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RESEARCH ARTICLE

Title: A Dental Perspective on the Rare Disorder of Acromesomelic Dysplasia - A Case Report.

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Abstract

Introduction: Acromesomelic dysplasia, the Maroteaux type is listed as a “rare disease” by the office of rare diseases (ORD) of the National Institute of Health (NIH). This means that acromesomelic dysplasia, Maroteaux type (AMDM), or a subtype of acromesomelic dysplasia affects less than 200,000 people. Acromesomelic dysplasias are skeletal disorders that disproportionately affect the middle and distal segments of the appendicular skeleton. As with the appendicular features, the vertebral features become more pronounced throughout childhood. The pelvis has a typical configuration, and the cranium may demonstrate frontal bossing, with a normal head circumference, hypoplastic maxilla and malocclusion.

Case report: The following case report discusses the medical and dental findings of acromesomelic dysplasia and their subsequent management.

Conclusion: Disorders like acromesomelic dysplasia have rarely been documented for any oral findings. Further documentation of such cases would be essential in establishing the full scope of the oral features. The findings enlisted here suggest that early preventive measures and intercepting any developing malocclusions would suffice the promotion of good oral health during infancy and childhood.

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Introduction:-

Acromesomelic dysplasia Maroteaux-type (AMDM), first described by Maroteaux in 1971, is a rare autosomal recessive osteochondrodysplasia.¹ This disorder is characterized by acromelia and mesomelia. Mesomelia describes the shortening of the bones of the forearms and lower legs relative to the upper parts of those limbs. Acromelia is the shortening of the bones of the hands and feet. Thus, the short stature of affected individuals is the result of unusually short forearms and abnormal shortening of bones of the lower leg.² About 50 cases of AMDM have been reported till date, most of them being the classical variety.³ This is an autosomal recessive skeletal dysplasia with a prevalence of ~1/1,000,000.⁴ A diagnosis of AMDM type is usually made on the basis of combined clinical, genetic and radiographic findings. Although this condition is usually diagnosed at birth and becomes more obvious in the first two years of life. Recently, Kant et al reported the mapping of a gene for AMDM to human chromosome 9p13-q12 by homozygosity mapping in four inbred families.⁵ Birth lengths and weights are normal, although mild shortening of long bones may be detected in some affected infants.⁴ In patients with AMDM, all skeletal elements are present, but they have abnormal rates of linear growth (Langer and Garrett 1980).⁶ By two years of age, radiographic skeletal changes are diagnostic for AMDM and include abnormal growth plates and short, misshapen bones in the extremities.⁴

Here in, we are reporting a case of a child affected with AMDM, along with dental findings and management.

Case report:-

A seven year old female child reported to our department, with the chief complaint of dental caries in relation to upper anterior and lower posterior teeth. Child showed stunted growth with normal intelligence. Previous medical records showed that she was diagnosed with AMDM at Indira Gandhi Institute of Child Health, Karnataka. Family

history revealed that she is daughter of a consanguineous couple from Karnataka with two normal siblings. Consanguineous couples have more chances of autosomal recessive disorders amongst siblings. The parents were of normal height and proportion. Pregnancy and perinatal period was normal. Short stature was noticed at two years of age. At the same time deformities of elbows and forearms were noticed (Fig.1).The deformities have been increasing since then. Developmental milestones were normal. There was no difficulty in vision or hearing.

At seven years of age, height and weight were 78 cm and 11.6 kg. On examination, the skull was brachicephalic with frontal bossing, occipital prominence and hypoplastic maxilla (fig 1). She had a broad, depressed nasal bridge which correlates to a class III facial profile and several carious teeth. There was extreme shortening of upper limbs more marked in forearms and hands. There was bowing of forearm bones leading to convexity of the radial border of forearm. Terminal extension at elbows was restricted. Pronation and supination were normal. Fingers were very short (Fig 2). The lower limbs though short were less as compared to upper limbs. The feet were short, flat and square with relatively large great toes and proximally placed second toe bilaterally. The nails were broad and short in both hands and feet (Fig 2). In 1997, S. Danda et al had reported similar features in two affected siblings.

Oral examination showed no soft tissue findings and hard tissue showed dental caries (Fig 3). Occlusion evaluation showed angels class 1 molar relation and mild lower anterior crowding.

Radiographic findings showed short, stubby long bones with flaring at the ends. Radius and ulna were more severely involved. There was bowing of radius and distal end of ulna was shorter than radius creating a gap between the distal end of ulna and carpals at the wrist. The metacarpals were short, broad with flared ends. The phalanges were short with shortening most marked in distal phalanges. Similar shortening was noted in metatarsals and phalanges of feet but was less marked (Fig 4). The first metatarsal and phalanges of great toe were broad. The borders of the vertebrae were regular with decreased height in the posterior part (posterior wedging). The clinical and radiographic features were diagnostic of acromesomelic dwarfism.

Dental radiographs showed lower anterior crowding, normal eruption sequence and caries in relation to upper anterior and lower posterior teeth (Fig 4). Strip crowns were given for upper central incisors and glass ionomer cement restoration was done for 1st and 2nd molars (Fig 3).



Fig 1 Child with acromesomelic dysplasia, note brachicephalic skull and hypoplastic maxilla.



Fig 2 Hands and feet of the individual. Note short and broad fingers, short and flat square shaped feet.



Fig 3 Oral examination and subsequent strip crowns in relation to 51 and 61.



Fig 4 Radiographs of the hand, feet, skull and teeth.

Discussion:-

The specific entity “acromesomelic dwarfism,” first reported in three children by Maroteaux et al. (1971), has subsequently been described in 140 individuals (e.g., Langer et al. 1977; Hall et al. 1980; Langer and Garrett 1980; Borrelli et al. 1983; Del Moral et al. 1989). The most comprehensive description of the skeletal features, provided by Langer and Garrett (1980), is based on radiological evaluations of 28 affected individuals. At birth, weight and length may be normal, although limbs may appear short; X-rays reveal short but not malformed appendicular bones. Disproportionate shortening of the limbs becomes more apparent during childhood. Although all appendicular skeletal elements are short, the distal and middle segments are generally more severely affected than the proximal segment.⁶ Acromesomelic dysplasia is characterized by markedly short hands and feet, dwarfism to variable degree and characteristic face with narrow palpebral fissures, short stubby nose and averted nostrils.⁷ The cranium may demonstrate frontal bossing, with a normal head circumference. Most affected individuals have no obvious facial dysmorphism; they have normal hair patterns and quantity, normal intelligence, and no evidence for other organ-system involvement.⁶ Literature is scarce regarding appropriate peri-operative management of patients with AMDM.⁸

There was a study done by Saadullah Khan, Raja Hussain Ali et al who conducted genotyping of six consanguineous families of Pakistani origin with AMDM. Sequence analysis of the gene NPR2 (Natriuretic peptide receptor- 2) identified a novel missense mutation (p.T907M) in five families, and a splice donor site mutation c.2986+2T>G in the other family.⁹ Berkowitz et al published a review of the pathophysiology of different types of dwarfs and their anesthetic implications.¹⁰ In both studies no relevant oral features were reported.

Our case presented most of the classical features of AMDM along with certain oral findings like retarded maxillary growth, minor anterior crowding and dental caries.

Conclusion:-

Such cases of acromesomelic dysplasia rarely come for dental treatment and the case which came to our department showed physical growth retardation in all aspect. Strip crowns were placed for restoring the anteriors. Special services that may be beneficial to affected children may include social support and other medical, social, and/or vocational services. Genetic counselling will be of benefit for affected individuals and their families. Dental features included lower anterior crowding and caries but there was no abnormality in the eruption sequence. Further follow up is required to check for developing malocclusion. Thus for such cases proper diagnostic evaluation should be done and they should be managed with proper care and concern so as not to hurt the child's self image.

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