ScreenPro FH Project

INFORMATION FOR PATIENTS

Dear Sir or Madam,

The results of clinico-biochemical testing indicate that you are likely a carrier of an illness called familial hypercholesterolemia. This condition results in genetically-determined metabolic dysfunction in the structure or formation of LDL receptors that are normally present on the surface of liver cells, and which mediate LDL-cholesterol uptake from the blood. LDL-cholesterol is the "bad" cholesterol that accumulates in the walls of blood vessels and significantly contributes to the development of atherosclerosis. Individuals with familial hypercholesterolemia manifest atherosclerosis at an early age (typically around age 35) in the form of coronary artery disease, myocardial infarction (heart attack), peripheral arterial disease, or cerebrovascular accident (stroke). Inherited differences in a person's DNA result in impaired function of LDL receptors, which leads to an accumulation of LDL-cholesterol in the blood. Patients with familial hypercholesterolemia always have significantly elevated levels of total and LDL-cholesterol; total cholesterol levels are often greater than 9 mmol / L. (Generally speaking, a normal total cholesterol value is below 5 mmol / L, and a normal LDL-cholesterol value is below 3 mmol / L.) Cholesterol can also accumulate in other tissues and manifest as eyelid xanthelasmas, tendinous xanthomas, or a whitish ring around the iris. Familial hypercholesterolemia is inherited through autosomal dominant transmission; this means that if one parent is affected, their children have a 50% probability of being affected, irrespective of sex. The incidence rate of familial hypercholesterolemia in the population is between 0.2 % and 0.4 %.

Familial hypercholesterolemia is a condition that can be treated! Dietary and lifestyle changes, together with medication to reduce blood cholesterol levels, can significantly reduce the incidence of early myocardial infarction. Until quite recently, familial hypercholesterolemia was diagnosed solely on the basis of clinical and biochemical signs of disease. Even today, such diagnostic criteria remain important. The most reliable diagnosis, however, is now based on a genotype analysis (i.e. identification of a specific type of mutation in the gene for the LDL receptor that is responsible for the dysfunction). This diagnostic tool will certainly be of great benefit in the future because it enables physicians to correctly diagnosis patients in a precise and timely manner. The international ScreenPro FH project was created in an effort to expand the number of accurately diagnosed and properly treated familial hypercholesterolemia patients, and to support the implementation of new diagnostic procedures in clinical practice. ScreenPro FH is currently active in more than 18 countries, including yours. The project's goal is to screen for and identify patients with familial hypercholesterolemia; to proactively screen and identify these patients' relatives; and above all to ensure their timely and effective treatment.

A network of regional centers has been created to provide expert assistance to patients with this condition. Upon receipt of your written consent, you will be enrolled in the ScreenPro FH project and asked to provide a blood sample that will undergo genetic testing to detect mutations in the gene that codes for LDL receptors. Blood relatives who have also provided written consent will undergo similar testing. Identification of the responsible mutation involves a very demanding and lengthy examination, which can lead to a delay in getting your results; we will inform you as soon as your results are available. If you test positive, you will be added to a database of all carriers of this metabolic disorder. Along with the previously described genetic testing (which involves nothing more than drawing an extra 5 ml tube of blood during a standard blood test), you must provide your written consent (the form for which is included with this document). You have the option to consult your general practitioner to answer any questions you might have. Your participation in the ScreenPro FH program is purely voluntary, and all information and testing results will be kept in accordance with the principles of medical confidentiality.

Adapted from the Czech Republic MedPed project.