

Lysosomal Myopathies

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Lysosomal myopathies are hereditary myopathies characterized morphologically by the presence of autophagic vacuoles. Autophagy is an intracellular bulk degradation process, which is used by all cells to eliminate waste materials. Autophagy is considered to be essential for myocytes and the lysosomal system becomes prominent in certain muscle diseases.

In muscle pathology, lysosomal abnormalities are seen in three types of vacuoles: rimmed vacuoles which are most likely a secondarily induced lysosomal abnormality, autophagic vacuoles, which are usually large and contain glycogen, seen specifically in acid maltase deficiency and autophagic vacuoles with unique sarcolemmal features with acetylcholinesterase activity (AVSF), which are seen in Danon disease and other related myopathies.

There are a number of hereditary muscle diseases that are characterized pathologically by the presence of rimmed vacuoles. All rimmed vacuolar myopathies are plausibly secondary lysosomal myopathies.

The most common one in Iran is hereditary inclusion body myopathy known as Nonaka myopathy or distal myopathy with rimmed vacuoles. Patients are characterized by distal myopathy with onset in early adulthood typically in the anterior compartment causing foot drop.

Acid maltase deficiency also known as Pompe disease or glycogenosis type II is an autosomal recessively inherited lysosomal storage disorder. Muscle biopsy reveals a vacuolar myopathy with glycogen accumulation and increased acid phosphatase activity. These features are non-specific and the diagnosis must be confirmed by enzyme assay either in muscle, fibroblasts or leukocytes. Acid maltase is a lysosomal enzyme encoded by a gene (GAA) on chromosome 17.

AVSF delineate five autophagic vacuolar myopathies. AVSF delineate five autophagic vacuolar myopathies. Danon disease, X-linked myopathy with excessive autophagy (XMEA), infantile autophagic vacuolar myopathy, adult-onset autophagic vacuolar myopathy with multiorgan involvement and X-linked congenital autophagic vacuolar myopathy.

Danon disease, an X-linked vacuolar cardiomyopathy and skeletal myopathy, was originally described as "lysosomal glycogen storage disease with normal acid maltase deficiency but had normal enzymatic activity. The primary defect resides in lysosome-associated membrane protein- 2 (LAMP-2), a lysosomal structural protein rather than a glycolytic enzyme. Danon disease is characterized clinically by the triad of hypertrophic cardiomyopathy, muscle weakness and mental retardation

Keywords: Lysosomal myopathy; Myopathy; Pompe; Acid maltase deficiency, Danon

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