CASE REPORTS

ARACHNODACTYLY AND ECTOPIA LENTIS IN A FATHER AND DAUGHTER

BY

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For many years after the original description of 'dolichosténomélie' by Marfan (1896), this peculiar type of local gigantism of the extremities received little general recognition until Ormond (1924, 1929) drew the attention of ophthalmologists to the frequent association of the skeletal deformities with congenital dislocation of the lenses.

Burch (1936), in a comprehensive review, was largely responsible for furthering recognition of the condition in America, and the number of cases that have been reported, mostly in the ophthalmological literature, during the past ten years is an indication that arachnodactyly is not of extreme rarity, and that many milder cases and 'formes frustes' have previously passed unrecognized. Stewart (1939) considers that the condition is not uncommon amongst mental defectives, although the intelligence of reported cases has generally been described as normal. Most of the reports have appeared in the French and German literature, but cases have been described in this country by Poynton (1903), Thursfield (1920), Poynton and Maurice (1923), Ormond and Williams (1924), Cameron (1924), Fowler (1924), Young (1929), Ormond (1929, 1930), Ellis (1931), Hudson (1932), King (1934), Sorsby (1935), Cockayne (1935), and Stewart (1939).

As the name arachnodactyly (Achard, 1902) implies, the most striking feature of the condition is usually the extreme length and slenderness of the digits. The description is not universally applicable, however, since some cases, e.g. the first of those here reported, whilst clearly being mild examples of arachnodactyly, have the arms and legs principally affected, whilst the hands and feet are practically normal. The increased length of the extremities results in a characteristic build, the patients commonly being considerably above the average height, but so slender and with so little subcutaneous fat that the weight is often greatly below the normal. Deformities of the spine, particularly scoliosis and kyphosis, and of the thorax (pigeon-breast, Trichterbrust) are common. Laxity of the ligaments tends to make the patients clumsy and graceless, whilst deformities of the feet (pes planus, hammer-toe) are the general rule. Other bony abnormalities such as spina bifida (Brock, 1927; Zuber, 1928; Weill, 1932) have been described, whilst in three instances (Ellis, 1931; Semah, 1938; Boudet et al., 1939) arachnodactyly has been associated with fragilitas ossium and blue sclerotics.

In about two-thirds of the cases the skull is dolichocephalic. The orbital ridges have sometimes been described as prominent and the jaw as prognathous. The teeth may be normal or long and slender and the palate high and arched.

Radiologically the bones of the extremities, particularly the phalanges, metacarpals and metatarsals, show the extreme length and slenderness observed clinically. Ossification is usually normal for age; extra epiphyses have been described in the digits, but these do not appear to be the rule. Apart from dolichocephaly, the skull shows no characteristic changes, and the sella turcica is usually normal, though it may be reduced in size (Young, 1929). Weill (1932) found calcification in the region of the pineal in one instance.

Congenital morbus cordis occurs sufficiently often (in one-third or more of the cases) to be regarded as an integral part of the complete syndrome. It was present in the second of my cases.

Many other additional congenital abnormalities have been described, either in association with arachnodactyly or occurring in other members of affected families. Some degree of webbing of the fingers is common; the ears are often prominent or malformed; the musculature is poorly developed.

The incidence of respiratory infections appears unusually high, and a number of patients (Hudson, 1933; Piper and Irvine-Jones, 1926; Börger, 1914) have succumbed to pneumonia. It is of interest that in the two latter cases, in which post-mortem examination was performed, congenital abnormalities of the lungs were found. Severe asthma occurred in the second case here reported.

Congenital dislocation of the lenses, giving rise to tremulous irides ('iridodonesis'), small miotic pupils and occasionally to recurrent attacks of glaucoma, is found in approximately 50 per cent. of cases of arachnodactyly (Ormond, 1930). The dislocated lenses are often opaque (as in case 1) and resistant to absorption (Hudson, 1933).

The following two cases illustrate most of the clinical features of arachnodactyly, and its association with ectopia lentis.

Case reports

FAMILY HISTORY. The patients to be described are Stephen V., aged thirty-two years, and his daughter, Ruth V. aged four years. Neither Stephen V.'s parents nor he and his wife Ellen V. were blood relations. Stephen V.'s paternal grandfather is said to have been blind (cause unknown). Stephen V.'s father died in Pembury Hospital in 1940 from cardiac failure following hyperpiesia; his eyes and limbs were normal and at autopsy there was no evidence of congenital morbus cordis. Stephen V. has four brothers and two sisters; one brother suffers from defective sight (cause unknown), but is otherwise normal, the others being normal both as regards sight and build.

Ellen V., aged thirty-one years (wife of Stephen V. and mother of Ruth V.), has been blind from birth. The right eye shows interstitial keratitis and opacity of the lens, whilst on the left there is advanced syphilitic choroiditis and pallor of the optic nerve. The Wassermann and Kahn reactions are still strongly positive, although she has been under treatment for congenital syphilis since 1937. She suffers from migrainous headaches and has had epileptic attacks on several occasions. Her heart shows left-sided enlargement; there is a short presystolic murmur and a blowing systolic murmur in the mitral area. There

has only been one pregnancy (Ruth V.), during the latter months of which she suffered from oedema.

Case 1. Stephen V., aged thirty-two years, has been practically blind from birth. He was brought up in a blind school until the age of sixteen and was subsequently trained as a basket-maker. At the age of ten an unsuccessful operation on his right eye was performed at the Brighton Eye Hospital. He says that he learnt to walk late and 'was always falling about'; he was thought to be abnormally tall and thin during later childhood and adolescence. He has good general health, and apart from the limitations imposed by his eyesight is normally active.

On examination he is of striking build, being tall (6 ft. 2 in.) and exceptionally slender, the length of the arms and legs being particularly noticeable. He takes a size 14½ collar and all ready-made clothes are much too loose for him. The hands and feet, however, are not abnormal and he can wear size 8 shoes.

The skull is not abnormal, except for slight prominence of the orbital ridges and somewhat prognathous jaw. The teeth, particularly those of the upper jaw, are long, widely-spaced and project forward. The ears are also rather prominent.

There are no abnormalities of the spine, thorax, heart, lungs or abdomen. There is poor muscular development, and very little subcutaneous fat, though the patient is actually stronger than his appearance suggests.

EYES. There is bilateral iridodonesis. Both lenses are dislocated. On the right the lens lies one sixth within the pupil and is opaque; on the left the lens is two-thirds within the pupil and is semi-opaque. Unaided vision on the right is one-sixtieth and on the left two-sixtieths.

WASSERMANN and KAHN reactions negative.

Case 2. Ruth V., a girl, aged four years at the time of examination (May, 1940), was born four weeks prematurely weighing six and a half pounds; delivery was normal. The child was thought to be of unusual length at birth, and though she sucked and cried normally, she always appeared thin. She was fed on a cow's milk formula. At no time had she been cyanotic. An umbilical hernia was strapped for the first year.

At nine months of age the child developed pertussis, and at eleven months was admitted to Pembury County Hospital with infantile eczema. Recurrent asthmatic attacks, which have become more severe and frequent as she has got older, began during the second year. They have been associated with the gradual appearance of the thoracic deformity which she now shows. There is no known food or other idiosyncrasy, and the asthmatic attacks occur both in winter and in summer. The child has been admitted to hospital six times on account of asthma since 1937.

During the first year it was noted that the child did not see normally. She cut two teeth at three months of age and had sixteen teeth at a year old. She spoke words at eighteen months and began to walk at two years old. She is still very liable to fall over.

On examination (May, 1940). A tall, extremely slender child, who is active but ungainly and unsteady on her feet. Height 44½ in. (normal for age 38 in.). Weight 35 pounds (normal for age 34 pounds, for height 45 pounds). Fig. 1 shows the characteristic length of the limbs and narrowness of the trunk, whilst the anxious expression of the face is also somewhat typical. The head measures 17.5 cm. in length and 12.5 cm. in breadth. The teeth are normal. The palate is high and arched. There is dorsal kyphosis and laxity of joint ligaments, particularly of the wrists, elbows and knees.

THE HANDS (fig. 2) measure 5½ in. from the tip of the middle finger to the radial styloid (normal control 4½ in.), and show the typical 'spider fingers.'

FIG. 1.—Ruth V., showing tall, slender build and deformity of thorax.

FIG. 2.—Ruth V. : Hands showing arachnodactyly.

THE FEET (fig. 3) are excessively long and slender. There is a hammer-toe deformity of the third and fourth toes on both sides.

THORAX. There is a well-marked pigeon-breast deformity of the chest, with flattening of the lower ribs anteriorly. Scattered wheezing expiratory râles are heard over both lungs.

HEART. The apex beat is palpable in the left fifth intercostal space immediately outside the nipple line, and the area of cardiac dullness extends half

an inch to the right of the right sternal border. There is a loud mid-diastolic murmur best heard at the apex, and also a harsh systolic murmur well heard in the third and fourth intercostal spaces immediately to the left of the sternum. The pulse rate is 100 to 110; there is a good response to exercise tolerance tests.

FIG. 3.—Ruth V.: Long, narrow feet and deformities of toes.

FIG. 4.—Ruth V.: Skiagraph of hands.

FIG. 5.—Ruth V.: Skiagraph of feet.

FIG. 6.—Ruth V.: Teleradiograph of heart, showing enlargement.

The orthodiagram confirms the cardiac enlargement, which is principally right-sided.

EYES. There is bilateral dislocation of the lens. The pupils dilate poorly, and there is bilateral iridodonesis. Visual tests show that acuity is moderately good.

MENTALITY. The child is mentally retarded, but educable, part at least of her backwardness being due to her prolonged hospitalization. She is moderately co-operative, understanding simple requests, and is beginning to speak in sentences. She is clean in habits.

The WASSERMANN and KAHN reactions and the MANTOUX TEST (1 in 1,000) are negative.

Discussion

The association of congenital abnormalities of the osseous system with congenital abnormalities of the eye is seen in other conditions than arachnodactyly. Examples are gargoylism (chondro-osteo-dystrophy and corneal opacities); congenital coloboma of the macula and apical dystrophy of hands and feet (Sorsby, 1935); punctate epiphyseal dysplasia associated with congenital cataract; and the Laurence-Moon-Biedl syndrome (polydactyly and retinitis pigmentosa, etc.). With the possible exception of punctate epiphyseal dysplasia, all these conditions may show a familial incidence and the ocular and osseous defects occur separately. In the case of the Laurence-Moon-Biedl syndrome, in which affected families have been studied in some detail, it has been suggested that a linkage of genes has taken place (Cockayne et al., 1935; Sorsby et al., 1939).

The two cases reported here illustrate the hereditary factor in both arachnodactyly and ectopia lentis, a 'forme fruste' of arachnodactyly with ectopia lentis in the father being followed by the appearance of the classical picture of arachnodactyly with ectopia lentis in the child. (The mother's blindness, though making the family a lamentable social phenomenon, is not regarded as being related to the condition in the child; there is no evidence that parental syphilis plays any part in the production of either arachnodactyly or ectopia lentis.) There was no consanguinity of the parents.

There are numerous reports in the literature in which arachnodactyly and ectopia lentis, combined or separately, have occurred in several siblings or in a parent and children. (Unfortunately few authors state whether or not there was consanguinity of the parents.)

Thus Achard (1902) described arachnodactyly in two sisters, their mother and maternal grandfather; Villard et al. (1931) reported a family consisting of a father (antecedents unknown) who suffered from ectopia lentis, a mother who was normal, and five children. Of the latter, the eldest boy and girl showed both arachnodactyly and ectopia lentis, a third child was normal, the fourth (a girl) had ectopia lentis only, and the fifth (a boy) was normal. Weve (1931) observed twenty-three cases of arachnodactyly in six families, in three of which there was certain evidence of hereditary transmission. The association of arachnodactyly and ectopia lentis was seen in a father, two sons, and a daughter. Weill (1932) observed both conditions in a mother and son, whilst in a second family the mother showed dislocation and calcification of the lenses associated with dwarfism, and her son ectopia lentis and a forme fruste of arachnodactyly. Young (1929) found arachnodactyly and associated congenital abnormalities in a mother, daughter and son. Harrison and Klainer (1939) give a family tree showing arachnodactyly in two siblings in one generation, and three in the next.

Although further familial studies are required definitely to establish the mode of inheritance and linkage of the two conditions, it seems probable that both arachnodactyly and ectopia lentis may be transmitted as Mendelian dominants, either separately or in association. There is no sex-linkage in either condition.

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