AN ACHONDROPLASIC TWIN.

BY

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In an interesting communication on the etiology of Mongolism, which appeared in the *British Medical Journal* of June 30th, 1928 (p. 1106), Dr. Hubert Armstrong incidentally mentions that this form of mental deficiency is becoming increasingly common. After twenty-three years' experience of Leicester school children I had independently come to the same conclusion, so that any inquiry that would throw light on the cause and possibly lead to the prevention of this distressing condition would, I am sure, be very welcome.

In his article Dr. Armstrong records the case of a Mongol twin which he had recently seen, and, on reviewing the facts relating to it and similar cases, he comes to the conclusion that Mongolism is due to some maternal influence exercised upon the ovum during its maturation. In connexion with this case of Mongol twin it appeared to me that it might be useful to consider the causation of another form of imperfect growth—namely, achondroplasia—especially having regard to an achondroplasic twin that I saw at my clinic recently. The notes of this case are as follows.

Family History.—The father is healthy and his height is 66 inches. There is no history of dwarfism in his family. The mother is healthy and is 64 inches in height (her father was 74 inches in height). The great-aunt on the maternal side is said to have had a son who grew to adult life but always remained a dwarf and in many respects resembled the patient. The mother's first child, a girl, was born in 1909; she is healthy and is 65 inches in height. The second child, a boy, was stillborn in June, 1916. The third and fourth children, two boys, were born in December, 1917—the twins referred to above. The fifth child, a boy, was born in August, 1919. He is healthy and is 62 inches in height, being about two inches above the average for his age. The sixth child was stillborn two years later.

Personal History.—Photograph No. 1 shows the twins aged 4½ years. The boy on the left was the first born and weighed 7½ lb. He is healthy and exceptionally tall for his age, being 60½ inches in height—that is, nearly the height of an average boy of 14 years. Apart from his stature the only thing noticeable about him is that whilst his hair as a whole is brown, he has a white patch about the size of a tennis ball, and his father has the same defect. The boy on the right weighed 7½ lb. at birth. "He was very different from his brother in appearance with his peculiar head and flat nose." For the first three months he was less vigorous than his brother. He is quite intelligent and healthy apart from his achondroplasic condition. He is now 40½ inches in height. Photographs 2 and 3 show the head to be relatively large, with well-developed frontal and parietal bones; the up-tilted nose; high arched palate (mouth breathing); the relatively long body; the short limbs with proximal segments shortened more than the distal; the short stumpy hands; the marked lumbar lordosis; the well-developed muscles and general thickness of the bones characteristic of achondroplasia.

It will be seen from the above notes that the first twin and the boy born at the subsequent pregnancy are exceptionally tall for their age, and it is probable that they will eventually be much taller than their parents. I have often noticed that a mother who is comparatively short, but who comes from a tall stock, as in this case, transmits the character of tallness to her male offspring. This fact partly accounts for the stature of the first twin.

Although the twins are of the same sex, their physical characters are so remarkably different that they cannot be described as homologous or identical twins. Consequently it may be taken for granted that they were developed from

two distinct ova. In these circumstances the placenta may remain permanently distinct, but even in such cases fusion of the placental areas eventually takes place. It is generally accepted that the anastomosis of the placental vessels may be such as to result in marked differences in the general development and vigour of the two foetuses. This disparity is often explained by saying that the stronger foetus has made a special claim on the blood supply, which has enabled it to develop at the expense of the other. In the present case there was no great disparity in weight at birth, there was no general atrophy, but the smaller child had some special defect of the cartilages and bones. Even granting that this foetus had a smaller supply of blood than the other, it appears to me improbable that this could have had such a specific and selective effect.

Various theories have been suggested to account for achondroplasia. The resemblance of achondroplasia to rickets is so close that at one time they were thought to be the same morbid process, but Gilford¹ states that the points of difference are so great that most authorities now regard the two conditions as two distinct diseases. Marie² holds that the dystrophy of cartilage is an effect and not the cause, and that the disease is a general dystrophy comparable to myxoedema. On the other hand, Castin³ has pointed out that the disease starts *in utero* before glandular activity begins, and so cannot be due to any defective internal secretion. He believes that it is toxic in origin and is akin to rickets. Pélouquin⁴ and others believe it to be due to maternal infection. Neither of these views, however, appears to me to be in harmony with a case of twins where one child is normal and the other achondroplasic. The question of possible relationship between achondroplasia and gigantism has been raised by Lannois,⁵ who reported an instance in which an achondroplasic had brothers who were giants. Since the interval between the births of the giants and the dwarf was very short, Lannois suspected the cause to be due to some dyscrasia on the part of the mother owing to frequent pregnancies. But this view does not seem tenable in the case of the achondroplasic twin, for the pregnancies were not very close together, and, further, the mother gave birth to an exceptionally healthy child at the same time as the achondroplasic one.

The question naturally arises whether achondroplasia is an inherited variation; to my mind this is probably the case, for there is no doubt that the condition is sometimes transmitted from parent to offspring. In 1913 I examined and photographed three dwarfs who were being exhibited at the Leicester Fair. The family consisted of father, mother, and daughter. The father was extremely small, though fairly well proportioned. The mother and daughter were typical achondroplasias.

Many similar cases have been recorded, but one published by Dr. Porter⁶ is extremely interesting. In this instance an octogenarian and

his two sons were affected, and the father and the brother of the former were also said to be achondroplasic.

In my experience achondroplasia is uncommon; its rarity is probably due to this useless variation being naturally eliminated in many cases by the death of the foetus owing to early ossification, or to the difficulties of parturition in the case of the pregnant achondroplasic woman.

REFERENCES.

- ¹ Hastings Gilford: *The Disorders of Post-natal Growth and Development*. - Emerson: *A System of Medicine*, by Osler and McCraw, vol. 1.
- ² Porter: *British Medical Journal*, 1907, i, p. 12.

FIG. 1

FIG. 2.

FIG. 3