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Poster Abstract: Clinical

A Case of Normal Pressure Hydrocephalus in Adult-Onset Pompe Disease

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OBJECTIVE

To describe a patient affected by glycogenosis type II (GSD II) and normal pressure hydrocephalus.

CASE REPORT

A 65-year-old man, who was diagnosed with Pompe disease when he was 50 years, developed memory loss and urinary urgency. His medical history showed severe left hip arthrosis with osteonecrosis which caused intense pain and functional limitations. The patient was also affected by chronic obstructive pulmonary disease, benign prostatic hyperplasia, and hyperamylasemia.

Examination revealed a lack of strength in the deltoid muscles (bilateral $F=3$), biceps brachii muscles (bilateral $F=4.5$), gluteus maximus muscles (bilateral $F=4.5$), bilateral iliopsoas muscles ($F=4$), and tibialis anterior muscles (bilateral $F=4$), hyperlordosis, right winged scapula, anserine gait, which was only possible with unilateral support, and an inability to completely abduct the arms. No extrapyramidal signs were observed except for postural instability, which might also be compatible with the weakness of pelvic muscles.

The patient underwent psychometric tests which revealed Mini-Mental-State-Examination score equal to 24 (adequate to age and schooling) and short memory loss. Brain MRI showed increased dimension of ventricular system with reduced subarachnoid cavities and periventricular hyperintensity, supporting normal pressure hydrocephalus and, moreover, atrophy of the midbrain, suggesting neurodegenerative disease. The neurosurgeon did not recommend any treatment. Angio-MRI of the cerebral arterial circle did not revealed any vascular anomalies.

The patient started Myozyme® 20 mg/kg every 2 weeks. After 1 year, he was able to walk unaided. Muscular strength was 5 in every muscle examined, no extrapyramidal signs were observed, and his cognitive status was stable. Actually, he is lame because of osteonecrosis of the left hip and has been referred to the orthopedic surgeon.

CONCLUSION

Although GSD II is considered to be a multisystemic disorder in which glycogen accumulation is present in several organs,[1] a case of GDS II together with normal pressure hydrocephalus has, to our knowledge, only been reported in an 8-month child.[2] In addition, the literature reported two cases out of 39 affected by GSD II associated with hydromyelia.[3]

In the above-mentioned case, the normal pressure hydrocephalus is probably a comorbidity rather than a peculiar manifestation of Pompe disease. However, a great benefit was seen in motor symptoms after treatment with Myozyme® and, moreover, no progression of cognitive deficit was observed.

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