# Myopathological features of McLeod Syndrome: a case report

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## Introduction

*McLeod syndrome (MLS) is a neurodegenerative disorder caused by mutations of the XK gene, encoding the XK protein, which carries the KX erythrocyte antigen. Its clinical features may include involuntary movements, psychiatric symptoms, cognitive decline, epilepsy and signs of peripheral nervous system involvement; also characteristic is the presence of thorny red blood cells, known as acanthocytes. We present a case of McLeod syndrome characterized by a myopathological picture of primitive myopathy and neurogenic atrophy.*

## Presentation and history

*A 63-year-old man came to our observation for the onset, four years prior to our observation, of choreic movements of the trunk and limbs. His clinical history was notable for generalized seizures since the age of 39 years that had been treated with phenobarbital, and for repeated findings of markedly increased serum CK levels (> 1000 U/L). His familial history showed similar signs and symptoms in his brother, who had died at the age of 59, and in a cousin (X-linked pattern). The patient was admitted to our Department for a full clinical and instrumental assessment.*

## Neurological examination

*Choreic gait, proximal muscle hypotrophy, diffuse hypotonia and areflexia; choreic movements of the face, trunk and limbs.*

## Electrophysiological testing

***ENMG**: Sensory-motor axonal polineuropathy*
***EEG:** diffuse epileptic abnormalities*

## Blood test

- **CPK 3136 U/l**
- *Blood group : **B Rh+ kk***
*(no espression of the Kell antigen)*
- *Peripheral blood smear:*
***acanthocytes** 20-25% (Fig.1)*
- *VPS13A gene: no mutations*

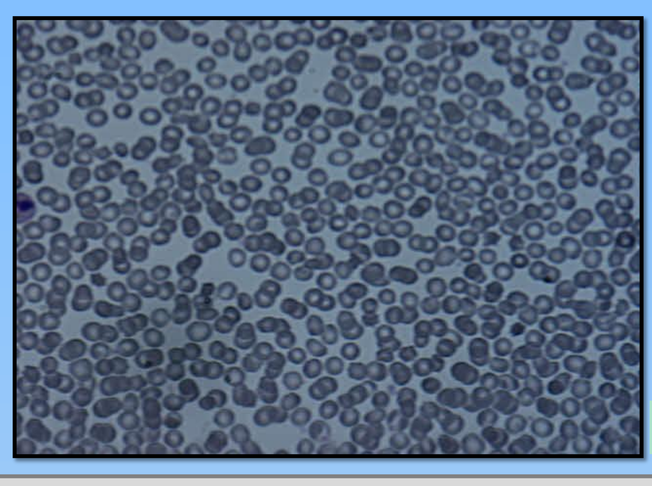

***Fig 1.*** **Peripheral Blood Smear**

## Muscle biopsy

*Muscle biopsy showed marked fiber size variability, with signs of necrosis and fibrosis, glycogen and lipid deposit at the intermyofibrillary and subsarcolemmal levels; type-grouping was evident.*

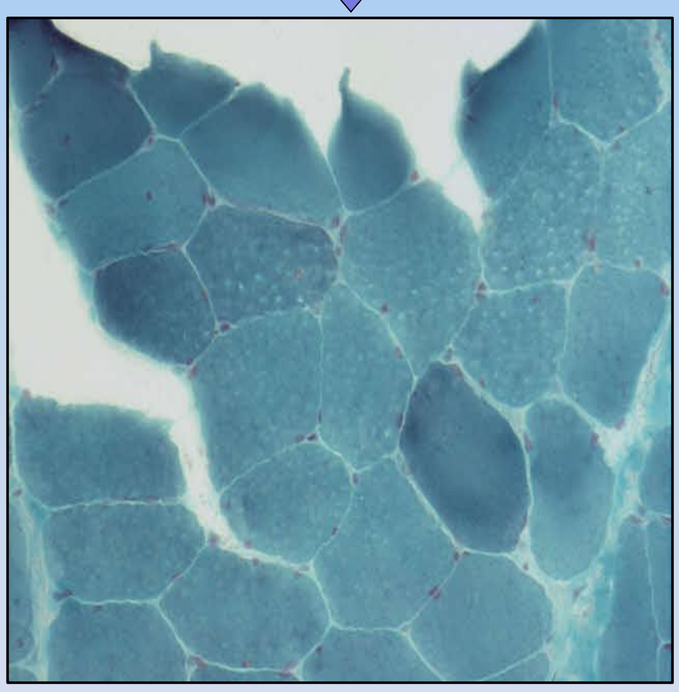

***Fig 2.*** **Muscle biopsy, Gomori (20X)**

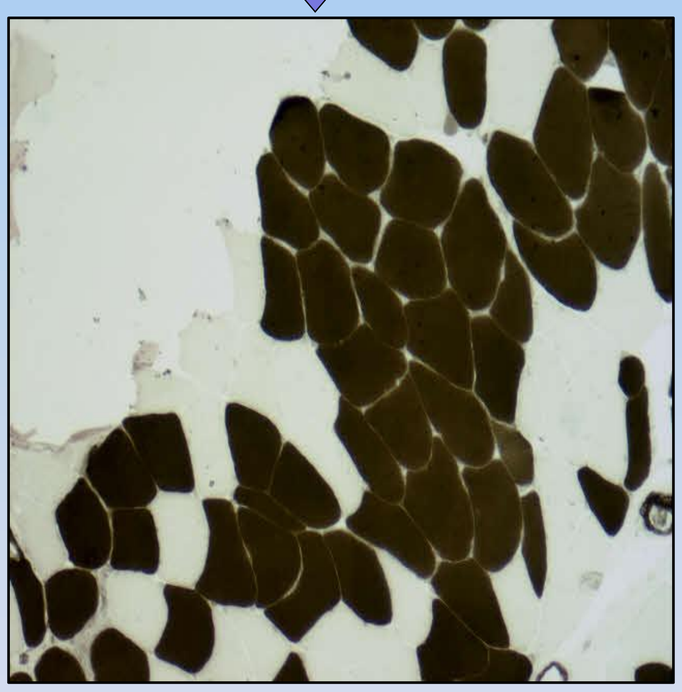

***Fig 3.*** **Muscle biopsy, ATP pH 4.3 (10X)**

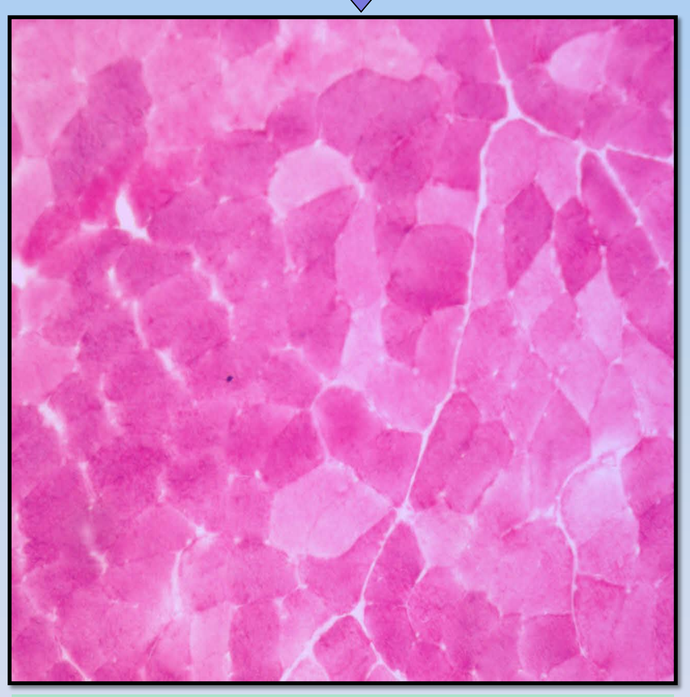

***Fig 4.*** **Muscle biopsy, PAS (10X)**

## Discussion & Conclusions

***Our case reveals a stringent clinical-myopathological correlation, as the primary and neurogenic muscle damage accounts to proximal weakness, CK increase and areflexia. While the clinical, laboratory and instrumental aspects strongly support a diagnosis of McLeod syndrome, genetic analyis (ongoing in our case) is needed in order to confirm the diagnosis.***

## Bibliography

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