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CASE REPORT

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Wilson's disease with Leu492Ser mutation and arylsulfatase A pseudodeficiency: just a coincidence?

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Abstract Wilson's disease (WD) is an autosomal recessive disorder of copper transport, related to mutations of the *ATP7B* gene (McKusick 277900). Here we report a new case of WD in which a rare mutation, Leu492Ser expressed for the first time in homozygosity, is associated with neurological presentation of the disease and arylsulfatase A pseudeficiency.

Key words Wilson's disease • Leu492Ser • Arylsulfatase A pseudodeficiency

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Introduction

Wilson's disease (WD) is an autosomal recessive disorder of copper transport that is clinically and biochemically well characterized and related to mutations of the *ATP7B* gene [1]. The frequency of this mutation in white populations is 0.56%. Here we report a new case of WD in which a rare mutation, the Leu492Ser, is expressed for the first time in homozygosity and is associated with neurological presentation of the disease. Moreover, in this patient, the mutation of the *ATP7B* gene is associated with arylsulfatase A pseudodeficiency, a condition characterized by low enzyme activity without impairment of sulfatide metabolism [2]. We speculate about the possible role of arylsulfatase A pseudodeficiency in this as well in other neurologic disorders.

Case report

The patient was a 30-year-old white man born from a consanguineous marriage (the parents were first cousins). He was healthy until 14 years of age, when he developed behavioral changes, psychic disturbances and mild dysarthria. He was treated with psychoactive drugs for two years. At 16 years of age, he suffered a cerebral trauma with fracture of the C6 vertebral body, followed by post-traumatic pontobulbar syndrome and cervical myelopathy. A few months later, in addition to pyramidal signs, he showed dystonic movements of the neck and limbs and progressive speech impairment.

He was referred to our Unit at the age of 22 years. Neurological examination showed dysarthria, severe hypotonia, dystonic postures (more pronounced in the neck and right limbs) and hyperactive deep tendon reflexes. Gait was severely impaired by dystonic movements.