Case Report

A Case Report On Hyperlipidemia In A Child

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Abstract

Hyperlipidemia is most commonly attributable to genetic disorders of lipid metabolism in the pediatric population, with familial hypercholesterolemia (FH) being a classic example that predisposes to premature atherosclerotic disease. We present a case of 5 years old child with FH evident by high cholesterol and LDL-C levels which responded to statins without causing any significant adverse effects. Besides statins other therapies can also be incorporated for lipid lowering like adding ezetimibe, PCSK-9 inhibitors and rarely used treatment modality called lipoprotein apheresis. Familial Hypercholesterolemia is a disorder which needs early diagnosis and initiation of prompt treatment to prevent early onset atherosclerosis and its complications. This case report is being published for its rarity and early diagnosis and management.

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Introduction

Hyperlipidemia is often secondary in nature in pediatric population; however, markedly elevated levels of low-density lipoproteins (LDL) are commonly attributed to primary, genetic forms of the disorder.[1] These conditions are strongly associated with the early development of atherosclerosis, and certain pediatric diseases can further accelerate this process, leading to premature coronary events. Familial hypercholesterolemia (FH) is a classic example of genetic hyperlipidemia, characterized by markedly elevated lipid levels, particularly LDL cholesterol. FH is the most common primary hyperlipidemia associated with cardiovascular abnormalities in childhood, often manifesting as early as the first decade of life.[2,7] Therefore, timely identification and management of familial hypercholesterolemia in children is pivotal.

We present a case of hypercholesterolemia with extremely high cholesterol levels in a 5-year-old child.

Case Report

A 5-year-old boy was brought by his consanguineous parents with a two-month history of whitish-yellow nodules on the gluteal region, dorsum of the left foot, and behind the left ear. Systemic symptoms such as chest pain, bone pain, or abdominal pain were not reported. There was no relevant past medical history. Family history revealed normal lipid levels in parents and an elder sibling. On examination, the child was thin and lean (weight 12 kg, height 100 cm). There were multiple xanthomata over both gluteal regions more on the right side (Fig 1), dorsum of the left foot (Fig 2) and back of the left pinna (Fig 3). Fundus examination was unremarkable with no evidence of juvenile arcus in the cornea, hepatosplenomegaly was absent, and cardiovascular, respiratory and neurological system evaluations were all within normal limits. His blood pressure recorded was 94/58 mm Hg. A lipid metabolism disorder was suspected, so he was investigated accordingly.

Baseline laboratory investigations, including hematology, liver functions, urinalysis, as well as cardiac evaluation, including ECG, chest X-ray, and echocardiogram, were normal. His serum cholesterol level was 1637mg/dl with the lipid profile revealing markedly elevated LDL, VLDL, triglycerides, an increased cholesterol-to-HDL ratio, and reduced HDL levels (Table 1). The child was started on lifestyle modifications, including a low-cholesterol diet (<200 mg/day), reduced fat intake (<7% of total calories), a diet rich in fruits and vegetables, regular gentle exercise (walking and cycling), and rosuvastatin 10 mg daily, in accordance with pediatric guideline recommendations and clinical judgment. After four months, serum cholesterol and LDL decreased to 1135 mg/dl and 853 mg/dl, respectively; however, xanthomas persisted; subsequently, the statin dose was up titrated to 20mg and ezetimibe 10mg was added, resulting after one month in further reductions in serum cholesterol (969 mg/dl), LDL (837 mg/dl), and VLDL (100 mg/dl).

As he was started on high intensity statins; so, he was also looked for any adverse effects of the drugs which included liver function tests, CPK and renal functions which were all within normal limits.

Discussion

Familial hypercholesterolemia (FH) is a genetic disorder characterized by high levels of low-density lipoprotein cholesterol (LDL-C), which ominously escalates the risk of cardiovascular disease (CVD) from a younger age. One of the major causative factors of FH is the mutations in the LDL receptor (LDLR) genes, which decreases the clearance of LDL-C from the blood.[1,7] Early onset of atherosclerosis in children with FH underscores the importance of early diagnosis and intervention.

The clinical manifestation of FH in children usually includes physical findings such as Xanthomas as observed in this case and acrus Juvenilis. These manifestations along with a positive family history for hypercholesterolemia help in early diagnosis. Accurate diagnosis and screening of other family members can

Table 1. Lipid Profile Reports Follow up visit wise

S.No.	Lipid Profile	Baseline visit Levels (mg/dl)	1st follow-up (After 4 months from baseline visit) Results(mg/dl)	2nd follow-up (1 month after first follow-up)	Normal Ranges (mg/dl)
1	Serum Cholesterol	1637	1135	969	<200
2	HDL	32	27	34	>60
3	LDL	1109	853	837	<135
4	VLDL	496	255	100	≤30
5	Cholesterol to HDL Ratio	51.15	42.03	28.50	<3.5

HDL=High density lipoproteins, LDL=Low Density lipoproteins, VLDL=Very Low-density lipoproteins

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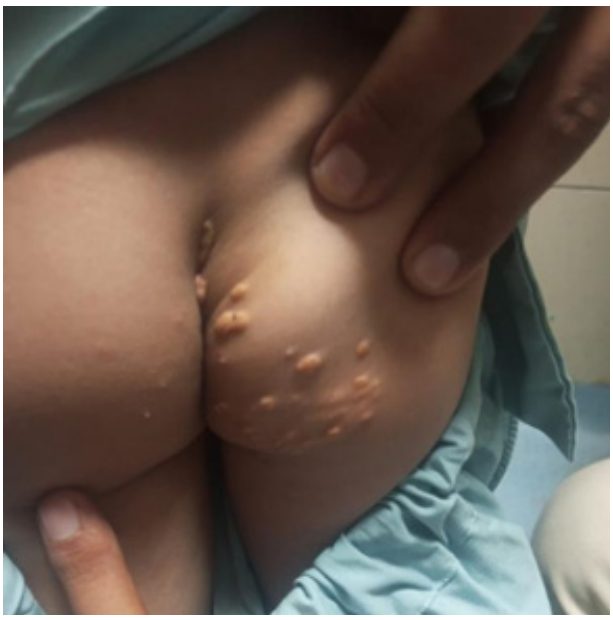

Figure 1: Xanthomas on the buttocks

Figure 2: Xanthoma on the dorsum of the left foot

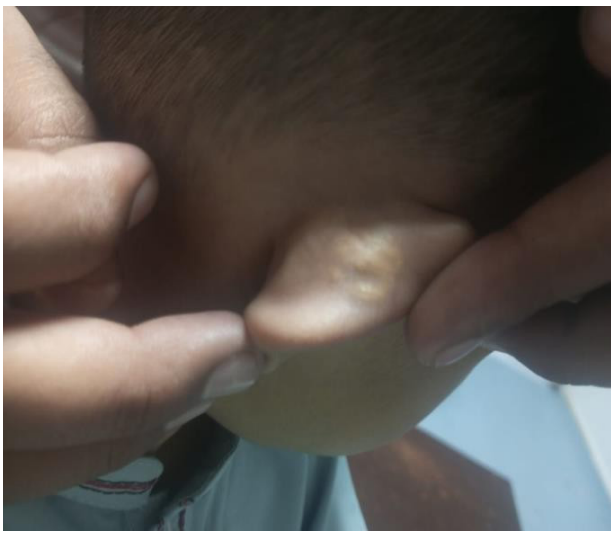

Figure 3: Xanthoma on the back of the pinna

be done through genetic testing.[3] The frequency of homozygosity for autosomal dominant traits such as FH is anticipated to be relatively high in Pakistan.[5] Genetic testing is rarely done in our setup due to a lack of resources.

The lifestyle modifications along with pharmacotherapy are the mainstay of management of FH in children. Dietary changes, including a low-cholesterol, low-fat diet, and increased physical activity, are vital but often not enough alone. There are several drugs used to reduce LDL-C levels; however, statins are the first line pharmacotherapy in this regard.[2] In this case, the patient was initially treated with Rosuvastatin, which significantly reduced LDL-C levels but did not completely normalize them. Rosuvastatin was initiated at a pediatric dose aligned with guideline safety[6], though not explicitly weight based. Guidelines recommend lipid reassessment at 6–8 weeks for dose adjustment. Due to limited resources, follow-up occurred at 4 months, which is acknowledged as a limitation.

Ezetimibe, a cholesterol absorption inhibitor, was added to the treatment regimen; further lowering LDL-C levels. Guidelines recommend maximizing statin dose before adding ezetimibe[6] but concerns about tolerance and adherence influenced decision-making. This combination therapy is supported by guidelines and has been promising in achieving target LDL-C levels in pediatric patients.[2,8]

Regular monitoring for potential adverse effects, such as liver enzyme elevations and muscle symptoms, is essential to ensure the safety of long-term statin use in children.

Those patients with FH who fail to achieve target LDL-C with conventional therapy need advanced treatment options which include PCSK-9 inhibitors and lipoprotein apheresis. Proprotein convertase subtilisin/kexin type 9 (PCSK9) inhibitors (evolocumab and alirocumab) are monoclonal antibodies that block the binding of proprotein PCSK9 to hepatic LDL receptors, thereby preventing receptor degradation, increasing receptor expression on the cell surface, and ultimately increasing LDL cholesterol clearance from the circulation. These agents have been proven to be well tolerated and effective in FH by lowering baseline LDL-C by 50% to 60%[7] and emerging as a miraculous agent in significantly lowering LDL-C levels in children.

Another novel drug Bempedoic acid works by inhibiting adenosine triphosphate (ATP)-citrate lyase (ACL) which then converts citrate to acetyl-CoA in the cholesterol synthesis pathway, leading to upregulation of LDLR, can be used as appurtenant to lipid lowering drugs specially the statins. It has been proven in multiple RCTs for its better tolerability as compared to statins and efficacy in dyslipidemias including familial hypercholesterolemia, specifically heterozygous familial hypercholesterolemia.[9] For a refractory hypercholesterolemia a more advanced, however very rarely available

option like Lipoprotein apheresis can be considered.[4]

The modest 25% LDL-C reduction highlights the challenges in managing homozygous FH in resource-limited settings. Discussion with the family included possibility of advanced therapies such as PCSK9 inhibitors or lipoprotein apheresis, though availability remained a barrier. The family expressed concern about the persistence of xanthomas and incomplete response but were motivated to continue therapy. They valued clear communication about treatment limitations and future options.

Conclusion

This case report underscores the importance of early diagnosis and aggressive management of FH in children to prevent the development of atherosclerosis and subsequent cardiovascular events. Reporting of this case is due to the rare nature of the disease and to bring awareness to its early diagnosis, prompt initiation of therapeutic measures to prevent early onset cardiovascular complications. A multidisciplinary approach, involving pediatricians, cardiologists, and dietitians, is pivotal for optimizing treatment outcomes. Ongoing research and advancements in genetic testing and pharmacotherapy offer promising opportunities to improve the management of familial hypercholesterolemia in pediatric patients.

References

1. Alonso R, de Isla LP, Muñiz-Grijalvo O, Diaz-Diaz JL, Mata P. Familial hypercholesterolaemia diagnosis and management. Eur Cardiol Rev 2018;13(1):14.
2. Sprecher DL, Schaefer EJ, Kent KM, Gregg RE, Zech LA, Hoeg JM, et al. Cardiovascular features of homozygous familial hypercholesterolemia: analysis of 16 patients. Am J Cardiol 1984;54:20-30.
3. Sadiq F, Shafi S, Sikonja J, Khan M, Ain Q, Khan MI, et al. Mapping of familial hypercholesterolemia and dyslipidemias basic management infrastructure in Pakistan: a cross-sectional study. Lancet Reg Health Southeast Asia 2023;12:100162.
4. Padmanabhan A, Connelly-Smith L, Aqui N, Balogun RA, Klingel R, Meyer E, et al. Guidelines on the use of therapeutic apheresis in clinical practice: Evidence-based approach from the Writing Committee of the American Society for Apheresis: the eighth special issue. J Clin Apher 2019;34(3):171-354.
5. Ahmed W, Whittall R, Riaz M, Ajmal M, Sadeque A, Ayub H, et al. The genetic spectrum of familial hypercholesterolemia in Pakistan. Clin Chim Acta 2013;421:219-25.
6. de Ferranti SD, Lyon S. Dyslipidemia in children and adolescents: Management. In: Fulton DR, Armsby C, editors. UpToDate [Internet]. Waltham (MA): UpToDate Inc.; 2025 Feb 27 [cited 2025 Aug 26]. Available from: https://www.uptodate.com/contents/dyslipidemia-in-children-and-adolescents-management
7. Arca M, Celant S, Olimpieri PP, Colatrella A, Tomassini L, D'Erasmo L, et al. Real-world effectiveness of PCSK9 inhibitors in reducing LDL-C in patients with familial hypercholesterolemia in Italy: A retrospective cohort study based on the AIFA monitoring registries. J Am Heart Assoc 2023;12(21):e026550.
8. Lee YJ, Cho JY, You SC, Lee YH, Yun KH, Cho YH, et al. Moderate-intensity statin with ezetimibe vs high-intensity statin in patients with diabetes and atherosclerotic cardiovascular disease in the RACING trial. Eur Heart J 2023;44(11):972-83.
9. Bostock B. The role of bempedoic acid in the management of dyslipidaemia. J Prescribing Pract 2021;3(12):484-8.

Authors' Contribution Statement

SUR contributed to the conception, design, acquisition, analysis, interpretation of data, drafting of the manuscript, critical review of the manuscript, and final approval of the version to be published. AUW contributed to the conception, design, acquisition, analysis, drafting of the manuscript, and critical review of the manuscript. MA contributed to the acquisition, analysis, interpretation of data, and drafting of the manuscript. RUJ contributed to the acquisition, analysis, interpretation of data, drafting of the manuscript, and critical review of the manuscript. SU contributed to the acquisition, analysis, interpretation of data, and drafting of the manuscript. MWJ contributed to the acquisition, analysis, interpretation of data, and drafting of the manuscript. MW contributed to the acquisition, analysis, interpretation of data, and drafting of the manuscript. All authors are accountable for their work and ensure the accuracy and integrity of the study.

Conflict of Interest

Authors declared no conflict on interest

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Data Sharing Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.