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Poster Abstract: Clinical

Pompe Disease and Normal Whole-Body Magnetic Resonance Imaging

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BACKGROUND

Pompe disease (PD) or glycogen storage disease type II, is an autosomal recessive disorder characterized by a deficiency in the lysosomal enzyme acid alpha-glucosidase. Several phenotypes have been described, ranging from rapidly progressive infantile forms to slowly progressive late-onset forms.

Recent studies have shown that magnetic resonance imaging (MRI), based principally on signal intensity changes resulting from fat infiltration into muscle and also from decreased muscle volume, can contribute significantly to a specific diagnosis in patients with several inherited muscular disorders, including PD, and contribute to an extensive evaluation of muscle alterations.

MRI in PD usually shows a very suggestive myopathic pattern with fatty infiltration and atrophy of muscles more evident in the spine extensors and pelvic girdle muscles, and consistent changes in the tongue and subscapularis.

METHODS

Our patient is a 9-year-old boy with PD who presented before the age of 4 years with high transaminases and moderately elevated phosphocreatinokinase on routine laboratory tests. A muscular biopsy performed when he was 7 years old showed abnormalities suggesting Pompe/Danon disease. Enzymatic levels for alpha-glucosidase and molecular studies confirmed the diagnosis of PD. The patient presently is still usually asymptomatic, describing muscular pain lasting a few hours or days about once every 2 months following more intensive play or running. Evaluation of muscular strength, pulmonary function, cardiac evaluation, and sleep test seem to be normal over time.

RESULTS

Three successive MRI examinations performed about a year apart in a 3T MRI machine with a whole-body muscular protocol consisting of T1 and STIR weighted sequences showed normal structural and volumetric muscle signal in all muscle groups without signs of fatty atrophy or edema, namely in the head and neck as well as in the axial muscles. Longitudinal semi-quantitative analysis between examinations showed no significant changes in the inter- and intra-muscle comparisons.

CONCLUSIONS

Although muscle MRI is considered to be a valuable diagnostic tool for the diagnosis of PD, a normal MRI may be found in some patients. Also, some reports have found that muscle atrophy in children may not have the same appearance on MRI to adults, lacking a high signal on T1WI. On the other hand, as our patient has successive normal muscular MRI patterns, but is mostly asymptomatic, we hypothesize that abnormal muscle MRI, although characteristic of PD, may correlate an advanced clinical disease.

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