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Poster Abstract: Diagnostic

Intracranial Arterial Abnormalities in Patients with Late-Onset Pompe Disease

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INTRODUCTION

Pompe disease is a rare metabolic disorder due to mutations in the gene encoding acid alpha-glucosidase (GAA), involved in glycogen degradation. Its impairment leads to glycogen accumulation in multiple tissues with a predilection for skeletal muscle, heart, and smooth muscle. Clinical presentation of late-onset Pompe disease (LOPD) is usually characterized by a proximal/axial muscle weakness and/or asymptomatic hyperCKemia that may be associated with signs and symptoms of respiratory impairment. Nevertheless, Pompe disease may be considered to be a multisystemic disorder since it has been demonstrated that there is a large involvement of different tissues and organs. Recently, cerebrovascular malformations have been demonstrated in LOPD patients, likely because of glycogen storage in the blood vessel walls. The aim of this study was to characterize the prevalence and type of intracranial arterial abnormalities in a cohort of LOPD patients.

MATERIALS AND METHODS

We studied 16 LOPD patients, aged 16–75 years. The diagnosis was achieved through biochemical and/or genetic analysis. Clinical aspects were characterized by presymptomatic hyperCKemia (4 patients), early respiratory impairment (1 patient) and proximal/axial muscle weakness (11 patients). Ten patients were under treatment with enzyme replacement therapy (ERT). A cerebral CT angiography, using MIP and VRT technique for 3D image reconstruction, was performed in all patients.

RESULTS

We found a cerebral aneurysm in 2 patients (dimension 2 mm and 4 mm) both located in the right middle cerebral artery, a fenestration of the basilar artery (1 patient), a vertebrobasilar dolichoectasia (5 patients) and some posterior circle anatomical variations (4 patients), either left or right vertebral artery dominance and posterior cerebral artery fetal origin. It is worthwhile mentioning that none of these patients manifested symptoms attributable to cerebrovascular alterations.

CONCLUSIONS

Our data confirmed the occurrence of cerebral artery abnormalities as a frequent finding in LOPD, also showing that they mainly involve the posterior circle. These abnormalities are rarely symptomatic, but, if not timely diagnosed, they could lead to life-threatening events, such as subarachnoid hemorrhage or brainstem compression. Therefore, in the light of these results, it seems reasonable to include a CT angiography or a MR angiography in the basic evaluation of LOPD patients. Early detection of cerebrovascular malformations could lead to an appropriate follow-up and, even, to a timely surgical approach.

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