Cardiology and Cardiovascular Medicine

Volume 1, Issue 2 Image Article

Incapacitating Xanthomas

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Received: 3 March 2017; **Accepted:** 09 March 2017; **Published:** 09 March 2017

Abstract

A 15 years old female presented with multiple gradually enlarging nodules over the limbs and buttock region for past 8 years. Patient was mainly concerned about swellings over her feet and hands, as these were so incapacitating that she was unable to do routine tasks like wearing shoes and writing. Her father and younger sibling had similar nodules but much smaller in size. Systemic examination was normal. Local examination revealed multiple yellowish brown, smooth surfaced painless xanthomas of varying size over gluteal region, bilateral knees,feet and hands. (figure 1) Xanthomas between thumb and index finger, a pathognomic finding in homozygous familial hypercholesterolemia(HoFH) was also seen. (Figure 1a and 1b) Blood investigations revealed deranged lipid profile (total cholesterol of 710 mg/dl, low density lipoprotein (LDL) cholesterol of 620 mg/dl, high density lipoprotein cholesterol of 35 mg/dl and triglycerides of 115 mg /dl). Rest of the blood investigations were normal. X-ray bilateral knees, feet and hands revealed soft tissue swellings corresponding to soft tissue swellings.Typical clinical manifestations at a young age, lipidogram and positive family history in two first degree relatives confirmed the

diagnosis of HoFH.[1] Genetic testing facility was not available Fortunately patient did not have any atherosclerotic or cardiac manifestations. She was started on high dose rosuvastatin and ezitimibe therapy.

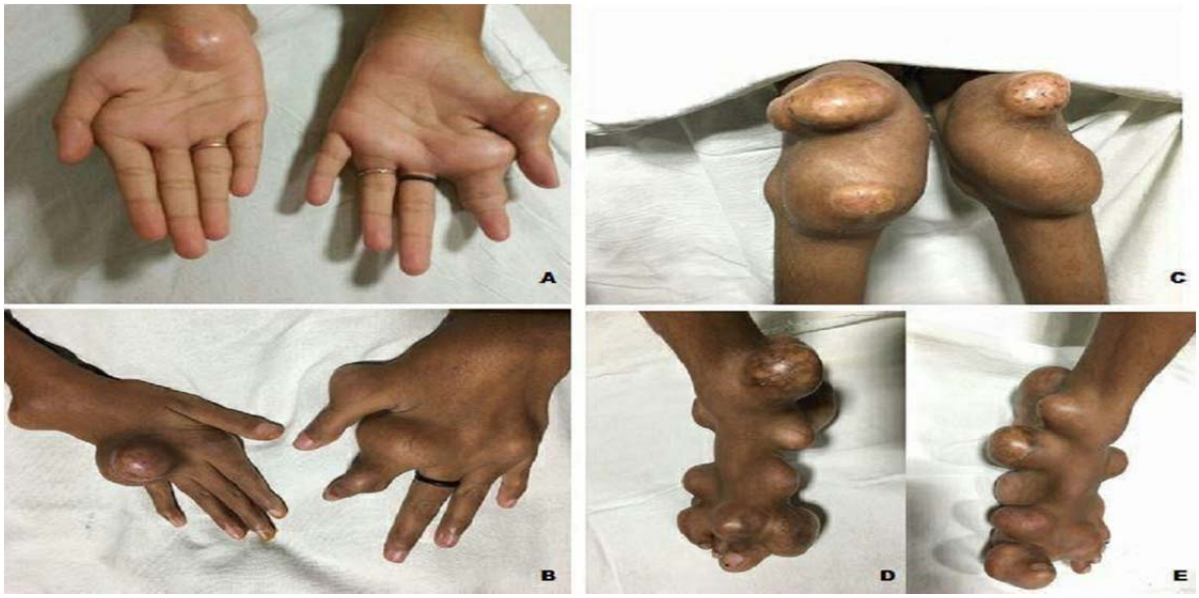

Figure 1: Image showing extensive tuberous and tendon xanthomas over a) dorsal aspect of both hands , b) palmar aspect of both hands , c) bilateral knees , d) right foot , e) left foot. Note the xanthomas between the index finger and thumb of left hand.

Conclusion

HoFH is a rare disease, occurring in 1:1,000,000 individuals.It results from mutation in the LDL receptor genes and results in premature atherosclerosis and xanthomas at a very young age.[2] Such excessive and incapacitating presentation of xanthomatosis adds clinical intriguity to the present case.

References

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