Pseudodominant inheritance of Wilson disease – case report

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Introduction: Wilson's disease (WD) is an inherited autosomal recessive disorder of a copper metabolism. The diagnosis of WD is usually made before age of 40, so pseudo-dominant inheritance is observed mostly in offsprings of WD patients, the risk is 4%. We describe a family in which WD was firstly diagnosed in a daughter of a father who was asymptomatic up to 60 years old. Case report: Proband was diagnosed with WD upon copper at age of 31 years when she had an acute hepatic failure. Her brother, age 37, was also diagnosed with WD. He was clinically asymptomatic but had abnormalities in laboratory liver tests. One year later WD was diagnosed in her father at age of 62 years. During hospitalization for an acute bronchitis, he was diagnosed with a liver cirrhosis and an esophageal varices. He has never suffered from any liver symptoms before. He has also decreased ceruloplasmin level, Kayser-Fleischer rings, neurological symptoms and brain MRI abnormalities characteristic for WD. Genetic showed the same mutation as his daughter has, but present only on one chromosome. Conclusion: This case report shows how important it is to obtain detailed family interviews after diagnosing WD in the proband. Information should be gained not only proband`s siblings, but also for other relatives although recognition of disease in the parents of WD is very rarely described.