Achondroplasia and Pregnancy

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ABSTRACT

A pregnant achondroplastic dwarf whose mother was also dwarfed, was delivered of a live infant with achondroplastic features. To the authors' knowledge, there has been no previously published report of the occurrence of dwarfism in three successive generations. This is probably because of a high perinatal loss due either to a lethal homozygous gene, hydrocephaly or to respiratory failure.

INTRODUCTION

Achondroplasia as a hereditary skeletal dystrophy was first described in 1791 by von Soemmerring and more recently by McKusick (1956) and Caffey (1966). However, pregnancy among achondroplastic dwarves, once regarded as an excitant of human curiosity, is not well-documented.

The present communication reports the reproductive performance of a pregnant dwarf, and the occurrence of dwarfism in three successive generations.

CASE REPORT

A 23-year-old primigravida was first seen in the antenatal clinic at 32-week gestation with the classical clinical features of achondroplasia – short limbs and short trident hands, short stature with a height of 120 cm, prominent forehead and a saddle nose. She walked with a waddling gait due to bowing of the legs and there was lumbar lordosis. The patient herself was delivered by Caesarean section 23 years ago and her mother was an achondroplastic dwarf with similar features (Fig. 1).

The blood pressure was 120/70 mm Hg and there was no ankle oedema. The fundal height corresponded to a 34-week gestation with longitudinal lie, and cephalic presentation. The foetal head was not engaged, and the foetal heart rate was normal. X-ray pelvimetry revealed gross reduction in pelvic dimensions with a brow presentation. An elective Caesarean section was performed at 38 weeks under general anaesthesia. A live male infant (Apgar score 10) weighing 2,470 gm was delivered; it showed achondroplastic features of short limbs, trident hands, stubby fingers, saddle nose, and a prominent forehead with a head circumference of 33 cm (Fig. 2). The neonatal course was uneventful and there was no respiratory difficulty. The mother and baby were both discharged on the tenth post-partum day.

DISCUSSION

Achondroplastic dwarfism occurs de novo in 80 percent of cases and is transmitted as an autosomal dominant inheritance (Murdock et al, 1969). Our patient was heterozygous since her mother was dwarfed and the father was of normal size; so too was the infant because the patient was dwarfed and her husband of normal stature.

Its rare occurrence in three successive generations is probably due to the tendency of dwarves to have few children, or to the fact that siblings of achondroplastic mothers are not necessarily dwarves, or to an elevated mortality rate particularly among those who are homozygous for the achondroplastic gene (Tyson et al, 1970). Caffey (1966) reported that of four achondroplastic infants, two died within the first six months. Noonan (1968) reported two stillbirths and two neonatal deaths among four achondroplastic dwarves. This high mortality rate is attributed to cardio-respiratory embarrassment from thoracic cage abnormalities, a complication which occurs more commonly with thanatrophic dwarfism or, in some cases,

Fig. 1. Patient with her newborn infant (left) and mother (right). A nurse stands behind

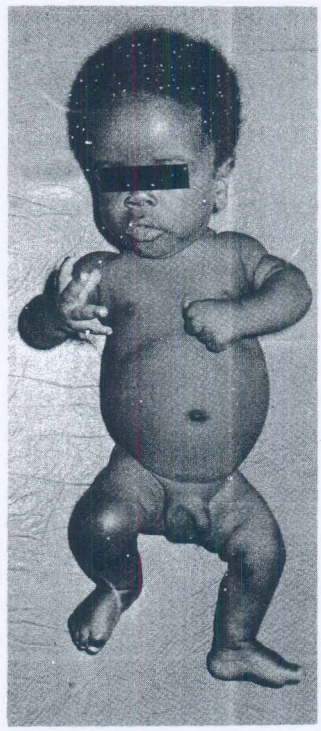

Fig. 2. Infant showing achondroplastic features of short limbs, trident hands, stubby fingers, saddle nose, and prominent forehead

Table: Maternal complications in 17 cases

	No.	Per Cent
Cephalopelvic disproportion	16	96
Pre-eclampsia	4	24
Hydramnios	2	12
Respiratory problems	6	36

to the occurrence of hydrocephalus secondary to obstruction of the cerebrospinal fluid flow at the foramen magnum (Cohen et al, 1967). These two complications were absent in our patient.

Despite the low birth weight of 2,470 gm which is similar to the mean birty weight among dwarves of 2,602 gm (Tyson et al, 1970) and 2,659 gm (Noonan, 1968), the head circumference was that of a normal baby. Consequently, the cephalopelvic disproportion was in fact due to reduced pelvic dimensions, and this is the commonest complication reported in the literature (Table). The alteration in pelvic configuration has been studied by Caffey (1958) who found that the greater sciatic notch is narrowed; the ilium is reduced in height; the sacrum is reduced in width; and the antero-posterior diameter of the pelvic inlet is diminished by forward displacement of the sacral promontory.

The development of pre-eclampsia in two of four patients reported by Lattanzi and Harger (1982) appears to support the contention of Tyson and colleagues (1970) that this complication is more likely to occur in achondroplastic dwarves as a result of compression of the inferior vena cava or renal vessels from prolonged intra-abdominal distension. However, of 17 cases found in the literature including the present case, only four have developed pre-eclampsia.

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