CASE REPORTS

ARACHNODACTYLY IN A CHINESE INFANT

BY

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Marfan in 1896 described the first case of arachnodactyly occurring in a girl of five-and-a-half years of age. This patient had long and slender hands and feet, decrease in soft tissue, contractures of fingers, relaxed ligaments and other associated congenital abnormalities. He called the condition 'pieds d'araignée,' because of the long, thin, spider-like feet. The term arachnodactyly ('spider digits') first used by Achard in 1902 has been retained although it describes only one of the many abnormalities in these cases. Other synonymous terms are hyperchondroplasia (Méry and Babonneix, 1902), 'a case of atavism' (Poynton, 1903), acromacrin (Pfaundler, 1914).

Pathogenesis

In 1920, Thursfield dealt briefly with the following two theories of causation of the disease: (1) That it is due to an endocrine disturbance, and (2) that it is in the nature of a primary muscular dystrophy. He thought that the balance of evidence was in favour of the latter theory, but later Ormond and Williams (1924) reviewed the above two theories in greater detail and were not inclined to favour either. They said that arachnodactyly is analogous to mongolism and not to cretinism, since there are usually associated with it several congenital malformations which are present in mongolism but not in cretinism, and as these congenital malformations have failed to react to endocrine therapy, they concluded that the condition could not be of endocrine origin. They have dismissed the second theory (that it is in the nature of a muscular dystrophy) as unsound because they found it difficult to correlate the characteristic symptom of muscular dystrophy, viz. the wasting, with the striking congenital abnormalities. Arachnodactyly is unassociated with a progressive course as occurs in typical muscular dystrophy. The developmental theory of this disorder, to which Ormond and Williams are inclined, is the most widely accepted theory at present. It is said to be due to a disturbance of mesoblastic growth, various mesodermal elements of the body structure having been affected in the early weeks of foetal life. With the exception of the eyes, only mesoblastic tissues are involved. Most probably the lens, which is of ectodermal origin, is secondarily affected by the weakening of the supporting tissues which most authors believe to be of mesodermal origin.

Clinical manifestations

Arachnodactyly or Marfan's syndrome is a congenital developmental anomaly characterized by an increase in the length of the long bones particularly distally. The abnormalities associated with arachnodactyly are not ordinarily detected until the second or third year of life. There are usually 'spider digits,' a slender skeletal development, a spur-like projection of the calcaneus which is especially characteristic, a long narrow thorax which is frequently funnel-shaped, and a high degree of asthenia. The joints and ligaments are abnormally weak, and grotesque displacements of the feet and ankles are possible. Occasionally contractures of ligaments, such as Dupuytren's contracture, occur. The face may have an appearance of maturity because of the decrease or absence of soft tissues; the bulbus oculi are deeply set because of the small amount of orbital fat. X-ray examination sometimes shows a premature development of the ossification centres and early disappearance of the epiphyseal lines.

There are also associated malformations and defects: those in the eyes (in about fifty per cent. of cases) include persistence of the pupillary membrane, tremulous iris, subluxation of the lens, nystagmus, myopia, colour-blindness. In the heart a patent foramen ovale has been reported. According to Piper and Irvine-Jones (1926) congenital defects of the heart form a classical combination. Club feet, flat feet and syndactyly have been reported. The associated structural defects are variable. They are deformities of the ear, webbing of the toes, kyphoscoliosis and thickening of the base of the skull. The intelligence is in most cases normal but may be defective.

Case report

A male Chinese Cantonese infant, aged seven months and fifteen days, was referred by the Lady Medical Officer in charge of the out-patient department, who stated that there was deformity of the skull with slight spasticity of the lower limbs, that on lifting the infant up with the hands under both axillae the limbs were crossed, that the knee-jerks were exaggerated and that the plantar reflex was an extensor response. A diagnosis of Little's disease had been considered.

History. The infant was born at full term by breech delivery on November 5, 1939, after a normal pregnancy. On November 17, it was noted that he was of good colour but thin, and that his hands and feet were bent upwards and rather stiff. The father said that the infant had the facial appearance of old age since birth. The grand-parents and parents showed none of the characteristics of arachnodactyly and no evidence of this disorder could be found in the two elder brothers. The infant was breast fed for five months and then given condensed milk and subsequently some rice, a little pork, beef and eggs.

Examination. The infant recognized both parents. His mental ability was fair. He followed simple orders from his parents. His face appeared prematurely aged and the forehead wrinkled (fig. 1). The head was asymmetrical. There was a prominent boss on the posterior part of the right frontal region with a corresponding prominence on the opposite occipital region conforming with the description of a plagiocephalic skull. There were also smaller bosses on the posterior part of the left frontal region and in both temporal

regions. The anterior fontanelle had closed, leaving a depression which measured 2 cm. anteroposteriorly and 1 cm. from side to side. Both ears were

FIG. 1.—Photograph of patient. Note the wrinkles on his forehead, deeply-set eyeballs, facial appearance of maturity, funnel-shaped chest and Harrison's sulcus.

FIG. 2.—Photograph of patient. Note the characteristic slender skeletal development, the long narrow thorax, the long hands and feet, the long fingers and toes.

large and soft and deficient in cartilage. The bulbus oculi were deeply set. The hard palate was deeply arched and the two upper and lower incisor teeth were present.

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He weighed 12 lb. 14 oz. and had the slender skeletal development characteristic of arachnodactyly (fig. 2). There was generalized absence of subcutaneous

FIG. 3.—Hand of patient marked B in the photograph contrasted with the hand of a normally formed Chinese infant, aged eight months, marked A.

FIG. 4.—Foot of patient marked B in the photograph contrasted with the foot of a normally formed Chinese infant, aged 8 months, marked A.

fat. The thorax was long and narrow. The chest was funnel-shaped and there was a Harrison's sulcus. He could neither sit up nor crawl. The musculature was poorly developed and there was a high degree of asthenia. The hands and

fingers and the feet and toes were long and slender (fig. 3 and 4). Pes planus and a spur-like projection of the calcaneus (fig. 5) were present. The left thumb showed a fixed flexion deformity at the metacarpo-phalangeal joint (fig. 6).

FIG. 5.—Foot of the patient showing spur-like projection of the calcaneus and pes planus.

There was unusual laxity of the ligaments at the knee and ankle-joints, enabling the infant to maintain the grotesque attitude of his legs seen in fig. 7.

There was congenital heart disease, manifested by diffuse praecordial pulsation, systolic murmurs over the pulmonary and tricuspid areas, and an accentuated pulmonary second sound.

The superficial and deep reflexes were present. The pupils were equal and

FIG. 6.—Left hand of patient showing the thumb contracted and flexed into the palm of the hand at the metacarpo-phalangeal joint.

small and reacted readily to light. The infant could recognize his parents and follow a bright light. An examination of the eyes was carried out under general anaesthesia by Dr. R. D. Williamson who reported: 'There is marked megal-

cornea with very high myopia. It is impossible to make out the details of the fundus and it cannot be established if the lenses are dislocated or not. The

FIG. 7.—Note the grotesque position of the legs adopted by the patient.

intra-ocular pressure in each eye is 15 mm. Hg (normal 15 to 25 mm. Hg).'

There was bilateral otitis media and the right testis was undescended.

The following measurements give a comparison between the patient and a normal Chinese infant aged eight months :—

	PATIENT AGED 7½ MONTHS	NORMAL CHINESE INFANT AGED 8 MONTHS
	All measurements in inches	
Length	28.4	24.0
Circumference of head	17.2	16.8
Arm	4.6	4.4
Forearm	4.4	3.6
Hand (from mid-point of wrist to tip of middle finger)	4.2	2.8
Thigh	5.4	6.0
Leg	5.0	5.4
Foot (from heel to tip of big toe)	5.0	4.2

The above measurements make clear the following points:—

1. The patient is 4.4 inches longer than the normal Chinese infant of eight months and his length is equal to the length of a normal American infant of 10½ months (Mead's chart compiled by Arthur I. Blau).

2. The length of the hand is only 0.2 inches less than the length of the forearm while the difference is 0.6 inches in the normal Chinese infant.

3. The length of the patient's foot is equal to the length of his leg whereas in the normal Chinese infant there is a difference of 1.2 inches.

X-ray examination by Dr. J. W. Winchester, Radiologist to the General Hospital, revealed a double scoliosis of the spinal column (fig. 8) and no abnormality in the appearance of the epiphysis was detected.

The Kahn test on the blood was negative. The patient had an attack of

bacillary dysentery of the Flexner type, and was discharged from hospital on July 22, 1940.

FIG. 8.—Skiagram of spine of patient showing double scoliosis.

Comment

This is the first case of arachnodactyly in a Chinese infant recorded in Malaya. The infant was first taken to hospital when he was five months old, at which time Little's disease was suspected. As it is said that the characteristic features of arachnodactyly are not usually noticed until the infant is two or three years old, we were fortunate in finding them, associated with ocular defects, in this patient at the age of seven-and-a-half months. Ocular defects are present in only half the cases, and are not essential for diagnosis. The familial and hereditary nature of the condition was not exemplified in this case, but isolated examples of the condition are known to occur. Some indication of the early presence of arachnodactyly can be gathered from the infant's welfare card where on the twelfth day it was stated that the infant was thin, and at three-and-a-half months that the infant was 'big but thin.' (The welfare nurse probably meant 'long and thin.') This infant's skull was asymmetrical and was of the plagiocephalic type. The patient also showed the classical combination of congenital morbus cordis with arachnodactyly (Piper and Irvine-Jones, 1926).

Summary

(1) A case of arachnodactyly is reported in a Chinese Cantonese infant of seven-and-a-half months.

(2) This is the first case of its kind reported in Malaya.

(3) Nearly all the characteristic features of the disease were recognized in this patient at an unusually early age.

(4) Particular features of the case are the appearance of premature senescence; the abnormal length of the infant as a whole and of the hands and feet in particular; and the presence of congenital morbus cordis.

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