Pregnancy in the achondroplastic patient: A case report

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SPAULDING (1942) once called a pregnant dwarf "an excitant of human curiosity" whereas others have considered them not only obstetric curiosities but surgical challenges. A case of pregnant achondroplasia, the most common form of disproportionate dwarfism, is described below.

Case Report

The patient was a 28-year-old Chinese, primigravida, who had been married for 1½ years. She had a normal menstrual pattern, the last normal menstrual period being on 8.4.70 - 14.4.70. Her expected date of delivery was on 15.1.71. The maturity at the first ante-natal visit (7.1.71) was 39 weeks' gestation. The patient was the eldest in a family of 5 children. Her husband, 2 brothers and 2 sisters and both parents were all normal in height and appearance.

On physical examination she presented the typical features of achondroplasia (see Figures 1 & 2). Her height was 4 feet 3½ inches (130.6 cm.)

She also had evidence of toxaemia with a blood pressure of 140/96 mm. of Hg. and oedema legs. The uterine size was about 39 weeks. The foetus was in cephalic presentation with the head unengaged. Both clinical and radiological pelvimetry revealed a grossly contracted pelvis. (see Figures 3 & 4). The typical features noted on X-ray of the pelvis were:—

(i) Deep sciatic notch
(ii) Small narrow sacrum
(iii) Reduction in height (the iliac bones appeared stubby)
(iv) Marked narrowing of the A-P diameter of inlet and outlet.

She was diagnosed as a case of pregnant achondroplasia with a severely contracted pelvis and mild pre-eclamptic toxaemia.

An elective lower segment Caesarean section was carried out on 10.1.71 and a live male baby weighing 2050 gm. was delivered. The latter showed no abnormality and was well when seen 6 months after delivery.

Discussion

Achondroplasia is a distinct entity resulting from an inborn error in the growth and development of cartilage (Maroteux and Lamy, 1964). Approximately 85-90% of all cases occur as a new dominant mutation. Only a few cases are transmitted by autosomal dominant inheritance since

Fig. 1 — Fig. 2: Photographs of patient showing features of Anchondroplasis (height = 130.6 cm).

effective reproductive efficiency is considerably reduced in this disorder (McKusick, 1966). Tyson (1970) found a high foetal wastage of 30% and explained the cause as being due to "defective germ plasm". Neonatal death rate is also high in achondroplastic infants, being due to respiratory embarrassment from thoracic cage abnormalities and neurological deficit, usually due to hydrocephalus. Morch (1941) reported 10 achondroplasics in 94,075 births (mutation rate of 1 : 24,000 gametes). Eight of these cases had normal parents.

The above patient probably represents a new mutation since the family history revealed no other members with achondroplasia. Parental age did not appear to play a role since both parents were quite young at the time of her birth.

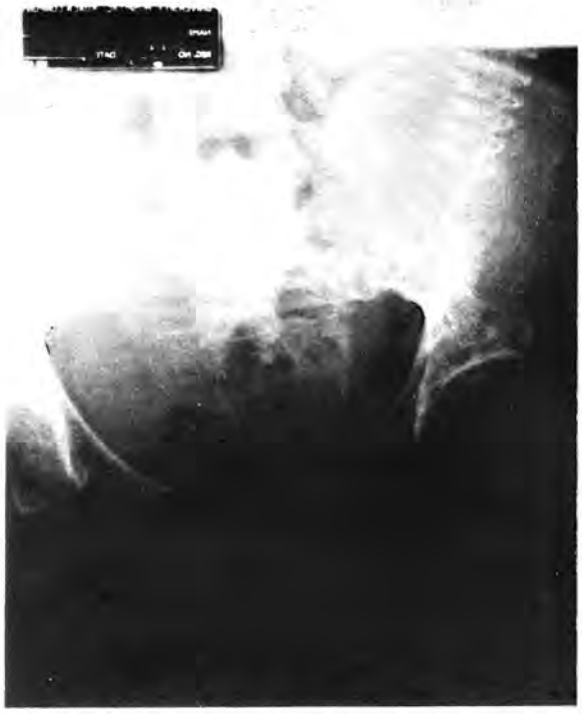
She was married to a normal spouse and as shown in the diagrammatic representation of an autosomal dominant factor, she has a 50% chance of giving birth to an achondroplasic child (Figure 5). The obstetrician is here obliged to make the patient aware of this fact, and help her to arrive at a mutually satisfactory decision as regards further pregnancies. This patient has followed our advice and is now on the contraceptive pill.

Tyson et al (1970) are of the opinion that the single most important objective in ante-natal care is the recognition of impaired cardio-respiratory function. In his series, 2 achondroplasts recalled living propped up in rocking chairs for the last 50 days of gestation because of respiratory embarrassment. This is associated with limited abdominal size. They, therefore, advised early delivery (35-37 weeks). This patient did not present this feature.

Although in Tyson's series of 25 cases there was no evidence of pre-eclamptic toxaemia, Gardiner (1970) has postulated its development in this sort of patients as due to possible pressure on inferior vena cava or renal vessels as a result of prolonged and excessive intra-abdominal distension.

The case under review is the second case of pregnant achondroplasia which the writer has managed. Both had mild pre-eclamptic toxaemia.

The mode of delivery in an achondroplasic



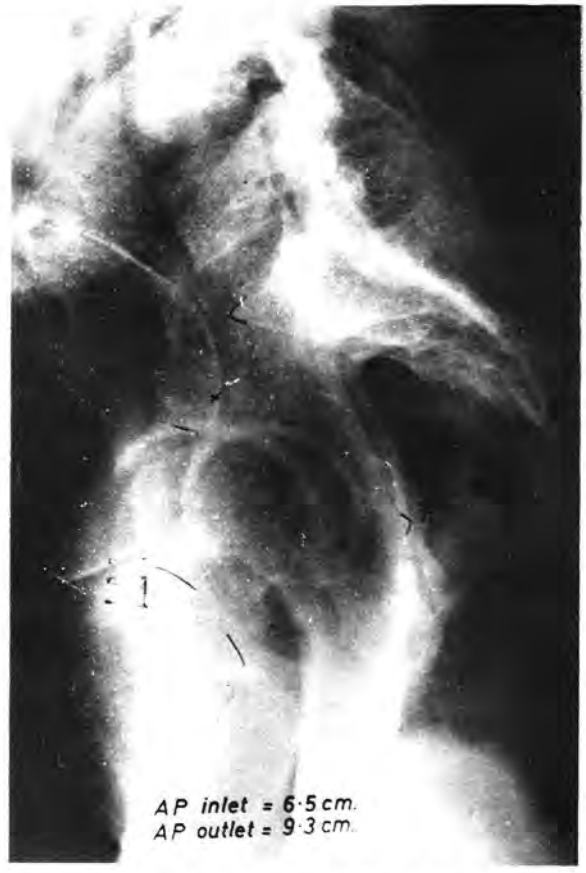


Fig. 3 — Fig. 4: X-ray pelvimetry showing gross pelvic contraction (AP and lateral views).

mother is invariably by lower segment Caesarean section because there is every likelihood of major cephalo-pelvic disproportion from severely contracted pelvis.

Summary

A case of achondroplasia in pregnancy is described. The mode of inheritance of this disease entity, the complications in pregnancy and the method of delivery are discussed.

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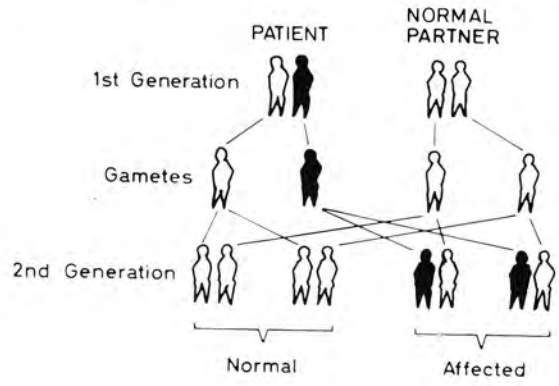

Figure 5 TRANSMISSION OF DISEASE INHERITED AS AN AUTOSOMAL DOMINANT FACTOR