Abstract

This thesis is a review with the topic of Marfan syndrome (MFS) that hasn’t been sufficiently described in Czech scientific literature yet. The first chapters pursue general informations and history of research of MFS, which was first described in 1896 and following research continued. During 1990s the main interest was to describe manifestations of Marfan syndrome and to find main genetic causes. Research continues until today and it is focused on searching specific mutations causing this syndrome.

The thesis furthermore describes clinical manifestations classical MFS and neonatal MFS, e.g. manifestations in many body systems as sketal, cardiovascular, pulmonar and other systems. Thesis is focused on process which enables to diagnose MFS. The main part of the thesis is a description of molecular nature and genetic aspects of this syndrome. The thesis describes in detail gene FBN1 and different types of mutationon which are responsible for formation of MFS. Attention is paid to genotype/phenotype correlation.

This thesis also includes suggestions for work with this topic (MFS) in teaching at high schools and grammar schools.