ACHONDROPLASIA IN THE FEMALE—OBSTETRIC OUTLOOK AND RADIOLOGICAL FEATURES

(Short Review of the Literature and a Case Report)

by

P. MADAN*, M.B., B.S., M.R.C.O.G.,

S. CHAWLA**, M.B., B.S., D.M.R.,

K. BERY**, M.B., B.S., D.M.R.D.,

and

S. VOHRA*, M.B., B.S., M.D.,

Achondroplasia has existed for centuries as is evident from ancient Egyptian art depicting two Gods of ancient Egypt, Ptah-Sokar and Bes as achondroplasiacs (Hunter and Jupe, 1959).

Due to their attractive and doll-like appearance when young, they were sold to the ladies of the court for large sums of money in the middle ages. The achondroplasiac dwarfs are normal in intelligence and often play the parts of clowns at fairs and circuses. Henré Tolouse Lautrec of "Moulin Rouge" fame was an achondroplasiac dwarf.

It is a generalized disease of the skeleton in which endochondral osteogenesis is retarded, whereas the periosteal osteogenesis is unaffected. This results in diminished longitudinal growth of the bones, while the diameter remains normal. The typical clinical appearance is of a dwarf with shortened, bowed extremities, a relatively elongated trunk, a large head with flattened nose and prominent buttocks (Caffey, 1956).

The exact etiology is unknown but an achondroplasialike disorder was produced in chicks by the injection of Insulin and Thallium into the yolk sac on the fifth day of incubation (Duraiswami, 1952).

The incidence of achondroplasia is rather low. Out of 1,30,030 pregnancy terminations studied by Jayant et al., (1961), there were only two cases of foetal achondroplasia. During an eight-year period in the Sloane Hospital for Women, 4 achondroplasiac infants were born out of 12,151 births (Caffey, 1958). Potter (1952) found 8 cases of achondroplasia out of 60,000 deliveries at the Chicago Lying-in Hospital. There were 4 achondroplasiac children born out of 26,309 deliveries at the Hadassah University Hospital, Israel, (Porat, Ehrenfeld and Brzezinski, 1956).

Prenatal diagnosis of the condition was made in the case reported by

*Departments of Obstetrics and Gynaecology and

** Radiology, Lady Hardinge Medical College & Hospital, New Delhi.

Received for publication on 5-4-63.

Shah (1962) when x-ray of the maternal abdomen was done, presumably for hydramnios and breech presentation.

Case Report

M. W., 28 years, female, primigravida, was admitted on 6-9-62 in the Lady Hardinge Hospital, with a history of full-term pregnancy, and labour pains since 4 hours. She came from a nearby village. She had received no antenatal care before admission.

On Examination. The patient was short-statured, her height was 108 cms. She had a large head with depressed nose, shortened upper and lower extremities and a relatively normal trunk. The hands showed trident deformity.

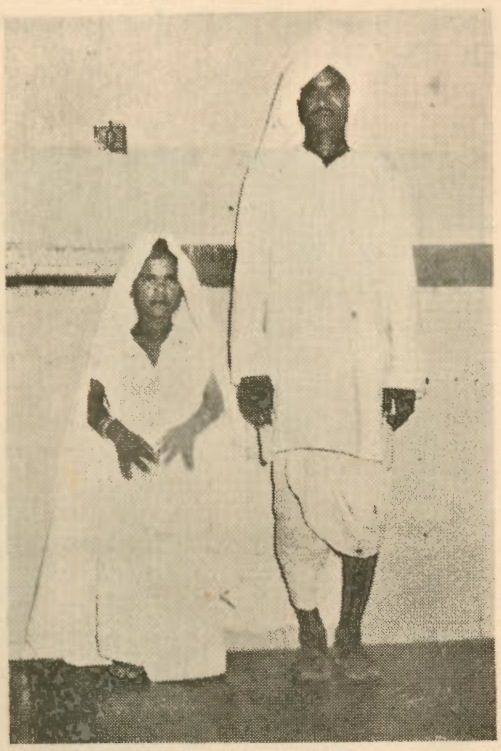

Fig. 1
Photograph showing achondroplasic female dwarf (108 cms.) with typical features—a large head, depressed nose and trident hands. The husband is normal.

Systemic examination revealed no abnormality.

Abdominal Examination. Height of the uterus was up to the level of the costal margin, cephalic end was presenting at the pelvic brim, and was free and mobile. Foetal heart sounds were heard and were of good intensity. Mild uterine contractions were felt.

Per Vaginam. The cervical canal admitted one finger and the cervix was partially effaced. The cephalic end presented above the pelvic brim and the bag of membranes was felt to be intact. The sacral promontory was very easily felt, markedly reducing the antero-posterior diameter of the pelvis, the diagonal conjugate being only 6.5 cms. The body of the sacrum was bulging forwards, the coccyx was small, curved anteriorly, making an acute angle with the sacrum. The ischial spines were felt very near the sacrum, thus reducing the width of the sacro-sciatic notch and space in the posterior segment of the pelvis. The pubic arch was narrow. Because of the obvious pelvic deformity and the patient being in labour, lower segment caesarean section was performed the same evening under gas + 0_2 anaesthesia. A female baby weighing

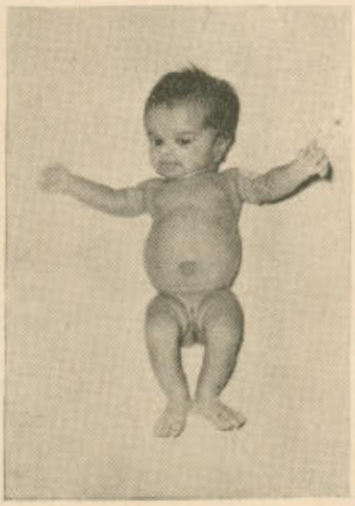

Fig. 2
Female baby with a normal trunk but shortened upper and lower extremities. The changes are more marked in the arms and the thighs, which show folds in the skin.

4 lbs. and 3 ozs. was delivered and cried immediately. The baby showed typical appearance of achondroplasia with talipes equino-varus.

Detailed radiological study of the mother and the baby were done. The mother had an uneventful post-operative period. The mother and the child were discharged after 4 weeks in good general condition.

There is no history of similar condition in the family. The husband was normal in stature (Fig. 1).

Radiological Features

The tubular bones are short but the calibre is normal. The cortex is thick with normal spongiosa and medullary canal. The tuberosities for muscular attachments are enlarged. The shortening is met with in all the bones of the limbs, including the metatarsal bones and the phalanges.

The epiphyses are normal but the ends of the diaphyses are over-developed and widened. Since the middle of the shaft is normal in diameter, this widening at the extremities of the bone gives it a typical dumb-bell appearance (Figs. 3 and 4).

Hands and feet are broad and stubby, the trident deformity is usually seen, as in our case, Fig. 4.

The pelvis is broad and flat with square ilia. The sacral promontory is rotated downwards and forwards and the coccyx is rotated upwards and backwards. The pelvic inlet is contracted partly because of the underdevelopment of the pubic and ischial bones and partly due to sacral tilt (Fig. 5).

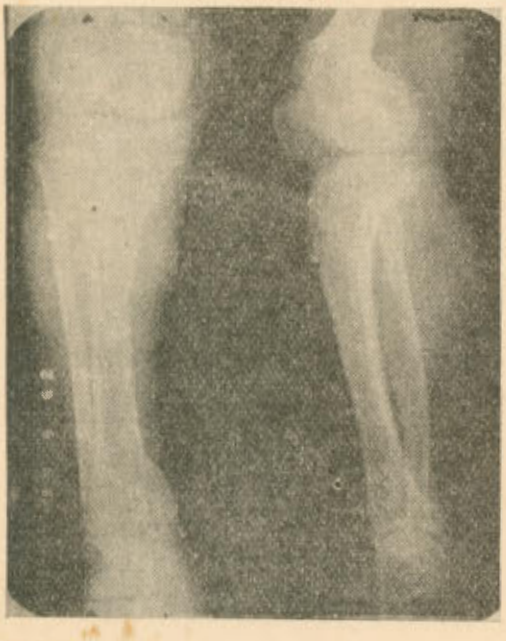

Fig. 3
Dumb-bell shaped short tibia with flaring at the ends.

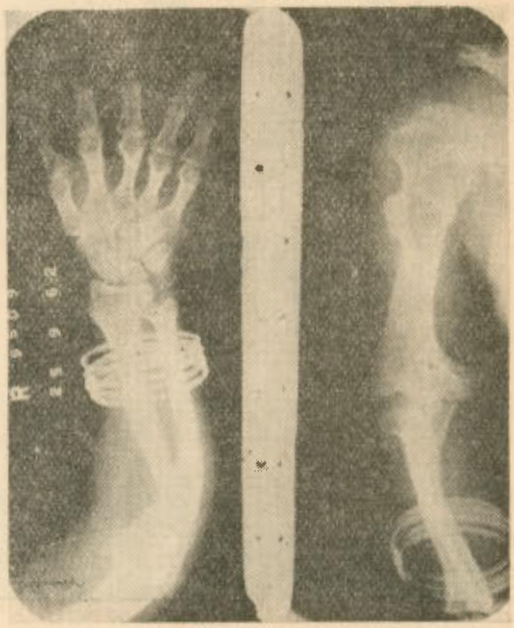

Fig. 4
Short humerus, radius and ulna with dumb-bell shaped flaring at the ends and prominent muscular attachments.

Fig. 5
Pelvis showing flaring of the ilia and marked contraction of the pelvic inlet.

The proportionately long spine characteristic of achondroplasia is due to the excessively thick cartilaginous discs. The vault of the skull appears too large as the base is constricted due to retarded growth of the cartilaginous bones specially the occipital and the sphenoid bones (Fig. 6). Hydrocephalus can occur due to

Fig. 6
Lateral skull showing a large vault of the skull with contracted base.

obstruction in the basal cisterns and aqueduct secondary to pressure by the constriction of the base of the skull (Dandy, 1921).

There is undergrowth of the ribs and sternum reducing the depth and girth of the thorax.

Comments

The condition is usually sporadic but instances of familial transmission have been recorded. Rischbeith and Barrington (1912) recorded 80 pedigrees in which 20 showed achondroplasia occurring in two or more generations. Their series suggests that the female sex is more predisposed than the male for they found amongst 126 cases, 70 females and 56 males.

According to Potter (1952), achondroplasiac dwarfs are most commonly born to normal parents. The birth of an affected child to an achondroplasiac couple is a rarity. Up to 1958, there have been only 5 cases of achondroplasiac infants, born to achondroplasiac parents, who survived (Potter and Coverstone, 1948; Porat, Ehrenfeld and Brzezinski, 1956, and Nattrass, 1958). Most of the achondroplasiacs are stillborn or die during the neonatal period, so the total number of affected people in any one country is small and the marriage of two achondroplasiac dwarfs is extremely rare.

There have been reports of normal parents producing a number of achondroplasiac children. Cohn and Weinberg (1956) reported a normal couple who had identical hydrocephalic achondroplasiac twins and the anomaly was repeated in the subsequent delivery of a single sibling. In between these two pregnancies, a normal child was born.

Most of the achondroplasiac dwarfs die in utero. During the first postnatal months mortality is high due

to hydrocephalus caused by constriction at the base of the skull.

Bony projections at the foramen magnum are thought to be one of the principal causes of foetal death. Cardiac and respiratory distress due to deformities of the ribs and sternum is another important cause of death. Out of 4 achondroplasiac infants born in Sloane Hospital for Women in 8 year period, two were stillborn, the other two, born prematurely, died during the first six months of postnatal life (Caffey, 1958). Out of 8 cases reported by Potter (1952) only 1 survived. Only 1 child out of the 4 cases seen at Hadassah University Hospital survived (Porat et al., 1956). Out of the 3 siblings reported by Cohn and Weinberg (1956) one died at 2 hours, other at 48 hours and the third was stillborn.

After the first year, however, achondroplasiac dwarfs usually lead healthy, asymptomatic lives save for the hazard of pregnancy and delivery. Maccallum (1932) recorded a necropsy on an achondroplasiac woman who died in her 76th year.

Since the genital organs are usually normal, the female may become pregnant and this fact makes the pelvic deformity of great importance as in this case, resulting in dystocia.

The severe antero-posterior contraction of the achondroplasiac pelvis constitutes an absolute obstacle for vaginal delivery. Good antenatal care and hospitalization of the patient during the last month of pregnancy is required in every pregnant achondroplasiac dwarf.

The majority of children born to achondroplasiac patients are normal. According to Mørch (1941) 108 achondroplasiac dwarfs produced 27 children, of whom 17 were normal and 10 were achondroplasiac.

The child born to the patient, being reported here, was clinically achondroplasiac and this was confirmed by radiological examination (Fig. 7).

Fig. 7
Changes of achondroplasia seen at the upper ends of humeri, femora and fibula.

Summary

i. The literature on achondroplasia is briefly reviewed.

ii. A case of achondroplasiac female dwarf is presented who was delivered of a similarly affected female infant by caesarean section.

iii. Obstetrical management and radiological features are described.

Acknowledgements

We are grateful to Dr. L. V. Phatak, Medical Superintendent, Lady Hardinge Hospital, for giving permission to publish the case. Our thanks are due to Mr. D. R. Sandhel of the Photographic Department for making prints, to Mrs. Raj Kanwal Thapar for the radiographs and to

Mrs. Mathur for typing the manuscript.

References

1. Caffey, J.: Pediatric X-Ray Diagnosis, ed. 1, Chicago, 1956, The Year Book Publishers Inc., p. 775.
2. Caffey, J.: Brennemann's Practice of Pediatrics, Maryland, 1958, Chapter 28, Vol. 4, W. F. Prior Company, Inc.
3. Cohn, S. and Weinberg, A.: Am. J. Obst & Gynec. **72:** 1346, 1956.
4. Dandy, W. E. Johns Hosp. Bull., **32:** 5, 1921.
5. Duraiswami, P. K.: J. Bone & Joint Surg. **34-B:** 646, 1952.
6. Hunter, D. and Jupe, M. H.: A Text Book of X-Ray Diagnosis by British Authors, ed. 3, Vol. IV, Philadelphia and London, 1959, W. B. Saunders Company, p. 433.
7. Jayant, K., Mehta, A. and Sanghvi, L. D.: J. Obst. & Gynec. India. **11:** 280, 1961.
8. Maccallum, W. G.: Text Book of Pathology, Philadelphia, 1932. W. B. Saunders Company.
9. Morch, E. T. Op. Dom. Biol. Hered. Human., Kbh., 3, 1941 quoted by Porat, E., Ehrenfeld, E. N. & Brzezinski (1956).
10. Nattrass, R.: Med. J. Australia, **2:** 196, 1958.
11. Porat, E., Ehrenfeld, E. N. and Brzezinski: J. Obst. & Gynec. Brit. Emp. **62:** 903, 1956.
12. Potter, E. L., and Coverstone, V. A: Am. J. Obst. & Gynec. **56:** 799, 1948.
13. Potter, E. L.: Pathology of the Fetus and Newborn, ed. 3, Chicago, 1952, The Year Book Publishers Inc., p. 442.
14. Rischbeith, H., and Barrington, A.: Eug. Lab. Mem., "Treasury of Human Inheritance", 15, parts 7 and 8, 1912, London, Dulau & Co. quoted by Hunter and Jupe.
15. Shah, L. C.: J. Obst. & Gynec. India. **12:** 523, 1962.