Case 1271

Pyknodysostosis with mandibular osteomyelitis
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Case Type: Clinical Cases
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Patient: 14 years, female

Clinical History:

The patient was admitted complaining of pain, swelling, and pus drainage at the right mandibular area for one week. She was bilaterally blind, and she had suffered multiple fractures at different ages.

Imaging Findings:

The patient was admitted complaining of pain, swelling, and pus drainage at the right mandibular area for one week. She was bilaterally blind, and she had suffered multiple fractures at different ages. She was only 140 centimetres tall. The right mandibular area had been draining pus for one year intermittently (Fig. 1). No apparent reason for the bone exposure could be elicited, and microbiological culture was negative for pathogens. Intraoral examination showed malodour, class II malocclusion, left 33-34 teeth transposition, multiple decayed and discoloured teeth, malposition, and delayed eruption the lower right first molar tooth was extracted. Orthopantomographic examination indicated unclear border radiolucency and sequestering at the right mandible, and a localised loss of lamina dura. The mandible was small with obtuse mandibular angles and retroggnathism, numerous unerupted teeth in the bicuspid region, some of which were supernumerary, and impacted molars (Fig. 2).

The craniofacial examination revealed open cranial sutures, Wormian bones, increased density and thickening of the basis of the skull, hypoplastic maxillary and frontal sinuses and mid-face hypoplasia in the lateral and antero-posterior skull radiographs.

The skeletal survey showed a generalised increase in bone density and multiple fractures (Fig. 3). Definite shortening of the long bones and the terminal phalanxes was observed. A three-phase bone scintigraphic study indicated slightly increased tracer uptake in the right mandibular angle (Fig. 4). The results of standard laboratory tests were within normal limits. The patient was treated surgically and appropriate antibiotics were administered.

Discussion:

Discussion Pyknodysostosis is a rare autosomal recessive disorder. It is considered to be a variation of osteopetrosis or even a cleidocranial dysostosis (1). The disorder was first described and named by Maroteaux and Lamy in 1962 (2). The differential diagnosis of pyknodysostosis includes cleidocranial dysostosis, osteopetrosis and acroosteolysis. The most commonly encountered problem in pyknodysostotic patients is fracture of the long bones resulting from relatively mild trauma. This patient had a history of multiple fractures of the extremities. Increased fragility of bone and osteomyelitis are common findings in osteopetrosis and pyknodysostosis due to replacement of the bone marrow by bony trabeculae (1,3). Three cases was reported concerning osteomyelitis of the mandible following tooth extraction in the literature (1,4). Ilankovan and Moos (3) suggested that the surgical procedures in the patient who had pyknodysostosis, enhance the possibility of infection, impaired wound healing, osteomyelitis and pathological fracture. Karkabi et al (5) concluded that increased frequency of osteomyelitis in pyknodysostosis after
tooth extraction was mainly due to impaired killing activity of monocytes. In this case, osteomyelitis developed after
 tooth extraction also, the patient’s lower first molar was extracted about one year ago. Extensive dental caries,
 enamel hypoplasia, delayed eruption and teeth malposition are the common oral findings of the disease (3).
 Histologically, the Haversian system appears very dense, but the features that differentiate pyknodysostosis from
 true osteopetrosis are the presence of demonstrable medullar cavities in the long bones and active medullary
 haemopoiesis (4). Anaemia or hepatosplenomegaly, as seen in osteopetrosis, are not features of pyknodysostosis.
 This patient had normal blood results. Osteopetrosis can result in constriction of the cranial foramina, with
 subsequent loss of hearing, optic nerve atrophy, facial nerve palsy, hydrocephalus, and mental retardation, none of
 which, however is a constant feature (1). This patient had a history of blindness at birth. There has been no case
 report including the bilateral congenital blindness associated with mandibular osteomyelitis and pyknodysostosis. A
 detailed evaluation of skull base foramina and optic canals was not performed in this case.

**Differential Diagnosis List:** Pyknodysostosis with mandibular osteomyelitis

**Final Diagnosis:** Pyknodysostosis with mandibular osteomyelitis

**References:**

Description: Lateral photograph showing an oral cutaneous fistula on the right side (arrow heads) and mandibular retrognathism. Origin:
Figure 2

Description: Panoramic radiograph showing fistulous tract with destruction of inferior border of right mandibular molar region (arrowheads) and obtuse mandible. The right mandibular first molar tooth is absent. Delayed eruption and teeth displacement is seen. Origin:
Figure 3

Description: Anteroposterior pelvis radiography showing generalised increased bone density and bilateral multiple old fractures at the collum femoris bilaterally (arrow heads). Origin:
Description: Scintigraphy showing slightly increased tracer uptake same in the right mandibular area (arrow heads) and other fracture areas. Origin: