BIRTH OF AN ACHONDROPLASIC INFANT TO AN ACHONDROPLASIC WOMAN

by

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It is believed that the goddess Ptah of ancient Egypt was an achondroplasic dwarf. The much valued daschunds are achondroplasic dogs selectively bred. The extreme rarity of the condition has been well documented. Morch as also Haldane assessed the frequency at 1 in 12000 births, while Slatus quotes a figure as high as 1:100,000. Cohn and Weinberg have postulated that the parents of achondroplasic dwarfs are normal in the large majority of cases. Potter and Coverstone affirm that the birth of an achondroplasic dwarf to a couple both of whom are affected is a rarity. Reviewing the literature, Morch (loc citras) observed that 108 chondrodystrophic parents together produced 27 children of whom 10 were affected. Death in early infancy is almost the rule in these unfortunate infants. Of the 8 achondroplasic neonates observed by Potter, there was only one survivor. At the Hadassah University Hospital, only one achondroplasic infant survived during a 15 year period. The present communication relates to the birth of an achondroplasic infant to an achondroplasic woman.

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Case Report

A 20 year old woman was admitted at term, 22 hours after rupture of the membranes. She was observed to be an achondroplastic dwarf. In view of the cephalopelvic disproportion due to contracted pelvis, a caesarean section was done and a male infant, weighing 1400 gms, was delivered. It was apparent that the infant also was achondroplasic. The head was relatively large, and the facies was typical with the characteristic depression at the root of the nose. The extremities were disproportionately short. The anthropometric measurements were as follows: length 14.7″; span 13.4″; upper segment 10″; lower segment 4.7″; ratio of the upper to the lower segment 2.13:1; circumference of the head 14.2″; circumference of chest 9.6″. X-rays of the extremities showed the classical changes of achondroplasia. The neonatal period was uneventful. Mother and child were discharged on the 21st postoperative day. The husband (first cousin of patient) and other members of her family were normal.

Comments

The rarity of "Hereditary" achondroplasia and the relative frequency of the "Sporadic" variety warrants comment. The consensus of opinion seems to be that the chondrodystrophy is transmitted as a mendelian dominant. Schaffer however states

that the disease may also be transmitted as a recessive. The affected person is almost always heterozygous. Since 50 per cent children of an affected individual will inherit the mutant gene, half the offspring may theoretically be expected to manifest the disease. Considerably less than the expected 50 per cent are clinically affected. Motulsky and Hecht point out that of every 100 persons carrying the given dominant gene, only 70 per cent will clinically express it. This factor also explains the "Skipping Phenomenon" met with in these cases. Sporadic cases are either due to a fresh mutation, or lack of expression of the gene in one of the parents. The rarity of achondroplasic infants being born to parents who are both affected, is attributed by Porat *et al.*, to the fact that the homozygous foetus is of low viability and gets aborted early in pregnancy. As is only to be expected, the entity has been reported in sibships. Cohn and Weinberg (loc citras) reported the birth of identical hydrocephalic achondroplasic dwarfs (twins) followed after several years by a single sibling presenting with the same syndrome.

Severe dystocia is to be expected in the delivery of achondroplasic women in view of the severe anteroposterior contraction of the pelvis. Porat *et al.*, (loc citras) draw attention to the frequency with which false labour pains occur in such patients. Deliveries associated with the birth of achondroplasic infants are of considerable obstetric significance. The incidence of breech presentations is high, and hydrocephalus when present, may lead to dystocia. Hydramnios is common. In fact Jensen attributes the entity to increased amniotic pressure consequent on hydramnios.

Shortness of stature is evident from birth. The extremities are short, the thighs and arms being more truncated than the legs and forearms. The ratio between the upper and lower segments exceeds the normal of 1.7:1. The disproportionately large head with relatively shortened base, broad face with characterestic depression at the root of the nose, as also the broad hand with fingers of equal length diverging from each other permits the diagnosis to be made even in the neonate. Mercer affirms that the head is both absolutely and relatively large. Mac Callum and Koller state that hydrocephalus is common. On the other hand Draps categorically states that hydrocephalus is extremely rare in achondroplasia.

There has been no uniformity of opinion as regards the high fatality rate of achondroplasic infants during early infancy. The abnormal mass of bone formed by synostosis of individual centres of ossification around the foramen magnum can cause compression of the spinal cord. Porat *et al.*, (loc citras) incriminate pulmonary hypoplasia which is common. Prematurity and coexistent anomalies if any, are probably contributor.

Achondroplasia is a classical example of a malformation caused by derangement in tissue differentiation. (Potter). As Schaffer (loc citras) points out, the basic defect lies in retardation and irregularity of cartilaginous and osseous growth. Microscopically, the cells of the epi-

physeal cartilage are large, are not arranged in rows, and show an undisciplined tendency to grow in all directions. (Boyd). The classical X-ray picture of the skeletal system is too well known to bear recapitulation.

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