CASE REPORT

A localised soft tissue and bone enlargement in an infant mandible

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SUMMARY

A 4-month-old infant was referred by a paediatrician for a rapidly growing swelling on the right side of the mandible of 1.5 months duration. His medical and family history was unremarkable. Palpation divulged a firm and tender enlargement with the overlying soft tissue showing no significant alteration in colour. CT scan revealed cortical irregularity involving the ramus on the right side along with right masseter muscle hypertrophy. Routine haematological investigation yielded values within normal limits except for a raised erythrocyte sedimentation rate. Histopathological examination of the tissue submitted following an open biopsy procedure showed reactive lamellar bone and trabeculae with fibrous marrow exhibiting inflammation. The final diagnosis of infantile cortical hyperostosis was clinched based on the clinicopathological correlation. A rare reactive bone dystrophy which could pose a certain diagnostic dilemma is addressed herewith.

BACKGROUND

Infantile cortical hyperostosis (ICH) also termed as Caffey's disease or Caffey Silverman syndrome, is a rare clinicopathological condition of uncertain aetiology and histogenesis.¹ It affects individuals below 5 months of age and involves bones such as the mandible, ribs, long bones of limbs, clavicle and scapula.² It is characterised by a triad of symptoms which include, soft tissue swelling, cortical bone thickening and irritability in behaviour.¹ It is usually a self-limiting disease with resolution without treatment in 6–9 months,² hence with a good prognosis. A number of reactive, benign and malignant lesions may manifest with periosteal bone reaction and mimic ICH, thus posing a considerable challenge for clinicians to arrive at a diagnosis. However, a thorough history, meticulous clinical examination, basic laboratory investigations and imaging techniques aid in the diagnosis. The awareness of this rare entity in infants would preclude invasive procedures and treatment regime.

CASE PRESENTATION

A 4-month-old infant, presented with a swelling on the right side of the mandible of 1.5 months duration as reported by the parents. The swelling was associated with pain of recent onset accompanied by irritability in behaviour. The infant was born of a full-term pregnancy and had normal growth and development. The history did not suggest of any trauma or injury to the patient or maternal infection during or postpregnancy. The medical records revealed a scheduled immunisation regimen which

was uneventful. There was no alteration in the child's feeding habits. Inspection revealed a 1 cm×2 cm swelling over the right angle of the mandible showing no significant alteration in the colour of the overlying skin or pus discharge (figure 1). Palpation revealed a well-defined, firm and tender enlargement without a local rise in temperature. The right submandibular lymph node was palpable.

INVESTIGATIONS

A CT scan of the affected area revealed cortical irregularities involving the mandibular ramus on the right side predominantly in the outer cortex. The right masseter muscle was bulky with a surrounding hypodense collection, measuring 3.7 cm×2.8 cm (figure 2). A three-dimensional reconstruction of the same showed cortical irregularities on the right ramus, with the left ramus being unaffected (figure 3). Routine haematological investigation revealed values within normal limits except for a raised erythrocyte sedimentation rate (ESR) of 42 mm/h.

Surgical exploration of the involved site performed under general anaesthesia revealed bone that was porous and flaky. Microscopic evaluation of the decalcified bone fragments showed trabeculae of woven bone interspersed with fibrous vascular marrow (figure 4). Some spicules of lamellar bone could also be appreciated. The trabeculae were lined by osteoblasts and occasional osteoclasts (figure 5) and the fibrovascular connective tissue

Figure 1 Extraoral swelling 1×2 cm in the right angle of the mandible.

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Figure 2 CT scan of the mandible (axial view) showing cortical irregularities involving the ramus of the mandible over the right side, predominantly in the outer cortex. The right masseter muscle is bulky with a surrounding hypodense collection, measuring 3.7 cm×2.8 cm.

displayed varying degrees of inflammation (figure 6). The lymph node was of normal architecture. Based on the clinical presentation supported by imaging and histological findings, the diagnosis of ICH was made. The chest and limb radiographs taken subsequently did not show any abnormality thus ruling out generalised involvement.

DIFFERENTIAL DIAGNOSIS

A number of conditions present with a swelling of the jaw in an infant. The conditions that were considered in the differential diagnosis based on the clinical findings included vaccinia osteomyelitis, ICH, monostotic fibrous dysplasia, cherubism and bone malignancy.

Vaccinia osteomyelitis, an interesting but rare form of osteomyelitis in children following vaccination with a live or attenuated viral vaccine like BCG³ was considered as the patient had received the vaccination as per schedule. This condition most commonly involves the bones of the extremities and less often other bones like the scapula, vertebrae, ribs, sternum and

Figure 3 Three-dimensional reconstruction of the CT scans showing cortical irregularity on the right ramus of the mandible.

Figure 4 Photomicrograph showing trabeculae of woven bone interspersed with fibrous vascular connective tissue (H&E stain, ×10).

clavicle. However, jaw involvement is very rare. Its clinical features include loss of function and limited motion with pain and swelling in the affected area⁴ as was seen in this case.

ICH, an inflammatory disorder of infants, which is characterised typically by a triad of manifestations namely bone changes, soft tissue swelling and irritability¹ was considered, as these features coincided with the presentation of our patient.

Cherubism, a benign fibro-osseous lesion resulting in progressive, painless, symmetrical expansion of the jaws in children was included in the differential diagnosis.⁵ However, a tender unilateral swelling of the jaw with no family history of a similar swelling did not go in favour of this lesion.

Monostotic fibrous dysplasia, which is a benign sporadic fibro-osseous lesion involving a single bone occasionally affecting the maxillofacial region was also taken into consideration. It typically causes a painless swelling of the affected area and is usually diagnosed in the second and third decade of life.⁶ However, the tenderness of the enlargement and the age of presentation in this case did not support monostotic fibrous dysplasia.

The possibility of a malignancy could not be ruled out because of the rapidly enlarging, tender lesion. Ewing sarcoma and osteosarcoma, two commonly occurring bone tumours in children were also thought of. The absence of small, blue round

Figure 5 Bony trabeculae lined by osteoblasts with occasional osteoclasts (H&E stain, ×40).

Figure 6 Inflammatory component in the connective tissue (H&E stain, $\times 10$).

cells and bone cells not displaying any atypical features precluded the diagnosis of malignancy.

Differentiating these lesions is of prime importance as the treatment regimes and the management of each of these entities vary considerably. Thorough histopathological investigations helped in clinching the final diagnosis.

TREATMENT

No treatment was rendered to the patient.

OUTCOME AND FOLLOW-UP

The child is under regular follow-up for the past 9 months and has shown improvement since then and is free of recurrence or new lesions.

DISCUSSION

ICH was first reported as a disease entity by Caffey and Silverman in 1945.¹ It is a rare condition that occurs in infants below 5 months of age.² There is no predilection for sex, race or social status and the exact aetiology of this condition is still unknown. However, some of the proposed causes are infections, immunological defects, primary arterial abnormalities and genetic abnormalities.⁷ Its characteristic triad of manifestation of soft tissue swelling, underlying cortical bone thickening and irritability in behaviour, was seen in our case. Other symptoms like fever, pseudoparalysis and dysphagia may also be present.²

This condition may be sporadic or familial in type.¹ Most cases are sporadic, with an average age of onset of about 9–11 weeks.² This was in accordance with our case where the parents noticed the lesion when the child was approximately 2 months old. The mandible is the bone most frequently affected in this type, and its involvement is almost always pathognomonic,⁸ as was true in this patient. However, the latter has an earlier onset, with the bones of the lower extremity being affected more often.²

Haematological examination of these patients may reveal anaemia, moderate leukocytosis, thrombocytosis, increased ESR, C reactive protein and alkaline phosphatase levels.⁹ None of these findings were noted in our case except for raised ESR.

Radiographical evaluation is crucial in the diagnosis of ICH. On radiographs the periosteal new bone formation with periosteal soft tissue thickening can be appreciated.⁸ This condition typically causes hyperostosis in the mandible and diaphysis of the tubular bones with the epiphysis and metaphysis being

spared.¹⁰ Bone scintigraphy is another useful diagnostic tool in the detection of this lesion where more radiotracer uptake is noted in the areas of periosteal reaction.⁸

The exact pathogenesis of this disease still eludes us but it is said to progress in three stages. In the early acute stage of the condition, inflammation of the periosteum occurs following which the periosteal envelope disrupts to blend with contiguous tissues (fasciae, muscle and tendon). In the subacute phase, the periosteum re-establishes itself, beneath which new subperiosteal bone formation takes place. Profuse layers of new bone are deposited causing thickening of the bone and the new layers become incorporated into the original cortex. Finally in the late remodelling stage, signs of inflammation disappear and excess bone is removed.¹¹

The clinical course of this disease is highly variable. It is usually self-limiting with resolution without treatment within a year.² Accordingly no treatment was administered in our case. But the inflammatory nature of ICH has led to treatment trials with anti-inflammatory agents such as steroids like prednisolone and non-steroidal anti-inflammatory drugs such as indomethacin, which have been reported to provide symptomatic relief to some patients.⁷ Despite its inherent self-limiting nature, recurring forms of this disease may occur and persist for many years. The condition may flare up and subside repeatedly, sometimes occurring at new sites. Continued episodes of the disease may eventually lead to deformities like interosseous bridging, dislocation, asymmetry and bowing of the bones.¹¹ The oral manifestations in severe cases include deformity of the mandible with severe malocclusions.² An inconsistent and unpredictable clinical course is one of the characteristic features of this condition.¹¹ Hence a regular follow-up is advisable.

Learning points

- Infantile cortical hyperostosis is a rare inflammatory disorder of infants, characterised by soft tissue swelling, cortical bone thickening and irritability in behaviour.
- A rare reactive bone dystrophy which could pose a certain diagnostic challenge could be addressed with by a careful clinical examination and relevant imaging techniques.
- This condition is usually self-limiting with resolution without treatment in 6–9 months. But a regular follow-up deems necessary.
- The awareness of this rare entity in infants would preclude invasive procedures and treatment regime.

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