Medical and Research Publications
International Open Access

Case Report **Journal of MAR Pediatrics (Volume 3 Issue 4)**

Autosomal Dominant Type of Acro Osteolysis

Venugopal Reddy I *[1], Bhaskar Shenoy [2], Sridevi Hegde [3]

1,2,3 Department of Pediatrics, Medical Genetics and Radiology Manipal Hospital, HAL airport road, Bangalore-560017. India.

Corresponding Author: Venugopal Reddy I, Associate Professor, Department of Cardiac Surgery, Imam Reza Hospital, Mashhad, University of Medical Sciences, Mashhad, Iran.

Received Date: January 28, 2023

Published Date: February 10, 2023

Introduction

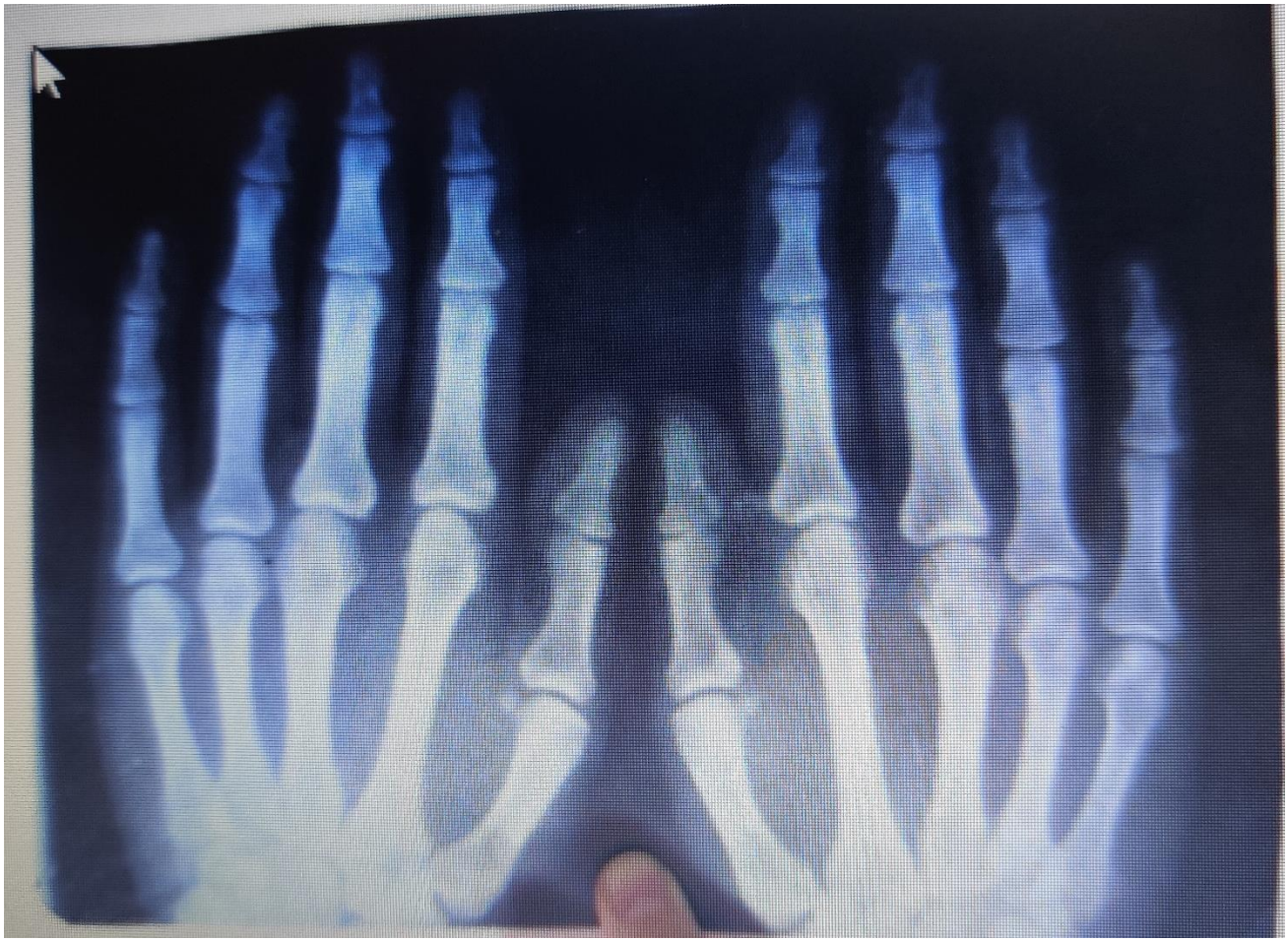
Autosomal dominant type of Acro osteolysis, is a rare autosomal dominant disorder with variable expressivity, characterized by short stature, progressively disappearing distal phalanges and crowded carpal bones. Failure of ossification of cranial sutures can be seen, with thickened cranial vault, absent frontal sinus, elongated Sella turcica, and progressive basilar impression. Hair is thick and straight, eyebrows and eyelashes prominent. Ears are low-set with prominent lobes. The nose is broad with anteverted nostrils. A resorption of alveolar process often induces an early loss of teeth, the vertebrae are biconcave, and osteopenia can lead to collapse and scoliosis is frequent. Several additional manifestations have been described in isolated cases: cystic kidneys, congenital heart disease, hydrocephalus, cleft palate, hepatosplenomegaly. While most patients have a normal intellectual quotient, some have mild mental retardation. Diagnosis is rarely made in childhood and pain is a frequent first manifestation, especially in the hands. The patients are weak, and traumatic fractures often occur. Osseous compression may result in decreased stature, and the basilar compression may be life-threatening. Biochemistry and osteodensitometry reveal excessive bone resorption, with early osteoporosis that might be treated by bisphosphonates. There are only 30 reported cases in the world literature.

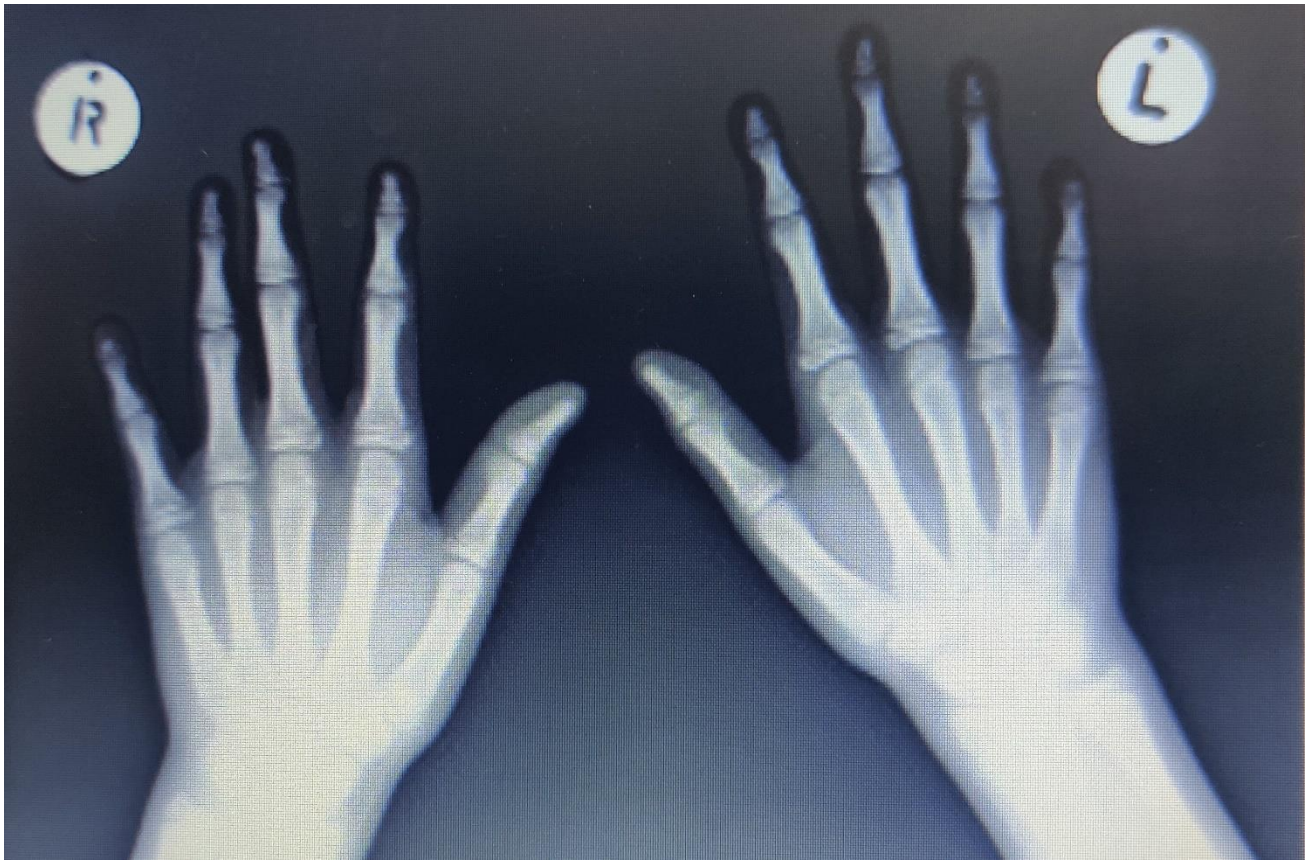
Clinical History

Child is referred to us in view of persistent respiratory difficulties from day 3 of life in the form of noisy breathing, recurrent episodes of cough and multiple congenital deformities were noted.

A 6 years and 5 months old first born female child, born to second degree consanguineous marriage couple at 37 weeks of gestation by a spontaneous vaginal delivery following an uneventful pregnancy with no significant antenatal and perinatal history. Mother is having crowded teeth without any major abnormality and no other family members effected.

Multiple congenital deformities were noted, characterized by distinctive facies, which was dysmorphic with a prominent premaxilla. There is hypertelorism and jaw is small with a thin upper lip. The palate is high arched and the uvula appeared rotated forwards and fixed to the palate. Teeth were abnormal and crowded. Anterior and posterior fontanelle are not closed with short stature (5th centile according to CDC). The arms are short with short stubby fingers with pseudo clubbing. Wide gap is present between first and second metatarsal space. Systemic examination: No abnormality detected.




R
L

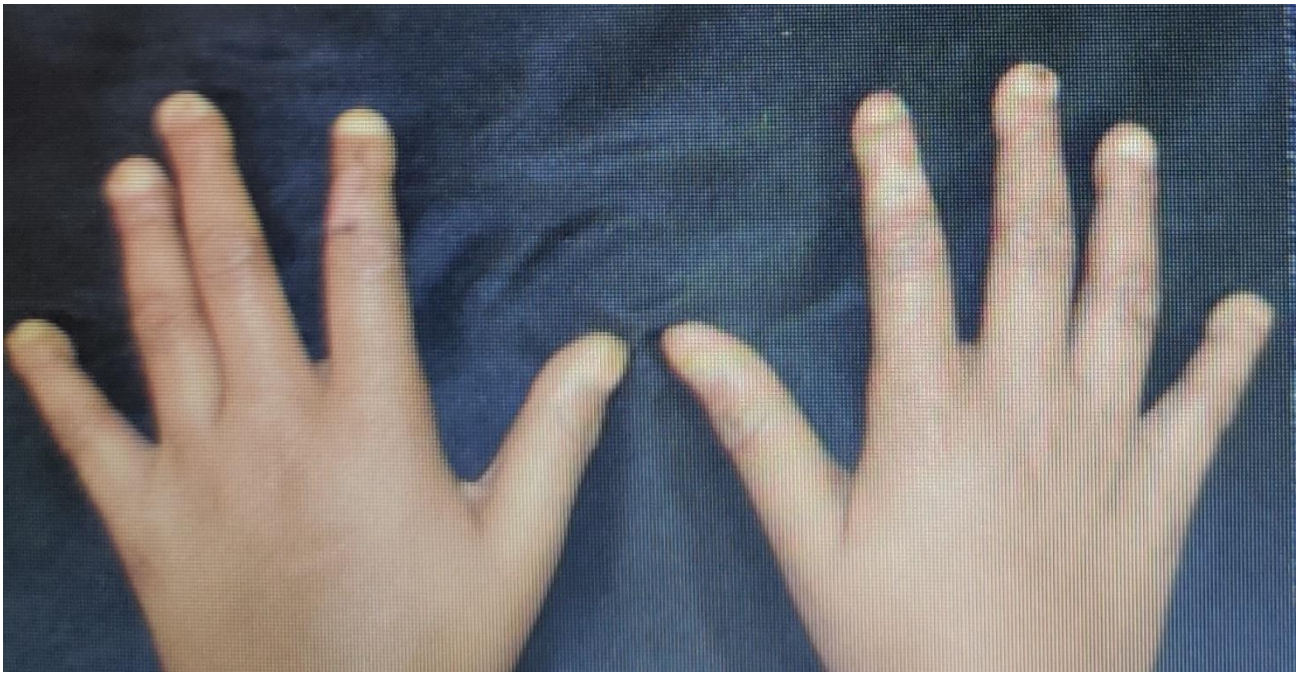


Figure 03. Radioghraph of both hands confirming longitudinal acro-osteolysis.

Radiological findings

The anterior and posterior fontanelle and pterions are open, non-fusion of sutures and few sutural (Wormian) bones noted. The frontal sinus and maxillary sinuses are not well pneumatized. The mandible angle is obtuse with malalignment of dentition in superior and inferior alveolar arch. Soft tissue opacity on posterior nasopharynx - suggestive of adenoid hypertrophy. No obvious fracture / dislocation seen in skull. X-ray of both hands confirming longitudinal acro-osteolysis with no evidence of fracture / dislocation. The bone age corresponds to 5 years (Delayed skeletal maturation) (GP method according to female standard 13). No evidence of fracture / dislocation.

Whole spinal X- rays (anteroposterior and lateral view) are normal.

Differential diagnosis

Acro-osteolysis refers to the dissolution of bone at the phalanges. It is commonly associated with scleroderma, sarcoidosis, neuropathic disorders, and rheumatoid or psoriatic syndromes. It can also be associated with nephropathy in childhood. The pathogenesis may be vascular, neurovascular, traumatic (due to burns and frostbite), toxic (due to PVC or ergot poisoning), metabolic or infective. It may also be idiopathic. The major diagnoses to rule out are Progeria, Pyknodysostosis and the Hajdu-Cheney

syndrome. This child does not display the characteristic facies, skin changes and premature ageing as seen in individuals with Progeria. Pyknodysostosis includes many of the features seen in this child but the differentiating factor between Pyknodysostosis and acro-osteolysis is that in Pyknodysostosis the bones are dense and the mandibular angle is almost 180 degrees. In Hajdu-Cheney syndrome skeletal abnormalities in the form of vertebral anomalies with joint dislocations, neurologic, cardiovascular and renal manifestations are more but in Acro osteolysis mainly bony changes are present.

Discussion

Most cases of autosomal dominant type of Acro osteolysis, are sporadic (12) but inheritance is likely to be autosomal dominant (9,10). Genes associated with this disorder are Notch-2, Cathepsin-K and Cathepsin –K etc(4,13). Although this is a congenital disorder, the correct diagnosis is rarely made until later childhood, when characteristic hand changes occur. The affected child has a distinctive facial appearance, short stature and delayed puberty (1,10). Intelligence is normal. The eyes slope downwards and there is tele canthus and synophrys. Optic atrophy can develop (5,10). The philtrum is long and the nostrils are anteverted (7). The mandible is small, the palate highly arched and the occiput prominent. The neck is short and the ears are low-set (13). Multiple Wormian bones occur in the skull (2,3,5). These are intra sutural bones occurring most frequently along the lambdoidal sutures (2,3). The presence of a few Wormian bones is normal up to the age of six months and only when multiple are they considered significant.

In acro-osteolysis, resorption of bone may occur at the phalangeal tufts, in the mid-portion of the phalanges or at the periarticular surfaces. Most of the rheumatological diseases cause erosions at the distal tufts and surrounding juxta-articular areas (2,3). Other characteristic radiological features such as progressive disappearance of bone is limited to the distal phalanges.

Classic Acro osteolysis develops in late childhood. The terminal phalanges tend to be small and triangular in appearance with an increase in the amount of surrounding soft tissue (9). Sometimes acro-osteolysis takes the form of transverse lytic defects across the shafts of the phalanges (8). The feet are less severely affected. Pain is a frequent manifestation, especially in the hands. A mnemonic commonly used for acro-osteolysis is PINCHFO. Pyknodysostosis, Psoriasis, Injury (thermal burn, frostbite), Neuropathy(diabetes), Collagen vascular disease (scleroderma, Raynaud's), Hyperparathyroidism, Familial (Hadju-Cheney, progeria), Occupational (polyvinyl exposure)(2,7).

Treatment of the disorder is symptomatic. No specific treatment is available for Acro osteolysis. Mainly supportive treatment – antiresorptive (Bisphosphonates) and symptomatic pain management (1). These treatments not cure the Acro osteolysis but delay the bone resorption and reduce the pain.

Reference

1. Hardegger,F. et al.'The syndrome of idiopathic Osteolysis',J Bone & Joint Surg ,67-b:89-91,1985

2. Cazalis,P. et al.'Acro-osteolyse de la main.',Ann Radiol,25:337-340 (*),1982

3. Lamy, M et al.'Acro-osteolyse dominate.',Arch Franc Ped ,18: 693-702 (*) ,1961

4. Ramos,F.J. et al. 'Further evidence that the Hajudu-Cheney syndrome and the "Serpentine fibula-polycystic kidney syndrome" are a single enitity' Am J Med Gent, 78:474-481 ,1998

5. Nishimura,G .et al.'Syringohydromyelia in Hajdu-Cheney syndrome.', Pediatr Radiol,26:59-61,1996

6. Kaplan, P et al. 'Cystic kidney diseases in Hajdu-Cheney syndrome.', Am J Med Genet, 30:175,1993

7. https://radiopaedia.org/articles/acro-osteolysis-mnemonic?lang=us

8. Pellegrini,V. et al. 'C.T findings in the Hajdu-Cheney syndrome.',Pediatr Radiol, 21: 304,1991

9. Diren, H.B. et. Al. 'The Hajdu-Cheney syndrome: a case report and review of the literature.;, Pediatr Radiol 20: 568-569 ,1990

10. Van den Houten B.R. et al.' The Hajdu-Cheney syndrome', Int J Oral Surg ,14 : 113-125 ,1985

11. Elias, A.N. et al. 'Herediatry Oseteodysplasia with Acro-Osteolysis(the Hajdu-Cheney Syndrome).', Am J Med , 65: 627-636,1978

12. Weleber, R.G. et al.'The Hajdu-Cheney syndrome.' J Pediatr ,88:243-249,1976

13. Silverman, F.N. et al. 'Acroosteolysis(Hajdu-Cheney syndrome).',Birth Defects OAS ,10 :106-123, 1974