# Unraveling the Puzzle: Pyknodysostosis A Rare Case Report

**Dr. Kusha Dhrangadhariya, [1] Dr. Siddharth Modi,[2] Dr. Angel Aghera, [3] Dr. Honey Raval [4]**

1. **Dr. Kusha Dhrangadhariya**
   PG Student, Oral Medicine and Radiology, Ahmedabad Dental College and Hospital, Near Hare Krishna Mandir, Bhadaj Ranchodpura Road, Ahmedabad, Gujarat 382115
2. **Dr. Siddharth Modi**
   Senior Lecturer, Oral Medicine and Radiology, Ahmedabad Dental College and Hospital, Near Hare Krishna Mandir, Bhadaj Ranchodpura Road, Ahmedabad, Gujarat 382115
3. **Dr. Angel Aghera**
   Reader, Oral Medicine and Radiology, Ahmedabad Dental College and Hospital, Near Hare Krishna Mandir, Bhadaj Ranchodpura Road, Ahmedabad, Gujarat 382115
4. **Dr. Honey Raval**
   PG Student, Oral Medicine and Radiology, Ahmedabad Dental College and Hospital, Near Hare Krishna Mandir, Bhadaj Ranchodpura Road, Ahmedabad, Gujarat 382115

**CORRESPONDING AUTHOR**

**Dr. Kusha Dhrangadhariya**

PG Student, Oral Medicine and Radiology,

Ahmedabad Dental College and Hospital, Near Hare Krishna Mandir,

Bhadaj Ranchodpura Road, Ahmedabad, Gujarat 382115

**Mobile -** +91-7622819001

**Email -** kushasoni21@gmail.com

**Abstract**

Pyknodysostosis is a rare autosomal recessive disorder, first described in 1962 by Maroteaux and Lamy. The disorder characterized by the postnatal onset of short limbs, short stature, and generalized hyperostosis along with acro-osteolysis with sclerosis of the terminal phalanges, a feature that is considered essentially pathognomonic. Other features include persistence of fontanelles, delayed closure of sutures, wormian bones, absence of frontal sinuses, and obtuse mandibular gonial angle with relative mandibular prognathism. Here, I present a case having classical features of pyknodysostosis.

**Keywords:** Pyknodysostosis, Autosomal recessive, Obtuse mandibular gonial angle

## INTRODUCTION

Pyknodysostosis is a rare sclerosing bone disorder and derives its's name from the Greek word "Pykons" which means 'dense' and is a condition of abnormally dense bone.[1] It is a rarely reported variation of osteopetrosis described for the first time in 1962.[5] Before 1962, this syndrome had been considered as "cleidocranial dysostosis", "a type of osteopetrosis" or a combination of these two Maroteaux and Lamy reported consistent variations of these disorders, which were felt to be sufficiently characteristic to constitute a new syndrome, which they termed Pyknodysostosis.[7] In 1996, the defective gene responsible for pyknodysostosis was

located, offering accurate diagnosis, carrier testing, and a more thorough understanding of this disorder. It is an autosomal recessive osteochondrodysplasia, usually diagnosed at an early age with incidence estimated to be 1.7 per 1 million births. Pyknodysostosis is a lysosomal storage disease of the bone caused by a mutation in the gene that codes the enzyme cathepsin K (CTSK).[2,9] The disease has also been named Toulouse-Lautrec syndrome, after the French artist Henri de Toulouse-Lautrec, who (it has been surmised) suffered from the disease.[1,3] In pyknodysostosis there is continuous endosteal bone formation without concomitant bone resorption and remodeling. The resulting bone is brittle and susceptible to fractures especially the long bones and vertebrae.[1] Although fractures of extremities are a frequent and inevitable element of the disease in the childhood, with time they become of minor importance as the affected individual realizes that certain activities should be avoided which can predispose to trauma and fracture.[1] The skulls of these patients are dolichocephalic with prominent frontal and occipital bossing.' Other skull anomalies seen on radiographic examination include an open anterior fontanelle, hypoplastic paranasal sinuses, wormian bones in the lambdoidal area, small facial bones, and hypoplasia of the angle of the mandible.[8] The trunk is not shortened, but often exhibits marked, pectus excavatum with underdeveloped breasts in women. The terminal digits of the fingers and toes are reduced and widened, presenting a characteristic drumstick appearance. Mild syndactyly has also been noted and on occasions, the distal phalanxes are absent. The clavicles are frequently aplastic at the acromial ends. Parrot like nose is a constant finding. Facial bones are usually under developed with pseudoprognathism. Obtuse angle of the mandible is an almost a constant finding.[1,5,6,8,10]

Oral and dental anomalies include an obtuse mandibular angle; underdeveloped facial bones, often with relative mandibular prognathism; persistence of deciduous teeth; premature or delayed eruption, malalignment; enamel hypoplasia; high arched and a grooved palate.[4]

## CASE REPORT

A 13 year old hindu female patient named residing at Ahmedabad belonging to middle socio-economic status came to Ahmedabad dental college and hospital with a chief complain of unaesthetic appearance since birth. Patient gave history of neonatal jaundice at the time of birth and was hospitalized at that time. Patient also had history of fracture of bones twice in her life at a very young age of one and a half year and two and a half year with minimal trauma.

Patients face was bilaterally asymmetrical. Frontal bossing, Slanting of right side of eye, Flattened zygoma with hypoplastic maxilla was seen. Facial profile was convex. Parrot beak shaped nose was seen. Base of the nose appeared broader. Nasal septum was deviated towards the left side. Hands and feet, digits, toes and clavicle appeared normal.

**Fig**: **1, 2, 3** Extra oral clinical photographs

Intraorally, maxillary and mandibular teeth crowding was present with anterior cross bite. High arched palate was seen with a groove present on hard palate running antero-posteriorly from incisive papilla to soft palate. Constricted maxillary arch was noticed.

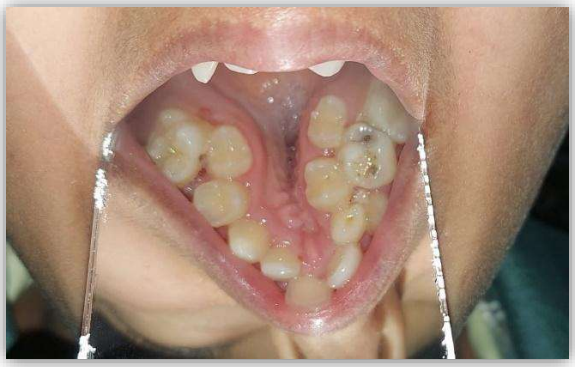

**Fig: 4, 5** Intraoral clinical photograph

Based on history and clinical examination, the provisional diagnosis was pyknodysostosis. Differential diagnosis includes cleidocranial dysostosis, crouzen syndrome.

Investigations included opg, PA skull and CBCT

An orthopantomograph showed retained deciduous teeth were present irt 65 and 73. All second premolars and second molars having open apex. Both right and left mandibular condyle appears hypoplastic. Maxilla and maxillary sinus appeared hypoplastic. Mandibular ramus height was reduced with obtuse mandibular angle.

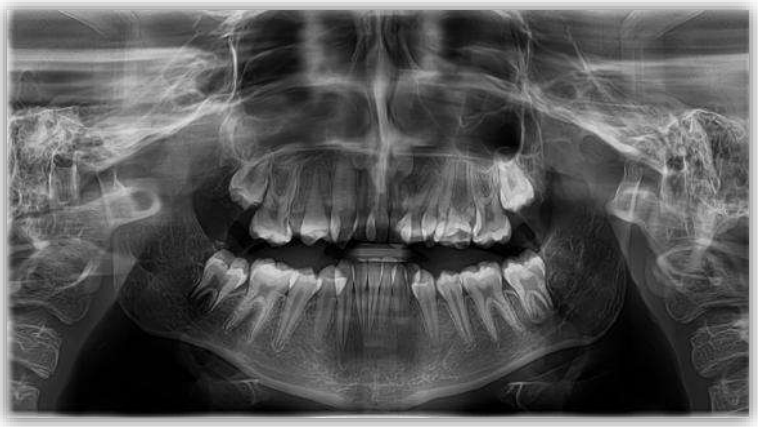

**Fig 6:** Panormic Radiograph (OPG)

PA skull view showed maxillary and frontal sinus extending to clavicle. Right side of maxillary sinus appeared more dense. Clavicle appears normal.

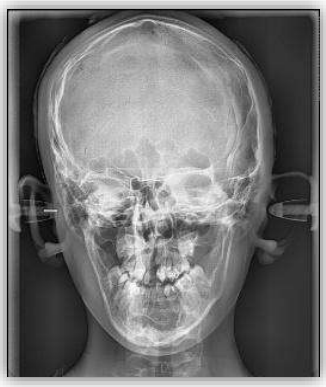

**Fig 7:** PA Skull

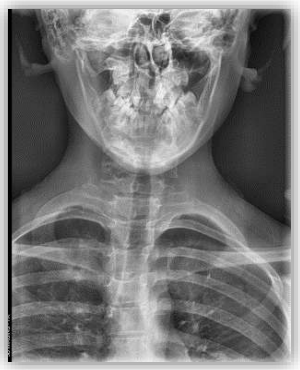

**Fig 8:** PA Chest

PA chest view showed normal development of clavicles.

Sections of CBCT shows palatal groove on hard palate. Maxillary arch is contricted. Right side of maxillary sinus appears filled and hypoplastic.

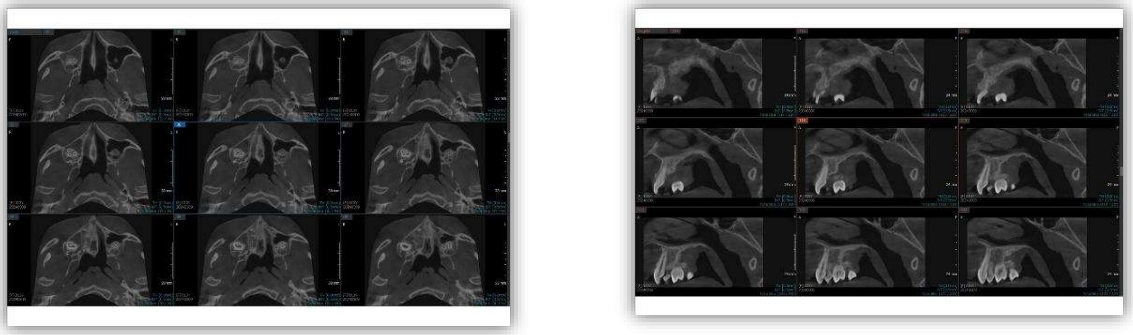

**Fig 9:** Cone beam computed tomography (CBCT)

## DISCUSSION

The case reported the physical and radiographic findings bore a striking resemblance to the classical features of pyknodysostosis as described in the review of literature.[1,5,6,8,10] Frontal bossing was seen. Base of the nose was broad with parrot beak shaped. The height of the ramus was reduced, with obtuse angle of the mandible. Patient was having history of long bone fracture. Intraorally, there was a central groove in the hard palate with constricted maxillary arch. Radiographs showed characteristic increase in the bone density of right maxillary sinus, hypoplasia of maxillary sinus and mandibular condyle, reduced ramus height and obtuse angle of the mandible. Clavicle appeared normal.

Pyknodysostosis should be differentiated from other syndromes having characteristic features closer to this syndrome which includes cleidocranial dysostosis, osteopetrosis, and crouzen syndrome.[1]

In Cleidocranial dysostosis the characteristic features of dense fragile bones and tapering of phalanges are absent.[1] Moreover, cleidocranial dysostosis and crouzen syndrome have an autosomal dominant inheritance where as pyknodystosis shows an autosomal recessive trait.[3,1] In crouzen syndrome, there is hypoplastic maxilla, deviated nasal septum and parrot beak shaped nose is seen same as pyknodysostosis.[6] Important features that differentiate pyknodysostosis from crouzen syndrome are palatal groove and history of fracture of long bones.[6]

## CONCLUSION

There is no specific treatment as of date for this disorder and treatment is supportive. . Crowding can be dealt with planned extraction of retained deciduous teeth. Orthodontic treatment is not recommended because low remodeling capacity of the bone puts the patient at high risk of orthodontic failure but in this case at present orthodontic treatment is running with controlled forces to reduce the chances of fracture. Life expectancy for a Pyknodysostosis patient is normal.

**Conflicts of Interest**

None

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