ACHONDROPLASIA

(Report of a Case)

by

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Congenital disorder of the skeleton in the foetus, showing short limbs (micromelia), large head and aprently normal trunk, is a rare abnormality. Kaufmann terms skeleton disorders in the foetus under the general term "Chondrodystophia Foetalis", while French calls it "Achondroplasia".

Achondroplasia consists of retardation of epiphyseal, but not of the peri-osteal bone-growth. It is primarily a faulty growth of cartilage and premature cessation of the endochondral ossification, resulting in short, thick extremities, large head, apparently normal-sized trunk, small pelvis (pelvis nana) and with deformities of the spine.

Little is known about the etiology of the disease, but according to Kaufmann, it is due to defect of bone-forming tissues occurring in the early weeks of the foetal life. It is always congenital, and heredity seems to play a part. It is commonly seen in twins. According to Abels, it is due to overaction of thyroid gland, while, according to Jensen, Wheeldon and Duken, changes in amniotic pressure play an important part.

The histologic picture shows disorderly arranged cartilage and columnar cells, increase in vascularity and in intercellular substance, and strands of fibrous bands from periosteum, separating the epiphysis from the disphyses. Some cases show hyperplasia of cartilage cells (chondrodystophia hyperplastica), in some cases there is hypoplasia of cartilage cells (chondro-dystophia hypoplastica), while some cases show soft cartilage (chondro-dystophia malacia).

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Case Report

A second para, aged 27 years, developed sudden hydramnios with breech presentation at 34th week. An X-ray of the abdomen was taken which revealed achondroplasia of the foetus". General condition of the patient was normal, TPR —N, BP-120/80 mm Hg., no odema of legs, urine-examination showed no abnormality, due date 7/1/61.

On 25/11/'61, she started getting very mild pains at 8 p.m., leaking membranes started at 11 p.m. She came to the hospital on 26/11/'61 at 5-30 a.m. with fully dilated cervix, and moderate pains, and, on examination, footling was presenting. She was delivered with breech extraction. There was slight difficulty in delivering the head, which was delivered by Mauricau-Smellie—Veit method. An episiotomy was done. A female child weighing 4 lb. 9 ozs. was born, took few gasps and died.

Placenta and membranes showed no abnormality; but the cord, near the umbilicus of the child, showed a big vesicle

about 2″ x 2″ size, filled with yellowish fluid (seen in the photograph).

Mother had uneventful recovery. There was no history of any abnormal child in the family of the parents. First child, aged 3 years, is normal.

The child showed the following characteristics:

Short and thick extremities; thighs and arms were shorter than legs and forearms, all fingers were thick, short and of equal length, legs showed varus curvature.

Head was large, circumference was 46 cms., sutures gaping and large fontanelles; hydrocephalus was present with prominent forehead, prominent chin, and depressed root of the nose.

The neck was thick and short, the trunk size was normal and funnel-shaped. The abdomen was prominent, ascites was present, vulva was odematous. The skin was coarse and loose. Spine showed no abnormalities on external examination.

Fig. 1

Discussion

There was a great deal of confusion about skeletal disorders in the foetus till it was cleared by E. Kaufmann. Achondroplasia was mistakenly diagnosed with cretinism and osteogenesis imperfecta congenita.

In cretinism there is no disproportion between extremities and trunk, signs of cretinism usually become noticeable after birth, and depression of the root of the nose appears later.

Osteogenesis imperfects congenita shows multiple fractures and cellus formation. Under X-ray, thin, subperiosteal portion and absence of trabecular appearance is characteristic.

Obstetric importance of such cases consists in the fact that it is associated with general odema, dropsy, hydrocephalus, breech presentation, and sometimes hydramnios; and thus there is difficult delivery of the child. But, as the literature shows and also seen in this case, as a rule, there is no great difficulty. But there are case-reports with history of difficult deliveries. Winkler and Ahlfeld reported a case of achondroplasia with difficult breech extraction. 'Gotingen Women's Clinic' also reported a case of achondroplasia with ascites and hydrocephalus, resulting in difficult delivery.

Prognosis. It is usually bad. These foetuses are usually still-born, or die in the first few days or during the first year. Early death is probably due to cord-compression from abnormal narrowness of the spinal canal or from abnormalities incompatible with life. If they survive for one year, the further duration of life is unaffected by the disease. The chondro-dystophic dwarf, while developing slowly, finally shows few deviations from the normal, as far as intellectual and sexual functions are concerned.

There is no *treatment*, except orthopedic correction of deformities.

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