Research Article

Familial hypercholesterolemia – case report

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Abstract

Familial hypercholesterolemia is an autosomal dominant disorder due to mutations in the LDL receptor gene. It is characterized by skin and tendon xanthomas, xanthelasma and arcus Cornea and associated with an increased risk of premature coronary heart disease. We report a case of 26 year old female who presented with xanthomas and elevated serum LDL cholesterol. Her elder sibling had similar complaints. This report is to emphasise the need to clinically recognize xanthomas and its association with elevated LDL-C. Screening of first-degree relatives and extended family members plays an important role in early detection and treatment.

Key words: familial hypercholesterolemia, low density lipoprotein cholesterol, xanthomas.

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INTRODUCTION

Familial hypercholesterolemia (FH) is an autosomal codominant disorder characterized by high serum low density lipoprotein cholesterol (LDL-C), tendon xanthomas, and prematurecoronary atherosclerosis. FH patients require lipid lowering drug therapy. If medical treatment is inefficient then LDLC apheresis and liver transplantation are needed.[1] The present article aims to report a case of a 26 year old female patient who developed xanthomas since the age of 13years and elevated serum LDL-C.

CASE REPORT

A 26 year old femalepatient who developed xanthomas since the age of 13 years, presented with history of multiple swellings over elbow and ankle. Her elder sibling had similar complaints. There was no history of diabetes mellitus, hypothyroidism, hepatic or renal disease. On physical examination, multiple nodules of varying size (2mm- 30mm) were observed on extensor surface of hands (Figure 1), elbow, buttocks, knee, ankle (Figure 2) and feet, suggestive of xanthomas. In addition, there was corneal arcus (Figure 3). Systemic examination was unremarkable. Investigations revealed total cholesterol of 660 mg/dl, LDL-C of551mg/dl, HDL-C of 40 mg/dl and triglyceride of 281 mg/dl. Completehemogram, blood sugar, renal function test, liver function test and thyroid function test were within normal limits. Chest X-ray and electrocardiogram, Echocardiogram, coronary angiogram were normal. Fine needle aspiration cytology from one of the nodule revealed foamy histiocytosis. Based on the above findings, a diagnosis of familial hypercholesterolemia, probablyhetereozygous was considered. She was started on rosuvastatin 40mg.

Figure 1

Figure 2

Figure 3

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DISCUSSION

Primary disorders of lipoprotein metabolism are due to genetic mutations. Secondary disorders of lipid metabolism occour in diabetes mellitus, hypothyroidism.[2] The heritable hyperlipidemia is of six types: I, IIa, IIb, III, IV and V. Subcutaneous xanthomas typically occur in patient with heritable hyperlipidemia[1] FH is an autosomal dominant genetic disorder due to mutations in the LDL receptor gene located on chromosome 19[3] According to the Frederickson's classification, this condition is categorized as a type II hyperlipoproteinemia[1] There are two types of familialhypercholesterolemia: the heterozygous form in which the patient has one normal allele and one mutated allele is the most common form with an incidence of 1 out of 500, whereas thehomozygous form in which the patient has two mutated alleles, an autosomalcodominant disorder, is rare with an incidence of approximately one in a million. Patients with heterozygous FH are usually diagnosed during adulthood and often respond well to medical therapy. Homozygous FH are often diagnosed earlyin childhood, do not respond well to medical therapy, and can progress rapidly to premature coronary artery disease[1] Several types of cutaneous xanthomas are recognized and associated with FH including xanthelasma, xanthomatendineum, and xanthomatuberosum. Other types of xanthomas, such as eruptive xanthomas, xanthomaplanum, palmar xanthomas, and tuberous xanthomas,are not usually associated with FH[4] Other clinical manifestations of FH include xanthomas located in the tongue and buccal mucosa, and arcusseniliscorneae.[5] Our patient had eruptive and tuberous cutaneous xanthomas, as well as Achilles tendon, lower leg extensor, hand extensor tendon xanthomas and arcusseniliscorneae, all classical, signs of FH. Our patient did not have any cardiac involvement. Simon Broome's diagnostic criteria for familial hypercholesterolemia says a definite diagnosis of familial hypercholesterolemia can be made if either the total cholesteroloncentration is above 7.5 mmol/liter in adults or the LDL cholesterol concentration is above4.9mmol/liter in adults and if tendinousxanthomas were present in the patient or a first degree Relative6-7Our patient and her elder sister had xanthomas with elevated LDL cholestrols. Hence this confirms the diagnosis of familialhypercholesterolemia in our patient. Treatment options available for FH are lipid lowering drugs like statins, bile acidsequestrants, apheresis and liver transplantation[1] Our patient was put on rosuvastatin 40 mg. After two month follow up there was significant regression of xanthomas and reduction in LDL-C levels.

CONCLUSION

The major complication of familial hypercholesterolemia is accelerated atherosclerosis, Hence, earlyrecognition of xanthomas and knowledge of its association with coronary artery disease is essential for every physician as early diagnosis and treatment can prevent premature deaths. All the relatives in the family should be screened for dyslipidemia. In this report, we highlight the typical clinical manifestation of familialhypercholesterolemia

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REFERENCES

1. Rader DJ,Hobbs HH. Disorders of lipoprotein metabolism. In: Fauci AS, Braunwald E, Kasper DL, Hauser SL, Longo DL, Jameson JL, *et al* editors. Harrison Principles of Internal Medicine. 17th ed, Vol.2, New York: McGraw Hill Inc; 2008.p.2416-429.
2. Manley RW, Weisgraber KH, Farese RV. Disorders of lipid metabolism. In: Wilson JD, Foster DW, Kronenberg HM, Larsen PR, editors. Williams textbook of endocrinology. 9th ed. Philadelphia: W.B. Saunders Co; 1998. p. 1099-153.
3. Nemati MH, Astaneh B. "Optimal management of familial hypercholesterolemia: treatment and management strategies," Vascular Health and Risk Management. 2010; 6(1):1079–1088.
4. Wolff K, Johnson RA, Suurmond D. Fitzpatrick's Color Atlas and Synopsis of Clinical Dermatology. Edn 5, McGraw-Hill Medical Publishing Division, New York, 2005.
5. Moyleand M, Tate B. "Homozygous familial hypercholesterolaemia presenting with cutaneous xanthomas: response to liver transplantation" .Australasian Journal of Dermatology. 2004; 45(4): 226–228.
6. Risk of fatal coronary heart disease in familial hypercholesterolemia Scientific steering Committee on behalf of the Simon Broome Register Group. BMJ.1991; 303:893-6.
7. Mortality in treated heterozygous familial hypercholesterolemia: implications for clinical management. Scientific Steering Committee on behalf of the Simon Broome Register Group. Atherosclerosis.1999; 142: 105-12.

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