

Therefore, the SoC group comprises clinicians from various disciplines and many different countries within Europe as well as the USA.

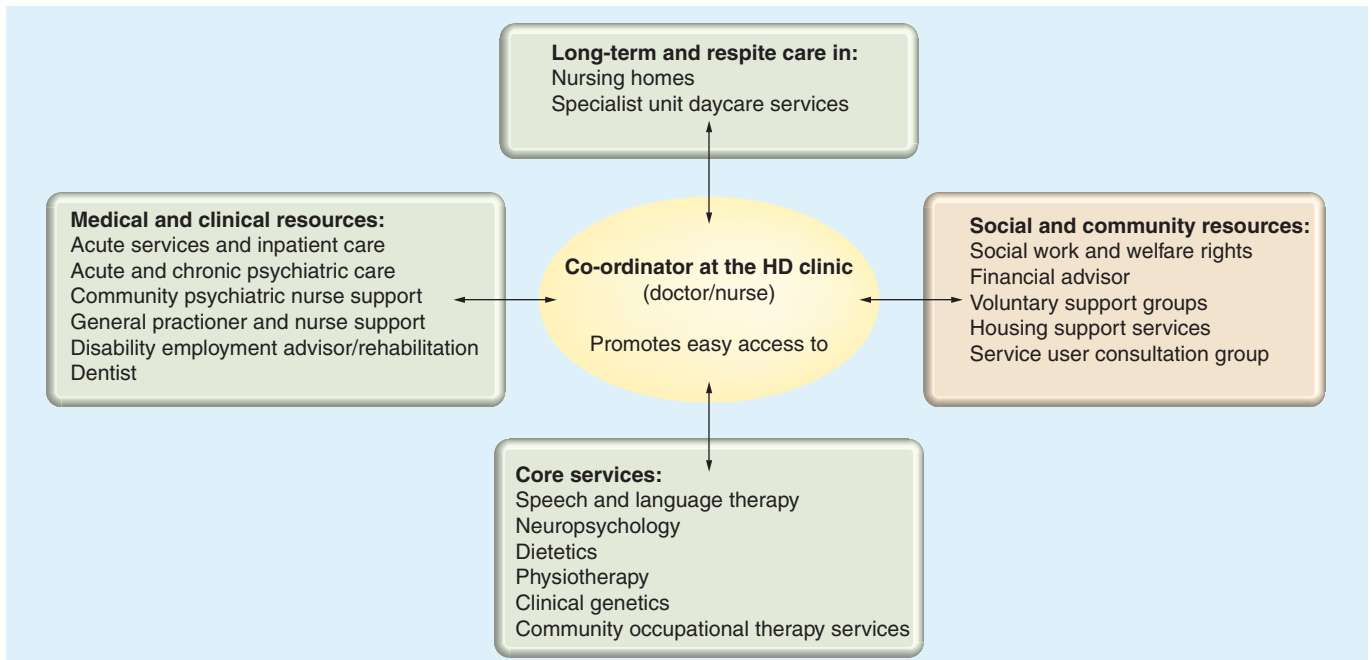


Figure 1. The care pathway within a managed care network.

HD: Huntington's disease.

Redrawn with permission from the European Huntington's Disease Network Standards of Care Working Group.

Doctors and specialist nurses from psychiatry, clinical genetics and neurology, physiotherapists, occupational therapists, dietitians, speech and language therapists and dentists have all contributed. As busy clinicians, it is not easy to allocate time to such a project as this, and we are grateful to the committed clinicians who have contributed thus far.

Recognizing that there are many aspects of HD care that require expert input, the SoC working party was divided into specialty groups: physiotherapists, speech and language therapists, dietitians, occupational therapists and dentists. These groups are not all inclusive of those within the MCN, and so several aspects of care have not been addressed by the group.

■ **Social work**

The essential role of social workers in the provision of community and financial support is recognized. These guidelines are designed for international application, and it is felt that intercountry differences were at their greatest in this area. This specialty has therefore not yet been addressed.

■ **Neuropsychology**

There is much evidence of the progressive cognitive and executive impairments in HD. To understand

the person with HD, a neuropsychology evaluation is required. Early deficits can result in family conflict, marital discord and loss of employment before a formal diagnosis of HD is made, usually based on neurological features. Knowledge of cognitive status which goes beyond the anosagnosia and the ubiquitous 'I'm fine!' of HD is imperative. The clarification of competence is essential for family care in dealing with finances, and in the planning and provision of care. Regular assessment is therefore required, but the frequency of such assessments will depend on the stage of disease. The EHDN is addressing the issues of which tools to use, and their frequency within a separate working group, and this area of care is therefore not addressed by the SoC group.

■ **Neurology**

Regular evaluation of the neurological features exhibited by an affected person gives knowledge about rate of progression and type of disease. Tools such as the UHDRS are useful since it addresses the essential elements of the clinical features, but its limitations are acknowledged. An EHDN working group is examining this with a view to introducing a modified UHDRS and perhaps other new tools to measure and evaluate

the motor features of HD. The neurological examination of HD does not require a formal neurological background and can be taught to other healthcare professionals with appropriate training and experience in clinical examination. These issues are being addressed by others within EHDN, and therefore not by the SoC group.

■ Other aspects

Other working groups within the EHDN are addressing treatment options (for instance useful drugs), assessment tools and quality of life measurement. The SoC group has concentrated their efforts in the clinical areas where support may benefit the patient, and where guidance was particularly lacking.

Not all countries have access to the services that are recommended, and not all patients need all aspects of the care described. An audit of the patient group at an international level is planned so that this can be gauged. Liaison with international family organizations allows us the opportunity to document the wishes of the Huntington's patient and to assess the impact of various treatment regimens. Of particular importance is our investigation of groups that are disadvantaged in their access to clinical review because of language or cultural barriers. In order to translate the benefits of the expert recommendations into optimal care, the group is devising a framework to widely disseminate those to clinicians, patients and their families.

The SoC group is creating a database of relevant literature on care issues, aids and other resources, and are to install these databases with EHDN for all to access. These, and country-specific sources of expert advice are planned for the EHDN website.

A pathway of care

■ Who cares?

An essence of care is to support and ameliorate the features it addresses. A long-standing problem for those affected by HD is that their disease crosses the boundaries of medical, psychiatric and social problems. Who is in charge? It is hoped that guidelines for care will address this with a clinic coordinator who is empowered to cross boundaries: this person need not be a clinician, but certainly someone with specialist knowledge of HD and its complexity.

■ What care?

The MCN illustrates the wide variety of expertise required to care for the families afflicted by HD. There are many areas where further work is required to clarify the extent of such involvement, and to assess the value for each family. Which professional should be at any clinic, and the detail of multidisciplinary working must be addressed at a local level. Audit of the work and research of the benefits or otherwise of the MCN should take place. In particular, the views of the families must be sought.

This group of papers is unique in this field. Collaborators from different countries, with different backgrounds but with a shared extraordinary commitment to the care of the HD families, have produced consensus papers in their fields. It is only since the advent of EHDN that there has been an ability to speak as one voice for HD. Research studies have been facilitated with rapid recruitment of participants in numbers adequate to lend validity to the studies. As yet, no large studies of management of HD have taken place that would lend themselves to the scrutiny of organizations such as NICE, or SIGN. The members of the SoC Working Group hope that these guideline papers will create firstly a better life with HD than before for the families, but also create a basis for audit, evaluation and further research.

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