CASE REPORTS

A CASE OF ARACHNODACTYLY

BY

R. M. STEWART, M.D., F.R.C.P.Ed.,

Medical Superintendent, Leavesden Hospital

According to Burch (1936) the total number of cases of arachnodactyly reported in the literature up to the year 1936 was in the neighbourhood of one hundred and twenty, and from an analysis of these he concludes that as a general rule the nervous system and the mentality are not affected, the general intelligence being often especially good and consistent with the possibilities of education.

In my experience this condition is by no means rare in the feeble-minded, who, indeed, afford a happy hunting-ground for nearly all the recognized forms of skeletal deformity. In the course of the last fifteen years I have seen five cases of arachnodactyly with associated mental defect, of which the following is an interesting example.

Clinical report

G. B., a male imbecile, aged sixteen years, was admitted to Leavesden Hospital on January 17, 1920.

History.—He was the third child in a family of five ; the second died of scarlet fever when nine years old ; the other three children were healthy. The father was healthy, but the mother suffered from cardiac disease and had an attack of rheumatic fever some months prior to the birth of the defective. Labour was normal and the patient was born deformed. No history of skeletal disease could be obtained in the antecedents.

State on admission.—The patient was an epileptic imbecile with a mental age of five years. He was clean, tidy and a useful ward worker. Unlike most epileptics, he was docile and good-tempered. His fits were frequent and severe.

PHYSICAL EXAMINATION.—September 16, 1924. Owing to his numerous skeletal deformities the patient's appearance was most unusual (fig. 1). He was sixty inches in height, with a head of unusual contour, a narrow chest, limbs of unusual length, a general absence of subcutaneous fat and a poorly developed musculature. The head measured fifty-three cm. in circumference ; breadth fourteen cm. ; length eighteen cm. ; cephalic index seventy-seven. The cranium was somewhat square, with a high forehead overhanging the face, prominent superciliary ridges and slight frontal bossing. The ears were large and stood out from the head in their upper halves. The nose had a sunken bridge with large, broad nostrils directed forwards and downwards. The chin

was large and the inferior maxilla presented peculiar spur-like processes projecting downwards from its angles. The eyes were rather wide apart, the palpebral fissures obliquely set, sloping downwards and outwards. The pupils were myotic and there was a bilateral correctopia, equal and opposite in the two eyes, the position being up and out. There was a slight degree of rotary nystagmus. The lids showed granules, typical of trachoma.

The upper limbs were abnormally long and thin, the elbow joints enlarged, the ulnar heads forming prominent swellings on the inner aspect of the antecubital fossae. Full extension of the elbows was not possible. On both sides the radius and ulna were curved antero-posteriorly and the upper third of the radius was bent inwards towards the ulna. The hands deviated to the ulnar side and the carpal joints were loose with bursae on their dorsal aspects. The

FIG. 1.—The patient shows thoracic deformity, elongated extremities, absence of hair and adipose tissue, and multiple herniae.

FIG. 2.—Photograph taken after death. The left hand of patient. Note the long and slender fingers.

metacarpal bones could be moved freely in all directions and the digits were remarkably long and slender. At the interphalangeal joints an extreme degree of flexion or extension was possible. In particular, the terminal phalanges, which were slightly hyperextended, could be bent upwards to a right angle. On each hand the minimus showed slight contracture at the first interphalangeal joint. The thumbs were double jointed. The nails were long, slender and curved from side to side (fig. 2). The lower limbs were also long, with small buttocks, poorly defined trochanters and abnormally mobile and small patellae, their transverse measurement being one and a quarter inches. But the thinness which characterized the upper limbs was not so evident in the lower; indeed, the legs had an appearance of solidity. The ankles and feet were broad and bent inwards in the neighbourhood of the base of the first metatarsal bone. The distal phalanges of the three outer toes were deflected towards the middle

line, and although all the toes were long they were not quite so striking as the fingers (fig. 3). The heads of the metatarsal bones were dislocated upwards on to the dorsum of the feet, and like the metacarpals could be displaced freely in all directions. Owing to the laxity of the joints of the lower limbs and the generalized hypotonia, the feet could be placed behind the patient's neck without causing him any discomfort.

The thorax was short and narrow. The sternum projected forwards with a deflection to the right and a depression at its lower end. There was considerable flattening of the chest wall, with retraction of the intercostal spaces. There was no rickety rosary. The waist was narrow ; the spine showed

FIG. 3.—Photograph taken after death showing the peculiar deformity of the feet and the excessive length of the toes.

lordosis and scoliosis. The upper central incisor teeth were large, widely spaced and inclined towards each other at their free edges ; the canines were hypoplastic. The palate was low and broad.

The neck was rather long and broad, with some enlargement of the cervical glands. Sexual development was normal, but there was no growth of hair on any part of the body except the scalp. The thyroid and lungs were normal. Examination of the heart revealed feeble sounds with a thrill and a presystolic murmur heard at the apex. The abdominal wall was lax and there were large double inguinal and umbilical herniae. By holding his breath, the patient was able to distend all three to about three times their normal size, an accomplishment which afforded him considerable pleasure. Laboratory tests were negative. A blood Wassermann reaction performed on September 29, 1924, was negative. The superficial and deep reflexes were normal.

Death from lobar pneumonia occurred on January 14, 1926. An autopsy was held twenty hours after death.

Autopsy.—In addition to frontal bossing, the calvarium showed marked thickening of its inner table, to which the dura mater was firmly adherent. The brain weighed forty-seven ounces and apart from a simple convolution pattern showed no naked eye abnormality. Both lobes of the left lung were in the stage of grey hepatization. The heart weighed seventeen ounces. The right auricle and ventricle were markedly dilated, the mitral valve segments were thickened, with a moderate degree of stenosis of the orifice. The hernial opening at the umbilicus admitted four fingers.

Discussion

The chief clinical features of arachnodactyly may be summarized as follows :

Skeletal defects.—In the majority of cases the skull is dolichocephalic with prominent supraorbital ridges, bossing of the frontal eminences, a broad and sunken nose, a narrow palatal arch and a massive chin. A certain degree of hypertelorism is not uncommon. The ears usually protrude from the head in their upper halves and show enlargement of their lobes. Usually normal, the teeth may be long and narrow, like the bones of the limbs. The face is thin and may present a prematurely old and pained expression. The limbs are excessively long and slender. Not only is there a real increase in the length of the extremities compared with that of children of the same age, but also a relative increase in comparison with the height. Thus, the extended arms may reach almost to the knees. The metacarpal bones and phalanges show a marked increase in length, the lengthening in some cases being relatively greatest in the terminal phalanges, giving to the fingers a delicate, spider-like appearance to which the condition owes its name. Roentgenograms may reveal the presence of epiphyses at the proximal and distal ends of the first and second metacarpals and all the phalanges. The feet also show an elongation due to the long and slender metatarsal bones and toes ; of the latter, the outer three often show an inward deflection of the terminal phalanges. Various thoracic deformities may be encountered. Of these ' trichterbrust ' is the most common. In some cases the thorax is pigeon-breasted, and in others narrowness and flattening of the chest wall may be seen. In nearly all cases there is some degree of kyphosis or scoliosis, to which there may be added winging of the scapulae. Accompanying these skeletal changes there is nearly always a laxity of joints and ligaments, more particularly those of the elbows, fingers and knees. This may be so marked as to permit of actual subluxation. Various writers describe contractures : there may be inability to supinate, to extend fully the thighs or the fingers.

General and visceral defects.—Emphasis is laid by nearly all writers on the poorly developed musculature and the striking degree of hypotonia, which may suggest amyotonia congenita. Subcutaneous fat is often notably deficient and its absence accentuates the unnaturally long arms and legs. In a significant proportion of cases cardiac disease is present. The defect may be of a congenital nature, such as patent foramen ovale, or valvular disease acquired sub-

sequent to an attack of endocarditis. In a few cases deformities of the blood vessels have been noted. As a rule the nervous system escapes implication and the general intelligence is usually described as normal.

Ocular defects.—In recent years the association of arachnodactyly with ocular changes has received much attention from the ophthalmologist. In about one-half of all the recorded cases ectopia lentis is present. The ectopia may be seen in all positions, though an upward dislocation is the commonest. Not infrequently the subluxation is incomplete and the lenses may be smaller than normal. Opacities of varying size and density have been described. Associated with ectopia lentis there are usually iridodonesis and contracted pupils; the latter dilate feebly under the influence of a mydriatic. A myopic elongation of the eye is usually present, and myopia as much as forty to sixty diopters has been recorded. Hypermetropia is rare.

On comparing the clinical features of a typical case of arachnodactyly with those found in the case recorded above, it will be seen that in the latter the majority of the cardinal signs were present. The presence of frontal bossing, prominent supraorbital ridges, sunken nose, malformed ears, massive chin, abnormally long hands and feet, thoracic and spinal deformities, cardiac disease and an extreme degree of muscular hypotonia left no room for doubt in the diagnosis. It is unfortunate that as the state of the lens escaped observation, it is not known whether a displacement was present, but the presence of bilateral correctopia, which is known frequently to accompany ectopia lentis, suggests that the employment of a mydriatic might have revealed this malformation. In certain features the appearance of the patient was suggestive of rickets, but no history of this condition was forthcoming. Unquestionably in its thoracic and spinal deformities and general laxity of ligaments and muscles arachnodactyly bears a superficial resemblance to rickets, but there appears to be no evidence for believing that rickets plays any part in the etiology of this condition.

Several features presented by the patient were not typical of arachnodactyly. According to Young (1929) in sixty-five per cent. of cases dolichocephaly is present; in the case here presented a cephalic index of seventy-seven indicated a mesocephalic type of head. The spur-like processes attached to the angles of the lower jaw, the marked enlargement of the elbow joints and the small patellae were unusual findings, and the presence of three large herniae which the patient could inflate at will were exceptional and no doubt an indication of the general lack of tone and poor development in the musculature of the body.

Various writers have suggested a causal relationship between the multiple deformities of arachnodactyly and modifications of the ductless glands, and in this connexion it is of some interest to note that although sexual development was normal, there was no growth of hair on the patient's face, trunk and limbs. Arachnodactyly, which can be both familial and hereditary, would appear to be a dystrophy in which there is a disturbance in the growth of mesoblastic tissues. The suspensory ligament of the lens is probably of ectodermal origin, and if a defect of this ligament is concerned in the production of ectopia lentis, then some explanation other than a fault in the mesodermal anlage must be found. In

the absence of histological evidence several theories have been suggested. Zentmayer (1936), for example, points out that the lens may owe its displacement to a rupture of the zonular fibres, such rupture being itself secondary to a myopic stretching of the coats of the eye. Another explanation, quoted by Zentmayer, rests on the assumption made by Collins and Hess that luxation can occur when the adhesions between the ciliary body and the margins of the lens are denser and less elastic on one side than on the other.

Another and broader conception places ectopia lentis with its associated skeletal defects in the category status dysraphicus. Originally defined by Bremer (1926), the characteristics of this condition are referable to a disturbance in the embryonal closing of the primitive central spinal canal. The range of disorders which may follow from this interference is wide and varies according to the degree of damage sustained by the cells of the medullary tube. In addition to a disproportion between the height and the span-measurement, the horizontally extended upper limbs exceeding the height of the patient, there may occur syringomyelia, spina bifida occulta, funnel-shaped chest, deformities of the fingers, muscular atony, acrocyanosis and anomalies of the mammary glands, the latter sometimes being associated with dislocation of the lens, Horner's syndrome and heterochromia iridis.

These defects are said to behave as Mendelian traits, and whilst this may also be true of ectopia lentis, the genetic factors underlying arachnodactyly are still in dispute and require further investigation, though there is certainly evidence for the view that the two conditions are linked by inheritance.

Summary

A case of arachnodactyly in an epileptic male imbecile is described. Among unusual features present were a mesocephalic cranium, spur-like processes attached to the angles of the inferior maxilla, enlargement of the elbow joints, small patellae, multiple herniae and an absence of hair on all parts of the body except the scalp.

REFERENCES

- Bremer, F. W. (1926). *Dtsch. Z. Nervenheilk.*, **95**, 1.
Burch, F. E. (1936). *Arch. Ophthal.*, Chicago, **15**, 645.
Young, M. L. (1929). *Arch. Dis. Childh.*, **4**, 190.
Zentmayer, W. (1936). *Arch. Ophthal.*, Chicago, **15**, 676.