ACRODYSOSTOSIS - A RARE SKELETAL DYSPLASIA

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ABSTRACT: Acrodysostosis is an extremely rare disorder characterized by short fingers and toes with peripheral dysostosis, nasal hypoplasia, and mental retardation. We present a case of a 21 year old girl came for CT chest examination. She had difficulty in learning with short stature, slow at learning with mild fever. Radiological findings included broad, short metacarpals and phalanges with cone-shaped epiphyses, hypertrophied first metatarsals, and thickening of the calvarium.
KEYWORDS: Peripheral dysostosis, V-shaped Epiphysis, Mental Retardation.

INTRODUCTION: Acrodysostosis is an extremely rare skeletal dysplasia characterized by abnormally short and malformed bones of the hands and feet, nasal hypoplasia, and mental retardation. Additional manifestations include short stature, and abnormalities of the spine, mandible, and skull[1, 2]. It was first described by Maroteaux and Malamut in 1968[3], and since then, around 50 cases have been reported in the literature. The etiology and prevalence are still unknown. Most cases are sporadic, although some cases have occurred in families, suggesting autosomal dominant inheritance[4, 10]-Although inherited as an autosomal dominant disorder most are new mutations.

The most unique and constant feature of acrodysostosis is generalized peripheral dysostosis. Severe shortening of the metacarpals, metatarsals, and phalanges are caused by prematurely-fused, cone-shaped epiphyses. Nasal hypoplasia and mental retardation are other cardinal features.[5, 6] More than 75% of reported patients have had intellectual deficits. Although associated neurologic defects, including hydrocephalus, optic nerve atrophy, seizures, choreoathetosis, and strabismus have been reported in some acrodysostosis patients[1], to date no explanation for mental retardation is apparent.

CASE REPORT: This 21 years old female presented with difficulty in learning, short stature with short stubby digits of hands and feet. Associated features were mental retardation, small upturned nose and flat face suggestive of acrodysostosis. On physical examination the height was 130 cm (10 cm below 3rd centile) and weight 35 kg (5 kg below 3rd percentile). Nasal bridge was depressed with mild micrognathism. Her lower limbs were short with large torso and short, broad hands and feet.

Figure 1

Figure 1. Photograph of 21 years old female having short stature, slightly depressed nasal bridge and micrognathia, dull expressionless face. Bilateral short stubby hands with normal length of upper limbs. Torso longer than the lower limbs.

There was short stature without any discrepancy in proximal or distal segments of the limbs. The fingers of hand and toes were short and stubby in nature **(Figure 2)**. Hair growth was normal. There were early signs of breast enlargement, well developed secondary sexual characteristics, and her sexual maturity rating was Tanner stage 4. Muscle strength was determined as grade 4 in both upper extremities, and deep tendon reflexes were normal. There were no sensory changes in the upper extremities. Mental age assessment was around 15 years.

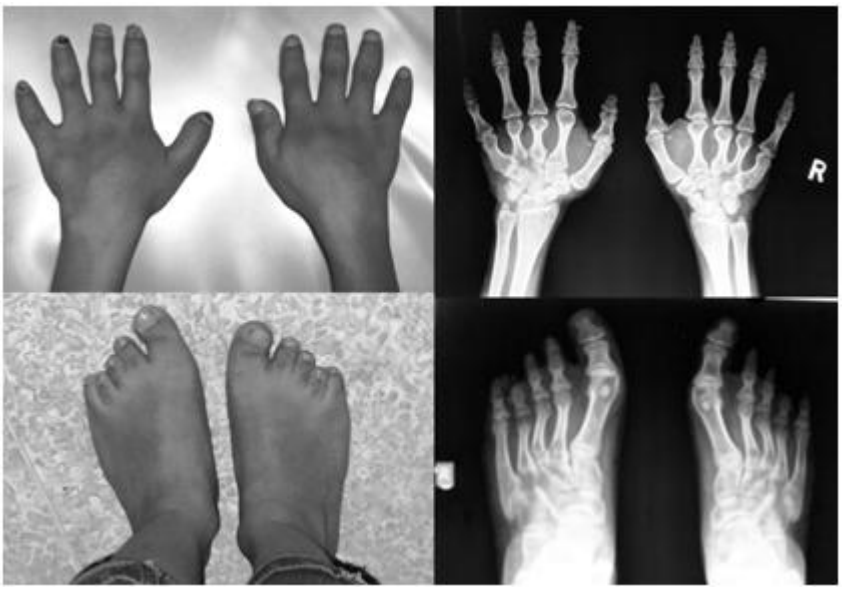

Figure 2

Figure 2. Pictures of hand and feet (a, b) showing short stubby hands (mesomelia), hyperplasia of great toes and hypoplastic 3rd and 4th digits of both feet. X ray AP views of both hands (c, d) showing short metacarpals, phalanges with distal tuft resorption (acro-osteolysis). First ray hyperplasia is seen in both feet with hypoplastic fourth metatarsals.

Serum calcium, electrolytes, alkaline phosphatase and full blood count were normal. Mild increase in TSH value with normal T3 and T4 were found.

Ultrasound scan of the abdomen did not reveal any abnormality.

A skeletal survey showed a bone age of >21 years. There was mild genu varus deformity and a modeling deformity of bones around the knee joints. Radiographs of the hands and feet showed short broad, short metacarpals, metatarsals and phalanges. There were inverted "V" shaped depressions with cone shaped epiphysis in the metacarpals and phalanges on both sides.

First ray hypertrophy involving both great toes with bifid terminal tufts was noted. Hypoplasia of fourth metatarsals was seen on both sides **(Figure 3)**.

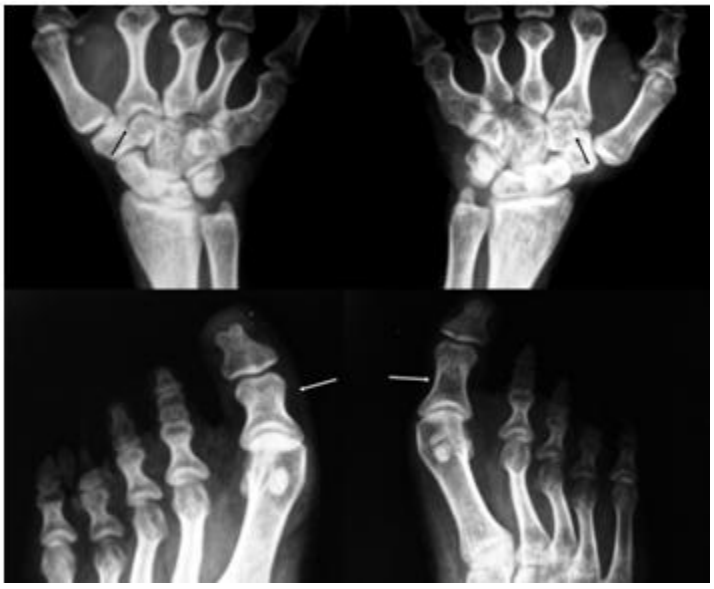

Figure 3 X-ray AP views both hands (a) showing short metacarpals with cone shaped (v shaped) epiphyses at carpometacarpal joints (black arrows). X-ray AP views both feet (b) First ray hyperplasia is seen in both feet (white arrows) with bifid terminal tufts.

Skull was not brachycephalic but the calvarium was thickened. Irregular thickening of the skull vault with focal hyperostosis of outer cortex is noted. Basal skull and cervical spine CT findings include spinal canal stenosis, mild platybasia with angulation and compression into the cervicomedullary junction, and downward displacement of the cerebellar tonsils (**Figure 4).**

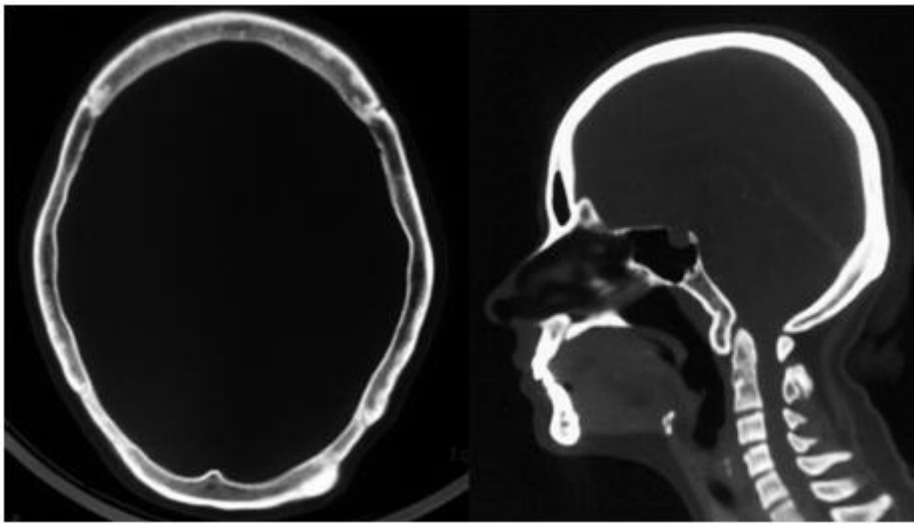

Figure 4. Axial CT Scan of skull vault (a) shows thickened calvarium with focal areas of hyperostosis. Coronal Reformat Cervical spine (b) shows mild platybasia with angulation at the CVJ junction and spinal canal stenosis.

The thoracic cage was cylindrical and elongated with splayed vertical anterior ends of the ribs. Early calcification of costal cartilages of ribs was seen. The sternum is protruding anteriorly giving "pigeon chest" appearance **(Figure 5).**

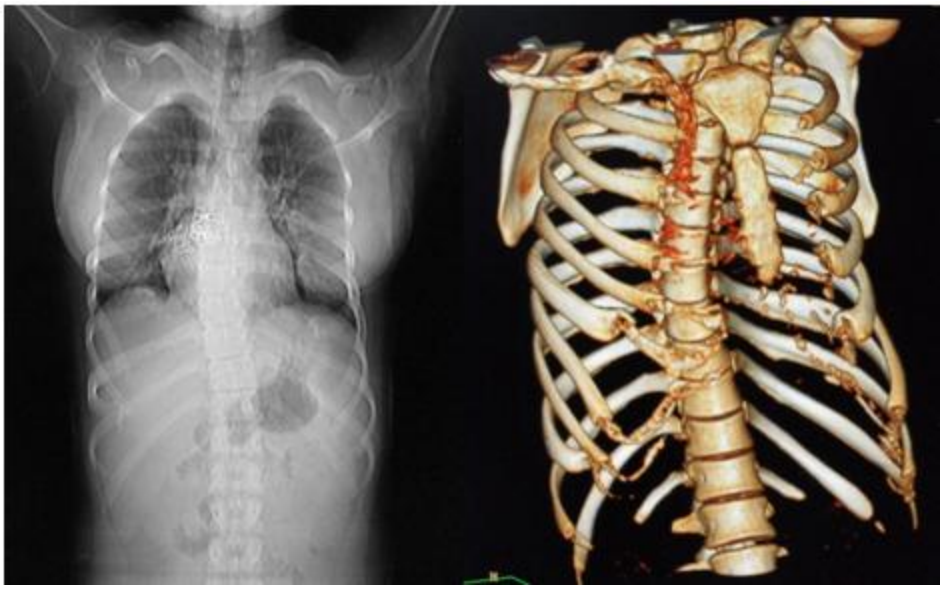

Figure 5. X-ray thoracoabdominal region (a) showing large elongated thoracic cage with scoliosis of the thoracic spine. (b) 3D-Reformat of thorax shows early costal cartilage calcifications with scoliosis.

In the dorsolumbar spine, vertebrae were of normal height. There was loss of normal caudal widening of the lumbar interpedicular distance without any signs of major spinal canal stenosis. Scoliosis of thoracic spine is seen with concavity towards the left **(Figure 6).**

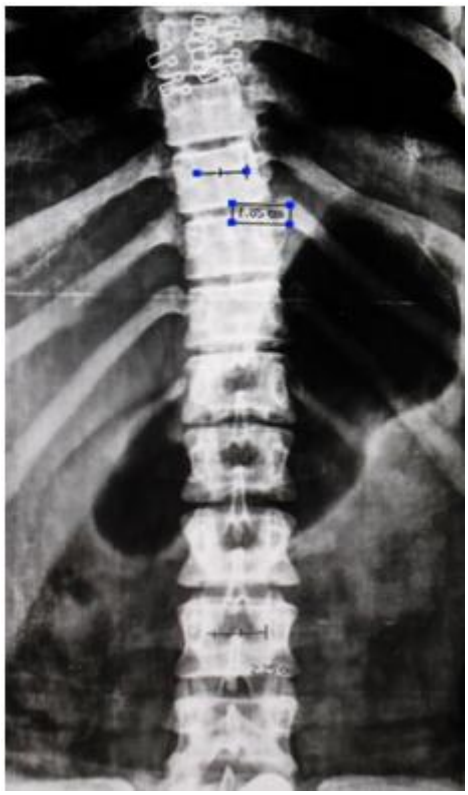

Figure 6: X-ray dorsolumbar spine AP view shows loss of normal caudal widening of the lumbar interpedicular distance with mild lumbar canal stenosis.

DISCUSSION: This 21 year old female presented with difficulty in learning, short stature with short stubby bones of hands and feet and cone shaped epiphyses in metatarsals, metacarpals and phalanges. Associated features seen in this patient were mental retardation, small upturned nose and flat face suggestive of acrodysostosis [4, 7].

Costochondral calcification is uncommon in people under 30 years of age. Premature costochondral calcification is associated with infections, mineral metabolism, thyroid disease, chronic renal failure, some malignancies and genetic factors[9]. In our 21 year old female case early costochondral junction calcification was noted.

Most patients with this condition do well except for arthritic complications and progressive restriction of movements of hands, elbows and spine[7, 11].The differential diagnosis of acrodysostosis are achondroplasia, hypothyroidism, pseudohypoparathyroidism, and pseudo-pseudohypoparathyroidism. Endocrine abnormalities have not been reported in the acrodysostosis syndrome[8]. There was no evidence of GH deficiency and hypothyroidism in our case, as patients having short stature due to hormonal imbalance can be treated with hormonal therapy[12].

CONCLUSIONS: Patient presenting with short stubby hands and feet, V-shaped epiphysis, first ray hyperplasia of first digit of foot, typical facial features and mental retardation, the possibility of acrodyostosis should be considered after ruling out other causes of short stature. Early diagnosis helps reduce life-threatening complications like spinal canal stenosis and to manage neurologic complications.

REFERENCES:

1. RobinowM,PfeifferRA,GorlinRJ,McKusickVA,Renuart AW, Johnson GF. Acrodysostosis. A syndrome of peripheral dysostosis, nasal hypoplasia, and mental retardation. Am J Dis Child 1971; 121:195-203.
2. Butler MG, Rames LJ, Wadlington WB. Acrodysostosis: report of a 13-year-old boy with review of literature and metacarpophalangeal pattern profile analysis. Am J Med Genet 1988; 30:971-80.
3. Maroteaux P, Malamut G. Acrodysostosis. Presse Med1968; 76:2189-92.
4. Jones KL. Smith's Recognizable patterns of human malformations. 6th ed. Philadelphia: Elsevier Saunders 2006.
5. Steiner RD, Pagon RA. Autosomal dominant transmission of acrodysostosis. Clinical Dysmorphology1992; 1:201.
6. Sujeewa Amarasena, MGK Samanlatha, MHAD de Silva, P Kolombage. A 12 year old girl with acrodysostosis: a rare cause of short stature. Sri Lanka Journal of Child Health.2009; 38:72-73.
7. Jung Min Ko, Kyu Sung Kwack, Sang-Hyun Kim, Hyon-JuKim. Acrodysostosis associated with symptomatic cervical spine stenosis. Journal of Genetic Medicine 2010; 7:145-50.
8. RC Ablow, YEHsia, IK Brandt. Acrodysostosis Coinciding with Pseudohypoparathyroidism and Pseudo-Pseudohypoparathyroidism. AJR 1977; 128:95-99.
9. Sariyildiz MA, Batmaz I, AydınF,Azboy I. Early onset idiopathic costochondral calcification can simulate costal Fracture. International Journal of Basic and Clinical Studies (IJBCS) 2012; 1(2):69-74.
10. Sheela SR, Ajai Perti, Grace Thomas. Acrodysostosis: Autosomal Dominant Transmission. Indian Pediatrics 2005; 42:822-26.
11. Shafeghati Y, Vakili G, Jannati J. Acrodysostosis; Report of a 21 years old Iranian patient. Journal of Rehabilitation 2005; 23:45-48.
12. Seino Y; Yamanaka Y; Shinohara M; Ikegami S; Koike M et al. Growth Hormone Therapy in Achondroplasia. Hormone Research 2000; 53(3); 53-6.

CASE REPORT

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