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MEDICAL MEMORANDUM

Marfan's Syndrome in two Nigerian Families

W. I. ADERELE

Department of Paediatrics, University College Hospital Ibadan, Nigeria

Summary

Aderele, W. I. (*1975*). *Nigerian Journal of Paediatrics*, **2** (**2**), 55. **Marfan's Syndrome in two Nigerian Families.** Eight cases of Marfan's Syndrome in two Nigerian families are described. The clinical features suggest dominant inheritance in one of the families, and a mutation in the other. None of the recognised cardiovascular or renal complications occurred in any of the cases studied.

ACCORDING to Dorrance (1947), Marfan's syndrome was first described in 1896. The syndrome is characterized by generalized connective tissue abnormality in the skeletal, cardiovascular and ocular systems. The cardinal manifestations include arachnodactyly, cardiac abnormalities, ectopia lentis and iridodenesis. Since its description by Marfan, there have been many reports of the syndrome from many parts of the world (Fischl and Ruthberg, 1951; Black and Landay, 1955; McKusick, 1955; Roark, 1959; and Sinclair, Kitchin and Turner, 1960). Most of these reports have however, come from Europe and America, and only a few cases have been reported from Africa (Forbes and Gordon, 1964; Adadevoh and Olatunbosun, 1965). The present communication describes eight cases of the syndrome in two Nigerian families.

Case Reports

Case 1 (*B.O., UCH No.* 296057)

Personal History:

This was a male child, aged 6 years, who presented at the University College Hospital (UCH), Ibadan, in August, 1972. The parents complained that since about the age of 2 years, the child had always found it difficult to see properly; he was said to be unable to pick up small objects from the floor. It was also noticed that since he started school 3 months previously, he was unable to read from the blackboard. The mother also complained of a sternal prominence which was first noticed when the child was one year old. Pregnancy, labour, delivery and neonatal period as well as the developmental milestones were normal.

The Family:

The pedigree of the patient's family is shown in Fig. 1. The parents and siblings of the propositus were examined by the author; information concerning other members of the pedigree was obtained from the mother of the propositus. The father was of normal stature. The mother, who is the father's only wife, is a very tall and lanky woman whose physical features are consistent with Marfan's syndrome. She complained of dimness of vision, and ophthalmic examination revealed bilateral subluxation of the lens. The patient has three siblings (two of

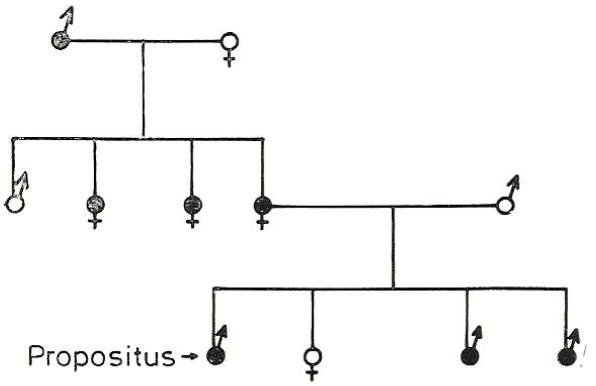

Fig. 1.

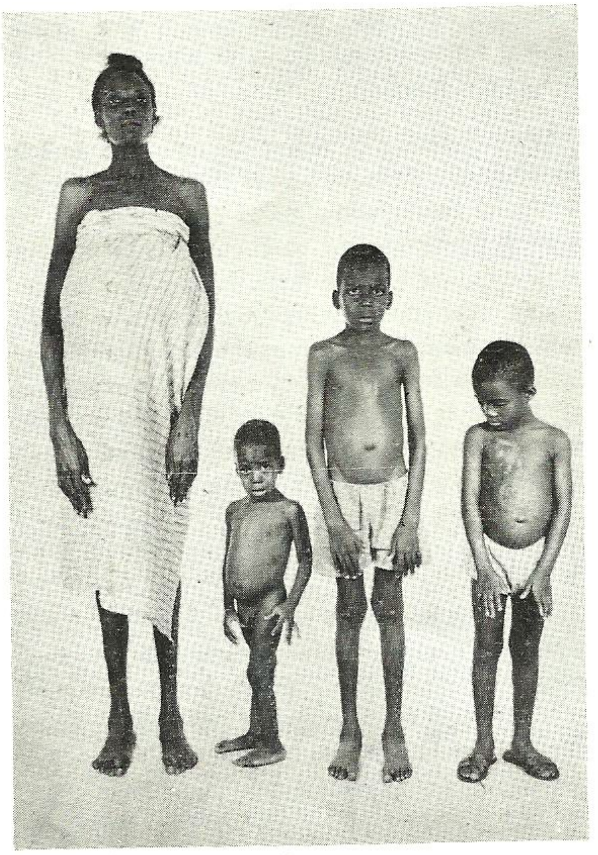

Fig. 2.

whom are shown with the patient and his mother in Fig. 2). Two of the three were males (aged 18 months and 6 months respectively) who, though symptomless, exhibited physical features (tallness, arachnodactyly, dolichocephaly and high arched palate) consistent with Marfan's syndrome. The third sibling, a girl aged 4 years, was normal. The deceased maternal grandfather is said to have been a very tall man who had similar physical features (including dimness of vision) as his daughter (propositus' mother). The propositus also had two maternal aunts with features consistent with Marfan's syndrome, and a normal maternal uncle.

Physical Examination:

(Figures in parenthesis refer to normal average values at similar age).

Height, 128 cm (115 cm); Crown-to-pubis, 53 cm (61 cm); pubis-to-heel, 75 cm (54 cm); arm span, 144 cm (112 cm); metacarpal index, 8.9.

Laboratory Investigations:

Haematocrit, 32 per cent; white cell count, 7,000/c.m.; haemoglobin electrophoresis, AS; serum electrolytes and urea, chest X-ray, electrocardiogram, urinalysis and intravenous pyelography were all normal.

Progress:

After series of examinations by an ophthalmologist, he was provided with special lenses. He has been wearing these with satisfaction, and when he was last seen in March, 1974, he was in good health.

Case 2 (T.A., UCH No. 296413)

Personal History:

This male child, aged seven months, also presented in August 1972. The mother gave a history of generalized body weakness since birth, and inability to control the head or to sit up unsupported. Neonatal period was uneventful.

The Family:

Both parents were middle-aged farmers. There were two siblings, five uncles and aunts. Examination of the entire family including the uncles and aunts revealed no features of Marfan's syndrome.

Physical Examination:

The infant was marasmic, weighing only 5.7 kg. He was severely hypotonic, and unable to sit up unsupported. The head was dolichocephalic. He had divergent squint and horizontal nystagmus. The pupils were very small and did not dilate with mydiatrics (mydriacyl and atropine). Other physical features included dull facies, a high arched palate, long, thin, spider-like fingers (Fig. 3) and toes, and hyper-extensibility of the joints. The feet showed bilateral calcanean spurs. There were no abnormalities in the cardiovascular and respiratory systems. Blood pressure was 80/30 mm Hg.

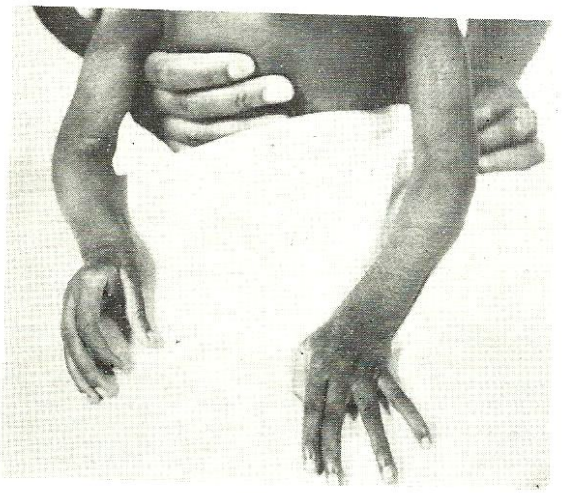

Fig. 3

Physical Measurements:

(Figures in parenthesis refer to normal average values at similar age).

Height, 74 cm (69 cm); crown-to-pubis, 39 cm (42.5 cm); pubis-to-heel, 35 cm (26.5 cm); arm span, 90 cm (66 cm); head circumference, 48 cm (46 cm); metacarpal index, 7.0 (5.3 ± 0.46).

Investigations

Haematocrit, 33 per cent; haemoglobin electrophoresis AS; serum electrolytes and urea, intravenous pyelography and chest X-rays were all normal.

Progress

The patient defaulted from attending the clinic for follow-up, and subsequent enquiries revealed that he died at home sometime in 1973.

Comment

The diagnosis of Marfan's syndrome is a clinical one, based primarily on certain accepted criteria, namely: arachnodactyly, high arched palate and dolichocephaly. These features were all present in the first patient. In addition, the child had physical measurements (arm span greater than the height by more than 7.5 cm; pubis-to-heel length greater than crown-to-pubis length by at least 5 cm; and a metacarpal index greater than 8.4), which according to Sinclair, Kitchin and Turner (1960), are classical features of the syndrome. Subluxation of the lens which is the commonest ocular defect in this syndrome also occurred in the patient.

The extreme hypotonia, arachnodactyly, hyperextensibility of the joints and bilateral calcanean spurs which were present in the second patient are also well-known features of Marfan's syndrome. Furthermore, the nystagmus and poorly reactive pupils, though less common features are well recognised ocular manifestations of the syndrome. The feature which at first, would appear not to conform to those of Marfan's syndrome are the body proportions and the metacarpal index. However, it is to be noted that the child was only seven months old, and at that age the average ratio, crown-to-pubis/pubis-to-heel, in normal children is 1.61 (Wilkins, 1960), whereas in the patient, the ratio was only 1.1. Similarly, the metacarpal index, though only 7, is in keeping with values obtained in cases of Marfan's syndrome presenting at a comparative age (Joseph and Meadow, 1969).

Among the cardiovascular abnormalities described in Marfan's syndrome, those affecting the aorta are frequent in adults (Dorrance, 1947; Goyette and Palmer, 1953; McKusick, 1955),

but in childhood, congenital cardiac abnormalities (septal defects, persistent ductus arteriosus and anomalies of the pulmonary artery) are common (Dorrance, 1947). None of the living patients described in this study has so far been found with a cardiac lesion, nor did the child who died have cardiovascular abnormalities.

The renal abnormalities associated with Marfan's syndrome have been well documented (Fischl and Ruthberg, 1951; Booth, Loughridge and Turner, 1957; Adadevoh and Olatunbosun, 1965). They include congenital defects, hydronephrosis and polycystic kidneys. Urinalysis and intravenous pyclography did not reveal any renal abnormalities in the cases described in this communication.

According to McKusick (1955), Marfan's syndrome may be inherited as an autosomal dominant trait or, it may occur sporadically. From a study of the two families in the present cases, both modes of inheritance seem to have occurred. In the first family, the propositus, his mother, maternal grandfather, three maternal aunts and two siblings, all had features of the syndrome. The mode of inheritance in this family appears to be an autosomal dominant type, the skeletal and ocular anomalies being caused by a single gene or gene complex. In contrast, a study of the second family revealed no features of the syndrome in any other member and this would be consistent with a sporadic occurrence of the syndrome, perhaps, as a result of mutation.

There is no specific treatment for Marfan's syndrome; its prognosis varies considerably according to the manifestations. The most dangerous and often fatal manifestations are those of cardiovascular and renal systems.

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