Duchenne muscular dystrophy

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Duchenne muscular dystrophy (DMD) is one of the most common types of hereditable muscular dystrophy. It is caused by the mutations of the dystrophin gene at the locus Xp21. This gene is responsible for encoding dystrophin – the structural protein of sarcolemma. The absence of dystrophin leads to the damage of muscular cells, which causes muscular weakness and gradual loss of endurance. The increase of free ionized intracellular calcium level due to the destabilization of sarcoplasmic membranes is the root of the problem, which leads to lysis of the muscular fibers.

Nowadays, two forms of this disease are distinguished: Duchenne muscular dystrophy (DMD) and Becker's muscular dystrophy (BMD). The last one is a mild form of a disease and usually is caused by the mutations, which do not disrupt the reading frame, so that defective dystrophin, which, however, retains partial functional activity, is synthesized. However, DMD is a more frequent disease: it strikes about 3 out of 10 000 males, as for BMD it is 3 out of 100 000.

This disease is X-linked recessive, so it's more common for men. Females with a single copy of defective gene may show only mild symptoms. Appreciable quantity of cases is caused by sporadic mutations. In other cases the defective gene is inherited from person's parents. The main symptoms of the disease are the following:

1) awkward manner of walking
2) Gowers's symptom
3) ascending muscular weakness
4) pseudohypertrophy of tongue and calf muscles
5) contraction of tendons
6) skeletal deformities
7) damage of heart muscle
8) dementia

There is no cure of DMD and BMD and the aim of treatment is to control symptoms and to improve the quality of life. But current researches in gene therapy give us promising results in the experiments on mice.