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Wilson France: a national database for Wilson's disease

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Introduction

Wilson's disease (WD) is a rare inherited disease with an efficient treatment if initiated early. Improving the knowledge of this disease is a priority of the French national Centre of Expertise for a better access to diagnosis and treatment. This national organisation created a database.

Objective

Improve the knowledge of WD by an epidemiological study on the French cohort.

Methods

We registered all patients followed by all the French centres working with the national Centre of Expertise.

Results

Since 2006, 281 patients (1-73 year old) were included in the Wilson France database (sex ratio: 1). Mean age at diagnosis was 19 years. First symptoms were neurological for 36% of the patients, hepatic for 38%, renal, psychiatric or hematologic for 11%. Fifteen percent were diagnosed after familial screening. At time of diagnosis, Kayser-Fleischer ring was observed in 95% of patients with neurological symptoms, in 55% of hepatic presentations and in 26% of the presymptomatic forms. Mean coeruloplasminemia was low (0.08 g/L) but 5% of patients had normal values (>0.2 g/L). Mean urinary copper was increased in 96% of the patients. Genetic investigation was not conclusive in 15.9 % of the families (only one or no mutation found). First treatment was D-Penicillamine in 85% of the cases and after a mean follow up of 15 years, the treatment was D-Penicillamine for 44.4% of the patients, Trientine for 14.4%, Zinc for 26.7%, association of chelator and zinc for 5.6 %; 5.6 % of the patients had liver transplantation.

Discussion

The database included approximately 1/3 of the Wilson disease patients in France. In order to improve the recruitment of Wilson's disease patients, coordination of all health professionals with a multidisciplinary approach is necessary. This work is realised in collaboration with Eurowilson database.

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