Familial hypercholesterolemia: a rare case of early diagnosis

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Introduction. Familial hypercholesterolemia (FH) is a common life-threatening genetic condition that causes high cholesterol and leads to a much higher-than-normal risk of coronary heart disease (CHD). The heterozygous type is found in about 1 out of 300–500 people, the homozygous type is quite rare in 1 out of 1 million people.

Objective. To analyze the clinical case of family hypercholesterolemia in sibs.

Methods. In 3 siblings (from triplets) at the age of 7 years randomly detected new-onset hypercholesterolemia 5.97 mmol/L, 5.65 mmol/L, 6.43 mmol/L. In a second study after 14 days, hypercholesterolemia persists (6.11 mmol/L, 5.67 mmol/L and 6.49 mmol/L, respectively). Two siblings (identical) had high cholesterol levels (4.060 mmol/L and 4.413 mmol/L) due to low density lipoprotein (LDL). The third child with the lowest level of hypercholesterolemia had normal LDL and a high level of high-density lipoprotein (HDL). No evidence of secondary hypercholesterolemia (diabetes mellitus, chronic renal insufficiency, hypothyroidism, cholestatic hepatitis, iatrogenic illness) was found.

A mother (42 years old) sticks to a strict diet with a reduced fat content, but hypercholesterolemia persists, statins therapy is not conducted, recommendations for examining children have not been received.

Results. Plasma LDL cholesterol level of 4.0 mmol/L or higher in follow-up blood test, provided parents with hypercholesterolemia, confirms the FH in two children from triplets.

At the same time, there are no external physical signs of the disease in children (xanthomas, corneal arch, xanthelasma). This, along with relatively low hypercholesterolemia, suggests a prognostically favorable Heterozygous Familial Hypercholesterolemia. Genetic screening for the presence of FH is not required to confirm the diagnosis but may be useful if the diagnosis is ambiguous.

A strict diet with a reduced fat content was recommended to patients, as well as supervision of a cardiologist and lipid screening. It was decided that at the age of 8–10 years, while maintaining LDL cholesterol > 4.0 mmol/l in follow-up blood test would be observed even on the recommended diet, the treatment with low doses of statins would be discussed.

Conclusion. Despite the prevalence of FH and the availability of effective treatment, FH is rarely diagnosed in children. This emphasizes the importance of lipid screening in childhood and cascading screening of all members of the patient 's family for the prevention of CHD.