GLYCOGENOSIS TYPE II – CASE REPORT

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Abstract

Cytolysis is commonly found in pediatric daily practice. Paper aim is to present the case of 9 years old boy admitted in Clinic II Pediatrics for cytolysis syndrome with unknown etiology.

Key words: glycogenosis type II, Pompe disease, child.

Background

Glycogen storage disease or glycogenosis type II(GSD II) is in fact a lysosomal storage disease (Pompe disease –juvenile form) is an autosomal recessive disorder with an incidence of 1/40 000live births. The disorder is a progressive, multisystemic, debilitating, and often fatal disorder. It was first defined in 1932 by Dutch pathologist Joannes C. Pompe in a seven-month-old girl who died of idiopathic cardiac hypertrophy and was found to have massive glycogen accumulation in many tissues, but predominantly skeletal and cardiac muscles. Infantile –onset Pompe disease is thought to be uniformly lethal without specific therapy. Affected infants present in the first few months of life with hypotonia, a generalized muscle weakness, feeding difficulties, macroglosia, hepatomegaly and a hypertrophic cardiomiopathy followed by death from cardiorespiratory failure or respiratory infection usually by 1 year of age. Juvenile and adult –onset disease (late onset forms) is characterized by a lack or absence of severe cardiac involvement and a less severe short-term prognosis. Symptoms can start at any age and are related to progressive dysfunction of skeletal muscles. The initial symptoms in some patients may be respiratory insufficiency manifested by somnolence, morning headache, orthopnoea and exertional dyspnoea.

Case report

We present a 9 years old boy admitted in our clinic for the evaluation of a cytolysis syndrome associated with somnolence and muscle pain. He is the unique child of a non-consanguineous healthy couple. The child was born at term, normally delivered after an uncomplicated pregnancy. He received natural alimentation for 4 months and after that with a milk formula. Diagnosed at 6 six years old (during a routine check) with cytolysis syndrome with unknown etiology is admitted in our clinic for the evaluation of this syndrome.

Clinical examination revealed a boy with 35 kg weight, 136 cm height, without any abnormal clinical findings, except muscle weakness.

Laboratory findings revealed elevated levels of serum creatine kinase 1263 u/l, aspartate aminotransferase 251 u/l, alanil aminotransferase 266 u/l, lactate dehydrogenase 635 u/l and aldolase 16.9 u/l. A chest X-ray, electrocardiography, echocardiography, electromyography and nervous conduction velocity were in normal parameters. Abdominal ultrasound findings include large gallbladder , and hypoechoic inhomogeneous liver structure. Muscle biopsy excludes an inflammatory miopathy.

After these laboratory findings we thought at Pompe disease. The next step was enzyme assay 39,3 nmol/h/mg (n= 51- 215), performed in Department of Cell Biology and Genetics, Erasmus University, Rotterdam .The confirmatory step for a diagnosis of Pompe disease is enzyme assay demonstrating deficient acid alpha- glucosidase.

For differential diagnosis many conditions were considered as follows:

1. Polymyositis: excluded by the biological investigations and muscle biopsy.
2. Duchenne and Becker muscular dystrophies: elevated levels of serum creatine kinase , aspartate aminotransferase , alanil aminotransferase , lactate dehydrogenase and aldolase were excluded by the pozitive enzyme assay for glycogenosis .
3. Danon disease: elevated levels of serum creatine kinase , aspartate aminotransferase , alanil aminotransferase , lactate dehydrogenase and aldolase but with normal enzyme activity.
4. Mitochondrial myopathy: excluded by the enzyme assay.

Also other muscular glycogenosis were considered but no positive enzyme dosing. Treatment options were limited to supportive or palliative care. For patients with the late-onset form of disease a high protein diet may be beneficial.

Discussion

Pompe disease affects patients of all ages and is always characterized by progressive degeneration of skeletal muscles (proximal and respiratory) and, in infants, cardiac muscle. The rate of progression varies, ranging from a rapidly progressive course that is usually fatal by one year of age, to a more variable but still relentless, progressive course resulting in significant morbidity and often premature mortality. Typically, when the disease is manifested early in infancy, the rate of progression is very rapid, and without treatment the prognosis is poor. Children and adults usually display more gradual and variable rates of disease progression; however, the prognosis often remains

unpredictable and poor. Recommended assessments are musculoskeletal tests (radiography, motor function test) cardiac tests (chest x-ray / MRI, electrocardiography, echocardiography), pulmonary/respiratory tests (spirometry, pulse oxymetry), laboratory cytolisis tests (serum creatine kinase, alanine aminotransferase, aspartate aminotransferase, lactate dehydrogenase).

Muscle weakness is a very common finding in pediatric practice, usually not considered to be a serious symptom.

References:

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