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Symphalangism, short stature, skeletal anomalies, and accessory testis: a new malformation syndrome

SUMMARY A 17-year-old Jewish Sephardi male is described with symphalangism, short stature, multiple skeletal anomalies, and an accessory testis, which appears to be a new malformation syndrome of possible genetic aetiology.

Harvey Cushing (1916), the well known neurosurgeon, investigated a family in which the affected members could not flex the proximal interphalangeal joints of the hand. He recognised that this malformation was genetically determined and coined the term symphalangism. Since then, a number of genetic syndromes have been described (McKusick, 1978) in which symphalangism is one of the major findings. Recently, we had the opportunity to study a young

man who had symphalangism and other multiple malformations not previously reported in conjunction with this hand abnormality. The purpose of this report is to give an account of what appears to be a new malformation syndrome.

Case report

A 17-year-old Jewish Sephardi male was referred to the Pediatric Endocrinology Clinic of the Sheba Medical Center for evaluation of short stature. The patient's past history was unremarkable.

Physical examination showed a short, well proportioned young man who measured 146 cm in height. His arm span was 139 cm, head circumference 56 cm, and he had a normal upper segment/lower segment ratio. Vital signs were normal. Positive physical findings included the following: short stature, mild acrocephaly, webbing about the upper and lower gums, absence of joint creases over the region of the dorsal surface of the proximal interphalangeal joints of the fingers, but not the thumbs, inability to flex the proximal interphalangeal joints of the hands (Fig. 1), and three testes, two on the right and one on the left (Fig. 2). No other abnormal physical findings were noted.

Radiographic studies showed mild acrocephaly (Fig. 3), lumbar scoliosis, malformed L5 vertebra, coxa valga, bone age of 14 years, and decrease in space with partial fusion of the proximal interphalangeal joints of digits 2, 3, 4, and 5 of the hands. Radiographic studies of the feet were normal.

Chromosome studies showed a normal male karyotype (46,XY). Serum growth hormone and testosterone levels were within normal limits, as were other routine laboratory tests.

Fig. 1 (a) Note the absence of skin creases over the region of the proximal interphalangeal joints of all fingers except the thumbs. (b) Probant unable to make a fist because of symphalangism of the proximal interphalangeal joints of the hands.

Fig. 2 Outline of palpable testes with two on the right and one on the left.

Fig. 3 X-ray of skull showing mild acrocephaly.

The patient appeared to be of normal intelligence, and stated that his father was approximately 180 cm tall and his mother was 170 cm tall. His parents were not related. He had 5 sibs, all of whom were of normal stature. No other family members had any of his malformations, according to the patient. The other family members were not available for study.

Discussion

Although the aetiology of the constellation of findings in this patient is unknown, there is some indication that they may be of genetic origin. Symphalangism by itself is almost always genetically determined (McKusick, 1978) and, when associated with other malformations to form a distinct syndrome, the cause is frequently genetic. Since other members of the proband's family were not available for study,

it is not possible to draw any conclusions as to the mode of transmission of this disorder.

Polyorchidism, though rare, is a well recognised finding and triorchidism is the most common (Gould and Pyle, 1956). Familial polyorchidism has been reported, though most instances represent isolated cases.

Since little is known about this syndrome, it is difficult to know its prognosis. There is no adequate treatment for symphalangism, and the presence of an extra testis does not usually require surgical removal. The cause of his short stature is not known.

It is hoped that the recognition of this syndrome by others will clarify the many questions that remain unanswered.

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Gastric adenocarcinoma due to ataxia-telangiectasia (Louis-Bar syndrome)

SUMMARY A 26-year-old male with ataxia-telangiectasia died with a gastric adenocarcinoma. Malignancy is a recognised complication of this condition, the majority of cases being reticuloendothelial. There have been three reports of gastric adenocarcinoma associated with ataxia-telangiectasia; this, however, is the first in the British published reports.

Case report

A 26-year-old male presented with weight loss, anorexia, and dyspepsia. Ataxia-telangiectasia had