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A Family of rare Genetic Disorders

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McLeod neuroacanthocytosis syndrome is a ultra rare genetic disorder that occurs almost exclusively in males.It affects brain and spinal cord. Affected individual develops jerky movements of arms, legs, dystonia of face ,tongue which ultimately leads to swallowing difficulty. Mc leod neuroacanthocytosis is caused by mutation in XK gene. People with this syndrome have reduced muscle mass , muscle strength , suffers from peripheral neuropathy along with dilated cardiomyopathy. The troublesome features of dystonia , inability to take care of oneself, anxiety , depression begins in mid adulthood It is a very rare genetic disorder with 150 cases reported worldwide. It is a X- linked recessive disorder. It is predominantly present in males with one X chromosome needs to be affected for males to manifest the condition. In females both X chromosomes should be affected in order to manifest the condition1It is of three types -Autosomal recessive chorea acanthocytosis, X linked McLeod syndrome, neurodegeneration with brain iron accumulation. Mean age of onset is 30 -40 years. The central nervous system abnormalities include choreiform movements, facial grimacing, generalized seizures , anxiety ,depression. It is diagnosed by clinical features , peripheral blood smear showing acanthocytes2.MRI brain will show atrophy of caudate nucleus, putamen. Genetic study is gold standard in classifying and diagnosing this condition. There is no treatment for his condition. It is managed on lines of conservative management.

Biography

Bhardwaj CM* completed his study in Resident internal medicine at SGT medical college hospital Gurgaon (122505) ,Haryana India