

A striking lipid phenotype: A case of homozygous familial hypercholesterolemia

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Abstract

Background: Homozygous familial hypercholesterolemia (HoFH) is a rare, severe genetic disorder characterized by markedly elevated low-density lipoprotein (LDL) cholesterol and premature atherosclerotic cardiovascular disease. Early recognition is crucial, as timely diagnosis and aggressive lipid-lowering therapy can significantly alter outcomes.

Case summary: A 9-year-old child born of consanguineous marriage presented with multiple skin lesions from 1 year of age. Initially, it involved the calf regions as linear, slightly raised lesions; over time, lesions gradually increased in size and number and spread to the ankles, knees, and elbows, with some coalescing into larger nodular swellings. He had been taking lipid-lowering medication for the last 5 years after consulting several physicians. Notable medical history included consanguineous marriage of his parents, no premature cardiovascular events among first-degree relatives, and presence of bilateral corneal arcus in his aunt. Examination revealed multiple tuberous xanthomas over the elbows and knees; tendinous xanthomas over the calf regions; and corneal arcus in both eyes. Laboratory assessment revealed high Total Cholesterol (highest 689 mg/dl) and high LDL-C (highest 624 mg/dl). Genetic testing came back positive for an LDL Receptor mutation. Both his father and mother had elevated LDL-C levels. Based on history, clinical findings and investigations, we diagnosed him as Homozygous Familial Hypercholesterolemia. Treatment included lifestyle advice such as less fat intake, more fruit and vegetable intake, exercise and maintaining a normal weight; pharmacological management included Rosuvastatin 20 mg once daily and Ezetimibe 10 mg once daily. During the first and second follow-up, his LDL-C and Total Cholesterol levels showed limited reduction, so the dosage was escalated to Rosuvastatin 80 mg and Ezetimibe 20 mg. We also counselled about other appropriate lipid-lowering therapies.

Conclusions: Familial Hypercholesterolemia is not only a disorder of elevated cholesterol—it is a lifelong cardiovascular risk condition. Timely diagnosis and intensive lipid-lowering therapy, together with screening of first-degree relatives, can substantially reduce the burden of premature cardiovascular disease. [*J Assoc Clin Endocrinol Diabetol Bangladesh*, 2026;5(Suppl 1): S82]

Keywords: Familial Hypercholesterolemia, Severe Hypercholesterolemia, HoFH, Genetic disorder, High LDL-C, Lipid lowering therapy

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