Case report

Gaucher disease type I complicated with Parkinson’s syndrome

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Abstract—Gaucher disease type I is the so-called non-neuronal adult form of the autosomally inherited lysosomal storage disease. The simultaneous occurrence of Gaucher disease with Parkinson’s syndrome has been reported to aggravate both disorders, leading to an unusually early onset and therapy resistance. Neurological alterations in Gaucher disease type I are mostly related to CNS bleeding and skeletal complications. The patient presented here was sensitive to combination therapy for 5 years.

Key words: Gaucher disease; Parkinson’s syndrome.

INTRODUCTION

Gaucher disease is a lysosomal lipid storage abnormality due to glucocerebrosidase enzyme deficiency inherited in an autosomal recessive way. As a consequence, cerebrosides resulting from the breakdown of membrane glycosphingolipids of degenerating cells all over the body cannot be digested. The accumulation of these lipid-containing metabolites in the macrophages causes their structural alteration, leading to the enlargement and functional impairment of the organs related to the reticuloendothelial system. Cytopenias, pathological fractures, and neurological alterations may develop.

Gaucher disease types II and III are characterized by mental retardation, seizures, and movement disorders, while in the adult type I disease such symptoms almost

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never develop. In Gaucher disease type I, CNS bleeding and nerve compression syndrome may occur in the course of the disease, related to thrombocytopenia and pathological bone fractures. We report here a case in which Parkinson’s syndrome and Gaucher disease occurred coincidentally.

CASE REPORT

A 62-year-old male patient was found to be thrombocytopenic on a routine medical check-up in September 1995. On physical examination, there was a mild palpable hepatosplenomegaly. The patient had no bleeding tendency. At that time, he complained of instability on walking and weakness in his legs. Bone marrow analysis revealed marked accumulation of Gaucher cells (Fig. 1). Measurement of enzyme chitotriosidase and beta-glucocerebrosidase activities confirmed the suspected diagnosis (Table 1). Specific Gaucher-type bone abnormalities were not detected. The patient was heterozygous for N370S [1] and V394L mutations and negative for 84GG mutation.

On neurological examination, there was no sign of a cranial nerve defect. The left arm showed dysmetry and dysdiadochokineses on pronation. There was no palsy of the lower extremities. Attenuation of reflexes on the left side and areflexia of the right patella were detected. Muscular tone was normal. Vibrational hypoesthesia of the right lower limbs and tactile hypoesthesia of the left upper extremity and the trunk were found. There was no coordination abnormality of the extremities. There was slight gait ataxia to the left. An MRI scan of the brain showed minimal

Figure 1. Gaucher cells in bone marrow (original magnification × 400; May Grünwald Giemsa stain).