Multiple pterygium syndrome*

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SUMMARY The multiple pterygium syndrome is a rare autosomal recessive condition characterised by arthrogryposis multiplex congenita, pterygia of the neck, fingers, and antecubital, popliteal, and intercrural areas, growth retardation, and facial, vertebral, and genital anomalies. We present two unrelated patients of 17 and 6 years of age, respectively, affected with this condition. We describe the natural history of their disorder since birth and review the spectrum of phenotypic variation of the multiple pterygium syndrome in 25 published cases.

Arthrogryposis multiplex congenita is a non-specific and aetiologically heterogeneous condition characterised by muscle wasting and joint contractures evident at birth, without signs of progressive neurological disease.¹ Many genetic and non-genetic disorders are known to result in arthrogryposis, having as their common denominator a pathogenetic mechanism involving decreased fetal joint movement.² One such entity is the multiple pterygium syndrome (MPS), which has been previously described under the non-specific names of arthrogryposis multiplex congenita³ or Bonnevie-Ullrich syndrome.^{4,5} This disorder was defined initially by Matolcsy⁶ and is characterised by growth retardation, multiple pterygia involving the neck, fingers, and antecubital, popliteal, and intercrural areas, flexion contractures of numerous joints, genital anomalies, and cleft palate.⁷

The multiple pterygium syndrome is rare and a recent review listed 20 patients in whom the diagnosis of MPS seemed certain.⁸ We report here two additional subjects with the syndrome, ascertained during a survey of patients with arthrogryposis multiplex congenita treated at the Children's Orthopedic Hospital of Caracas.

Case reports

CASE 1

A girl was born on 7.11.62 after an uncomplicated term pregnancy and vaginal delivery. There was no history of medication taken by the mother during gestation. The mother was 32 years old and the

father 35 years old at the time of birth; both were healthy and unrelated. This was the mother's tenth pregnancy the first four of which were by another husband. All the previous pregnancies produced normal liveborn children. There was no family history of spontaneous abortions or congenital defects.

At birth the infant had flexion contractures of the elbows, wrists, fingers, and left knee, as well as bilateral pes talus valgus with rocker-bottom feet. At 3 years of age she was able to sit but could not stand. Dislocation of the right patella, shortening of the left femur, and contracture of the left hip adductor muscles were detected at this time. At 6 years of age she was able to stand with support but could not walk. Wasting of the muscles of the limbs was noted at this time. Pterygia were noted in the neck and popliteal regions, more marked on the left side. Flexion contractures of the fingers, adducted and apposed thumbs, and soft tissue syndactyly between the proximal phalanges were also evident. Later she began to develop dorsolumbar scoliosis with left convexity. The left popliteal pterygium was partially resected, the dislocated right patella was replaced, the webbing of the first interdigital spaces was resected in both hands, and bilateral first metacarpophalangeal arthrodesis was performed to allow better function of the persistently adducted thumbs.

Physical examination at the age of 17½ showed a young female, 140.5 cm tall, able to stand and walk with an awkward gait and marked left sided limp. The head was normocephalic, and there was a low posterior hairline and large, cup-shaped, low set ears (fig 1). She had an antimongoloid slant of the palpebral fissures, mild ptosis of the right lid, prominent nasal bridge, downward turned mouth,

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FIG 1 Case 1 at 17½ years old.

and micrognathia. The palate was high and arched and had a submucous cleft. The neck was short with an obvious pterygium and limited extension and lateral movements. There was dorsolumbar scoliosis with left convexity of moderate degree. The shoulder joints had normal mobility except for limited external rotation, with a maximum excursion of 20°. The elbows had flexion contractures with limited pronosupination and extension. There was limited extension of the wrists, wasting of the thenar, hypothenar, and interosseous muscles, hypoplastic palmar creases, camptodactyly and flexion deformity of the fingers, and proximal soft tissue syndactyly between the digits. The first interdigital webbing had been operated on and the thumbs were abducted because of the arthrodeses performed years before (fig 2).

The lower limbs had muscle wasting, more evident on the left side which was shorter. The left popliteal pterygium had been operated upon twice and a femoral osteotomy had been performed to counterbalance the flexion contracture of that knee; its extension, however, was limited to 135°. The right popliteal fossa had no flexion creases and the pterygium was softer and milder and did not impede knee extension. Both feet had prominent calcaneus bones and rocker-bottom appearance (fig 2). She had no crural pterygia or umbilical or inguinal herniae. Sexual development was normal, menarche having occurred at 13 years of age. The external genitalia showed hypoplasia of the labia majora. Mentality was normal.

Skeletal x-rays showed a normal skull, fusion of the cervical vertebrae from C4 to C6, thoracolumbar

FIG 2 Case 1. Proximal soft tissue syndactyly between fingers, flexion contractures of the digits, atrophy of thenar and hypothenar eminences. Webbing of first interdigital spaces was operated on and arthrodeses performed to abduct the thumbs. Bottom, prominent calcaneus and rocker-bottom right foot.

FIG 3 Case 1. X-rays of the spine at ages 1 month (left), 7 years (middle), and 16 years (right), showing progressive scoliosis.

scoliosis of left convexity, and narrowing of the intervertebral discs in the upper thoracic spine. The ribs were normal (fig 3). The left femur was shortened. Vertical talus was evident in both feet.

CASE 2

A boy was born on 2.6.74 after an uncomplicated term pregnancy and vaginal delivery. The mother was 19 years old and the father 20 years old at his birth. Both were healthy and denied consanguinity, although they and their parents were all born in the same small village in rural Venezuela. This was the second pregnancy of the mother, the first one having ended in a first trimester spontaneous abortion. Four subsequent pregnancies after the birth of the proband produced normal liveborn children. There was no family history of birth defects.

At birth the infant was noted to have a short neck with pterygium, multiple joint contractures, coxa vara, pes talus valgus, and pterygia of the axillae and antecubital and popliteal fossae. He has been followed up at the Children's Orthopedic Hospital of Caracas since the age of 2 years, at which time attempts were made to correct the talus deformity of the feet and the flexion contractures of the knees surgically.

Physical examination at 6 years of age showed a male child with severe joint contractures of the shoulders, elbows, wrists, fingers, and knees. He could not stand without support for long, although he managed to walk short distances with an awkward gait with flexed hips and knees. Height was 97.6 cm and head circumference 50.4 cm. He had a low posterior hairline, low set posteriorly rotated ears, antimongoloid slant of the palpebral fissures, long philtrum, downward turned mouth, and micrognathia. The palate was high and arched but there was no cleft. There was cervical pterygium and the

neck was short and stiff. The shoulder girdle muscles were wasted and axillary pterygium inhibited full abduction of the shoulder joints. He had, in addition, antecubital pterygia, flexion contracture of the elbows, soft tissue syndactyly between the fingers, and flexion deformity of the digits with flexed and apposed thumbs (figs 4, 5).

The spine had a thoracolumbar scoliosis of right convexity and exaggerated lumbar lordosis. There was wasting of the lower limb muscles and flexion contractures of the knees. The popliteal pterygia had been repaired at the age of 4 years, but they were still evident. Both feet had prominent calcaneus bones and rocker-bottom appearance (fig 5). Bilateral intercrural webs were present, the scrotum was hypoplastic, and the testes undescended. No umbilical or inguinal herniae were detected.

Skeletal x-rays showed dorsolumbar scoliosis, fusion of the thoracic vertebrae from T1 to T3, normal ribs, and bilateral vertical talus (fig 6). Banded chromosome studies showed a normal 46,XY karyotype. Mentality was normal.

FIG 4 Case 2 at 6 years of age.

FIG 5 Case 2. Top, flexion contractures of the fingers. Bottom, popliteal pterygium, muscular wasting, and rocker-bottom foot.

FIG 6 Case 2. Top, fusion of T1 to T3 and mild scoliosis. Bottom, vertical talus is evident.

Discussion

The clinical findings in these two patients are consistent with the diagnosis of multiple pterygium syndrome. The main features of this condition were initially set forth by Matolcsy⁶ and its phenotypic spectrum was described in a recent review of 20

TABLE Physical signs in 25 patients with MPS

<i>Present in all patients</i>
Neck pterygium
Growth retardation
<i>Present in 50% or more of patients</i>
Popliteal pterygium
Antecubital pterygium
Intercrural pterygium
Multiple flexion contractures
Flexion deformity of the fingers
Soft tissue syndactyly of the fingers
Talipes equinovarus
Rocker-bottom feet
Epicanthal folds
Antimongoloid slant of the palpebral fissures
Eyelid ptosis
Low set ears
Micrognathia
Long philtrum
Cryptorchidism
Hypoplastic labia majora
<i>Present in less than 50% of patients</i>
Axillary pterygium
Cleft palate
Downward slanting mouth
Umbilical hernia
Inguinal hernia
Hypoplastic nipples
Rib or vertebral anomalies
Scoliosis or lordosis
Congenital hip dislocation
Mental retardation

affected patients. The table lists the pattern of malformations present in 25 subjects with MPS from published reports, including our two patients.^{3 4 6 8 16 21} It is apparent that the range of phenotypic anomalies is wide. The present delineation of the syndrome is probably somewhat biased towards the more severely affected since these are more likely to get medical attention and to be described in published reports.

Most cases have been sporadic, although four instances of familial occurrence in sibs have been reported,^{3 6 16 21} as well as one instance of parental consanguinity.⁴ The parents of case 2 of this report denied being consanguineous. However, they and the four grandparents of the proband were born in the same small rural village, raising the possibility that there could be some unknown degree of kinship. The information on this syndrome presently available is consistent with autosomal recessive inheritance.

In one patient a 47,XXY/48,XXXY mosaicism was found.¹⁹ This finding is probably coincidental, since no chromosomal abnormalities were observed in at least six cases in which cytogenetic studies were performed (our case 2,^{8 15 16 21}).

The underlying abnormality in this disorder is unknown. In some cases biopsy showed muscle degeneration and disorganisation of myofibrils,¹¹⁻¹³ However, this was not found in a necropsied case,²⁰ and, moreover, in two additional patients no histological abnormality was detected in skin,

skeletal muscle fibres, peripheral nerves, or anterior horn cells.²¹ The proposed pathogenetic mechanism involving decreased fetal joint movement remains a possibility still to be proved.

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