

Homozygous familial hypercholesterolaemia in two sisters misdiagnosed as rheumatoid arthritis

Mahmoud H Ayesh (Haj Yousef),¹ Ayham Shatnawi,² Mai Khdeir,² Diala Khaldoun Alawneh²

¹Department of Medicine, Jordan University of Science and Technology, Irbid, Jordan
²Jordan University of Science and Technology, Irbid, Jordan

Correspondence to
Dr Mahmoud H Ayesh (Haj Yousef),
ayeshmahmoud@hotmail.com

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DESCRIPTION

A female patient aged 25 years and her younger sister aged 23 years were referred to the haematology clinic at our hospital suffering from iron-deficiency anaemia. Before taking the medical history of the two patients, the older patient told the treating physician (the author) that she and her sister had been experiencing yellow swellings on their joints since they were 10 years of age. The patients were told that the cause of the swellings was rheumatoid arthritis (RA), but despite treatment for RA, the swellings increased in size. No related signs were observed among relatives, including the father and mother.

On physical examination, both sisters had xanthoma of the hands, elbows, knees and ankles (figures 1–3). Lipid profiles of both patients showed high cholesterol (22 and 23 mmol/L) and low-density lipoprotein (LDL) levels (21 and 22 mmol/L), and normal levels of triglyceride (1.36 and 1.46 mmol/L) and HDL (0.8 and 0.85 mmol/L), respectively. The father and mother had high cholesterol (10.54 and 9.23 mmol/L) and LDL (8.1 and 6.7 mmol/L) levels, respectively, consistent with heterozygous familial hypercholesterolaemia (HeFH). We found parents to be first cousins.



Figure 1 Tendon xanthoma involving the hands of both patients.



Figure 2 Tuberous xanthoma of the elbows of the younger sister.



Figure 3 Tuberous xanthoma of the knees of the younger sister.

A diagnosis of homozygous familial hypercholesterolaemia (HoFH) was made based on early age onset of xanthomas with high LDL levels, supported by HeFH diagnosis in both parents. No molecular studies were available.¹

HoFH, a rare genetic disorder with a frequency of 1:3 000 000 to 1:1 000 000, is characterised by premature accelerated atherosclerosis of the cardiovascular system that affects the aortic root and the coronary ostia. In young patients, early symptoms and signs are associated with aortic stenosis and regurgitation. Treatment options for HoFH include lifestyle modifications, the administration of lipid-lowering medications, such as statins, and LDL apheresis. Recently, lomitapide and PCSK9 monoclonal antibody inhibitors have been used.¹



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Learning points

- ▶ Homozygous familial hypercholesterolaemia (HoFH) is characterised by high plasma low-density lipoprotein levels and xanthomas before 10 years of age. It can be misdiagnosed as rheumatoid arthritis.
- ▶ HoFH is an important risk factor for premature coronary artery and atherosclerosis disease in children and young adults.
- ▶ HoFH early symptoms and signs are often associated with aortic valve-related disease in young patients.

Contributors MHA worked up the case and wrote the case report. AS, MK and DKA reviewed the case report.

Competing interests None declared.

Patient consent Obtained.

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