Case Reports

Multiple pterygium syndrome (Escobar syndrome)

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Multiple Pterygium Syndrome (MPS) is a rare condition with less than 20 cases reported in the world literature up to 1980[1]. Characteristic features include pterygia of the neck, axilla, antecubital and popliteal areas, numerous flexion contractures of the joints and genital abnormalities. Bussiere has first described these unusual malformations in 1902. To our knowledge MPS has not been documented in Sri Lanka.

Case history

An 8-month-old boy was admitted to our unit with a history of multiple skeletal deformities present since birth. He is the second child born to non-consanguineous parents. The elder sib is a 5-year-old healthy girl. The mother is a 29-year-old housewife and the father is a 35-year-old clerk. Except for mild hyperemesis during the first trimester for which no specific treatment had been taken, the antenatal period was uneventful. The baby was delivered vaginally at term and the birth weight was 2.4 kg. The baby hasn't had any medical problems up to the time of admission.

Examination revealed a pleasant well nourished baby with slight asymmetry of the head (Figure 1).

Other abnormalities were low set ears, epicanthic folds, ptosis of the left eye, prominent nasal bridge and long philtrum. He also had pterygium of the neck. Pectus excavatum and marked scoliosis and lordosis were also noted (Figure 2).

Figure 1

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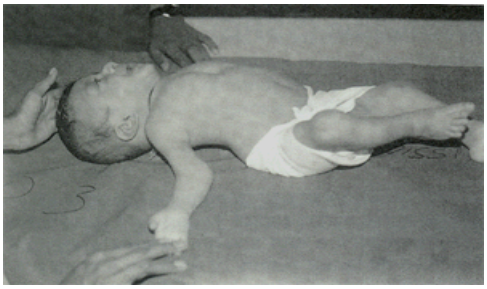

Figure 2

Nipples were hypoplastic and there were flexure contractures of both upper and lower limbs, with pterygia in the antecubital and popliteal regions (Figure 3)

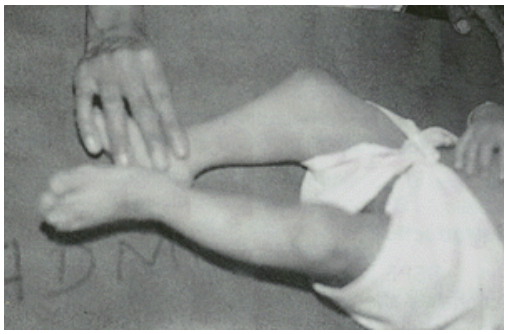

Figure 3

There were flexion deformities of the fingers and rocker bottom feet. The baby also had cryptorchidism. Cardiovascular and respiratory systems were normal.

The pattern of malformations seen in this patient was compatible with Multiple Pterygium Syndrome.

Table 1 lists the clinical findings of those patients from the literature.

Discussion

These data suggest that some patients with MPS may have few features which may go unnoticed. The most consistent malformations present in this syndrome have been pterygia of the neck (100%), antecubital and popliteal areas (90%), joint flexion deformities and foot deformities (74%) and these were found in our patient. Although most cases have been sporadic, occurrences of

affected sibs have been reported[2], and the disease is apparently transmitted as an autosomal recessive trait. The differential diagnosis of this syndrome should always include arthrogryposis multiplex congenita and popliteal pterygium syndrome. Patients with popliteal pterygium syndrome have additional features like cleft lip and lip pits, rib or vertebral anomalies and mental retardation, and the condition is transmitted as an autosomal dominant trait. Patients with MPS have normal intelligence and treatment includes physiotherapy and corrective surgery.

Table 1 - Clinical findings in this patient and similar syndromes

Clinical findings	*Present case*	*Multiple pterygium syndrome*	*Popliteal pterygium syndrome*
Orofacial			
Epicanthic folds or hyperteleroism	+	13/19	
Long philtrum	+	13/19	
Antimongoloid palpebral fissures	+	12/19	
Low set ears	+	12/19	
Micrognathia		11/19	+
Eyelid ptosis	+	10/19	
Cleft palate		7/17	+
Down-turned corners of the mouth	+	7/19	
Ankyloblephron filiforme			+
Cleft lip or lip pits			+
Syngnathia			+
Musculoskeletal			
Neck pterygium	+	19/19	
Multiple flexion contractures	+	15/19	
Axillary pterygium		10/19	
Rib or vertebral anomalies		8/19	+
Scoliosis or lordosis	+	6/19	
Umbilical hernias		5/19	
Congenital hip dislocation	+	4/19	
Hypoplastic nipples	+	2/19	+
Limbs			
Antecubital pterygium	+	17/19	
Popliteal pterygium	+	17/19	+
Flexion deformities of fingers	+	16/19	
Soft tissue syndactyly of fingers		14/19	+
Talipes equinovarus		14/19	+
Rocker-bottom feet	+	10/19	+
Genital			
Cryptorchidism	+	6/10	+
Intercrural ptrygium		12/19	+
Hypoplastic or absent labia majora		5/9	+
Inguinal hemia		5/19	+
Others			
Growth retardation	+	19/19	+
Mental retardation		2/19	+

References

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2. Srivastava RN: Arthrogryposis multiplex congenita: case reports of two siblings. *Clin Pediatr* 1968; **7**: 691-4.