Geneticheck - Genetic Report

Gene

CLCN1

Associated Diseases

Myotonia Congenita, Autosomal Recessive
Myotonia Congenita, Autosomal Dominant
Thomsen And Becker Disease

Phenotype

Skeletal muscle hypertrophy
Hypertrophy (increase in size) of muscle cells (as opposed to hyperplasia, which refers to an increase in the number of muscle cells).

Muscle weakness
Reduced strength of muscles.

Autosomal dominant inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in heterozygotes. In the context of medical genetics, an autosomal dominant disorder is caused when a single copy of the mutant allele is present. Males and females are affected equally, and can both transmit the disorder with a risk of 50% for each child of inheriting the mutant allele.

Muscle stiffness
A condition in which muscles cannot be moved quickly without accompanying pain or spasm.

Myalgia
Pain in muscle.

Dysphagia
Difficulty in swallowing.

EMG abnormality
Abnormal results of investigations using electromyography (EMG).

Lid lag on downgaze
Delayed descent of the upper eyelid on downgaze. Also described by some authors as von Graefe sign.

EMG: myotonic runs
Spontaneous, repetitive electrical activity demonstrated by electromyography (EMG).

Muscle hypertrophy of the lower extremities
Muscle hypertrophy primarily affecting the legs.

Handgrip myotonia
Difficulty releasing one's grip associated with prolonged first handgrip relaxation times.

Percussion myotonia
A localized myotonic contraction in a muscle in reaction to percussion (tapping with the examiner's finger, a rubber percussion hammer, or a similar object).

Autosomal recessive inheritance
A mode of inheritance that is observed for traits related to a gene encoded on one of the autosomes (i.e., the human chromosomes 1-22) in which a trait manifests in individuals with two pathogenic alleles, either homozygotes (two copies of the same mutant allele) or compound heterozygotes (whereby each copy of a gene has a distinct mutant allele).

Myotonia with warm-up phenomenon
Myotonia that occurs after a period of rest and decreases with continuing exercise.

Myotonia
An involuntary and painless delay in the relaxation of skeletal muscle following contraction or electrical stimulation.