McLeod neuroacanthocytosis syndrome

Summary / Zusammenfassung

McLeod syndrome (MIM 314850) is an X-linked multi-system disorder with hematological, neuromuscular, and CNS involvement. It is characterized by absent expression of the Kx RBC antigen, weak expression of Kell RBC antigens, and acanthocytosis.

Neuromuscular manifestations include myopathy, sensory-motor axonal neuropathy, and cardiomyopathy. CNS manifestations resemble Huntington’s disease, and comprise a choreatic movement disorder, neuropsychiatric abnormalities, and generalized epileptic seizures. McLeod syndrome is caused by mutations of the XK gene encoding the XK protein, a putative membrane transport protein of yet unknown function that contains the Kx erythrocyte antigen. Available data suggest an important role of the XK/Kell-complex in apoptosis regulation.

We perform several studies (magnetic resonance imaging, volumetry, and spectroscopy, brain pathology, muscle histology, hematology) to study the clinical and pathological characteristics, and to elucidate the pathophysiology of the disorder in the setting of an international network of neuroacanthocytosis specialists.

Publications / Publikationen


Keywords / Suchbegriffe
McLeod, neuroacanthocytosis, acanthocyte, CNS, PNS, chorea syndrome

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Funding Source(s) / Unterstützt durch
Mutaxia Foundation

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Duration of Project / Projektdauer
Jan 2003 to Dec 2015