Huntington's disease–like syndrome: a case report

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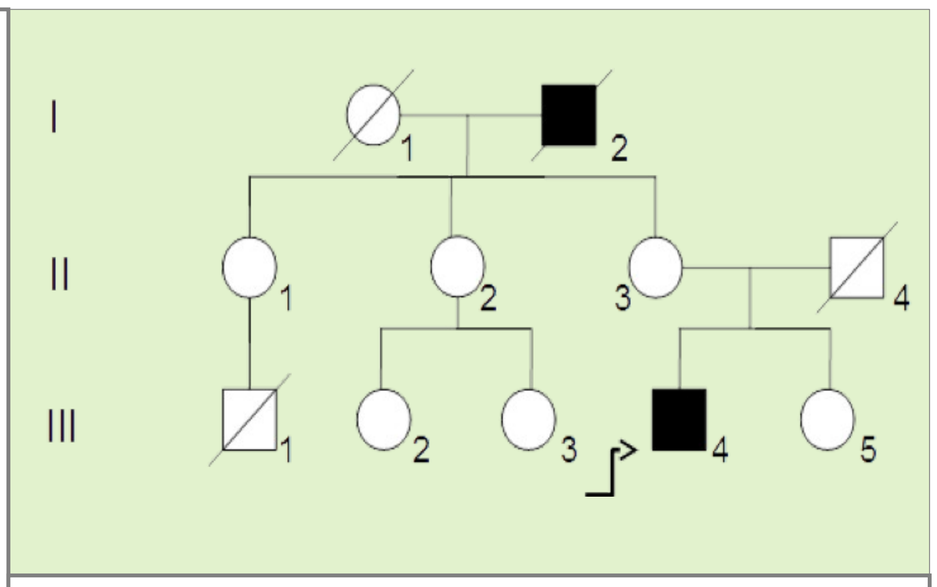
History and clinical examination

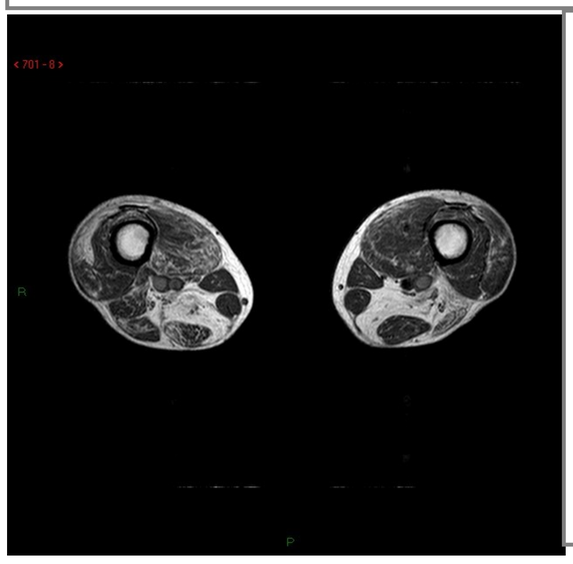
Anxiety and compulsive behavior with motor and vocal tics since childhood. At age 45 choreic movements of the limbs and trunk, and onset of apathy and depression. At age 48 postural instability and frequent falls. At age 52 orolingual dystonia, progressive impairment of speech and eating, increased irritability and attention deficit, compulsive scratching behavior with skin lesions.
Limb hypotonia and muscle wasting with reduced deep tendon reflexes, normal strength and mild lower limb hypopallesthesia.



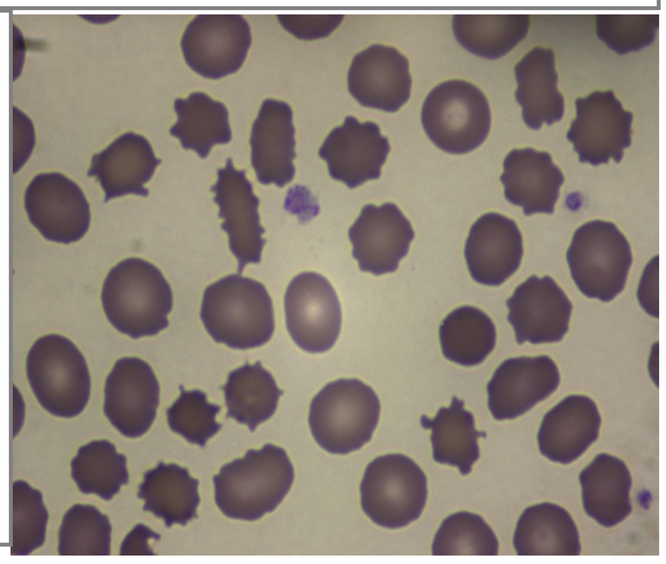
Family history of chorea and depression in the maternal grandfather.

Diagnostic work up

-Blood tests: normocytic anemia, increase of liver enzymes, CK elevation.
-EKG, echocardiography: normal.
-Brain MRI: caudate atrophy, no iron accumulation.
-EMG and nerve conduction study: sensory-motor axonal polyneuropathy of lower limbs.



- Muscle MRI (lower limbs): multi-segmental, asymmetrical fatty substitution.
- Peripheral blood smear: acanthocytes.
- RBC phenotype: negative Kpa/Kpb, reduced Kell antigen, consistent with "McLeod blood group phenotype".
- Genetic analysis of XK gene: positive.



Comment McLeod Syndrome (MLS) is an extremely rare, X-linked recessive neurologic disorder, caused by the absence of XK gene protein product. It has a wide phenotypic variability, and the symptoms include compensated anemia with acanthocytosis, sensory-motor polyneuropathy with primitive and secondary muscle atrophy, chorea, parkinsonism, epilepsy, and cardiomyopathy. We report the first Italian case of MLS caused by a small deletion in exon 3 of the XK gene. This mutation leads to a shift in the reading frame with the creation of a stop codon at position 301, causing translation of a truncated protein product. It has only been described in five other patients, one coming from USA with Anglo-Saxon descent, two Chilean brothers of German descent and two Japanese brothers, all presenting with different clinical features, with striking phenotypic variability even between members of the same family.

References Danek, A. et al. *McLeod neuroacanthocytosis: genotype and phenotype.* Ann. Neurol. 50, 755–764 (2001). Miranda, M. et al. *Phenotypic variability of a distinct deletion in McLeod syndrome.* Mov. Disord. 22, 1358–1361 (2007). Wada, M. *An unusual phenotype of McLeod syndrome with late onset axonal neuropathy.* J. Neurol. Neurosurg. Psychiatry 74, 1697–1698 (2003).

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