

MULTIPLE PTERYGIUM SYNDROME (ESCOBAR SYNDROME) – A CASE REPORT

A Goh, K W Lim, V Rajalingam

ABSTRACT

A 4-year-old boy with Escobar Syndrome, a rare syndrome of sporadic or autosomal recessive inheritance is described. The main features include small stature, multiple pterygia of the neck, axilla, antecubital area, a typical facies and limb abnormalities with normal intelligence.

Keywords: Escobar syndrome

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INTRODUCTION

The clinical and laboratory features of the multiple pterygium syndrome (MPS) have most often been described in the literature under the diagnosis of arthrogyposis multiplex congenita, Bonnevie – Ullrich Syndrome, pterygium syndrome and, more recently, multiple pterygium syndrome⁽¹⁾. The pterygium syndromes in themselves are a heterogeneous group with sporadic⁽²⁾, autosomal recessive⁽³⁾ and autosomal dominant⁽⁴⁾ inheritance described. The Escobar syndrome or multiple pterygium syndrome is a rare syndrome with about 50 cases noted thus far⁽⁵⁾. The unusual collection of malformations was originally described by Brussiere in 1902. This disorder was fully delineated as a distinct entity by Escobar et al in 1978⁽¹⁾.

CASE REPORT

TKM is a 4-year-old Chinese boy delivered full-term by caesarean section for abnormal lie to parents of a non-consanguineous marriage. It was an uncomplicated pregnancy. Birth weight was 2950 gm. There was no family history of congenital anomalies. The child has two older siblings, brother aged 18 years, and sister, aged 16, who are both well. The developmental milestones of this child were normal as ascertained from his mother. He was able to sit by 6 months of age, walk and talk by 14 months of age.

He was first referred to the paediatric outpatient clinic for dysmorphic features and limb deformities at the age of 1½ years. However, they defaulted further follow-up. He was referred back at the age of 3 years by the orthopaedic surgeon who had been seeing him for his limb deformities.

Physical examination revealed a small pleasant 4-year-old boy. His head circumference was 48 cm (50th percentile), height 91 cm (10th percentile) and weight 11 kg (3rd percentile). He had a mild facial asymmetry, the left being more hypoplastic than the right. He had a past history of a left torticollis from a congenital sternomastoid tumour which could account for the facial

asymmetry. He had a typical facies with epicanthal folds, micrognathia, a high-arched palate and low-set ears (Fig 1). He had a low hairline. Pterygia of the neck and axilla were present (Fig 2). Camptodactyly with partial syndactyly of the fingers were seen together with wasting of the upper limb muscles (Fig 3 and 4). Fixed flexion deformity of the hips and rocker bottom feet were present as well (Fig 5). Both testes were descended. He also had streak-like pigmentation of the skin over the buttocks. He was fully ambulatory despite the limb deformities. Intelligence was normal.

X-rays of the limbs showed no bony deformities. Chromosomal culture done was normal.

Fig 1 – Typical facies of Escobar syndrome with epicanthal folds, micrognathia and low-set ears



Department of Paediatrics
Tan Tock Seng Hospital
345 Jalan Tan Tock Seng
Singapore 1130

A Goh, MBBS, M Med (Paed)
Registrar

K W Lim, MBBS, M Med (Paed), FAMS
Consultant

Department of Paediatrics
Singapore General Hospital
Outram Road
Singapore 0316

V Rajalingam, MBBS
Resident

Correspondence to: Dr A Goh

Fig 2 – Low hairline with pterygia of the neck

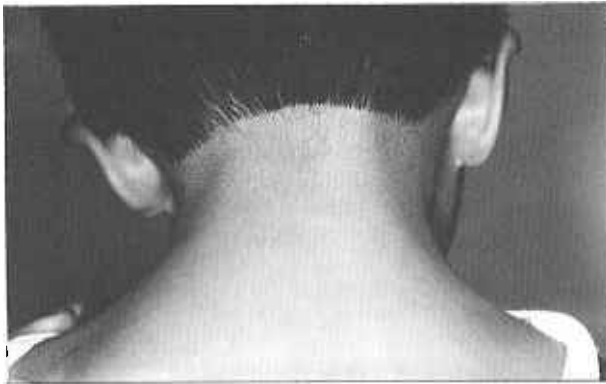


Fig 3 – Palmer view showing camptodactyly with partial syndactyly of the fingers with evidence of wasting

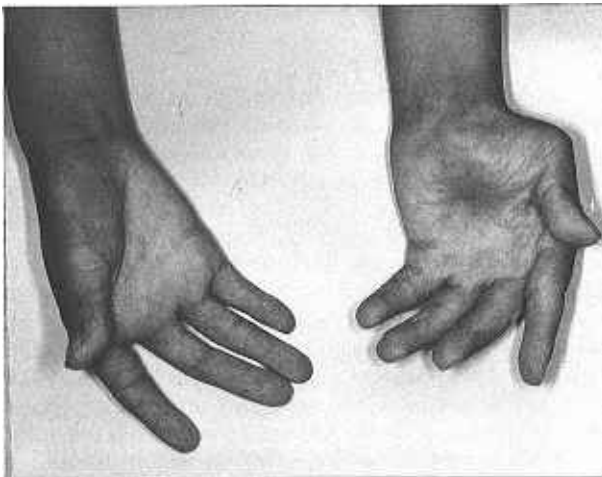
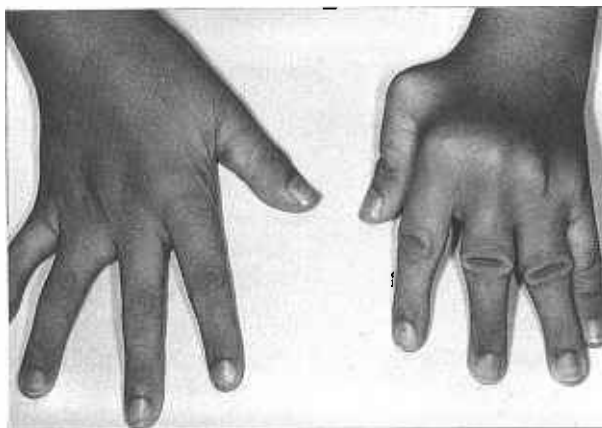


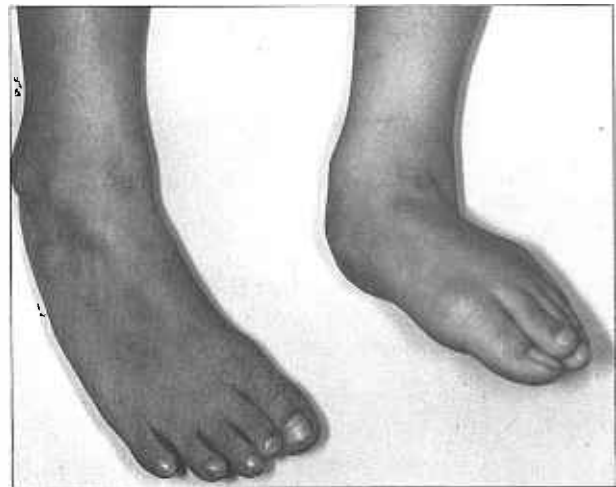
Fig 4 – Dorsal view showing similar abnormalities as in Fig 3.



DISCUSSION

The pattern of malformations seen in this patient represents the multiple pterygium syndrome. The syndrome when first described was characterised by growth retardation; pterygia of the neck, axilla, antecubital, popliteal, digital and intercrural areas; numerous joint flexion contractures; genital anomalies and cleft palate. Associated features as described by Matolcsy in 1936 in 3 siblings were ptosis of the eyelids, epicanthal folds, cleft palate, low-set ears, retrognathia, downward-turned corners of the mouth, syndactyly and camptodactyly of the fingers, talipes equinovarus and rocker-bottom feet.

Fig 5 – Rocker bottom feet



The most consistent malformations seen in MPS are⁽¹⁾
(a) pterygia of the neck (100%), antecubital (90%), popliteal areas (90%),
(b) syndactyly (74%) and camptodactyly (84%) of fingers,
(c) numerous joint flexion contractures (74%) and
(d) foot deformities (74%)

Other occasional abnormalities include umbilical hernia (26%), inguinal hernia (26%), congenital hip dislocation (21%) and hypoplastic nipples (11%)⁽¹⁾. Isolated case reports include abnormalities such as spina bifida occulta, cutis laxa, hydrocephaly, platyspondyly, clitoromegaly, ventricular septal defects and pectus excavatum⁽¹⁾. In our patient, an unusual streak-like pigmentation of the skin was seen which may be yet another feature associated with the syndrome.

The aetiology of the pterygia is unknown, but biopsy shows the presence of muscle degeneration and disorganisation of the myofibrils⁽⁶⁾, a finding compatible with muscles not in use.

Most cases of MPS are sporadic. In this patient, his parents are non-consanguinous and there is no family history of congenital malformations. However, familial cases have been reported⁽⁷⁾. Intrafamilial variation in phenotypic appearance may occur but a multiple pterygia is usually present.

In these patients with MPS, chromosome abnormalities are absent⁽¹⁾ and intelligence is normal⁽⁵⁾.

The differential diagnosis of this syndrome is the popliteal pterygium syndrome and the antecubital pterygium syndrome reported by Wallis et al⁽¹⁴⁾. In the popliteal pterygium syndrome, pterygia of the neck, antecubital area and axilla are not present but include features such as cleft lip, lip pits, syngnathia and ankyloblepharon⁽¹⁾. The features of the antecubital pterygium syndrome include bilateral antecubital webbing, absent skin creases over dorsal surface of the distal interphalangeal joints of the fingers and absent long head of triceps. Pterygia of the neck and axillary areas are absent⁽⁴⁾. Both these syndromes are inherited in an autosomal dominant manner.

The prominence of the limb deformities in multiple pterygium syndrome makes it imperative that the management should be multidisciplinary in nature, which includes the services of a physician, orthopaedic surgeon, physiotherapist and plastic surgeon. The long term complications of patients with multiple pterygium syndrome include hearing loss and subfertility in males⁽⁷⁾. However additional longitudinal studies are required to determine life expectancy, further medical problems and outcome of surgical and therapeutic intervention.

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For further information, please contact:

Dr James Goh
Department of Orthopaedic Surgery
National University Hospital
Lower Kent Ridge Road
Singapore 0511
Tel: 7724424
Fax: 7780720