Thomsen syndrome

Thomsen syndrome, syn. *Myotonia congenita, Ataxia muscularis*	
Clinical picture	Difficulty sucking and stiff facial expressions
Cause	autosomal dominantly inherited myotonia syndrome
Diagnostics	clinical, laboratory, family history, genetic examination
Prognosis	Good, the disease can affect chewing, swallowing or walking
Classification and references	
ICD-10	G71.1
MeSH ID	D009224
OMIM	160800 255700
orphaned	ORPHA206973
MedlinePlus	001424

Thomsen's syndrome (also *myotonia congenita* or *ataxia muscularis*) is an autosomal dominantly inherited syndrome of **myotonia**, manifested already after birth by **difficult sucking** and **stiff facial expressions**. After muscle contraction, there is no relaxation, the musculature is significantly hypertrophic (due to repeated muscle contractions) with increased irritability. The sick are not capable of faster movements. On the whole, however, the disease **does not cause** significant problems for them.

A rarer, autosomal recessively inherited form of congenital myopathy is called **Becker disease** (OMIM: 255700 ; *it is not a form of Becker muscular dystrophy, but a different clinical entity*) and is caused by a mutation in the *CLCN1* gene.

Links

External links

https://www.akutne.cz/algorithm/cs/321--/

Related Articles

- Statin myopathy
- Myopathy
- Myasthenia gravis
- Myopathic syndrome
- Myotonic syndrome

References

AMBLER, Zdeněk. *Základy neurologie : [učebnice pro lékařské fakulty].* 7. edition. Galén, c2011. ISBN 9788072627073.

- VOKURKA, Martin – HUGO, Jan. *Velký lékařský slovník.* 9. edition. Maxdorf, 2009. 1159 pp. ISBN 978-80-7345-202-5.