

High Cholesterol: *It's All in the Family*

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CardioCase Presentation

Luke's Lipids



Luke, a 32-year-old computer salesman and a recent immigrant to Canada, presents with palpitations. He is a non-smoker.

His family history includes a father who died suddenly at the age of 42 and one brother who underwent coronary bypass at age 40.

On examination, he has occasional extrasystoles, but the exam is otherwise unremarkable.

His blood pressure is 128/76 mmHg and he has tendon xanthomata (Figure 1).

A lipid profile is ordered (Table 1).

Table 1

Luke's lipid profile

- Total cholesterol: 9.0 mmol/L
- HDL: 1.0 mmol/L
- LDL: 7.4 mmol/L
- Triglycerides: 1.2 mmol/L

HDL: High-density lipoprotein
LDL: Low-density lipoprotein

What's Your CardioCase Diagnosis?

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CardioCase Discussion



Figure 1. Tendon xanthomata on dorsum of hands typical for heterozygous familial hypercholesterolemia.

A closer look at Luke


Luke is diagnosed with familial hypercholesterolemia (FH). FH is an autosomal dominant condition with defective low-density lipoprotein (LDL) receptor activity. The prevalence of this condition in the general population is one in 500.

A typical sign of FH is the presence of tendon xanthomata. FH is associated with early development of atherosclerosis and coronary heart disease.

What is the treatment?

The physician should recommend family screening. Luke should also be referred to a cardiologist for further assessment.

Aggressive lipid-lowering therapy is indicated with an LDL target < 2.5 mmol/L.

The first-line therapy is a statin. The statin can be combined with ezetimibe or fibrates if needed. 

About the author

Dr. Fodor is a professor emeritus of medicine and head of research, Prevention and Rehabilitation Centre, University of Ottawa Heart Institute, Ottawa, Ontario.