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Intractable bimaxillary osteomyelitissecondary to pyknodysostosis

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Abstract
Pyknodysostosis is a rare, sclerosing bone disorder with an autosomal dominant trait. The syndrome as such is not recognized easily and given the diffuse osteosclerosis prevalent, complications such as osteomyelitis and pathological fracture are not uncommon. However, given the diversity and aggressiveness of the oral microbiota, maxillofacial infections pose a much larger and imminent threat. We present a case of intractable bimaxillary osteomyelitis in a38 year old male patient- misdiagnosed as osteopetrotic and review the clinical and radiographic findings.

Keywords: Pyknodysostosis, osteomyelitis, pathologic fracture.

1. Introduction
The term *pyknodysostosis*, denotes *pyknos* (thick or dense), *dys* (defective), and ostosis *(bone)* [2]. Prior to its discovery as a separate entity by Maroteaux and Lamy in 1962, the condition was classified in the same subgroup as Albers-Schonberg disease, deemed to be an atypical presentation of cleido-cranial dysostosis and osteopetrosis. The disorder, also known as Toulouse-Lautrec syndrome, was named after the famous French artist who was thought to be afflicted with pyknodysostosis.

Parental consanguinity has been recognized and accepted as primary etiology. However, it is inherited as an autosomal recessive trait, with mutation in the cathepsin-K gene and is characterized by failure in resorption and remodelling of primary spongiosa by osteoclasts, often resulting in resulting in excessive mineralization [1]. The resulting bone is brittle and pathological fractures and osteomyelitis have been known to occur. Since the bone marrow is displaced by newly formed bone, infections of the oral cavity have also been known to result in osteomyelitis that is very difficult to control, and on some occasions, even death [2, 3].

Clinical presentation of pyknodysostosis is varied and includes dysplasia of the skull with frontal and parietal bossing, open fontanelles, hypoplastic paranasal sinuses, with partial or total aplasia of the terminal phalanges. Morphologically, the skull appears to be disproportionate when compared to the facial bones. The eyes often appear exophthalmic, and the nose has frequently been described as “parrot beak”. Various other orthopedic presentations include pectus excavatum, bowing of long bones, bilateral genu valgum, *Clavicular hypoplasia*, kyphosis and scoliosis [4, 2, 5, 3].

An increase in bone fragility, increased bone density and osteomyelitis are common characteristics in both pyknodysostosis as well as osteopetrosis. They are both characterized bythe same basic histopathologic picture- replacement of the bone marrow by bony trabeculae. In view of this, the chief differentiating factor is perhaps the presence of medullary cavity as well asactive medullary hematopoiesis [1, 5, 3]. Another differentiating factor is in the fact that osteopetrosis is known to cause constriction of the cranial foramens, with subsequent disorders in hearing and facial expressions, optic nerve atrophy, hydrocephalus and mental retardation. Anemia and hepatosplenomegaly, suggestive of extramedullary hematopoiesis, are present in osteopetrosis [5, 3].

The purpose of this report is to present a case of pyknodysostosis presenting with classical craniofacial findings and intractable recurrent bimaxillary chronic suppurative osteomyelitis subsequent to multiple dental extractions and sequestrectomies. In the present case, the patient was misdiagnosed as osteopetrotic for 20 years, but after thorough clinical examination, we

were able to confirm the diagnosis of pyknodysostosis.

2. Report of a case

A 38 year old male, 4 feet 3 inches tall was referred to the maxillofacial service at our University hospital for evaluation and treatment of draining extra oral fistulas – one in the left submandibular region and another at the right infra orbital rim. (Fig 1 A, B) He walked with a Cane. He was micrognathic, had frontal and parietal bossing, a hypoplastic clavicle, a parrot-like nose, exophthalmos, a hypoplastic mid face, an asymmetric mandible and spoke with a nasal twang.

In his history, the patient stated that he had been well until the age of ten years – after which he had been diagnosed as osteopetrotic. The patient's first admission was at the age of 17 – for fracture of his right and left ulna and radius. After which, the patient underwent numerous sad missions for fractures of his pelvis, and right fibula. Given his osteopetrotic history, all of these injuries were treated with closed reduction and subsequent immobilization. The fractures were all noted to heal within normal periods, but most with mal-union. Approximately 2 years ago, the patient underwent extraction of multiple decayed teeth in his upper and lower jaw. Despite being on antibiotics, six months later, the patient noted pain and swelling in relation to the right side of the mandible. Numerous decayed teeth were extracted, sequestra removed and multiple attempts at rehabilitation were carried out. Maxillary extractions had also been carried out since then, with relatively minor problems. Within a year's time, an extra oral fistula developed, which remained persistent despite numerous attempts at curettage.

On presentation to our department, intraoral examination revealed a small, "V" shaped, grooved high palatal vault, severe malocclusion, the region of the left parasymphysis, body and ramus ofthe mandible were denuded of mucosa, with decayed, necrotic stumps of bone protruding intothe oral cavity - irritating the lateral rim of the tongue where significant swelling of the surrounding mucosa was obvious. (Fig 2 A) The suppuration thus extended to involve the entire left half of the mandible, with pathological fracture of the left parasymphysis. (Fig 2 B) On further examination, there appeared to be an additional area of denuded, necrotic bone with anoro-antral communication in his right maxilla in the region of the first molar.

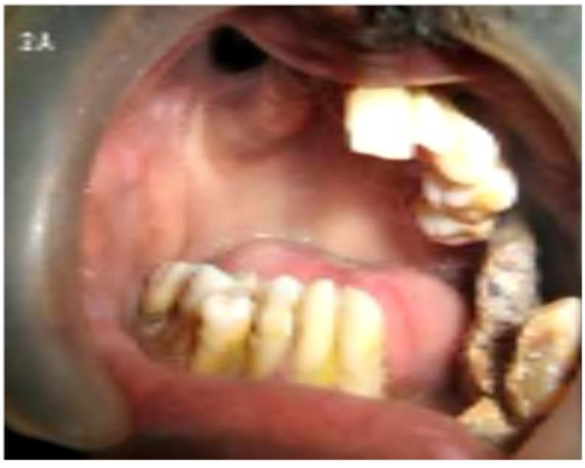


Fig 1 A: Front view of the patient with the infra orbital draining sinus

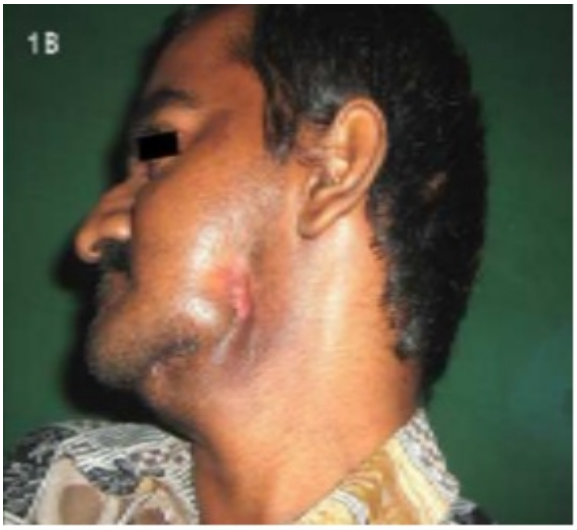


Fig 1 B: Lateral view with sub-mandibular sinus

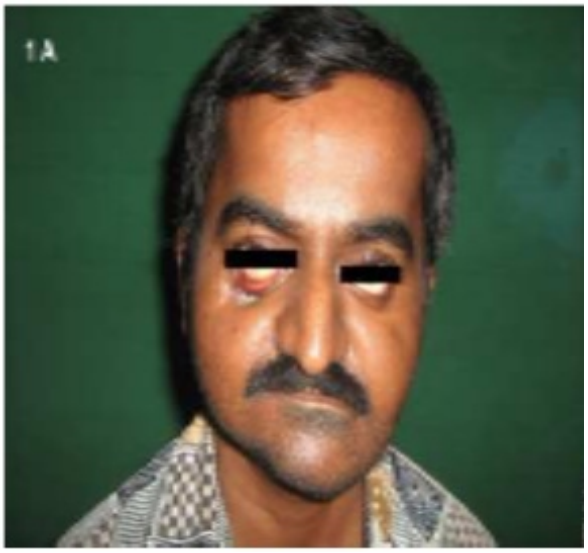


Fig 2 A: mandibular osteomyelitis with pathologic fracture (left), oro-antral Communication(right) and a midline grooved palate.

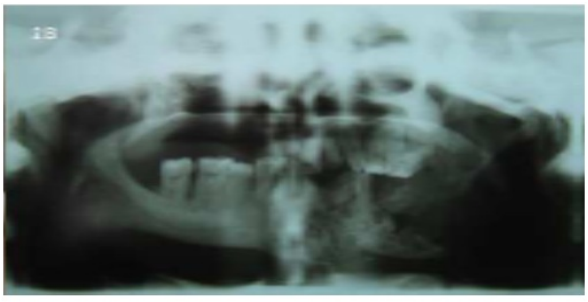
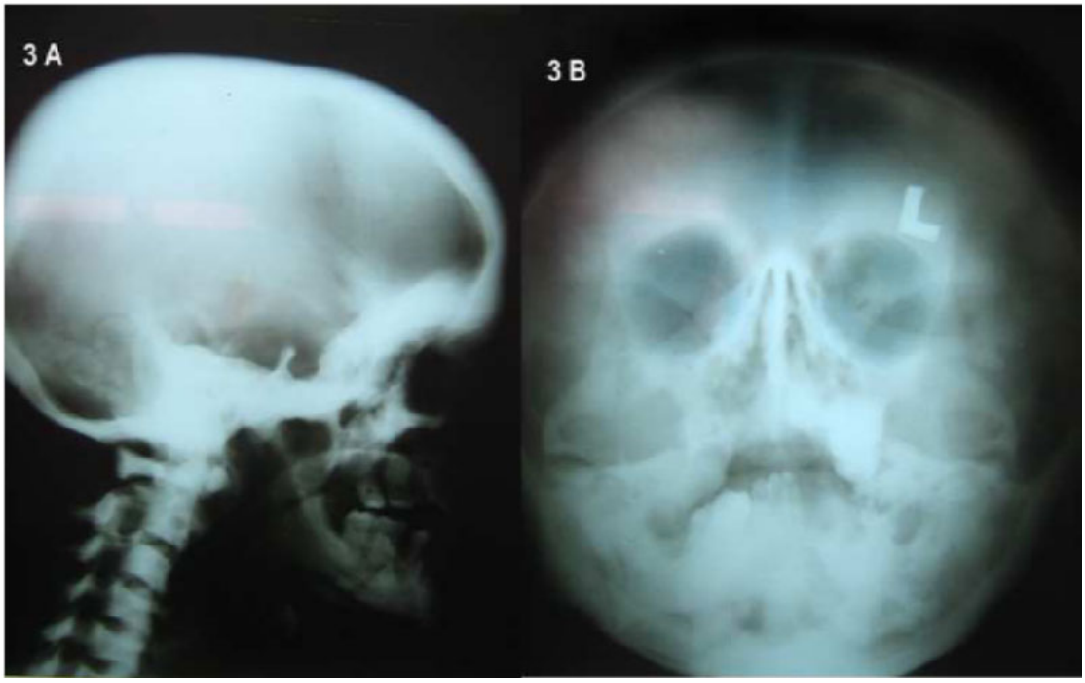


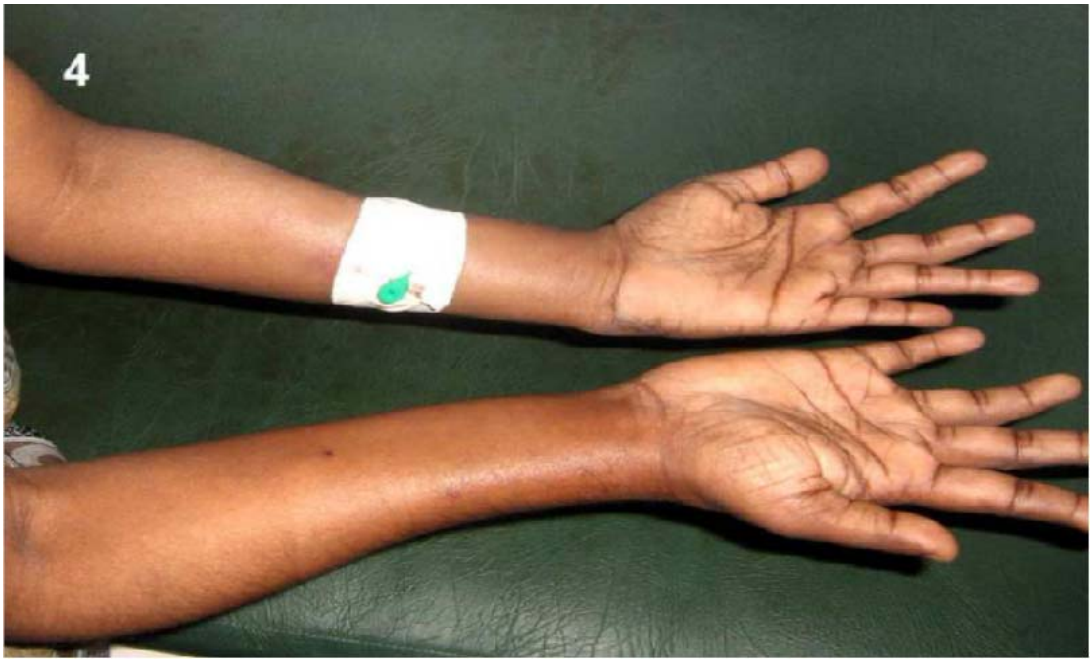
Fig 2 B: Panoramic radiograph

3. Radiographic findings

Skull radiographs showed open sutures and fontanelles, hypoplastic mid facial bones and an obtuse mandibular gonial angle, sclerosis of the skull base and the maxillary sinus lacked normal pneumatization. (Fig 3 A, B) In addition the orthopantomograph revealed an abnormal mandible on the left; with considerable modelling deformity, altered texture with mixed sclerotic and radiolucent texture to it. There is an area

of necrosis extending from the symphysis across the left body of the mandible, extending into the ramus-condyle unit with pathological fracture in two separate regions – most likely in keeping with chronic suppurative osteomyelitis. There are smaller areas of radiolucency suggestive of erosions and the sclerosis representing longstanding insult- perhaps resulting in osteoblastic response. (Fig 2 B) The patient was advised selective surgical debridement under local anaesthetic, but, was unfortunately lost to review.





4. Discussion

Clinical features of pyknodysostosis have been known to overlap the more common osteopetrosis and cleidocranial dysostosis. It is believed that the first case description was as early as 1923 by Montanari in an 18 year old patient who has an unusual variant of achondroplasia [6]; however, it was not until 1962 that Maroteaux and Lamy defined the characteristic features of pyknodysostosis. Thereafter, diagnosis of pyknodysostosis has been reported in patients from the age of 9 months onwards [6, 7].

In our case, from his early adulthood, the patient was diagnosed as osteopetrotic. The disorder was re-diagnosed as osteopetrotic when he was treated for fractured radius and ulna bones non-operatively and subsequently in the following years for closed reduction of multiple fractures. In keeping with the same diagnosis, treatment was carried out in all 4 episodes of osteomyelitis of jaws complicating tooth extractions or repeated sequestrectomy of both maxilla (right side) and mandible (left side) beginning approximately 2 years ago.

However, on clinical and radiological examination, various features typical of pyknodysostosis were observed which made it possible to establish this diagnosis over osteopetrosis. First, the patient's dwarfism (4 feet, 3 inches) is a characteristic feature of pyknodysostosis that is quite rare in osteopetrosis

where the stature is normal. Soliman *et al*, in their review describe this to becaused by an increase in bone volume of the sella turcica, which causes compression on the pituitary gland, causing hypoplasia of this gland, and a deficient production of growth hormone [8].

Frontal and parietal bossing encountered in the patient is more prevalent in pyknodysostosis than in osteopetrosis. Clinically, along with the complete absence of nerve compression or anemia, hepatosplenomegaly and normal serum alkaline phosphatase and calcium supported our diagnosis of pyknodysostosis. Midface hypoplasia, and groovy midline of the palate along with hypoplasia of teeth and hypercementosis of the patient are also commonly noted features of pyknodysostosis. The patient's radiological evaluation also revealed open fontanelles, failure of closure of cranial sutures, lack of pneumatization of paranasal sinuses, obtuse mandibular angle in conjunction with pseudoprognathism, and increased osseous radiodensity. None of the patient's relatives had any features of the disorder. But a point of interest is that, although outpatient exhibited classical signs of pyknodysostosis, there appeared to be no abnormality in his the length of his arms and the hands' phalanges were of normal adult length. (Fig 4)

Although there are several reports of osteomyelitis developing in a clinical setting of

Pyknodysostosis [9, 10, 5, 3] none appear to be as severe or as mutilating- as seen in our case. The patients' age, repeated extractions, multiple sequestrectomies, poor oral hygiene - all without adequate antibiotic prophylaxis could have all been individual contributing factors to his condition. However, at the same time, it is prudent to remember that osteomyelitis in pyknodysostosis is considered refractory because of diffuse osteosclerosis, and given the aggressiveness of the oral microbiota, maxillofacial infections pose an imminent threat. Gradual elimination of medullary spaces in the jaws severely reduces and compromises the reserves ofthe patient's local immune defences against infection, sometimes even leading to death [3].

In conclusion, as authors, we would like to stress the importance of recognizing the potential threat this condition presents and perhaps the need to aggressively control osteomyelitis in a clinical setting.

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