Juvenile Pompe

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Pompe disease (Acid Maltase deficiency) has been traditionally classified into 3 forms: infantile, childhood and adult. But in fact Patients are distributed in a spectrum of signs and symptoms between severe and lethal infantile form to adult type with minor problem in childhood and mild progressive muscular weakness.

Some authorities use “Childhood” or “Juvenile” pompe in view of child’s age at the beginning of the first clinical manifestations. Others utilize this term to specify patients without cardiomyopathy and with exclusive skeletal muscle involvement.

Anyway the various signs, symptoms and severity of this disease represent a continuum, which depending on the residual activity of the enzyme Acid Maltose, determine the ultimate prognosis of the patient.

Young patients with Juvenile pompe may present with a mild to moderate delay in motor development .This retard in achieving normal motor milestones is usually interpreted by parents as laziness or lack of pleasure to move. Another common neglected point in the past history is lack of physical endurance and avoidance to walk or run long distances. With progression of the disease Trendelenburg gait and lumbar hyperlordosis appears. Rising from sitting position and climbing stairs becomes difficult and children hold their thighs with their arms to achieve this movement (Gower’s sign). These signs and symptoms remind us Duchene Muscular Dystrophy. Truncal muscle weakness usually leads to scoliosis. Some patients develop “rigid spine” and this clinical presentation is very similar to congenital muscular dystrophies with a rigid spine .In some cases, involvement of respiratory muscles,particularly diaphragm, result in nocturnal hypoventilation causing typical symptoms such as fatigue, night sweat and lack of appetite. These respiratory symptoms can precede the skeletal muscle weakness.

Regarding potential medical treatment of pompe disease and overlapping clinical presentation with other muscular dystrophies, we must always think of this disease when we are confronted with a myopathy in a child or young adult.

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