

Familial Dyslipidemia; What Type of Management In Children? A Report of One Case

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Case Report

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Abstract: Dyslipidemias are among the most common hereditary diseases, their early diagnosis is essential for the health of the child in adulthood. Exact knowledge of family history is crucial for primary diagnosis as well as for the overall assessment. We report the case of an 11-year-old girl, with family history of dyslipidemia, revascularized ischemic heart disease before the age of 50 years. She had xanthomas since the age of 3, and a biological dyslipidemia found at the age of 5. Initially started on colestyramine 4g / day then atorvastatin 40 mg and ezetimibe 10 mg / day. The diagnosis of familial hypercholesterolemia of genetic appearance, type IIa was made. Lipid metabolism abnormalities are significant risk factor for the early development of arteriosclerotic lesions and should be diagnosed and treated as early as possible. The management of dyslipidemia in children is complex including an increase in physical activities, the prevention of risk factors and a medical treatment with statins as a first choice since the age of 8 years.

Keywords: Familial hypercholesterolemia- cardiovascular risk-statins.

INTRODUCTION

Dyslipidemias are among the most common hereditary diseases, early diagnosis is crucial for the child's health in adulthood. Exact knowledge of family history is essential for primary diagnosis as well as for the overall assessment. The treatment of dyslipidemias is complex including the stimulation of physical activities, the prevention of risk factors as well as diet and drugs. We report the case of an 11-year-old girl.

OBSERVATIONS

G. S from Youssoufia (Morocco), aged 11 years and 2 months, has since the age of 3 multiples xanthelasms at elbows, knees and Xanthoma tendinosum. The initial biological assessment when she was 5, showed a total cholesterol level of 9 g / l. she initially started colestyramine 4g / d. After one year, she was prescribed atorvastatin 40mg / d and ezetimibe 10mg / d. However, she didn't take any of them for 2 years because of her lack of resources. During the Past Medical History (PMH) taking, we note the existence of dyslipidemia, High blood pressure (HBP) and revascularized ischemic heart disease in the father, paternal uncle and aunt before the age of 50. Currently, she reports palpitations, both exercise and rest dyspnea, intermittent claudication without chest pain. At medical examination, the ankle-brachial pressure index (ABPI) is normal on both sides at 1, she has xanthelasma in the knees (Figure 1), elbows (Figure 2) and Xanthoma tendinosum (fingers, toes, Achilles tendons) (Figure 3), the TANNER scale is considered at B2P1 without stunting. Blood tests found: total cholesterol= 8.65g / l;

LDL = 6.21g / l; triglycerid = 1.29g / l. Cardiovascular outcome assessment: echocardiography reveals a slightly altered and thickened tricuspid aortic valve with moderate aortic insufficiency; minimal mitral insufficiency with no left atrial enlargement. We were unable to have the results of Doppler ultrasonography of the lower extremity arteries. The diagnosis of family hypercholesterolemia of genetic appearance, type IIa is made. The patient is put on pravastatin 10 mg / day and ezetimibe 10 mg / day with dietary restrictions and recommendations of physical activities.



Fig-1: xanthelasma of knees



Fig-2: xanthelasma of elbow



Fig-3: Xanthoma tendinosum

DISCUSSION

Lipid metabolism disorders are a heterogeneous group of diseases characterized by abnormal plasma levels of cholesterol and/or triglycerides. Most dyslipidemias are a significant risk factor for the development of arteriosclerosis and should therefore be taken seriously in children, although they are almost always asymptomatic [1].

Most patients with dyslipidemia are asymptomatic in childhood; fatty deposits in the skin, xanthelasma, may be the first symptoms of familial hypercholesterolemia (Figure 1). Medical family history taking has a key role. All first degree relatives are to be included [3].

High plasma cholesterol levels do not cause acute complications, but they are a risk factor for early atherosclerosis. We fear in particular, complications of homozygous familial hypercholesterolemia, in which acute myocardial infarction is described with fatal outcome already in childhood [2]. Very high triglyceride levels ($> 10\text{mmol/l}$) can cause acute symptoms such as; acute abdominal pain, gastrointestinal bleeding and acute pancreatitis, the prognosis of which can be severe. Hypertriglyceridemia, on the other hand, is not a risk factor for early atherosclerosis.

Almost all children who have dyslipidemia need a diet and / or medication. However, it is difficult to decide starting from which values the medical treatment becomes necessary. The results of the diet are not satisfactory even applied strictly in familial hypercholesterolemia.

Statins are the gold standard for the treatment of familial hypercholesterolemia. Simvastatin, lovastatin, atorvastatin, pravastatin, fluvastatin and rosuvastatin are prescribed in children in USA and Europe from the age of 10 years, except the pravastatin which is allowed only from the age of 8 years [4]. For some, rosuvastatin can be started as early as 6 years old; in Australia for example; atorvastatin can be used from 6 years old, especially in severe hypercholesterolemia [5]. The efficiency of statins has been confirmed, even in the period of puberty [6-8].

The treatment can be started at low dose and titrated according to the lowering of LDL cholesterol. There is no set threshold, but an expert consensus recommends LDL cholesterol $<3.5\text{mmol/l}$ at age of 10 or a 50% reduction between the age of 8-10 years, especially for children with important cardiovascular risk factors [4, 9-12]. The combination of ezetimibe and colestyramine might be helpful in some patients [10, 11, 13, 14].

It is advisable to do blood tests every 6 months and then every year after stabilization. When treated with statins, transaminases and creatine kinase should be checked. In case of a very high cholesterol level and critical family history, stress ECG, echocardiography and Doppler ultrasonography of supra-aortic trunks are indicated [15-19].

CONCLUSION

Lipid metabolism abnormalities are a significant risk factor for the early development of arteriosclerotic lesions and should be diagnosed and treated as early as possible.

The management of dyslipidemia in children is complex including an increase in physical activities, the prevention of risk factors and medical treatment with statins as a first choice.

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