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ACANTHOCYTOSIS AND HYPERCKEMIA

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A fourteen-year-old boy was referred to neuromuscular clinic for the investigation of hyperckemia (serum creatine kinase: 4000 IU/L; n: 0-170)detected during laboratory examinations. He was thefirst child of consanguineous parents. His psychomotor development was normal and he had no symptoms of a neuromuscular disease in the past. Detailed history taking did not reveal any signs of involuntary movements, bradykinesia and social problems. Neurologic examination showed normal muscle power in upper and lower extremities, and no signs of muscle atrophy. Deep tendon reflexes could not be elicited. Nerve conduction studies were normal but electromyography revealed combined neurogenic and myogenic potentials in lower extremity muscles. A Muscle biopsy did not show any pathology and was interpreted normal. Targeted customized Mendeliome panel[1] next generation sequencing revealed a homozygous splice site mutation in the vacuolar protein sorting-associated protein (*VPS13A)*, NM_015186.3, c.6095+1G>C.

Hyperckemia is a condition characterized by elevated levels of enzyme called creatine kinase in blood. Chorea-acanthocytosis is an autosomal recessive caused by mutations in the VPS13A gene, which encodes the protein “chorein”. The disease is characterized by chorea, dystonias mainly involving the face, parkinsonism, vocal tics, epilepsy, social disinhibition and distal muscle wasting. The mean age of onset is 35 years.[2] Neuropsychiatric symptoms are also common and may precede movement disorders. Acanthocytes usually constitute 5 to 50 percent of circulating red blood cell circulation. They may be absent or may appear late in the course of the disease.[3] Most patients have elevated serum creatine kinase levels but the cause of creatine kinase elevation is unknown. Nerve conduction studies may be normal, but may show sensory axonal neuropathy in some cases. Electromyography may show myogenic or neurogenic potentials. Retrospectively, after the genetic diagnosis, we could confirm the presence of acanthocytes (Fig. 1). Our patient had no neurological complaints and no neurological

abnormalities. Thus, peripheral blood smear may give important diagnostic clues in cases of idiopathic hyperckemia. Whole exome sequencing is also a preferable diagnostic modality in cases of idiopathic hyperckemia but has challenges in counselling of the family in a clinically asymptomatic case in the context of a progressive neurologic disorder.

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Figure Legends

Figure 1: Peripheral blood smear of the patient showing acanthocytosis