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AN ATYPICAL PRESENTATION OF POMPE DISEASE

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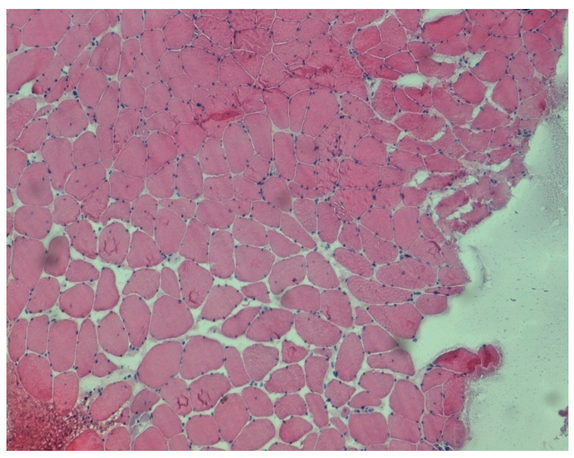
Here we present the case of a 72-year-old man with an atypical presentation of Pompe disease, at the beginning charactherized by axonal neuropathy and personal history of polycystic kidney disease. Subsequently the patient has developed progressive weakness in the lower limbs and difficulty walking.
Creatine kinase was mild elevated. EMG revealed a myopathic and neuropathic pattern.

MUSCLE BIOPSY

Family history reveals a sister with polycystic kidney disease and unspecific proximal myophaty

Muscle biopsy showed rare fibres with cytoplasmic vacuols mostly skewed oriented, sometimes with rimmed like appearance. Scattered atrophyc or degenerated fibres, acid phosphatase staining showed intacellular positive spots in a discrete number of cells.



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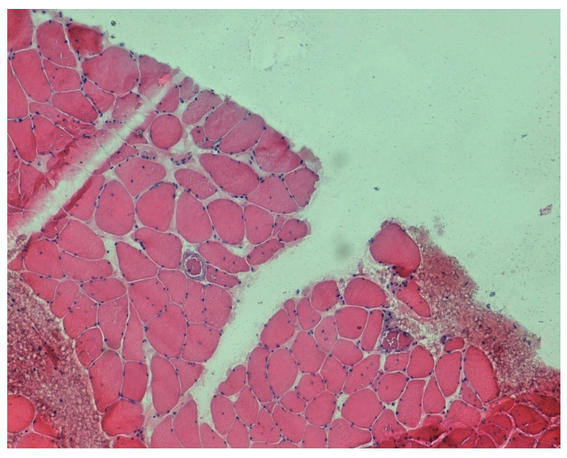

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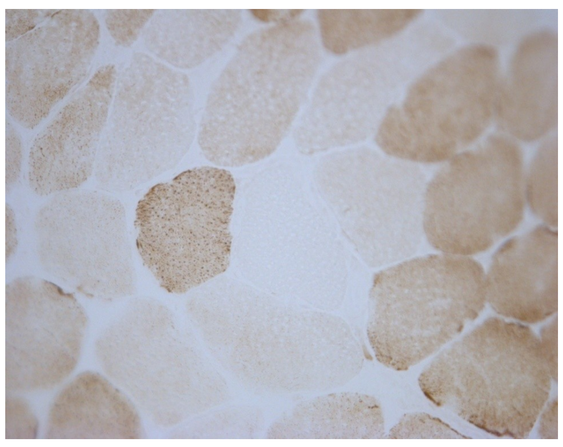

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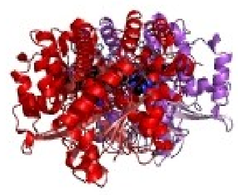

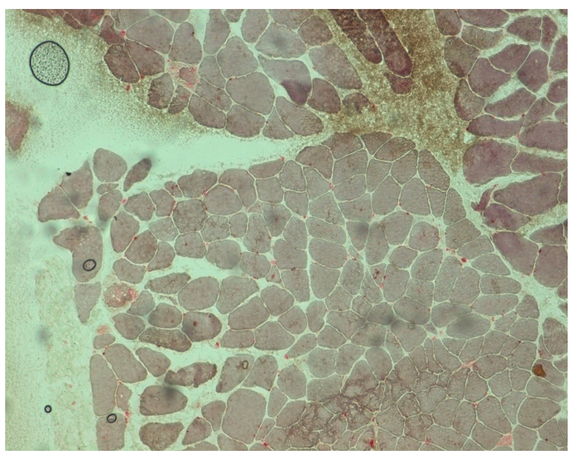

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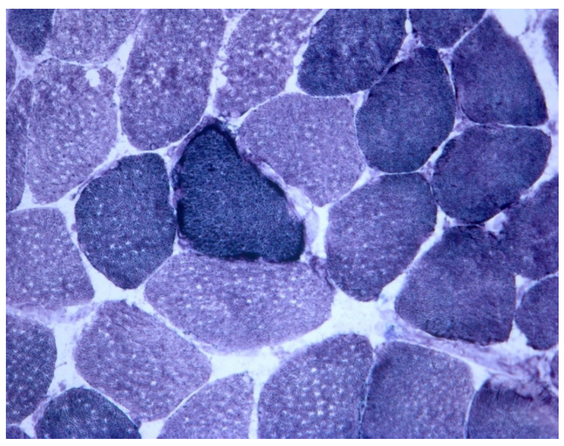
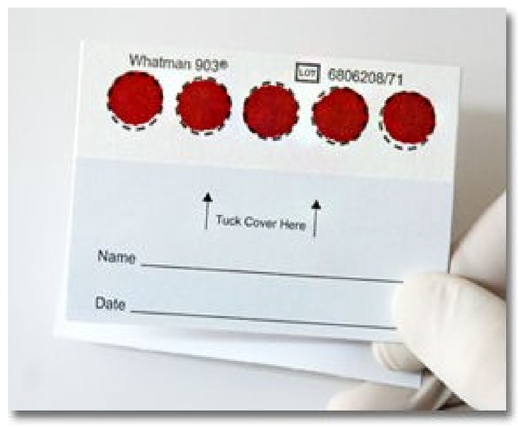


SDH



DBS showed absent activity of acid alpha-glucosidase enzyme

DISCUSSION: Pompe disease is an autosomal recessive metabolic disorder which causes an accumulation of glycogen in the lysosomes due to deficiency of the lysosomal acid alpha-glucosidase enzyme. The clinical spectrum ranges from fatal hypertrophic cardiomyopathy and skeletal muscle myopathy in infants to relatively attenuated forms in adults, which manifest as a progressive myopathy without cardiac involvement. Our case represents an atypical presentation of Pompe disease with initially mild myophatic signs and slow progression of muscle weakness in a patient with important comorbidity.