Wilson's Disease National Standards Document

Wilson's disease (WD), as with other Rare Diseases, requires a coordinated, cross-specialty approach by clinicians with expert knowledge and experience to ensure the best chance of long-term health.

A comprehensive WD service should be based on the 4 key principles listed below:

- Provision of **resources** offering a holistic view managing all aspects of WD over a patient lifetime by utilising a multidisciplinary approach with the requisite Clinical expertise
- Employment of a **consistent clinical approach** to the diagnosis and management of WD with ready access to robust genetic and biochemical analysis
- Implementation of **education and training** in primary and secondary care to enable early and accurate diagnosis of WD
- Support **research** into all aspects of WD including the development of new therapies and improving the lifelong care of patients, including data capture for the national interest

Basic Service Requirements

In line with the UK Rare Disease Strategy, a Specialised Service for WD should incorporate the following key features:

- **Centre of Excellence (C of E)** comprising Core Specialists recognised as leaders in the management of WD, providing up to date advice and guidance to patients, fellow Clinicians and General Practitioners.
- **C of E to support an infrastructure of satellite hospitals** to improve early diagnosis and provision of lifetime care to the distributed WD population to agreed clinical standards.
- **C of E to review all patients** at agreed periods to monitor and coordinate care and that the service provided is to specified clinical and service standards. Trientine patients are mandated for annual review at C of E. There should be a clear audit trail of monitoring of patients with WD.

A core feature of any Centre of Excellence should be the capability to provide a **coordinated, multi-disciplinary service** that addresses the immediate medical diagnosis and the management of lifelong care for WD patients. Important constituents include:

- **Hepatologists** - key role in diagnosis and treatment as the liver is the primary site of the genetic defect; Core members. Hepatologists will also have the necessary links to transplant centres (liver transplantation may be required in certain circumstances).
- **Neurologists** with specific expertise in movement disorders - crucial in the diagnosis and management of Wilson's disease since neurological manifestations are common, potentially debilitating and irreversible if detected late; Core members.

- **Clinical Biochemists** - expertise is fundamental for the laboratory monitoring of WD; Core members.
- The link between Biochemistry, Hepatology and Neurology is critical, including links between Adult and Paedatric departments in each.
- **Neuropsychiatrists** - as psychiatric manifestations are common, affected patients should have access to the Specialist Services available within regional Neuroscience Centres. In addition access to a **Clinical Psychologist** is key for paediatric patients including during transition to adulthood.
- Integrated access to other clinical specialties that may be involved but less frequently in the care of patients with WD including **Ophthalmology, Dermatology and Bone Medicine Specialists.**
- **Specialist nurse support** for patients with WD, their families and/or carers, is desirable to address issues specific to WD and who will be responsive to problems requiring specialist clinical input.
- **Provision of Genetic counselling** to patients, siblings and other family members, specifically the ready access to Clinical Genetics Services.
- **Provision of allied health services** including speech and language therapy, physiotherapy and occupational therapy.
- **Provision of patient support** (WD Support Group UK).

C of E should deliver a fully coordinated service utilising **multi-disciplinary clinics** with the following features:

- **Care coordination services, adequate administrative resources** for managing patient care plans and medical services when patients attend multi-disciplinary clinics and support lifelong care when required.
- **Recognising the need to provide advice** where applicable in relation to social care, occupational therapy, speech and language and swallowing therapy, disability benefits and driving
- **Medical baseline review**, which documents patient symptoms and can be used to assess disease progression, consistent with recognised expert guidance, e.g. EASL Clinical Practice Guidelines: Wilson's disease (2012).
- Ideally a **Patient Care Plan,** which should include treatment plans for any issues identified in the medical baseline review and how and who will monitor them. Any concerns relating to social service matters that might impact on the overall management of the patient's health should also be included. The Care Plan should also identify potential risks and problems that the patient should be aware of during their treatment.
- **Follow up medical assessment consistent with recognised expert guidance**, e.g. EASL Clinical Practice Guidelines: Wilson's disease (2012).

- **Annual patient (and where appropriate, family) review** evaluating the quality of care and health of patient, preferably against actions or strategies documented in the patient care plan.
- **Coordination and liaison with paediatric services** to manage the transition of adolescent patients to adult services.
- **Advice and medical care for female patients with WD** who are planning a family and support for pregnant woman during and after their pregnancy.
- **Medical administrative and communication resources** to collect patient information from satellite hospitals for patient reviews and to assist in providing advice and guidance to manage the distributed WD population.
- Transparent links between the C of E and respective satellite units, and their respective roles.

Medical Research

In common with many rare diseases patients with WD often suffer from an incorrect or delayed diagnosis. Progression of the disease over a patient's lifetime may require more complex care**.**

C of E should coordinate and lead research for example in the following areas:

- **Improving Early diagnosis**, developing new tools and techniques to make an accurate diagnosis in shorter timescales across all age groups.
- **Supporting trials into new drug treatments.**
- **Medical Genetics: review the use of new genetic approaches** that might be used in the treatment of WD and assist in treating wider health issues associated with WD such as mental illness.
- **Improving the management of clinical issues** that influence the quality of life for patients with WD e.g. joint and skin problems.
- **Support the development of registry initiatives such as the Public Health England NCARDRS service to** hold data on WD individuals to support clinical practice, audit, service planning and research.

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[1] Version 1.2 Wilson's Disease BASL Special Interest Group