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**RESEARCH ARTICLE**

# A VERY RARE CASE OF PYCNODYSOSTOSIS WITH CONCURRENT OSTEOMYELITIS OF MANDIBLE

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**ABSTRACT**

Pycnodysostosis is a rare clinical entity, first described in 1962 by Maroteaux and Lamy. It is an autosomal recessive genetic disorder characterized by osteosclerosis, brittle bones, short stature, delayed closure of the fontanels, wide lambdoidal sutures and premature synostosis of the coronal suture, clavicular dysplasia spondylolysis and acroosteolysis. This paper presents a very rare case of a patient with Pycnodysostosis, with concurrent osteomyelitis and review the relevant literature.



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## INTRODUCTION

Pycnodysostosis is also known as Toulouse-Lautrec's disease (Christos, 2002). Robert Weismann-Netter in 1962 is first credited with the description of the disorder. Pycnodysostosis (PYCD) is an autosomal recessive osteosclerotic skeletal dysplasia characterized by susceptibility to fractures, short stature, delayed closure of the fontanels, wide lamboidal sutures and premature synostosis of the coronal suture, craniofacial abnormalities clavicular dysplasia spondylolysis, and acroosteolysis (Maroteaux and Lamy, 1962; Warman *et al*., 2011). Oral findings include delayed tooth formation and eruption process, short and poorly shaped roots, micrognathia with an obtuse mandibular angle, deep and narrow palates, and, consequently, malocclusion, including cross-bite, anterior open bite, and crowding.

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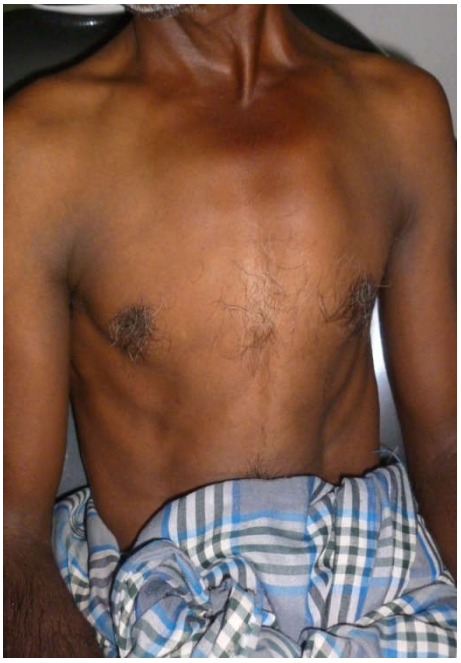
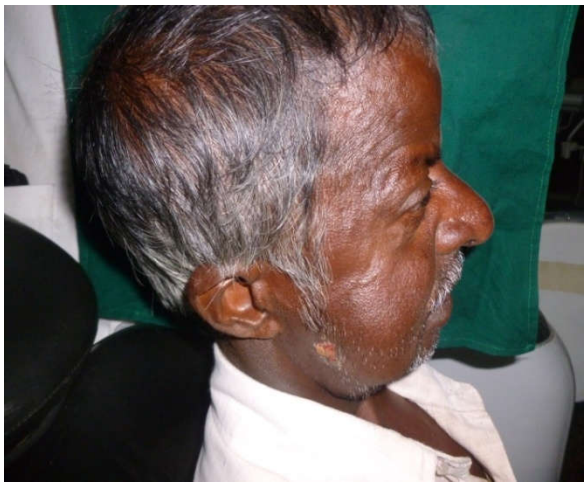
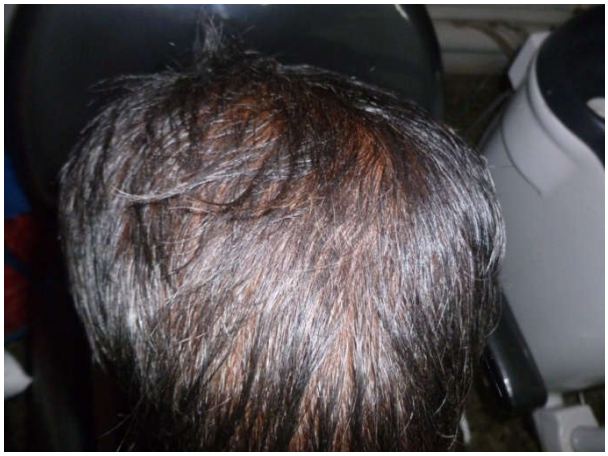
Poor oral hygiene, periodontal disease, and dental caries are common. Affected patients are susceptible to pathological fractures and osteomyelitis of the maxillary bones (Rajeev puri *et al*., 2013). The objective of this study was to report the general and craniofacial features, risk factors of pathological bone fractures and chances of osteomyelitis of the jaw bone in a patient with PYCD.

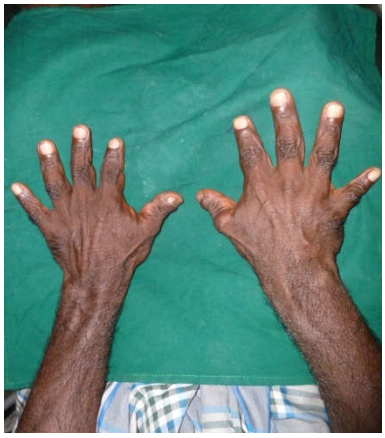
### Case report

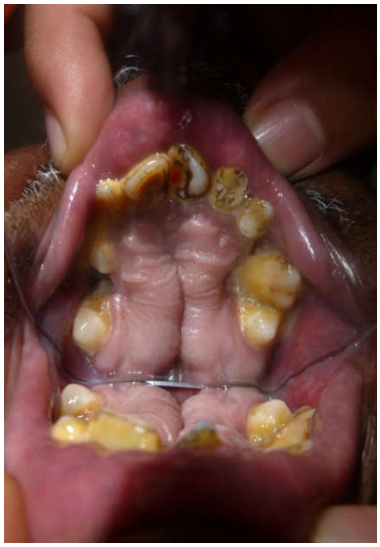
A 35 – year old male patient reported to the Department of Oral Medicine and Radiology, with a complaint of pain and swelling in his right lower back tooth region since a month (Fig.1 & Fig.2). Patient gave a history of extraction done on the lower right back tooth region three months back and later noticed swelling associated with pain in the same region. The swelling was sudden in onset, rapidly progressed in size with no signs of any regression.





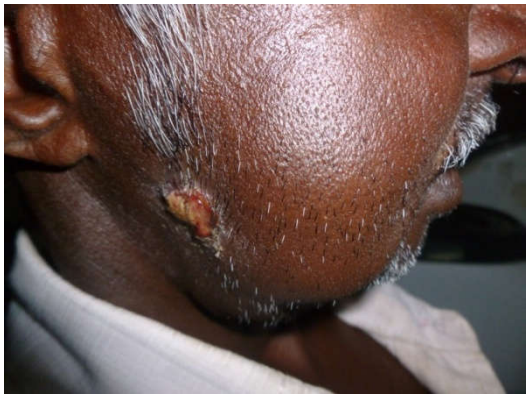


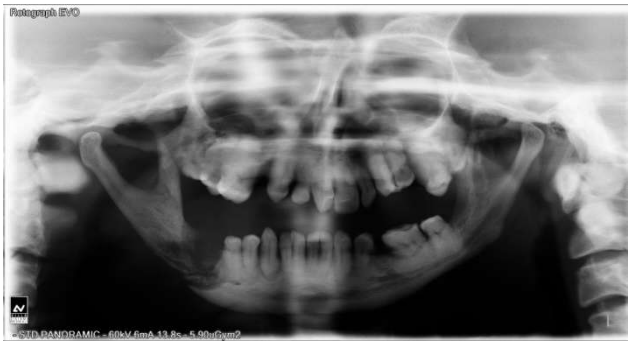


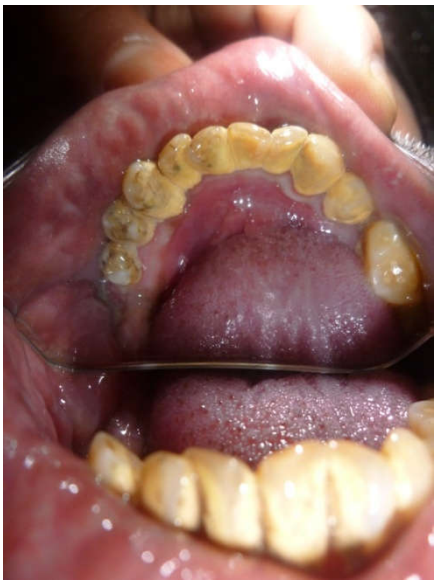


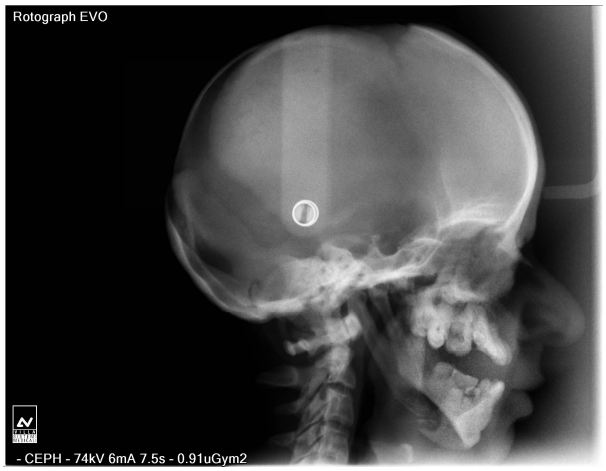
The pain was continuous, dull in nature, which relieved on intake of medication. The Medical history was contributing to the present complaint, where he reports history of fracture of his leg bones following minor fall. General physical examination found the patient appearing more aged compared to his actual age, severely wrinkled skin, very short stature with exophthalmic orbits and short digits (Fig.3). He had a slightly tilted rib cage (Fig.4) with altered gait and a brachycephalic head (Fig.5) Poorly nourished, poorly built, well oriented to time and place with normal vital signs. Examination of the swelling- was diffuse, solitary, present at the right lower third of the face measuring approximately 3 x 4 cm.

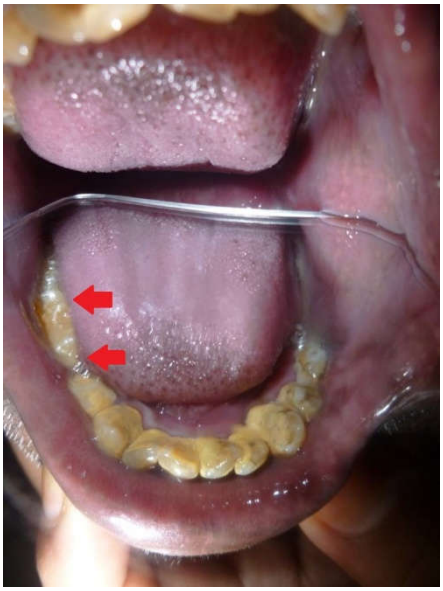
It extended antero – posteriorly from the right corner of the mouth to the angle of the mandible and superior– inferiorly from 1 cm inferior to the Zygoma to 2 cm below the inferior border of the mandible.

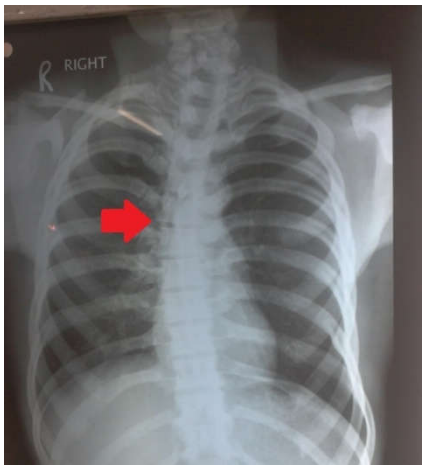


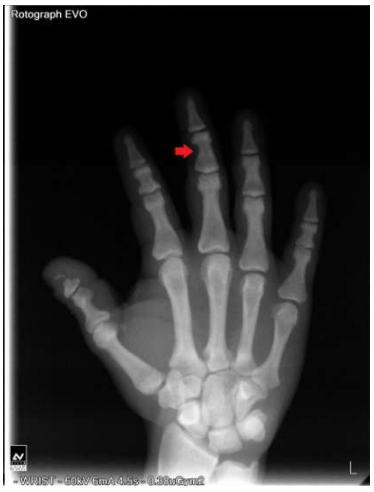




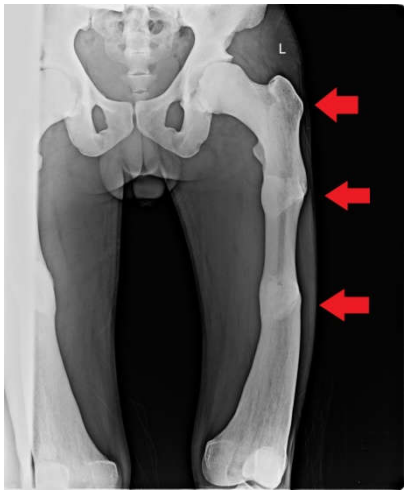


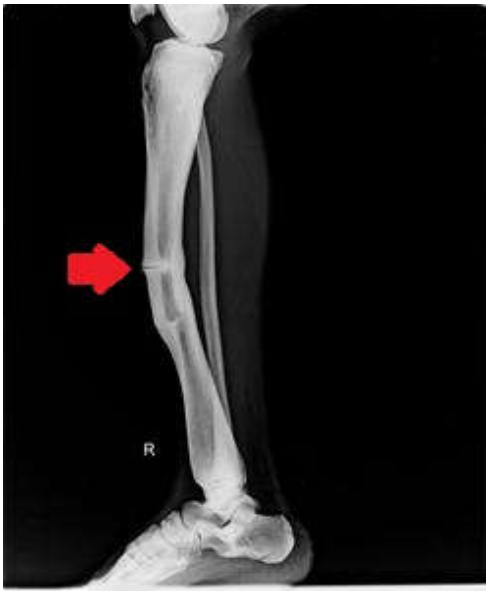



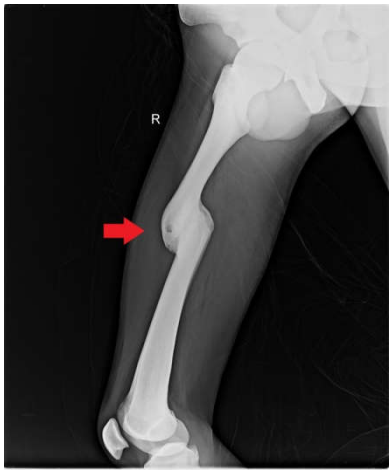


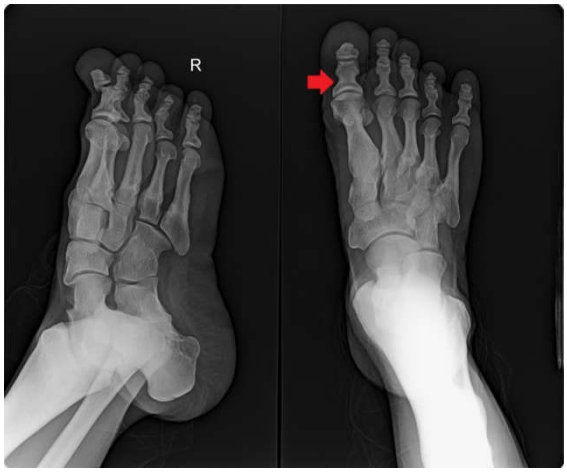



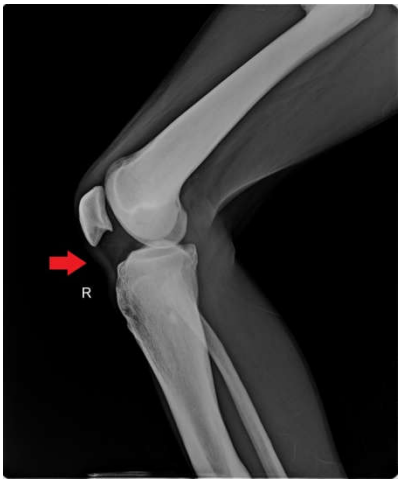

The surface of the swelling had central crustation with active sinus opening with pus discharge (Fig.6). On palpation it was firm in consistency, severly tender, fixed to the underlying bone, with evidence of serosanguinous discharge.. Right submandibular lymph node was palpable, mobile, and tender. Mouth opening was normal with slight deviation of the jaw to the right, associated with but poor oral hygiene.

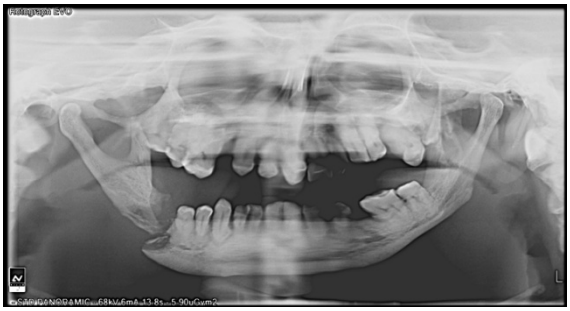


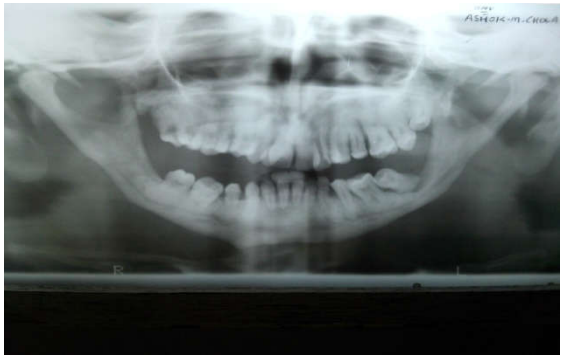





The maxilla and mandible were hypoplastic, with a narrow–arched palate, midline groove was also observed in the palate, suggestive of a submucous cleft (Fig.7). The teeth were periodontally compromised, with severe malocclusion (Fig.8).Examination of alveolus in the region of 46-48 region, presented with a non healing ulcer of the extraction site, measuring approx 2x3cm in size covered with pseudo-membranous slough with active sign of pus discharge (Fig.9). On palpation severe tenderness of the ulcer and the surrounding swelling areas of the mandible noticed with altered neural sensations around the lesion.

**Table.**

| Features | Pycnodysostosis | Cleido-cranial dysplaria | Osteopetrosis |
|---|---|---|---|
| Base of skull | Dense | Normal /rarely dense | Dense |
| Cranial sutures | open | Normal | Normal |
| Paranasal sinuses | Unaerated or closed | Normal | Unaerated |
| mandible | Loss of angle | Normal | Normal |
| Clavicle | Present, sometimes dysplastic | Absent or dysplastic | Present and normal |
| Hand & feet | Aplastic tufts, short phalanges, overriding nails | Normal | Normal |
| Pelvis | Coxa plana | normal | Coxa vara |
| Spontaneous fracture | Present | absent | Present |
| Bone texture | Dense without obliteration of intramedullary canals | normal | Dense with obliteration of intramedullary canals |
| Blood findings | Normal | normal | Aplastic anemia |
| Genetic | recessive | dominant | Dominant, anaemic type: recessive |
| Stature | Short | Normal | Normal |

A provisional diagnosis of chronic suppurative osteomyelitis of the mandible was considered, further examination included panoramic and lateral skull radiographs. OPG findings revealed partially edentulous arches, with short dentitions associated with generalized hypercementosis. There was generalized horizontal & vertical bone loss with decreased vertical heights of the mandible. The gonial angle of the mandible was found flattened bilaterally with the evident of narrow & sharper coronoid and condylar processes. Distal to 45 tooth, there was an ill-defined diffuse saucerised complete radiolucency seen which was extending inferiorly to the lower border of the mandible along with pathological fracture. Bony erosions of the alveolar crest were also seen and the mandibular canal was intact (Fig.10).

Lateral skull radiograph showed persistent opening of the fontanelles & occipitalis. Cortical thickening was also seen at the frontal & occipital bones (Fig.11). The variation in General physical appearance of the patient, instigated to take additional radiographs in the other regions of the body like chest, hand-wrist, and long bones of leg, pelvis & feet. Chest radiograph showed scoliosis of the spine & slight curvature of the rib cage towards right side (Fig.12).Hand-wrist radiograph showed curvature & resorption of the phalanges (Fig.13). Right & left femur bones showed mal-union & remodeling of the bones in different regions (Fig.14 & Fig.15). There were no bony changes in pelvic region. Tibial radiograph showed resorption in the knee joint region (Fig.16) and pathological fracture lines & bowing of the bones (Fig.17) and also feet radiograph was taken which showed resorption of the phalanges (Fig.18) as same as seen in the hand-wrist radiograph. Based on all the clinical & radiological findings, a diagnosis of Chronic Sclerosing Osteomyelitis of right mandible was given associated with Pycnodysostosis Syndrome. The patient was advised antibiotic & analgesic and recalled after 10 days for surgical treatment procedure, but he did not report back.

After 2 months, patient came back to the department with chief complaint of recurrent pain and swelling in the same region. On local examination, there was complete absence of pus discharge from the previous ulcerative lesion with persistent swelling. OPG showed complete pathological fracture of the right side of the body of mandible with displacement of the segments at the 46 tooth region (Fig.19). Later patient was unavailable for treatment and follow up.

## DISCUSSION

Pycnodysostosis is an autosomal-recessive disorder of osteoclast dysfunction due to mutation of cathepsin K gene causing osteosclerosis. This disorder is seen in 1.7 per 1 million birth. In this disorder, sclerosing activity takes place in the bone structure due to a genetic defect, located on chromosome 1q21. This syndrome consists of 12 different mutations that produce mutational changes in a lysosomal cystine protease, cathepsin K, the expression of which is reduced in the osteoclast cells in the bone. This protease is responsible for degrading collagen type 1, which constitutes 95% of the organic bone matrix (Daniela alves Pereira, 2008). The bones become more brittle, showing slow remodeling & arising high bone density.

The general clinical & oro-facial features of Pycnodysostosis are short stature, fractures of long bones, brachycephaly with frontal & partial bossing, exopthalmic eyes with bluish sclera, underdeveloped facial bones, dental anomalies, poor oral hygiene, mucous cleft in the palate, micrognathia, short & broad digits of hand & feet, scoliosis of the trunk, narrow & curved rib cage are seen (Quais Mujawar *et al*., 2009; www.ajronline.org; Anne *et al*., 1998; Nirupama *et al*., 2013). Radiological findings of long bones may show some degree of widening because of several regions of low bone remodeling. Skull and facial radiographs shows open anterior fontanel, flattened mandibular angle & undeveloped paranasal sinuses. Even vertical height of the mandible & maxilla is decreased with presence of hypercementosis in a short & sharp rooted teeth (Fig.20).

Hand-wrist & feet radiographs show partially or totally aplastic phalanges (Quais Mujawar *et al*., 2009; Ben Barnard and Wim Hiddema, 2012). Cranio-facial analysis demonstrates antero-posterior hypoplasia of maxilla and low anterior face height due to vertical hypoplasia of maxilla and mandible. This analysis also confirmed a mild skeletal class II relationship. The absence of mandibular angle & long soft palate, causing pharyngeal narrowing (Hugo *et al*., 2012). In the present case report, patient was found to have all the clinical & radiological findings as same as Pycnodysostosis syndrome, which helped us to reach the final diagnosis along with diagnosis of chronic osteomyelitis of right mandible. Few authors are of the opinion that maxillary segment is more vulnerable for fracture , later causing osteomyelitis because of the bone remodeling in the

midline palatine suture which results in a generalized increase in bone density and volume, osteosclerosis, reduced medullary space, and increase brittleness (Marcio Vieira Ortegosa *et al*., 2013). Forceful teeth extraction or serial extraction cases can causes fracture of the mandibular bone and later leading to osteomyelitis. In the present case, patient gave history of forceful teeth extraction of his right lower jaw region with non healing sockets later progressed to osteomyelitis of the mandible.

There is no specific treatment for this disorder. Preventive and supportive care is needed to minimize tendencies of bone fracture. Dental hygiene care and fluoride application for children is helpful for protecting dental caries. Fixed orthodontic treatment is contraindicated for this type of patient, which can lead to bone fractures (Hugo *et al*., 2012; Marcio Vieira Ortegosa *et al*., 2013). Proper surgical sequestrectomy and curettage is needed for the treatment of osteomyelitis. Spinal anesthesia is the best option for this patient before undergoing any surgical treatment because intubation for general anesthesia is difficult due to high arched palate, mandibular hypoplasia & large protruded tongue, which made it Mallampati Grade IV (Rajeev puri *et al*., 2013).

On the basis of few similar clinical & radiological features, Pycnodysostosis can be misdiagnosed with Cleidocranial Dysostosis and Osteopetrosis. The Cathepsin K Gene analysis is the gold standard for final diagnosis of this disorder. But in the year 1969, Z Emami Ahari *et al* differentiated these 3 bone disorders (Emami-Ahari *et al*., 1969) on the basis of clinical & radiological features in a tabular form:-

**Conclusion**

Pycnodysostosis is a bone disorder which is susceptible for bone fracture with a minimal trauma and further can cause bone infection, like osteomyelitis. The morbidity and mortality chances are higher in this disorder. There is no such treatment protocol still invented which can cure this disorder because it's a genetic autosomal recessive disorder. So therefore, prevention from any trauma or injury to the bone can give a good prognostic result.

## REFERENCES


Anne, C. 1998. O'Conne, BDentSc, Michael. Brenon, Clair A. FrancomMano. Pycnodysostosis: orofacial manifestations in two pediatric patients. American Academy of Pediatric Dentistry - 20;3.

Ben Barnard, Wim Hiddema. 2012. *The South African Journal of Radiology*, 16(2):74-76

Christos, S. 2002. bartsocas, Pycnodysostosis: Toulouse-Lautrec's and Aesop's disease. Hormones. 1 (4):260-262

Daniela alves Pereira, Leonardo berini aytes, Cosme gay escoda. Pycnodysostosis. A report of 3 clinical cases. *Med Oral Patol Oral Cir Bucal*., 2008 Oct;13(10):E633-5

Emami-Ahari, Z., Zarbi, M. and Javid, B. and Shiraz. 1969. *The Journal of Bone and Joint Surgery*. May; 51B;(2).

Hugo P. Caracas, Paulo S. Figueiredo, Heliana Dantas Mestrinho, Ana Carolina Acevedo and Andre F. Leite. Pycnodysostosis with craniosynostosis: Case Report of the Craniofacial and Oral features. *Clinical Dysmorphology*., 2012, 21: 19-21

Marcio Vieira Ortegosa, De´bora Romeo Bertola, Meire Aguena, Maria Rita Passos-Bueno, Chong Ae Kim, Maria Estela Justamante de Faria. Challenges in the Orthodontic treatment of a patient with Pycnodysostosis. The Cleft *Palate-Craniofacial Journal,* 2013 00(00) pp. 000–000

Muthukrishnan, N. and Shetty, M.V.K Pycnodysostosis* report of a case. www.ajronline.org .VOL. 114, No. 2

Nirupama C, Sarasakavitha D, Shivakumar Palanivelu, Bala Guhan. Pycnodysostosis: A case report of rare Entity. *Journal of Indian Academy of Oral medicine and radiology*, April-June 2013;25(2):00-00

Quais Mujawar, Ravi Naganoor, Harsha patil, Achyut Narayan Thobbi, Sadashiva Ukkali and Naushad Malagi. Pycnodysostosis with unusual findings: a case report. *Cases Journal,* 2009, 2:6544

Rajeev puri, Arpita saxena, Awak Mittal, Zia Arshad, Yogita Dwivedi, Trilok chand. Pycnodysostosis: An Anaesthetic Approach to this rare genetic disorder. Case Reports in Anesthesiology 2013. Article ID 716756;

Yang Xue, Tao Cai, Songtao Shi, Weiguang Wang, Yanli Zhang, Tianqiu Mao and Xiaohong Duan. Clinical and animal research findings in Pycnodysostosis and gene mutations of cathepsin K from 1996 to 2011. *Orphanet Journal of Rare Diseases*, 2011, 6:20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