Pyknodysostosis : A case report with review of literature

Abstract:

The postnatal emergence of short limbs, small stature, and global hyperostosis, combined with acro-osteolysis with sclerosis of the terminal phalanges, are the hallmarks of a rare autosomal recessive ailment called pyknodysostosis. This condition is virtually pathognomonic. The wormian bones, obtuse mandibular gonial angle with relative mandibular prognathism, persistence of fontanelles, delayed closure of sutures, and absence of frontal sinuses are other characteristics. Here, we describe a case of pyknodysostosis that was discovered to exhibit the traditional characteristics both during the general physical examination and intraoral examination.

Key-words: Pyknodysostosis, Dysmorphy, Frontal bossing, Deafness

Introduction:

The rare clinical condition known as pyknodysostosis was initially reported by Maroteaux and Lamy in 1962. Toulouse-Lautrec syndrome is another term for the illness, named for the French artist Henri de Toulouse-Lautrec, who is thought to have had the condition. The identification of the faulty gene causing pyknodysostosis in 1996 allowed for a more precise diagnosis, carrier screening, and comprehensive comprehension of this condition. The occurrence of this autosomal recessive condition, which is predicted to affect 1.7 out of every million newborns, is typically detected at a young age.[1]

Midface hypoplasia, prominent cheekbones, high nasal bridge, frontal bossing, beaked nose, mandibular projection or retraction, and micrognathia linked to malocclusion, including open bite and crossbite, are among the facial features.[2-4]

Case Report:

A 55 year old male reported to the Department of oral medicine and Radiology complain with discoloration of teeth since 5-6 months and wants them to cleans it .Personal history reveals patients brushes with neem sticks and not brushes regularly

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Clinical findings:

Under physical examination, the following conditions were noted: dwarfism, brachydactyly with dysplastic nails, sandal gap deformity of the toes [Figure 1A and B], enlarged skull with frontal and occipital bossing, midfacial hypoplasia, beak-shaped nose, proptosis with downward-slanting eyes, blue sclera, overhanging columella, and hypoplastic, micrographic maxilla. It felt soft, pliable, and sensitive to the touch. Pectus carinatum (pigeon chest) refers to a chest wall deformity in which the sternum protrudes anteriorly Figure 1C and D],intraorally staining of teeth with slight high arch palate Figure [1E and 1F] 1A

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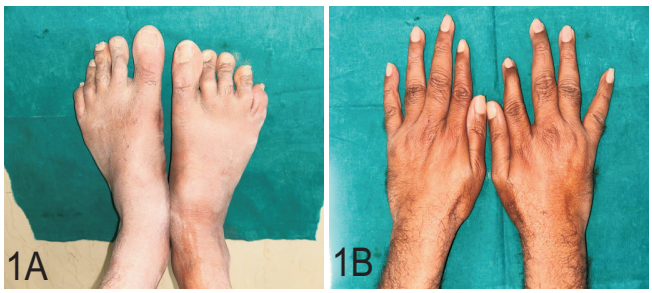

Figure 1: (A) Brachydactyly of hands and dysplastic nails. (B) Brachydactyly of feet, dysplastic nails, and sandal gap deformity

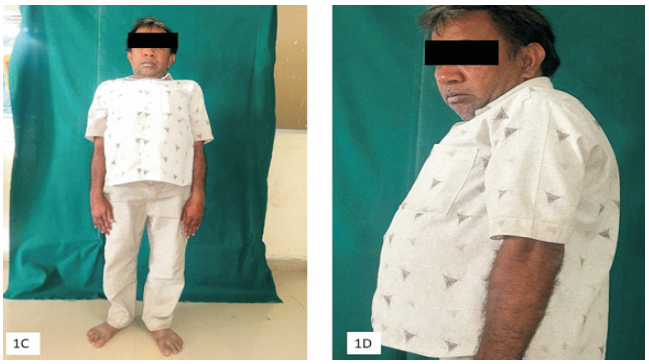

Figure 1c and 1d frontal bossing, midfacial hypoplasia and pigeon chest

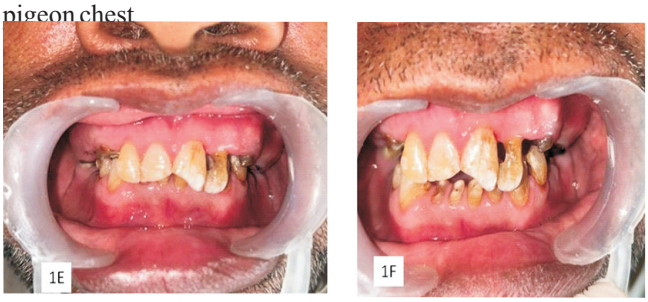

Figure 1Eand 1F Intraorally staining of teeth ,with slight high arch palate

Diagnosis assessment:

Serum creatinine, serum calcium, serum phosphorus, serum alkaline phosphatase, kidney function test, liver function test, and total blood count were among the blood studies performed. The lateral cephalogram [Figure 2A] and PA (Postero-Anterior) skull [Figure 2B] showed a wide cranial vault with non-pneumatized paranasal sinuses, frontal and occipital bossing, open anterior and posterior fontanelles, and hypoplastic facial bones. Exams using radiography, such as the orthopantomagram (OPG) showing reduced mandibular height, obtuse gonial angle, and. [Figure2C], revealed diffuse sclerosis. obtuse mandibular angle. Hand-wrist [Figure2D] and foot radiographs displayed marked with acro-osteolysis. Postero anterior view of lower limbs showed sclerotic tibia and fibula [Figure2E]

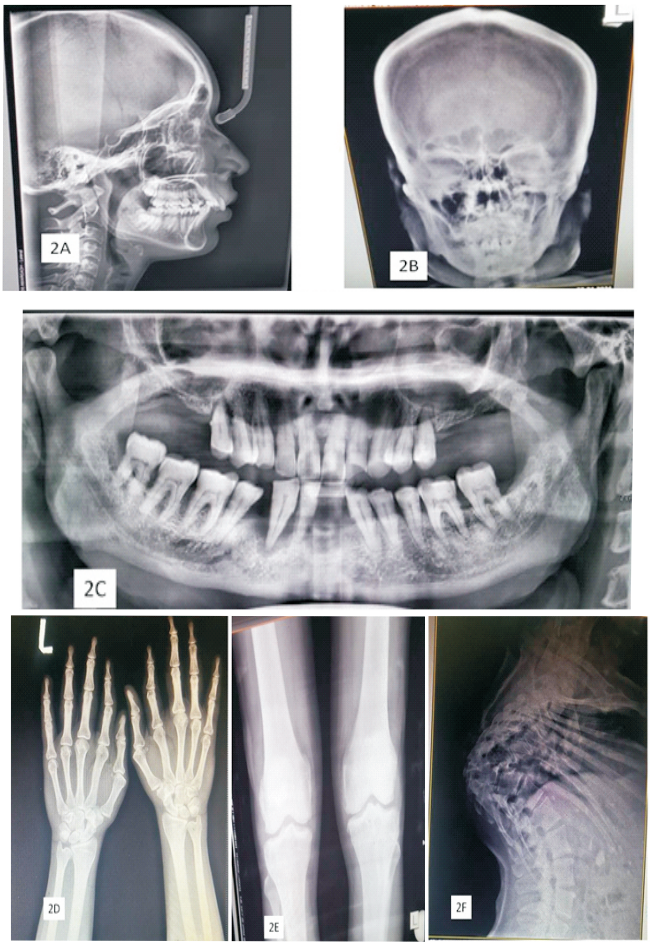

Fig 2 A and 2B implies frontal and occipital bossing, open anterior and posterior fontanelles and the the orthopantomagram (OPG) showing reduced mandibular height, obtuse gonial angle, and with revealed diffuse sclerosis Fig 2 C. Hand-wrist [Figure2D] and foot radiographs displayed marked with acro-osteolysis. Postero anterior view of lower limbs showed sclerotic tibia and fibula [Figure2E] Figure 2F pigeon chest reveals a mild anterior protrusion of sternum deformity of chest wall.

Discussion:

An alteration in the gene that codes for the bone-forming enzyme cathepsin K results in pycnodysostosis, a hereditary bone condition. In order for osteoclasts—normal bone cells—to reabsorb into the bone and regenerate new bone, they need this enzyme. Because pycnodysostosis patients lack cathepsin K, their osteoclasts' usual activity is impaired, making it difficult for the patients to appropriately reabsorb

the organic matrix, a component of bone.. Remodeling is another name for this process, which is essential to good bone health. Due to this inadequate re-absorption mechanism, the bones of people with pycnodysostosis are unusually thick and brittle.

The differential diagnosis of pyknodysostosis should take into account a number of bone disorders, including osteopetrosis, acroosteolysis, cleidocranial dysostosis, and osteogenesis imperfecta. Whereas pycnodysostosis is autosomally recessive, cleidocranial dysostosis is inherited by autosomal dominant heredity.

The short stature with dwarfism is a constant feature; it was present in our case and also in 90.32% according to data from the literature ranging from (-1.5 DS to -5 DS) [2, 3] Through our review of the literature, we found the following main features [2]; open fontanels and cranial sutures ; grooved palate; midfacial hypoplasia ; acro-osteolysis of distal phalanges ; narrow palate ; open bite . Our patient had the same characteristics except parietal bossing and open fontanels. The main radiographic maxillofacial features were : obtuse mandibular angle , large head with frontal, parietal and occipital bossing Life expectancy for a pycnodysostosis patient is normal[3]

The CTSK gene mutation analysis is the confirming test for pycnodysostosis, however clinical characteristics and radiographs are the main sources of diagnosis. Numerous new cathepsin K gene mutations in pycnodysostosis patients have been documented in the literature. Due to the distinctive phenotypical appearance of proportionate dwarfism and dysmorphic facial features, the disease is typically diagnosed at an early age.[6] However, due to a propensity for infections and fractures, it is occasionally (as in our case) discovered too late. With chronic open fontanelles and cranial sutures, it presents similarly to pycnodysostosis. However, it invariably affects the clavicle, which is a bone that is rarely impacted in pycnodysostosis.[5] Moreover, it does not lead to an overall increase in bone density.[6] Many fractures are seen in people with osteogenesis imperfecta, but they are more severe than in those with pycnodysostosis, and they are accompanied by characteristics such choanal atresia.

Characteristic clinical and radiographic features of pycnodysostosis are summarized in Table 1.[5-10]

Table 1. Characteristic clinical and radiographic features of pycnodysostosis[5-10]

Clinical features	Radiographic findings
General	Short stature with proportionate dwarfism
	Generalized osteosclerosis
	Multiple fractures of varying ages with sparing of the medullary cavities of long bones
Head and neck	Large head with frontal and parietal bossing
	Sclerotic calvarium and skull base
	Hypoplastic facial bones with dysmorphic features
	Incomplete pneumatisation of the paranasal sinuses
	Persistent open anterior fontanelle
	Obtuse mandibular angle
	Micrognathia
	Dental abnormalities
	Snoring and stridorous breathing
Thorax	Narrow chest, Aplastic acromion and/or distal clavicles
	Pectus excavatum
Spine and pelvis	Kyphoscoliosis
	Increased lumbar lordosis
	Dense vertebral bodies with sparing of the transverse processes
	Failure of complete segmentation of the atlas, axis and lower lumbar vertebrae
	Spoon-shaped vertebral bodies
Limbs	Joint hypermobility, Widening of the distal femur

The radiological and clinical findings listed above serve as the main basis for the diagnosis. Nevertheless, an examination of cathepsin K gene mutation.The gold standard confirmatory test is still[5,6]. Since there are no particular choices for therapy, the most crucial components of clinical care are fracture management and prevention. Preventing problems can also be achieved with good dental care and routine examinations.[5]

Clinical and radiological findings stated above are the main basis for the diagnosis. The most reliable confirming test, meanwhile, is still a cathepsin K gene mutation analysis[5, 6]

No specific treatment options exist, so treatment is supportive, with fracture prevention and management constituting the most important aspects of clinical care. Additionally useful in avoiding problems are good oral hygiene and routine examinations. It is crucial to take precautions to avoid or lessen the likelihood of bone fractures since they provide the biggest risk to individuals with Pycnodysostosis. Exercise and safe, low-impact activities are among of these preventative measures, as is handling an afflicted youngster with caution. For those who are impacted because of different dental defects, dental hygiene and routine examinations are extremely beneficial.[5]

At present, no specific treatment is present for PYCD (Pycnodysostosis) patients, and only symptomatic and preventive treatment is practiced. However, there are no serious complications and life expectancy is good in such patients.[5]

Data from the literature present the occurrence of this disorder in the Indian population in the last 5–6 years [Table2] with different positive finding[11-22]

Table2: Clinical features of the patients with Pycnodysostosis in Indian Population.

Cases in Indian population							
Publication	**Number of cases**	**Mean age/ sex**	**Consanguinity**	**Open fontanelles**	**Acro-osteolysis**	**Malocclusion and dental manifestation**	**Osteomyelitis**
Bathi et al 2000[11] *Int. J. Oral Maxillofac. Surg.*	2 cases	38 both male	-	Open anterior and posterior fontanelles	Present	Present	Present
Rajan *et al.* January 201512] *Medical Journal Armed Forces India*	1 case	12/ female	Absent	Anterior fontanelle open	Present	Present	-
Rohit *et al.* January 2015[13] *Journal of Clinical and Diagnostic Research*	2 cases	38 1-male 1-female	Present in female case	Not commented	Present	Present	Present
Bhardwaj *et al.* November 2015[14] *Journal of Indian Academy of Oral Medicine and Research*	1 case	50/male	Absent	*Wormian bone involved*	Present	Present	-
Mandal *et al.* March 2016[15] *Clinical Dysmorphology*	5 cases	5 4-male 1-female	Present in 1 case	Open anterior and posterior fontanelles	Present in all 5	Present	-
Muralidhara *et al.* December 2016[16] *International Journal of Research in Medical Sciences*	1 case	35/male	Not commented	Open anterior and posterior fontanelles	Absent	Present	Present
Nandhini.S *et al.* 2017[17] *Journal of Advanced Pharmacy Education and Research*	1 case	20/ female	Not commented	Anterior open fontanelles	Present	Present	-
Dhameliya *et al.*, 2017[18] *Contemporary Clinical Dentistry*	1 case	25/ female	Not commented	Anterior open fontanelles	Present	Present	Present
Bhoyar *et al.* Dec 2020[19] *Contemporary Clinical Dentistry*	1 case	58/male	Absent	Open anterior and posterior fontanelles	Present	Present	Present
A. Sharma *et al.* *Journal of Oral Biology and Craniofacial Research* (2021)[20]	1 case	35/male	Present	Open anterior fontanelles	Present	Present	-
Yadav *Journal of Primary Care Dentistry and Oral Health(2023)*[21]	1 cases	21/male	Absent	Open anterior fontanelles	Present	Present	-
Patil et al *Journal of Indian Academy of Oral Medicine and Research(2024)*[22]	1 cases	20/Female	Absent	Open anterior and posterior fontanelles	Present	Present	Present

Conclusion:

The foundation for diagnosing this illness is the clinical and radiological characteristics. Making the diagnosis as soon as feasible helps to plan the course of therapy and gives patients a higher quality of life.

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