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Type I Sialidosis: A Clinical, Biochemical and Neuroradiological Study

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Key Words
Sialidosis · Macular cherry-red spot · Neuraminidase · Sialyloligosaccharides · Nuclear magnetic resonance imaging

Abstract
We report biochemical, morphological and neuroradiological findings in a 40-year-old woman affected with type I sialidosis. The clinical symptoms, consisting of a cerebellar syndrome, were first noted at the age of 17 years. The macular cherry-red spot was first observed after 23 years of disease. A CT scan performed at 21 years of age showed enlargement of the fourth ventricle. Nuclear magnetic resonance imaging of the brain performed at the age of 40 showed severe atrophy of the cerebellum and pontine region; atrophy of cerebral hemispheres and of the corpus callosum was also observed. We emphasize the prolonged course of illness in this patient, observed over a long period of time. Of particular interest is the neuroradiological study showing our findings both at the beginning of the disease and after 20 years.

Introduction

Sialidosis is a rare autosomal recessive lysosomal storage disorder due to isolated α-neuraminidase (sialidase) deficiency leading to a defect in the degradation of glycoproteins and accumulation of sialic-acid-containing oligosaccharides and glycopeptides (MIM 256550). Two types can be distinguished: type I sialidosis refers to a slowly progressive syndrome characterized by decreased visual acuity in childhood or juvenile age, macular cherry-red spot, action myoclonus and grand mal seizures; type II sialidosis comprises the severe infantile and congenital phenotypes [1]. Patients with the congenital form show severe non-immune hydrops fetalis and ascites [2], while all patients with the infantile form have a dysmorphic gargoylic aspect, visceromegaly, dysostosis multiplex, macular cherry-red spot and mental retardation. Renal involvement has also been described [3, 4]. Biochemical analysis shows neuraminidase deficiency; however, a residual activity in type I is generally found [1]. The two forms belong to the same complementation group as shown in 1980 [5] and are probably due to different mutations within the same gene.

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