

WILSON'S DISEASE: CLINICAL EVOLUTION OF MOLDOVAN PATIENTS

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Introduction. Wilson's disease (WD) is an autosomal recessive genetic disorder associated with a high mortality and disability rate. WD manifests as chronic liver disease and/or neurological impairment due to accumulation of copper in several tissues, principally in the liver and brain (Fig.1). Early diagnosis and therapy can result in a good prognosis of WD.

Purpose. To analyze the clinical and laboratory evolutions of WD and the effects of the standard treatments in Moldovan patients with WD.

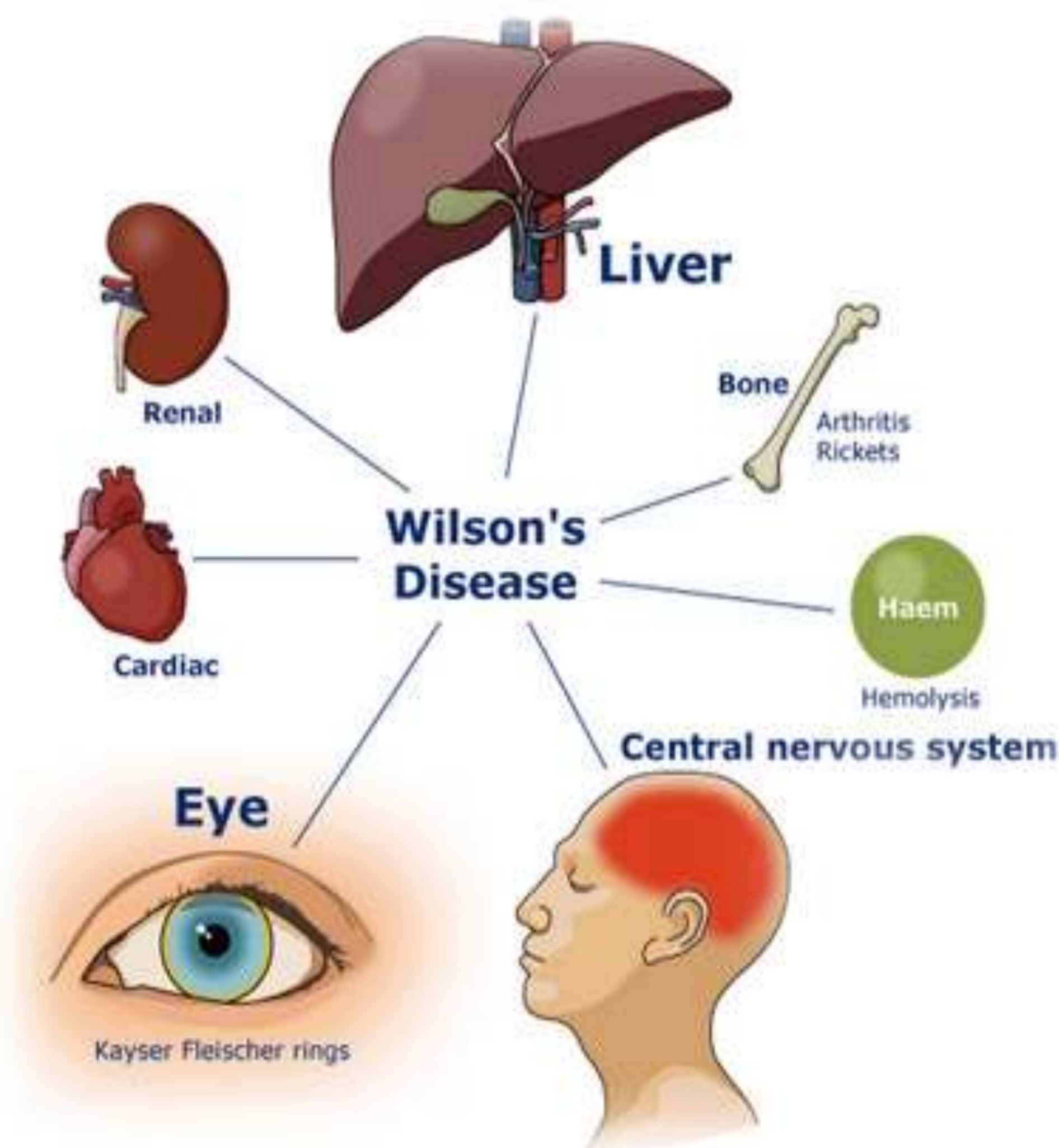


Fig. 1

Material and methods. 15 patients (6 females and 9 males) with WD were evaluated retrospectively between 2018-2021. (Fig.2)

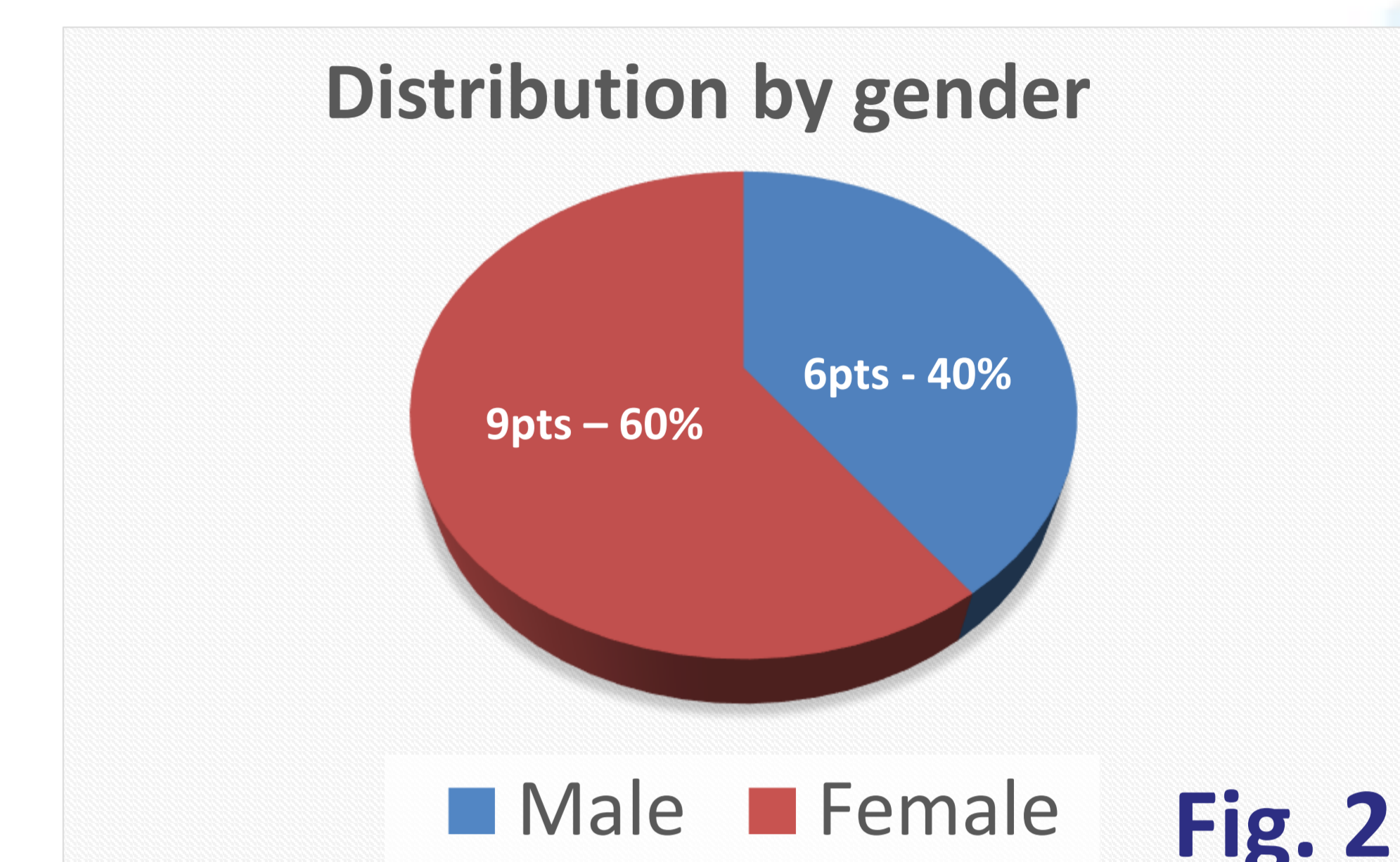


Fig. 2

Results. The median age at diagnosis was 22 years (2 – 36 years). The distribution by clinical phenotypes is presented in Tab.1.

Tab.1

Distribution by clinical phenotypes				
Neurologic	Hepatic	Mixed	Kayser-Fleischer	Asymptomatic
4 pts – 26,7%	3 pts – 20%	3 pts – 20%	1 pt – 6,7%	4 pts – 26,7%

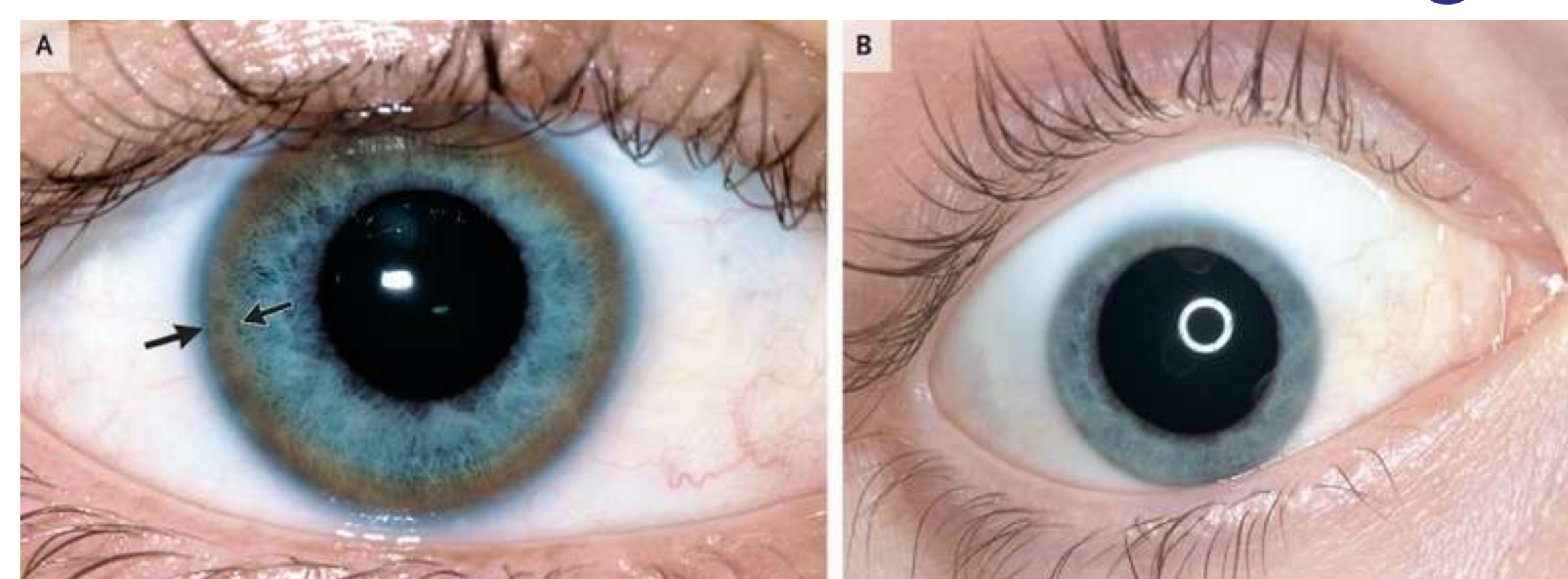


Fig. 3

Fibrosis analysis (by Fibroscan) revealed that: 6 patients - F2, 2 - F4, 2 - steatosis, 1 - F0. The treatment consisted of D-penicillamine associated with pyridoxine for all patients. At the end of the study, all treated hepatic patients were asymptomatic. (Fig.3)

Conclusions. The study suggests that Wilson's disease must be ruled out in children older than two years presenting with abnormal levels of hepatic enzymes because of the heterogeneity of symptoms and the encouraging treatment results obtained so far.