genetica

Humangenetisches Labor & genetische Beratungsstelle

Hyperkaliämische period. Paralyse, HyperPP1 (SCN4A)

Genetik und Klinik

(Deutsch noch nicht verfügbar)
Hyperkalemic periodic paralysis type 1 (hyperPP1) is characterized by attacks of flaccid limb weakness (which may also include weakness of the muscles of the eyes, throat, and trunk), hyperkalemia (serum potassium concentration >5 mmol/L) or an increase of serum potassium concentration of at least 1.5 mmol/L during an attack of weakness and/or provoking/worsening of an attack by oral potassium intake, normal serum potassium and muscle strength between attacks, onset before age 20 years, and absence of paramyotonia (muscle stiffness aggravated by cold and exercise). The attacks of flaccid muscle weakness usually begin in the first decade of life. Initially infrequent, the attacks then increase in frequency and severity over time until approximately age 50 years, after which the frequency of attacks declines considerably. Potassium-rich food or rest after exercise may precipitate an attack. A cold environment and emotional stress provoke or worsen the attacks. A spontaneous attack commonly starts in the morning before breakfast, lasts for 15 minutes to one hour, and then disappears. Cardiac arrhythmia or respiratory insufficiency usually does not occur during attacks. Between attacks, hyperPP1 is usually associated with mild myotonia (muscle stiffness) that does not impede voluntary movements. Many older affected individuals develop a chronic progressive myopathy.

HyperPP1 is inherited in an autosomal dominant manner. Most individuals with hyperPP1 have an affected parent; the proportion of cases caused by de novo mutations is unknown. Each child of an individual with hyperPP1 has a 50% chance of inheriting the mutation. Prenatal diagnosis for pregnancies at increased risk is possible if the disease-causing mutation in the family has been identified; however, requests for prenatal testing for conditions such as hyperPP1 that do not affect intellect and have some treatment available are not common.

Dienstleistung

Auftrag: Gezielte Hotspot-Mutationsanalyse im SCN4A-Gen

Fachbereich: Neurologie

Methode: PCR und Sequenzierung der Exone 13 und 24 des SCN4A-Gens

Gen(e): SCN4A

Untersuchungsmaterial

Probe: Venöses Blut

Probengefäss: EDTA- oder Heparin-Röhrchen

Menge: 1-5 ml

Praktische informationen

Zustellung: A-Post

Dauer: 2-3 Wochen

Preis (TP): Bei medizinischer Indikation gemäss Tarif Analysenliste

Bemerkung: -