EASL publishes first European Clinical Practice Guidelines for Wilson's disease

7 February 2012

Geneva, Switzerland: The first European Clinical Practice Guidelines (CPGs) for the diagnosis and management of Wilson's disease are published today by the European Association for the Study of the Liver (EASL) on the EASL website -- www.easl.eu.(1) Developed to assist physicians and healthcare providers in the clinical decision making process, the guidelines describe best practice for the diagnosis and treatment of patients with Wilson's disease -- a rare genetic(2) disorder that, if left untreated, is fatal.

Approximately one in 30,000 people worldwide are affected by Wilson's disease -- a condition in which copper is not excreted by the body effectively, leading to excess copper build up, liver failure and damage to the brain. While Wilson's disease may manifest at any age, the majority of patients present between the ages of 5 and 35.

Lead author Professor Peter Ferenci said: "The clinical presentation of Wilson's disease can vary widely, but it must be considered in any patient who presents with a combination of unexplained liver disease and neurological or neuropsychiatric disorders. In the absence of Kayser-Fleischer rings(3) -- which are typical, but not always present -- the guidelines recommend measurement of urinary copper excretion and hepatic parenchymal copper as diagnostic methods of choice. Notably, age alone should not be the basis for eliminating a diagnosis of Wilson's disease."

The CPGs, based on a systematic review of existing literature, provide best practice diagnosis and treatment protocols with an emphasis on:

- Clinical presentation and prognosis
- Diagnostic strategies (e.g. serum ceruloplasmin, basal 24-hour urinary copper excretion, genetic analysis)
- Importance of family screening
- Treatment options (e.g. chelating agents, zinc, liver transplantation)

With treatment, prolonged survival has become the norm for Wilson's disease patients. The guidelines recommend chelating agents -- drugs that bind to copper and remove it from the body (D-penicillamine or trientine) -- as the initial treatment for symptomatic patients and that, unless liver transplantation is performed, treatment is maintained for life.

Professor Roderick Houwen added: "Unfortunately, as there are no optimally designed randomized controlled trials conducted in Wilson's disease, there is a lack of high-quality evidence to estimate the relative treatment effects of the available drugs. Our evaluation is mostly based on large case series that have been reported in recent decades, which highlights a clear need to conduct more robust randomized controlled trials to better understand treatment for this rare condition."

Professor Mark Thurz, EASL Secretary General, added: "EASL is dedicated to promoting hepatology research and education to improve the worldwide treatment of liver disease. Its series of Clinical Practice Guidelines aims to promote best practice to drive better clinical outcomes and inform both the scientific community and the wider public of the latest developments in the field. We hope these new Wilson's disease guidelines provide clinicians with the most up-to-date, evidence based methods for the management of affected patients."

The Wilson's disease CPGs will be published in the March issue (Volume 56, No. 3) of the *Journal of Hepatology* -- EASL's official journal.

More information: 1. Ferenci P, et al (2011) Wilson's disease: EASL Clinical Practice Guidelines. European Association for the Study of the Liver. Available at www.easl.eu/_clinical-practice-guideline

2. Due to mutations of the ATP7B gene on chromosome 13.

3. Kayser-Fleischer rings are a golden-brown discolouring of the eye's corneal rim. The cornea is the transparent front part of the eye that covers the iris, pupil, and anterior chamber.

Provided by European Association for the Study of the Liver