Case Report

Case report on Caffeys disease in an adolescent with a familial aspect

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ABSTRACT

Caffey's disease is a bone disorder that most often occurs in babies. It is inherited in an autosomal dominant pattern but not all people who inherit the genetic change develop signs and symptoms. Most people with Caffey's disease have no further problems related to the disorder after early childhood. Occasionally, another episode of hyperostosis occurs years later. The peculiarity about this case was its presence in the adolescent stage of the child's growth. This condition is mostly underreported due to its regression in childhood itself, and hence we have reported this unique case. The parents presented with complaints of a different etiology and with complaints of bone pains and deformity since birth. He was diagnosed primarily with Caffey's disease after ruling out other diagnoses. There is very little literature on Caffey's disease due to its regression in early childhood.

Key words: Adolescent, Caffey's disease, Familial, Genetic, Pediatrics

Caffey's disease is characterized by massive subperiosteal new bone formation (usually involving the diaphyses of the long bones as well as the ribs, mandible, scapulae, and clavicles) typically associated with fever, joint swelling, and pain in children, with onset between birth and 5 months and spontaneous resolution by age 2 years [1,2]. Episodes of the recurrence of the manifestations of Caffey disease have been reported multiple times in individuals with the classic infantile presentation. Although the etiology of Caffey's disease is not completely understood, familial and sporadic forms appear to exist [3]. Generally, it is treated by sole acetaminophen having a good outcome [4].

CASE PRESENTATION

We report a case of an adolescent who was brought by his parents to the pediatric outpatient department with complaints of cough, fever, loose stools, and vomiting episodes for 2 days. While examining, it was noticed that there was a gross deformity of his right forearm. The swelling measures approximately 20 cm×8 cm×7.5 cm currently. On asking for further details, it was observed that the father was afflicted by similar deformities as well as the child's paternal grandparents and the child's sibling. The mother is not afflicted by the same [Figure 1].

The mother explained that she noticed mild swelling on his arms soon after birth. The child would complain or cry intermittently with waxing and waning of the swelling which increased gradually in episodes with tenderness and signs of inflammation intermittently. There was no gross reduction of mobility noted at present. She proclaimed that he was examined for the same, and the findings were normal. No reports are available regarding the same.

Other differentials such as hypervitaminosis were ruled out because he was having a normal diet and there was no excessive ingestion of the vitamin in any form. Bone infection was thought to be unlikely because of the lack of "toxicity" with normal vital signs other than a low-grade temperature despite multiple sites of involvement. The absence of significant bone tenderness and the radiological picture was against a diagnosis of multifocal osteomyelitis. A bone tumor again was felt to be very unlikely because of the multiple sites of involvement (primary bone tumors are usually single), and the absence of lytic lesions on X-rays seen in most secondary bone tumors. There was no involvement of the mandible or clavicle in the child. There is no reported increase in the swellings of the bone currently. It was then confirmed that he most possibly could be a case of Caffey's disease per the clinical and radiological pieces of evidence. An X-ray was performed of the afflicted arm and knee and was confirmed by a radiologist. The patient was recently assessed (follow-up) in the outpatient department. The child had no complaints of pain and was able to carry out all his daily activities without any disruption.

DISCUSSION

Caffey's disease is a self-limiting disorder also known as Infantile Cortical Hyperostosis. A trio of systemic symptoms (irritability and

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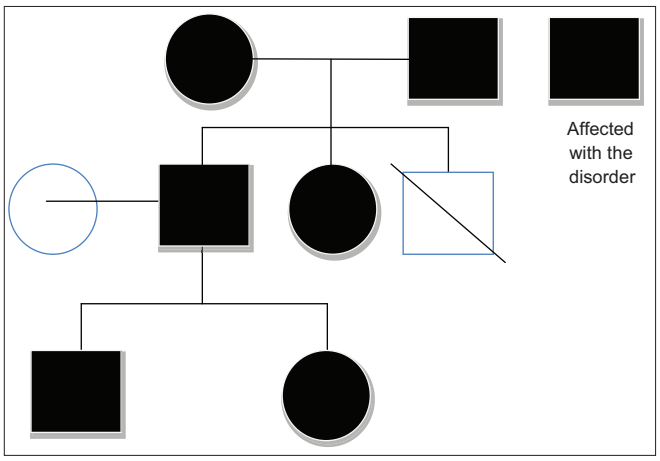

Figure 1: Pedigree chart of the patient

fever), soft-tissue swelling, and underlying cortical bone thickening characterize it [1]. Caffey and Silverman described it as a disease entity for the 1st time in 1945. The majority of instances are sporadic, but there have been a few familial cases with autosomal dominant and recessive patterns described. Infections, immunological problems, and genetic anomalies are among the proposed reasons. The identification of a gene locus (gene COLIAL, 17q21) in three unrelated families with the autosomal dominant inheritance that encodes the Alfa-1 chain of kind I collagen has raised some issues about whether certain cases constitute a kind of collagenopathy such as osteogenesis imperfect [2].

The case report highlights the challenges and complexities of diagnosing and treating Caffey's disease. Early recognition and diagnosis are critical for the successful management of the disease, but the rarity of the condition and the similarity of its symptoms to other bone disorders can make diagnosis difficult. The case reported the presentation of Caffey's disease in adolescence and the presence of the condition in the paternal family confirmed the genetic aspect of the disease. It also led to an insight into the hereditary passage of the disease to the next generation and enlightened us regarding the complexity of genetic disorders. Treatment may involve a combination of pain management, physical therapy, surgery, and genetic counseling to address the underlying cause of the disease. The diagnosis of this case proved difficult due to a lack of adequate data during the infancy of the child.

The most valuable diagnostic study is radiography. The distinctive feature is cortical new bone development (cortical hyperostosis) underlying areas of soft-tissue edema. While no laboratory tests are specific for Caffey's disease diagnosis, the following essential differential diagnoses must be ruled out: osteomyelitis, chronic hypervitaminosis A, bone tumor, scurvy, child maltreatment, and extended PGE1 infusion. Knowing about this unusual illness and its normal clinicopathological profile will keep the patient from being subjected to unneeded testing [3].

CONCLUSION

Caffey's disease is a rare but serious genetic disorder that affects bone growth in children. Early diagnosis and treatment are important for managing symptoms and improving the quality of life for affected individuals as well as genetic counseling in cases once diagnosed. Further research is needed to better understand the underlying mechanisms of the disease and to develop more effective treatments.

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