Becker muscular dystrophy

Becker Muscular Dystrophy (*BMD*) is a genetic disease of the muscles of the human body. This is an X-linked gonosomal recessive inheritance , which is why they are mainly male (99.9%). Women are often carriers , ie. the mutated gene is contained in their DNA, but its recessiveness causes it not to show. Becker muscular dystrophy is generally a milder form of Duchenne Muscular Dystrophy (DMD). The incidence of BMD in the population is about 1: 18,000 .

Genetics

Muscular dystrophy is caused by the deletion of a certain part of a chromosome. The distribution of deletions is typical for the spectral repeat region (up to 46%). Becker muscular dystrophy accounts for about 15% of locus mutations .

The fundamental genetic difference between the allelic phenotypes of DMD and BMD is that DMD is genetically lethal , while in BMD, male reproductive capacity is maintained at a high percentage. Therefore, the frequency of inherited mutations is very high and newly created mutations are minimal. Both forms of dystrophy differ in onset and course, not in severity.

A genetic test to detect this disease is expensive because the mutation occurs on one of the largest human genes.

Causes

At the molecular level, the causes are different. One of the most common causes of dystrophy is a low concentration or abnormal form of plectin protein in muscle tissue, which is caused by a mutation in the appropriate X chromosome gene . In the muscles, the desmino-plectin system does not bind properly .

Symptoms

Becker muscular dystrophy can be detected at the molecular level as early as the prenatal period.

In childhood or early *puberty* (around 11 years), symptoms begin to appear:

- general physical weakness (the disabled person is not physically fit, has below-average results in sports,...),
- muscle cramps.

During *adolescence* , the symptoms escalate. Appear:

- difficulty walking fast , walking up stairs, running,
- difficulties in lifting heavy loads,
- loss of muscle mass (shoulder, upper and lower limbs),
- weakening of mimic and masticatory muscles → problem with food intake,
- weakening of the sphincters → spontaneous leakage of urine or stool (incontinence),
- inability to walk from 40. – 50. year,
- respiratory problems, heart problems and other symptoms associated with smooth muscle weakness .

Treatment

The treatment of the cause is currently unknown.

The course of the disease can be slowed down in the following ways:

- regular rehabilitation (impact exercise does not make sense),
- muscle massages,
- stretching splints (stretching and exercising a muscle in sleep).

Links

https://www.wikiskripta.eu/index.php?curid=69228

Related Articles

- Duchenne muscular dystrophy
- Daltonism
- Hemophilia

External links

- KAČÍRKOVÁ, Jitka. *Becker Muscular Dystrophy (BMD)* [online]. [feeling. 2016-10-23]. < http://www.amd-mda.cz/nervosvalova-onemocneni/diagnozy-typy-nervosvalovych-onemocneni/beckerova-svalova-dystrofie-bmd >.

References

- NUSSBAUM, Robert L, Roderick R MCINNES and Huntington F WILLARD, et al. *Clinical Genetics: Thompson & Thompson.* 6th edition. Prague: Triton, 2004. 492 pp. ISBN 80-7254-475-6 .
- LÜLLMANN-RAUCH, Renate. *Histology.* 1st edition. Prague: Grada, 2012. 576 pp. ISBN 978-80-247-3729-4 .

References

1. ↑ KAČÍRKOVÁ, Jitka. *Becker Muscular Dystrophy (BMD)* [online]. Association of Muscular Dystrophics in the Czech Republic, © 2016. [feeling. 2017-01-13]. < http://www.amd-mda.cz/nervosvalova-onemocneni/diagnozy-typy-nervosvalovych-onemocneni/beckerova-svalova-dystrofie-bmd >.
2. ↑ NUSSBAUM, Robert L, Roderick R MCINNES and Huntington F WILLARD, et al. *Clinical Genetics: Thompson & Thompson.* 6th edition. Prague: Triton, 2004. 492 pp. ISBN 80-7254-475-6 .
3. ↑ NUSSBAUM, Robert L, Roderick R MCINNES and Huntington F WILLARD, et al. *Clinical Genetics: Thompson & Thompson.* 6th edition. Prague: Triton, 2004. 492 pp. ISBN 80-7254-475-6 .

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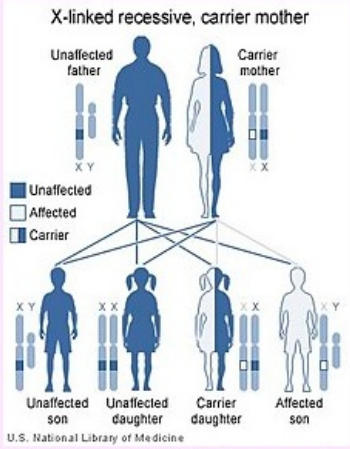

X-linked recessive inheritance, carrier mother

Cause	**plectin** gene mutations
Examination in the Czech Republic	list of workplaces
Incidence in the world	1:18 000 in the population

Classifications and references

ICD-10	G71.0
MeSH ID	D020388
OMIM	300376
orphanet	ORPHA98895
MedlinePlus	000706
Medscape	313417