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Rare Case of Adult Onset Pompe's Disease

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RARE CASE OF ADULT ONSET POMPE'S DISEASE IN A PATIENT WITH TYPE 1 DIABETES MELLITUS

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Introduction

- Pompe's disease is a lysosomal storage disease characterized by accumulation of glycogen primarily in muscle tissue.
- Infantile and late onset are the two common forms of this disease.
- We present a case of adult onset disease in a patient with type 1 diabetes mellitus (DM) with involvement of muscle, liver and bone.

Case History

- A 28-year-old male was being followed up for type 1 diabetes mellitus. He had a long standing history of mild muscle weakness.
- Family history : Unknown muscle disorder in father and cardiomyopathy in grandfather.
- Previous work up showed elevated CPK levels of 1340 (reference range < 130IU/L) along with elevated transaminases.
- Muscle biopsy - Autophagic activity producing rimmed vacuole like structures in many of the muscle fibers, with increased staining of cytoplasm with Periodic acid Schiff.
- He was lost to follow up and about 10 years later decided to undergo further evaluation , which showed persistently elevated CPK levels.
- EMG studies which showed generalized myopathic process.
- Serum acid maltase level was found to be low.
- On Glucosidase aide alpha gene sequencing, he was found to have c.-32-13T>G and c.1655T>C (p. L552P) mutation.

Laboratory Data

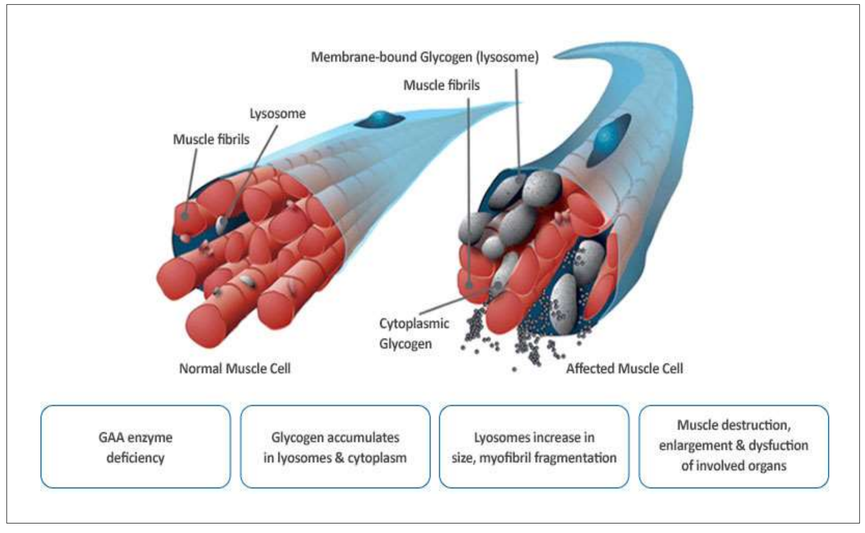
Laboratory data before diagnosis

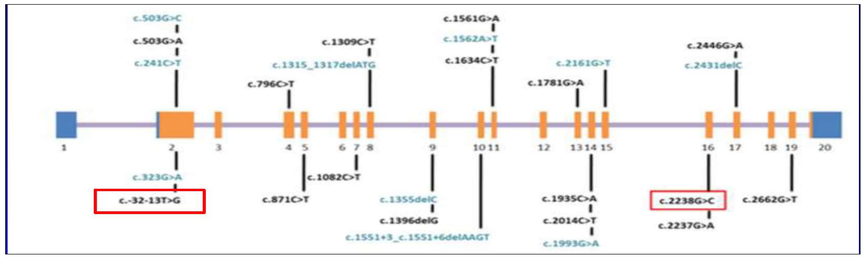
DATE	AST (Ref 15-35 IU/L)	ALT (Ref 30-65 IU/L)	CPK (Ref < 130 IU/L)
July 2017	138	178	1860
March 2009	224	210	1340
March 2008	249	184	1343
June 2007	216	191	
November 2006	164	175	

Laboratory data after initiation of enzyme replacement

DATE	AST (Ref 15-35 IU/L)	ALT (Ref 30-65 IU/L)	CPK (Ref < 130 IU/L)
July 2018	90	139	1125

Pathophysiology of Pompe's disease





Common mutation in adult onset Pompe's disease

Learning Points

- Late-onset Pompe's disease is a rare, progressive, autosomal recessive disorder; disease progression and symptomatology are variable between individuals, which delays the diagnosis.
- Use of ERT has substantially altered outcomes for the patients, underscoring the importance of early diagnosis with high index of suspicion.
- Additional impact of type 1 DM is of concern in our patient.
- Since glycogen hepatopathy is associated with uncontrolled Type 1 DM, tight control of diabetes may help to reduce the impact of acid alpha glucosidase deficiency.
- Improvement of bone density after ERT suggests need for further studies to elucidate the pathophysiology of bone involvement in this disorder.

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