Multiple Pterygium Syndrome (Escobar Syndrome) with Atrial Septal Defect

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Abstract: The authors present the first case of multiple pterygium syndrome (OMIM # 265000) from Gujarat, a rare syndrome characterized by multiple pterygia, facial and skeletal anomalies. A 9-year-old female child born of consanguineous marriage, with features of arthrogryposis multiplex, multiple pterygia, hypoplastic genitalia and skeletal anomalies presented with pneumonia. A previously unreported association of Atrial Septal defect was discovered on routine 2D echocardiography, which is important for prognostication and follow-up. [Gosai M et al NJIRM 2013; 4(1) : 162-165]

Key Words: Multiple Pterygium Syndrome, Escobar Syndrome, Arthrogryposis multiplex, Atrial Septal Defect, hypoplastic genitalia

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Introduction: Multiple pterygium syndrome is a rare syndrome originally described by Bussiere in 1902 and fully delineated as a distinct entity by Escobar and colleagues in 1978 [1].It has also been described as Escobar Syndrome, Nonlethal Type Pterygium Syndrome, Pterygium Colli Syndrome, Bonnevie - Ullrich syndrome and Pterygium Universale [2,3].

Case Report: A 9 -year-old girl, born to consanguineous parents, presented with fever, cough and headache. She had dysmorphic features and multiple joint contractures. She was diagnosed to have pneumonia with chest x-ray showing right hilar shadow and was started on antibiotics. After her condition was stabilized, she was investigated for her congenital anomalies. The girl was born after an uneventful antenatal period by normal vaginal delivery. There was no history of any medication administered during pregnancy. There was no family history of any similar congenital anomaly. One male sibling had expired due to congenital heart disease at 20 days of life. The family belonged to Muslim community. At birth, the child was noticed to have multiple joint contractures .They had showed a local doctor, who advised physiotherapy.

On examination, the child was conscious and cooperative. She had hypertelorism, antimongoloid slant, ptosis, epicanthal fold, low set ears, low hair line, furrowing of tongue, webbing (pterygium) of neck, micrognathia, high arched palate and short stature. There were multiple pterygia of the neck, axilla, cubital fossae and the popliteal region and fat pads on proximal interphalangeal joints. Bilateral knee flexion contractures and vertical talus deformities were seen.

Figue 1: Dysmorphic facies

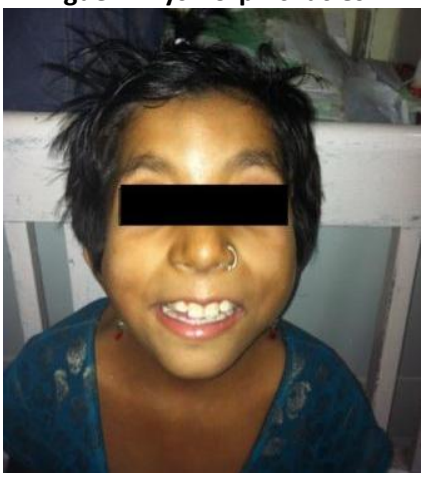
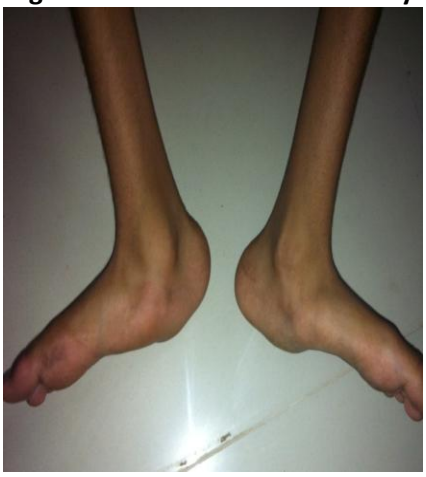


Figure 2 : Vertical talus deformity



There also were elbow flexion contractures with limited hip abduction, which were all suggestive of arthrogryposis multiplex congenita. Genitalia were hypoplastic (absent labia majora). The nipples were widely spaced.

There was no mental retardation and the child was attending school. On ophthalmological examination, there was refractive error (myopia). Fundus examination and hearing were normal. USG abdomen and chest were normal. 2 D Echo showed *ostium secundum atrial septal defect (ASD)* with left to right shunt. X-ray skull showed widened sella turcica and X-ray chest abnormal (paddle shaped) ribs. Barr body testing was negative. Karyotyping was normal (44XX). The child underwent surgery for release of hip and knee contractures and orthopedic rehabilitation.

Figure 3 : Hypoplastic genitalia

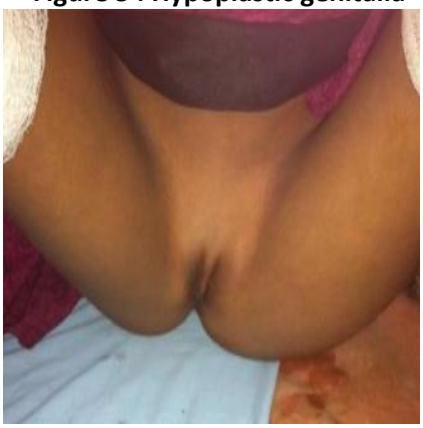
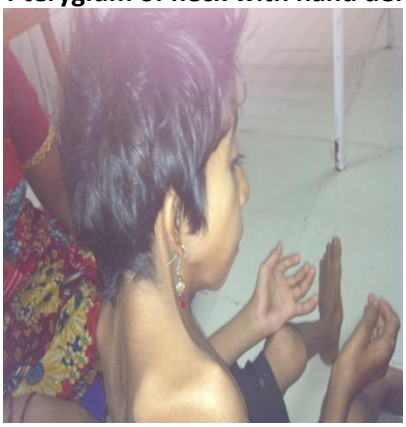


Figure 4: Pterygium of neck with hand deformities



Discussion: The pattern of malformations seen in this patient represents the multiple pterygium syndrome. The syndrome when first described was characterized 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 described are 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.

The most consistent malformations seen in Multiple Pterygium Syndrome are [3].
(a) pterygia of the neck (100%), antecubital (90%) and popliteal areas (90%)
(b) syndactyly (74%) and camptodactyly (84%) of fingers,
(c) numerous joint flexion contractures (74%) and
(d) foot deformities (74%)

Other occasional malabnormalities include umbilical hernia (26%), inguinal hernia (26%), congenital hip dislocation (21%) and hypoplastic nipples (11%). Isolated case reports include abnormalities such as spina bifida oculta, cutis laxa, hydrocephaly, platyspondyly, clitoromegaly, ventricular septal defects and pectus excavatum [3]. In our patient an unusual atrial septal defect and overriding of second toes were seen which may be yet other features associated with the syndrome. Most cases of Multiple Pterygium Syndrome are sporadic. However, familial cases have been reported. In these patients with Multiple Pterygium Syndrome, intelligence is normal.

Till date, in indexed Indian studies, there have been 4 cases from South India [4, 5]. This patient is the first report from North India. To conclude, 2D Echocardiography and genetic counselling should be done in all cases of Multiple Pterygium Syndrome to rule out cardiac defects. Also early vigorous physiotherapy and surgery is indicated to retain greatest joint mobility and ambulation.

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