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Images of Interest / Imagens de Interesse

Soft Tissue Swelling in an Infant: What is the Diagnosis?

Edema dos Tecidos Moles em Lactente: Qual o Diagnóstico?

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Abstract

Caffey's disease also known as Infantile Cortical Hyperostosis is a rare, self-limited disease, that usually appears between birth and 6-months-old. It can be inherited as an autosomal dominant trait. However, sporadic forms and incomplete penetrance have been reported. The diagnosis is made by the triad of soft tissue swelling and underlying cortical bone thickening (hyperostosis), systemic symptoms and the changes in the X-ray. Molecular genetic testing is also available. Treatment is symptomatic.
The authors describe one infant with Caffey's disease and it's management.

Keywords

Caffey's disease; Infantile cortical hyperostosis; Collagen type 1.

Resumo

A doença de Caffey, também denominada Hiperostose Cortical Infantil, é uma doença rara e autolimitada, que se manifesta habitualmente entre o nascimento e os 6 meses. Frequentemente é transmitida de forma autossómica dominante, embora existam casos descritos de penetrância incompleta e de formas esporádicas. É caracterizada pela tríade de edema dos tecidos moles, hiperostose e sinais/sintomas sistémicos. O diagnóstico é realizado através destes achados clínicos e pela imagem típica em radiografia de hiperostose subperiosteal massiva, estando disponível a confirmação por estudos genéticos. O tratamento é sintomático.
Os autores apresentam o caso de uma criança com doença de Caffey e a sua evolução.

Palavras-chave

Doença de Caffey; Hiperostose cortical infantil; Colagénio tipo 1.

Case Description

A four-month-old boy, with no prenatal or neonatal relevant history, was evaluated on the pediatric orthopedic department, due to bilateral leg swelling and pain. Complaints started unilaterally in the first month of life, progressing over time, with analgesic drug requirement. The X-ray showed periosteal thickening and tibial bowing (Figure 1).
In this case, diagnosis of Caffey's disease was made by clinical and imaging findings. Genetic studies were available, but refused by the family. Symptomatic treatment was undertaken with non-steroidal anti-inflammatory drugs and paracetamol. He remains under surveillance, currently with 10 years old, without complaints and with no physical limitations (Figure 2).

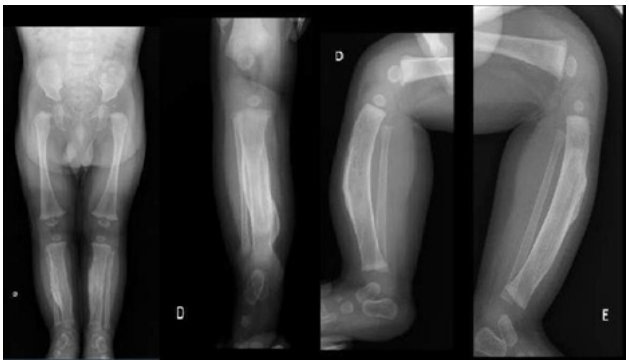

Figure 1 – Radiographs of lower limbs (age: six months) showing symmetrical periosteal reaction of the tibial diaphysis and bowing.

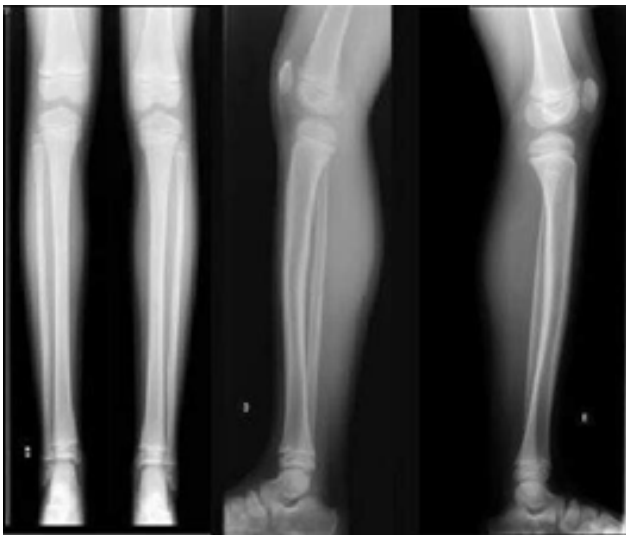

Figure 2 – Leg radiograph (age: eight years) shows normalization of tibial cortical thickness; the right tibia still shows a slight flexion deformity.

Learning Points of Discussion

Caffey's disease, also known as Infantile Cortical Hyperostosis (ICH), is a rare[1] bone disorder, characterized by painful soft tissue swelling and underlying cortical bone thickening (hyperostosis) that can be associated with systemic symptoms such as fever and irritability.[1-3] The massive subperiosteal new bone formation involves the mandible, clavicles, scapula, ribs and the diaphysis of long bones (most often the ulna)[1-4]. Unfrequently, the ilia, calvarium, carpus, tarsus, phalanges and vertebral bodies are affected.[5]

ICH is a type I collagenopathy and it is inherited as an autosomal dominant trait 3 associated with a mutation in the gene encoding the alpha-one chain type I collagen on chromosome 17q21 (COL1A1).[2,3] However, sporadic forms and incomplete penetrance have been reported.[3] There is no gender or racial predilection.[1]

This disorder usually presents between birth and six months of age,[1,2] although it can be diagnosed by prenatal ultrasound from 35 weeks of gestation (WG).[3] This is distinct from the Caffey dysplasia, with poor prognosis, which appears earlier than 35 WG.[1,3]

The diagnosis is made by clinical features and radiological findings.[1] Radiography in the acute and subacute phases reveals layers of subperiosteal bone deposition (either solid or multilamellar[6]), leading to osseous expansion characterized by dense cortical thickening and soft tissue swelling.[7,8] The pattern of involvement localized solely to the diaphysis of long bones typically leads to spindle-shaped bones.[9] Persistent skeletal deformities, such as bowing, marrow cavities, and residual streaks of incompletely absorbed cortical remnants, may be observed.[7] The involvement can be symmetrical.[2] Lytic skull lesions were described.[10] Radiological findings may endure for years after clinical resolution.[11,12]

Laboratory tests show elevation of the inflammatory parameters.[1,3] Molecular genetic testing is available.[3]

The differential diagnosis includes osteomyelitis, chronic hypervitaminosis A, hyperphosphatemic familial tumoral calcinosis, bone malignancies, scurvy, child abuse, storage diseases, prolonged prostaglandin E1 infusion.[1-3]

Usually it has a spontaneous remission, so treatment is limited to symptom management to relieve pain and decrease edema.[1,3] In some patients, bone lesions can recur at their original sites or other locations and the disease can have an unpredictable clinical course with remissions and relapses.[1-3]

The patient should be followed to adulthood due to the possibility of mandibular asymmetry, scoliosis due to persistent synostosis of the ribs, persistent synostosis of limb bones, short stature, leg length discrepancy, bowing of long bones, joint extensibility, increased risk of fractures and inguinal hernias.[3,11]

Ethical Disclosures / Divulgações Éticas

Conflicts of interest: The authors have no conflicts of interest to declare.

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Confidentiality of data: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Protecção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos responsáveis da Comissão de Investigação Clínica e Ética e de acordo com a Declaração de Helsínquia da Associação Médica Mundial.

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