

Atherosclerotic cardiovascular diseases are still the most common cause of death in Europe, despite new diagnostic methods and treatment. Although many accelerators of the atherosclerotic process are known, only LDL-cholesterol is considered to be the causal risk factor for atherosclerosis. Familial hypercholesterolaemia (FH) is an autosomal dominantly inherited disease whose carriers have had a high level of LDL-cholesterol since childhood due to reduced amount or function of LDL receptors. Determination of the causative mutation is not always possible and the diagnosis is established by using some scoring systems which take into account personal and family history and some typical signs (e.g. tendon xanthomas) in addition to LDL-cholesterol value. The treatment of FH is lifelong and to achieve the LDL-cholesterol target, combination therapy (ezetimibe, modern biologic therapy) in addition to statins, is often necessary. However statins are always the mainstay of the treatment.

By retrospective analysis of data from 1236 patients diagnosed with FH, we confirmed the cardiovascular risk of these patients is different depending on the presence of other risk factors. At the highest cardiovascular risk were individuals with combination of risk factors - high level of LDL cholesterol and total cholesterol, as well as high level of triglycerides and lipoprotein Lp(a) and the presence of other diseases (arterial hypertension, diabetes mellitus). The treatment can be complicated by presence of statin intolerance. In a cohort of 300 patients, we have proven statin intolerance is an overestimated diagnosis, but on the other hand it can be a significant barrier in treatment. Therefore, other active agents, including biological treatment are being developed and have shown an excellent lipid lowering effect with a favourable safety profile in clinical trials. The effect and safety of monoclonal antibodies, PCSK9 inhibitors, we confirmed in assessment of first 314 patients treated with PCSK9 inhibitors in our center. Not only the availability of modern therapy, but also the rich clinical experience and access to newer diagnostic procedures are the reasons why patients diagnosed with FH should be followed up in a specialized center. The benefit of this long term and systematic care, defined by achieving better results was confirmed in the analysis of 1236 patients mentioned above. The international ScreenPro project has also shown that international cooperation between countries with different levels of medical care has a positive impact on improving the diagnosis and treatment of these patients.

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