Caffey Disease in Infancy
A diagnostic dilemma for primary care physicians

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ABSTRACT: Caffey disease is a rare and self-limiting condition characterised by cortical hyperostosis with inflammation of adjacent fascia and muscles. It usually presents in infancy and clinical features include hyperirritability, acute inflammation with swelling of overlying soft tissues and subperiosteal new bone formation. Awareness of the existence of this rare condition and its typical clinical and radiological profile will avoid unnecessary investigations and treatment and help the physician to explain its good prognosis to parents of affected children. We report a three-month-old male infant who presented to the Outpatient Paediatrics Department at Moti Lal Nehru Medical College, Allahabad, India, in 2018 with a right shoulder mass, decreased upper limb movements and irritability. The patient was treated with ibuprofen and paracetamol. Irritability and limitation of movement improved over a treatment period of two weeks.

Keywords: Caffey Disease; Infant; Prostaglandin E1; Thrombocytosis; Case Report; India.

Case Report

A three-month-old afebrile male infant presented to the Outpatient Paediatrics Department at Moti Lal Nehru Medical College, Allahabad, India, in 2018 with a history of mild irritability, a right shoulder mass and decreased upper extremity movement for the previous two weeks. There were no symptoms associated with the respiratory, gastrointestinal or urinary systems. He was born full-term at 39 completed weeks of gestation via normal vaginal delivery with a birth weight of 2.5 kg, a length of 49 cm and a head circumference of 34.5 cm. His Apgar scores were 6 and 8 at 1 and 5 minutes, respectively. The infant was vaccinated according to his age with no significant antenatal or postnatal history. There was no history of prostaglandin E1 infusion to maintain ductal patency during the neonatal period. He was the first child of the couple and there was no family history of similar complaints. The infant was exclusively breast-fed since birth with no vitamin supplementation. There was no history of trauma, intramuscular injections, lethargy or convulsions. Child abuse was not suspected as he was the outcome of a planned pregnancy. Upon examination, the patient was active and alert, afebrile, mildly irritable and had no dysmorphic facial features. Anthropometric examination was normal for his age. A right shoulder mass with soft tissue swelling over the body of scapula was noted [Figure 1]. The infant

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had decreased upper extremity movements with pain during passive movement of the right upper extremity. However, deep tendon reflexes were well-preserved and there was no swelling or tenderness over other bones. The remainder of the systemic examination was within normal limits. X-ray examination of the affected shoulder revealed diffuse thick irregular ossification along the cortex of the scapula involving both anterior and posterior surfaces with diffuse thickening of the scapula with a thickened cortex [Figure 2]. Ultrasonography of the shoulder showed that the adjacent muscles around the scapula were relatively bulky with altered echogenicity without any evidence of fluid collections [Figure 3]. Biochemical and haematological investigations were mostly unremarkable except for an elevated C-reactive protein level [Table 1]. Genetic analysis could not be done as it was not available at the centre and the parents refused further investigations as it is a self-limiting condition.

On the basis of the clinical findings, elevated C-reactive protein and radiology, the infant was diagnosed with Caffey disease. Subsequently, he was treated with ibuprofen and paracetamol. Irritability and limitation of movement improved over a treatment period of two weeks. He was doing well on follow-up.

Discussion

Caffey disease is a rare, self-limiting disorder of early infancy with an onset within the first six months of life and usually resolves without sequelae by two years of age. It is often sporadic with occasional reports of being autosomal dominant or recessive. It is characterised by a triad of systemic symptoms (e.g. irritability and fever), soft tissue swelling and underlying cortical bone thickening. The mandible is the most commonly affected site followed by scapula, clavicle, ribs and long bones.

Laboratory findings include elevated erythrocyte sedimentation rate and in some cases high alkaline phosphatase, thrombocytosis, anaemia and elevated
immunoglobulin levels.5 There are no specific laboratory tests for Caffey disease. Radiology is the most valuable tool in diagnosing Caffey disease as it shows layers of periosteal new bone formation with cortical thickening.

However, its diagnosis may be delayed as it mimics many other conditions such as osteomyelitis, hypervitaminosis A, syphilis, scurvy, bone tumours, prolonged prostaglandin E infusion and child abuse.6 As the current patient was thriving and there was an absence of early bone radiological signs and a normal white blood cell count, osteomyelitis was ruled out. There was no history of vitamin A supplementation as national programmes in India recommend routine vitamin A supplementation from nine months onwards. Negative venereal disease research laboratory tests for both the mother and infant made syphilis an unlikely diagnosis. Malignancy and scurvy were also excluded based on normal radiography/ultrasonography and haematological tests. In addition, the infant did not have history of prostaglandin infusion during the neonatal period to maintain ductal patency.

After excluding various differential diagnoses and given the characteristic features of Caffey disease that were present in the current case (irritability, a right shoulder mass and decreased upper extremity movements with periosteal new bone formation), the patient was diagnosed with Caffey disease.

**Conclusion**

Awareness of the existence of Caffey disease and its typical clinical and radiological profile as well as its self-limiting nature will avoid unnecessary investigations and treatment in resource-limited countries such as India.

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