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Achogdrogenesis type II (langer-saldino) — A case report

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Summary

Achondrogenesis is a lethal form of congenital chondrodystophy characterised by extreme micromelia. Definitive clinical and radiographic criteria have been established to differentiate Type II Achondrogenesis (Langer-Saldino) from Type I Achondrogenesis (Parenti-Fraccaro). The mode of inheritance is autosomal recessive for both types.

We are presenting a case of Type II Achondrogenesis, a still born male to consanguinous parents. The clinical features included an enlarged head, protuberant abdomen and short stubby limbs. The mother had earlier delivered two still born males presumably with similar features. Radiographic characteristics of absence of rib fractures and well ossified iliac bones with concave medial margins and absent or deficient ossification of the sacrum, ischiae, and pubic bones differentiated Type II Achondrogenesis from Type I Achondrogenesis.

Résumé

Achondrogénése est une forme Lethale de Chondrodystophie congénitale caractérisé en Micromelie. La definition de critères clinicales et radiographiques est constitué pour differencier Achondrogénése Type II (Langer-Saldino) d' Achondrogénése Type I (Parenti-Fraccaro). La méthode inherité est sous forme recessive autosomal pour les deuse types.

Nous avons presenté un cas d' Achondrogénése Type II un nouveau né décè, son parents etaient familié. L' aspect clinical contenait cephalomegalie, ventre protuberante et extremités court. Chezlamèse IL y avait deux deliveres précoce de deuse nouveause né décès (masculin) ont eté presumé de même aspect. Les caractères Radiographiques d' absence de fraction costale, et bon ossification d' os Iliaque avec un hord medial concave, et absence on deficit d' ossification de Sacrum, Ischiae et Pelvis tous les antécédents avaient differencié le Type Achondrogénése II de Type I.

Introduction

Achondrogenesis is a lethal form of congenital chondrodystrophy characterised by extreme micromelia. Since 1936 when Parenti first described the condition under the name "Anosteogenesis", literature on achondrogenesis continues to accumulate[1,2].

Definitive clinical and radiographic criteria have been established to differentiate Type I Achondrogenesis (Parenti-Fraccaro) from Type II Achondrogenesis (Langer-Saldino). The mode of inheritance is said to be autosomal recessive for both types. The histopathology of achondrogenesis is characterised by hypercellular cartilage at growth plates with irregular column formation of cartilage cells and marked lack of progression through proliferation, maturation and eventual mineralization of the intercellular matrix of the cartilage[2,1].

Case report

A 30 year old, Sudanese female, married to a second cousin was admitted to Khartoum Teaching Hospital in Sudan, as a case of Polyhydramnios, this being her fifth pregnancy. She delivered at term a still born male in breech presentation after artificial rupture of membranes was performed. The placenta appeared normal. The mother noticed that the male infant with short limbs and bulky body resembled two previous products of conception. The first and fourth pregnancies were associated with polyhydramnios and the subsequent delivery of male infants with bulky bodies and short limbs. Both husband and wife were of normal stature. All laboratory investigations performed on the couple did not reveal a disease pattern.

Fig. 1: Grossly edematous, still born male with short limbs, depressed nasal bridge and upturned nostrils

The case under consideration was a still born male, grossly edematous and weighed 9 pounds (Fig. 1). The skull circumference and heel-crown length measured 36 cm. and 34 cm. respectively. The anterior fontanelle was widely open, the eyes wide apart and no blue colouration of the sclera was observed. The nasal bridge was depressed and the nostrils up-turned. The ears were normally set but edematous. The neck was short and a hump on its posterior aspect was seen. The chest looked narrow. Both upper limbs were short and study and each hand consisted of five fingers. The abdomen was protuberant. The umbilicus was normal in length and contained two arteries and one vein. The genitalia was normal. The lower limbs were short and curved inwards and each foot consisted of five toes. A nasogastric tube could be passed normally down to the stomach and some amnion could be aspirated. The anus was meconium stained. There was no family history of a similar condition on either the paternal or maternal side.

Radiological features

There is generalised retardation of skeletal ossification resulting in severe short limb stature. Soft tissue shadows are very prominent (Fig. 2). The skull shows a relatively enlarged calvarium with a wide anterior frontanelle. The chest is short and the lung fields atelectatic. The ribs are short, flat and have flared and cupped anterior ends. No fractures of the ribs are identified. There is marked flaring and widening of the proximal ends of the humeri and distal ends of the radii. Some of the long bones of extremities show constriction of the diaphyses and small spurs in the metaphyses. The scapulae are small, irregular and poorly ossified. The tubular bones of hands and feet are barely visible. The femora are short and broad with slight incurving of the proximal ends. The iliac bones are small, well ossified with concave medial and inferior margins ending in a short pointed ischial spine. The ischiae are barely perceptible. The pubic bones are not seen. The sacral bones are poorly ossified. There is inadequate ossification of the vertebral bodies which are faintly visible in the lumbar region. The intervertebral disc spaces are relatively increased. There is no distal narrowing of the interpendicular spaces.

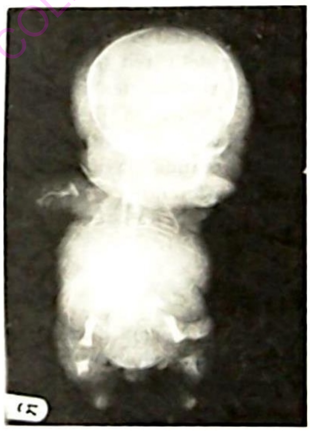

Fig. 2: Frontal skiagram of the child showing prominent soft tissue shadows, short, stubby long bones and absent ischiae and pubic bone

Discussion

Achondrogenesis is a lethal form of congenital dwarfism that has very specific clinical, radiological and pathological characteristics differentiating it from other forms of dwarfism namely thanatophoric dwarfism, achondroplasia, asphyxiating thoracic dystrophy, diastrophic and metatrophic dwarfism to name a few[1-4]. According to Yang's classification the case under study is one of Langer-Saldino's type of heritable lethal achondrogenesis i.e. Type II characterised by relatively better ossification of the

calvarium, absence of rib fractures, diaphyseal constriction of long bones with flared and cupped ends, well ossified iliac bones with concave medial margins and absent or deficient ossification of the sacrum, ischiae and pubic bones. Achondrogenesis Type I, also referred to as Parenti-Fraccaro's type, is characterised by disproportionately large heads, poor ossification of the calvarium, multiple rib fractures, short broad bones of extremities with prominent bony spurs at the border of the metaphyses and poorly ossified iliac bones.

Conclusion

We have presented a case of Achondrogenesis Type II (Langer-Saldino's type) which is a rare distinct entity with characteristic radiographic findings. Histopathological features include degeneration of the proliferating epiphyseal and diaphyseal cartilage. Death is attributed to atelectasis caused by the narrow chest. The cause of this condition is unknown.

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