1249

Clinical Presentation and Outcome of Patients with Severe Form of Wilson's Disease from a Monocentric Cohort of Liver Reference Center

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Abstract Text

Background:

Wilson's disease (WD) is a rare autosomal recessive disorder of copper metabolism, leading to liver cirrhosis and neurologic disorders. Even if there is an efficient chelating treatment, liver transplantation (LT) is the treatment for patients with severe presentation or with severe worsening under well-conducted therapy.

Aim:

The aim was to access clinical presentation and outcome of patient's with severe form of Wilson's disease in a monocentric cohort of liver reference center.

Methods:

This is a retrospective analysis of all patients with diagnosis of WD in a liver reference center (Paul-Brousse Hospital, Villejuif, France). Patients were evaluated clinically, biologically, morphologically and genetically. Severe hepatic forms were defined by prothrombin time <50%.

Results:

We included 107 patients with hepatic symptoms of WD, 54 (50.5%) females and 53 (49.5%) males, from 1974 to 2016. The mean follow up was 15 years [extr: 1-44 years]. 57 (53.3%) had neurological symptoms associated with the hepatic symptoms at admission. The mean age at diagnosis was 20.1 (±10.54) years with a difference between the age concerning the patients with hepatic symptoms (17±8.6 years) and mixed symptoms (23±11.4 years) (p=0.0081). 73 patients (68%) had cirrhosis at diagnosis. A total of 34 patients (32%) were transplanted, at a mean age of 27 (±12.2) years. 50 patients (47%) presented with severe hepatic form of the disease. Concerning 21 of these 50 patients (42%), a LT was indicated after a mean interval of 142 days [extr: 1-720 days]. 8 of these patients had at presentation a fulminant liver failure and had been transplanted in a mean interval of 5 days [ext: 1-21 days]. Among other patients who required LT, 8 (23.5%) had severe neurological symptoms and 5 (14%) had liver cancer. Excluding patients with coma due to encephalopathy, all other patients with severe forms received copper chelation treatment. 26 of the 50 patients (52%) with severe hepatic forms improved with copper chelators and didn't required LT. None of the patients transplanted for severe hepatic forms died during follow-up (mean follow-up: 17 years [extr: 1-27 years]). Among the 26 patients who improved (mean follow-up: 17 years [extr: 2-32 years]), one of them died in a context of cerebral hemorrhagic stroke. One of the 8 patients transplanted for severe neurological symptoms died of severe infection. There was no specific genetic mutation associated with the severity of the disease.

Conclusion:

At presentation, two thirds of patients with WD referred to a liver reference center had cirrhosis and 47% presented with a severe hepatic form. For 42% of severe hepatic forms, a LT has been requiered.The prognosis of these severe forms of WD is excellent for patients in charge in a specialized center.

Disclosures

The following people have nothing to disclose: Martha Darce, Emmanuel M. Gonzales

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