Acrodysostosis: A case report.

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Abstract:

Acrodysostosis is a rare syndrome characterised by peripheral dysostosis (gross shortening of hands and feet), mental retardation, hypoplasia of nose and hypoplasia of maxilla. An 11 year old female child presented to Civil Hospital Ahmedabad with complaints of short stubby fingers. On detailed general examination, patient had dysmorphic facial features, depressed nasal bridge, upturned nose, gingival hyperplasia and dental malocclusion with skeletal survey suggestive of hypoplastic nasal bone, shortening of metacarpal, metatarsal bone & phalangeal bones and early fusion of epiphysis of distal end of radius and ulna ; all pointing towards the diagnosis of acrodysostosis. Any patient presenting with short stature, a short stubby fingers, and acrodysostosis should be suspected. No specific therapy is available.

Key Words: Acrodysostosis, Nasal hypoplasia, Short stubby fingers.

Introduction:

Acrodysostosis, also known as Arkless-Graham syndrome or Maroteaux Malamut syndrome, is a rare congenital malformation syndrome which involves shortening of the interphalangeal joints of the hands and feet, intellectual disability in approximately 90% of affected children, and peculiar facies. Other common abnormalities include short head (as measured front to back), small broad upturned nose with flat nasal bridge, protruding jaw, increased bone age, intrauterine growth retardation, juvenile arthritis and short stature. Further abnormalities of the skin, genitals, teeth, and skeleton may occur.

Though most cases occur sporadically, it is believed to be an autosomal dominant condition. Both males and females are affected. The disorder has been associated with the older age of parents at the time of conception.

Case Report:

An 11 year old female child was referred to us for evaluation of psychomotor delay. Child was born to a 35 year old father and 26 year old primigravida mother. There is no history of consanguinity. She started to walk at 2 years of age. Height of child is 136 cm (25th centile) and head circumference is 57 cm (50th centile). She had dysmorphic facies with a broad depressed nasal bridge. Menarche was achieved at 10 years of age. She also had gingival hyperplasia and dental malocclusion. The striking feature was

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short stubby fingers with broad hands. (Image 1) The toes were also short.

A detailed skeletal survey was done which should broad and short metacarpals and phalanges with cone shaped epiphysis, hypoplastic nasal bone, early fusion of epiphysis of distal end of radius and ulna. (Image 2)

Image 1 Short stubby fingers with broad hand

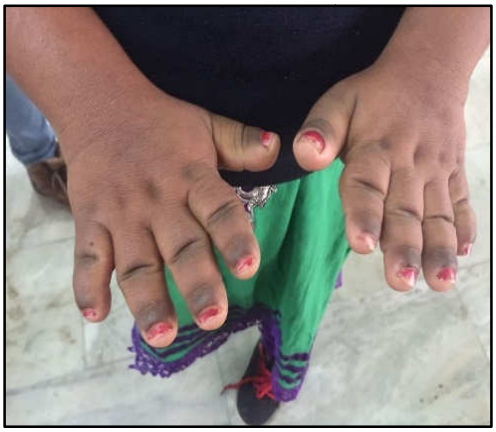
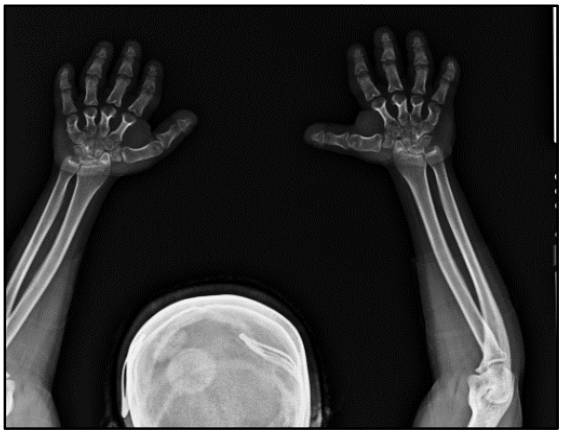


Image 2 Short metacarpals & Fusion of distal epiphysis of radius & ulna



Fundus examination was normal. Hearing of child was normal. Ultrasonography of abdomen and MRI study of brain was normal. Routine investigation and serum calcium, phosphate, alkaline phosphatase levels were normal. Thyroid function test was normal. There was no significant family history.

Based on clinical feature with evidence of skeletal survey, diagnosis of acrodysostosis was confirmed.

Discussion:

Acrodysostosis is a rare syndrome characterised by peripheral dysostosis (gross shortening of hands and feet), mental retardation, hypoplasia of nose and hypoplasia of maxilla. [2] Many individuals have developmental delay and Intellectual disability. Patients with acrodysostosis may have hormonal resistance [3] especially Thyroid and parathyroid hormones and based on that they are of two types. Type 1 is caused by mutation of PRKAR1A gene associated with hormone resistance and type 2 is caused by mutation in PDF 4 D gene and not associated with hormone resistance. Other physical findings include [4] middle ear infections (otitis media), loss of hearing, obesity, pigmented navie, blue eyes, broad hands. Individuals may eventually develop arthritic changes in hands which can lead to problems involving hand skill and coordination.

This disorder would be present since birth but is recognized till few years after birth. Male and females are affected equally.

Some individuals develop resistance to other hormones such as Para Thyroid Hormone & Thyroid Stimulating Hormone. In Some affected males have hypospadiasis, cryptorchidism may be present. It should be differentiated from Albright hereditary osteodystrophy.

Treatment:

Patient was given calcium and vitamin D supplements and was asked to come for regular follow up to monitor growth and development of the patient.

Treatment is based on specific symptoms. There are no standardized treatment protocols or guidelines for affected individuals due to rarity of the disease. No specific therapy for the disease is available. Surgery may be performed to correct specific abnormalities such as underdeveloped (hypo plastic) and/or abnormally prominent jaws (prognathism). In some cases, dental braces may be required to correct misaligned teeth (malocclusion). In addition, in some cases, physical therapy may also be required. Thyroid hormone supplementation, growth hormone and vitamin D supplements may contribute to improve growth and prevent obesity. In case of delayed sexual development, GNRH analogues may be tried.

Conclusion:

Any patient presenting with short stature, a short stubby fingers, and acrodysostosis should be suspected. A diagnosis of acrodysostosis is based upon identification of characteristic symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests including X-rays. Early intervention is important to ensure that children with acrodysostosis reach their full potential. Special services that may be beneficial to affected children may include special remedial education, social support, and/or other medical, social, and/or vocational services.

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