HEREDITARY SCOLIOSIS

BY

HUGH G. GARLAND, M.D., M.R.C.P.

HONORARY PHYSICIAN, LEEDS PUBLIC DISPENSARY AND HOSPITAL

There are now many well-recognized skeletal deformities in man which are inherited according to Mendelian law, and a large proportion of these are inherited as Mendelian dominants—that is, each affected person has an affected parent and an affected grandparent, and on the average half the children of an affected parent are themselves affected; among the commoner skeletal defects inherited in this way is brachydactyly.

One of the commonest deformities seen in hospital practice is scoliosis, the causes of which are numerous and often clearly understood, but there seems to be considerable doubt as to the importance played by heredity in the production of this condition. In 1896 Tubby (*Deformities*, p. 104) considered the hereditary factor to be common and important, and he quoted two families from his own practice in each of which several members were affected by scoliosis. Most authoritative articles on the subject mention the hereditary factor, but it is usually dismissed in a cursory manner. The discovery, then, of a family in which scoliosis is seen in five generations is important, especially as the deformity appears to be primary—that is, there is no recognizable disease of the nervous system (for we know that scoliosis is a frequent accompaniment of such hereditary diseases as Friedreich's ataxia and hypertrophic neuritis) or of the vertebrae.

As will be explained in the detailed description of the family the deformity in these cases has not been associated with any disability, and, indeed, several affected adults were not aware of their abnormality until I pointed it out. The pattern of the deformity is remarkably constant in all the affected persons, and it takes the form of the usual distortion of the thorax resulting from scoliosis. Briefly, there is a prominence of the ribs and costal cartilages of one side, affecting the lower more than the upper, the opposite side being flattened; the sternum, being rotated about its vertical axis, appears to be narrow; on the side of the prominence, which is well marked in the chondro-sternal junctions, the clavicle is raised, the shoulder being higher and straighter; the opposite clavicle is lower and not so prominent as its fellow; the angle of Louis is prominent. The degree of this distortion of the chest will, of course, depend on the position of the axis about which vertebral rotation occurs. In all these cases the scoliosis is either slight or not apparent on inspection, but it can always be seen by *x*-ray examination. The concavity is usually to the left in the lower dorsal and lumbar regions.

A,II,1 and A,I,1 and 2 are dead, but I,3, an intelligent man, who has appreciated his abnormality all his life and who has been carefully examined, assures me that his father and grandfather had exactly the same deformity, though it caused no disability; he says that his father, who died aged 65, was "affected on the left side." I,3 is a man of 64, and shows marked prominence of the left lower costal cartilages, starting at the third. His sisters and cousins have not been examined, but are said to be normal; I have written to I,1, who confirms that he has no deformity; I,5 and 6 say the same. I,2 died of cancer aged 55, and I,4 died aged 40. Nothing is definitely known of D,I,1, and D,I,2 is said to be normal. D,I,3 has been examined. She is a woman of 42 who has had a "pigeon breast" all her life; the left side of the chest bulges forward, although in this case the upper part of the chest is the most affected. D,I,4 was killed, aged 24, but the father is quite certain that the left side of the chest was affected. D,I,5 died aged 9 months and

nothing is known of him. D,I,6 died at the age of 36, and is said to have been normal. D,I,7 is 34, and has been seen; he knew nothing of his deformity, and again the prominence is on the left side. D,I,8, aged 32, has also been examined; she was said to be normal, and admitted that she had not noticed her deformity until it was pointed out to her. In this case the prominence is on the right side and is slight; there is no scoliosis obvious to the naked eye. She, like several of the others, has always had difficulty in getting her clothes to fit at the shoulders. D,I,9, aged 30, has been examined and is normal. D,I,10 has also been examined; in her case the right side is prominent, and she knows that the deformity has been present since birth. D,II,1 and 2 have not been seen, but are said to be normal. D,II,3, 4, and 5 were also said to be normal, but D,II,3, aged 12, was found to be affected on examination, the prominence

being right-sided and confined to the lower ribs; scoliosis was only demonstrable by *x*-ray examination. D,II,6, aged 15 months, has been examined, and is apparently normal.

It is interesting to note that I,3 shows two other congenital abnormalities. One is an accessory nipple on the left side, and the other is a "shagreen patch," of considerable size and similar to those so commonly seen in cases of epiloia, which lies symmetrically over the lower dorsal region. Similar abnormalities have not been seen in the other affected persons.

It will be seen that the deformity runs directly through five generations with the single exception of D,I,6; but this person is dead, and as D,I,8 had failed to notice her own deformity it is quite possible that D,I,6 was affected. This being the case, and considering the last three generations, there are sixteen children of affected parents; of these, eight are affected and three found to be normal on examination. In the case of the remaining five it is impossible to make a definite statement, but it is probable that the majority were unaffected. This gives as close an approximation to the 1:1 ratio as can be expected with such small numbers.

Here, then, is an example of scoliosis inherited as a simple Mendelian dominant, but the exact pathological anatomy is obscure. Radiograms have shown no structural defect in the vertebral bodies or intervertebral disks, nor have these patients shown any evidence of disease of the nervous system. So far as I know this is the first recorded example of scoliosis inherited as a Mendelian dominant, and it may give the clue to many cases of scoliosis of hitherto unknown origin.

It is proposed to hold an international congress of electro-radio-biology in Venice in September to study exclusively the biological phenomena caused by *x* rays and radium, independently of their application to medicine and therapy. The subjects to be discussed include the probable influences on organic and organized matter of the various rays, radiations, or radio-active salts in organic combinations, electric states of the atmosphere and their effect on biological conditions, and the question of the possible inheritance of such modifications. Further information may be obtained from Dr. Giocondo Protti, S. Gregorio 173, Canal Grande, Venice.