

Duchenn's muscular atrophy

Duchenne muscular dystrophy is a congenital inherited disease characterized by loss of active muscle mass. It is an X-linked recessive disease. The first symptoms appear in the first few years of life of affected boys. It occurs at a frequency of 1:3 000 births.

Genetics

DMD is caused by mutation genes responsible for the production of a specific structural protein of the sarcolemma called **dystrophin**. Muscular dystrophies with its abnormality are called **dystrophinopathies**.

The gene for DMD is located on the short arm of the chromosome X, was mapped in the 1980s, the whole process of mapping was simpler than for e.g. cystic fibrosis precisely because it was known to be a sex-linked disease. In Duchenne muscular dystrophy, there is a complete cessation of dystrophin formation. In families where DMD is known to occur, prenatal diagnosis is possible, thus, if prevention is followed, the next affected individual in the family may not be born at all.

A milder form of the disease is **Becker muscular dystrophy** (1:18,000), where dystrophin is produced in small amounts and is damaged, but the onset of the disease is later and the overall progression slower (it is a different type of mutation in the dystrophin gene).

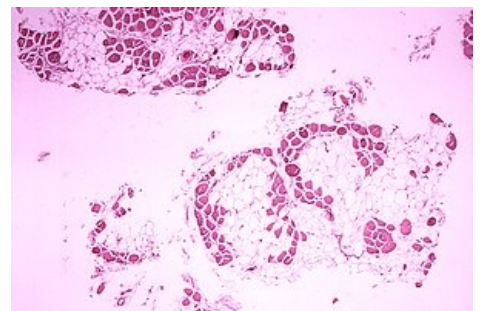
Disease progression

- The boy is born without any symptoms and his development is completely normal for the first few months.
- Around the age of 2-6 years, the first symptoms appear, dystrophin deficiency begins to manifest and as a result, dying muscle fibres begin to be replaced by connective tissue.
- Gradually, the affected boys begin to have problems with walking, running, getting up from lying or sitting (the pelvic girdle muscles are the first to show damage), and the typical myopathic syndrome of the lower limbs gradually develops. Compensatory pseudohypertrophy occurs, which is visible on the compensatorily enlarged calves of boys.
- Subsequently (around the age of 12), wheelchair confinement occurs. The brachial plexus muscles of the upper limbs are also affected and kyphoscoliosis develops.
- In addition to the skeletal muscles, the heart muscle is also affected, which can result in dilated cardiomyopathy with subsequent heart failure.
- Other accompanying symptoms include restricted breathing (due to respiratory muscle involvement and may progress to the point where artificial pulmonary ventilation is required).
- Death occurs around the age of 30, and may be caused by severe recurrent pneumonia or heart failure.

Diagnostics

The first suspicion is usually made by a paediatrician.

- To exclude this, serum creatine kinase (CK) levels may be tested, which is one of the indicators of muscle disease. When muscle cells are damaged, it is CK that enters the serum. Boys with DMD have extremely high serum CK levels at birth, when there are no signs of the disease. Serum aminotransferases (ALT, AST) are often elevated.
- For confirmation, a sample of biopsy muscle is taken and according to the degenerated cells + increased fat and connective tissue content, the disease can be diagnosed.
- Other investigative methods used in the diagnosis and monitoring of the disease progression are EMG, MRI or invasive monitoring of dystrophin content directly in the muscle.



Duchenne-muscular-dystrophy

Treatment

For DMD today, there is only compensatory treatment in the form of physiotherapy or antibiotics given against frequent pneumonias. There is no causal treatment.

Living with DMD

- **Education and upbringing** - Boys with DMD have problems with learning, memory, maintaining attention, and with their emotions; some experts believe this is a result of dystrophin deficiency.
- **Mobility** - compensatory aids such as crutches, wheelchairs or pushchairs are needed to increase mobility.
- **Diet** - is an integral part of compensating for the disease, as excess weight would lead to a rapid worsening of symptoms.
- **Speech** - many boys with DMD have significant speech or communication impairments but do not lose visual perception or manual skills - many DMD patients have made a living in the arts.

Links

Related articles

- Myopathy
- Thomsen syndrome
- Myotonia
- Myastenia gravis
- Myopathic syndrome
- Myotonic syndrome

External links

- wikipedia:en:Duchenne muscular dystrophy
- Muscular dystrophy Duchenne/Becker (<https://www.parentpripation/description.htm>)

Used literature

- MAREŠ, Jaroslav – KOČÁREK, Eduard – SEDLÁČEK, Zdeněk. *Praktikum z molekulární genetiky*. - edition. -. 72 pp.
- AMBLER, Zdeněk. *Základy neurologie : [učebnice pro lékařské fakulty]*. 7. edition. Praha : Galén, c2011. ISBN 9788072627073.