Abstract

Duchenne muscular dystrophy is one of the most frequent and very severe congenital myopathies, affecting mainly boys. The disease is caused by a mutation in the gene encoding the dystrophin protein. The gene is located in the muscle tissue cells on the inner side of the sarcolemma. Dystrophin provides a link between the actin filaments and the extracellular matrix. It is important for the proper functioning of muscles during contraction and relaxation. As explained in this thesis, the production of dystrophin is of critical importance already at the muscle tissue development stage. The *DMD* gene expression also affects the expression of the other genes which play a key role in the right development and growth of muscle tissue. Mutations in the *DMD* gene cause changes in the signalling pathway genes such as PKA, thus affecting the expression control of other genes. *Mdx* mice used in DMD studies show abnormalities at prenatal stages, which are manifested through wrong organisation of microtubules and location of muscular cell nuclei, and a general increase in the number of fast myosin fibres (FMyHC). The absence of dystrophin also has an adverse effect on the satellite stem cells. The signalling pathway required for the correct spindle apparatus orientation is damaged. The wrong orientation causes the asymmetric cell division to fail, which leads to the premature “ageing” of cells.

Keywords: dystrophin, Duchenne muscular dystrophy, DGC complex, muscle tissue, myogenesis, *mdx* mouse