Escobar Syndrome with Monodactyly: A Rare Case Report

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INTRODUCTION

Multiple pterygium syndrome (MPS) is a very rare genetic disorder. There have been very few cases of this disorder reported in the medical literature. In India, till now, four case reports are published and none is from north India.

Mutations in the CHRNG gene which are transmitted predominantly in an autosomal recessive pattern are considered as the cause for this condition, though other modes of inheritance have also been suggested. Depending on the severity of the disease, two forms of MPS can be differentiated as nonlethal and lethal. Nonlethal form (Escobar syndrome) does not worsen after birth while lethal MPS is fatal before birth or very soon after birth.

The central manifestation of this disorder is the presence of multiple pterygia, fixed joint contractures, characteristic facies in case of nonlethal form while fetuses with lethal form may develop hydrops fetalis, cystic hygroma, arthrogryposis, intestinal malrotation, microcephaly, and congenital diaphragmatic hernia in addition to features of nonlethal form.

We want to report this case because it not only has clinical manifestations of Escobar syndrome but also has few new features such as monodactyly and hypoplastic pectoralis muscle.

CASE REPORT

We present a case of a 7-year-old boy, hailing from a remote village of north India, born to nonconsanguineous parents with one normal elder sibling. The mother was a 29-year-old lady and the father was 33 years old. The kid was born after an unsupervised, uneventful antenatal period by normal vaginal delivery. There was no family history of any congenital anomaly.

At birth, the child was noticed to have the anomalies like left-hand monodactyly and elbow contracture with which he presented to us. As the age progressed, his inability to completely extend his elbow and lack of left hand grip made them visit a local hospital from where he was referred to tertiary care center.

On examination, the child had characteristic facies with a sad mask-like face, left hemifacial hypoplasia, bilateral ptosis, downward slanting eyes, low set ears, low posterior and posterio-lateral hairline, a high arched palate, furrowed tongue, and a narrow chin. He was having normal speech and hearing. He was having normal intelligence and his milestones were appropriate as per age.

The pectoralis muscles of the right side were absent on clinical examination. On examination of the left arm, there was pterygium of cubital fossa with absent carpal bones and monodactyly of the left hand.

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DISCUSSION

MPS was first described by J. A. Bussiere in 1902 in Annales d’hygiene et de medicine from Pondicherry whereas Escobar et al. compiled the various clinical manifestations of the nonlethal form and the disease was termed as Escobar syndrome in 1982.[5]

In people with Escobar Syndrome, most consistent clinical features are pterygium of neck (100%), antecubital fossa (90%) and popliteal fossa (90%), camptodactyly (84%) and syndactyly (74%) of fingers, numerous flexion contractures of joints (74%), and foot deformities (74%). Other features included are umbilical hernia, lordosis, scoliosis, lung hypoplasia, cleft lip, cleft palate, furrowed tongue, peculiar spoon shape tongue, ptosis, cryptorchism, small clitoris, and hypoplastic labia majora, distinctive facial features including ptosis, down slanting palpebral fissures, epicanthal folds, a small jaw, and low-set ears.

Along with the above symptoms, fetuses affected with lethal MPS may develop hydrops fetalis, cystic hygroma, severe arthrogryposis, hypoplasia of vital organs such as heart, lung, or brain, intestinal malrotation, kidney abnormalities, a cleft palate, microcephaly, and congenital diaphragmatic hernia.

It is important to differentiate this condition clinically from seemingly similar syndromes such as arthrogryposis multiplex congenita and popliteal pterygium syndrome [Table 1].

The mode of inheritance is usually autosomal recessive and rarely autosomal dominant. The pattern of inheritance in our report appears to be autosomal recessive, as none of his parents or his sibling is affected. The gene responsible has been identified as the CHRNG gene which codes for the gamma subunit of the acetylcholine receptor required for the communication between nerve cells and muscle cells in the developing fetus.

Spranger et al. reported that two cases with autosomal recessive MPS, type Escobar, who described multiple pterygia, severe contractures, short stature, and external genital anomalies are prominent features. A striking feature was severe muscular atrophy.

Shalev et al. reported a large consanguineous Arab family in which 9 members had some of the typical features of Escobar syndrome.

As quoted by Rajput et al., In India, other than their study, to date, only three case reports of Escobar syndrome are published, two of them are from south India and one is from central part of India.

![Figure 1: Characteristic sad face with bilateral ptosis and right side pectoral muscle atrophy](image1)

![Figure 2: X-ray of the left upper limb showing elbow joint deformity and hand anomaly](image2)

| Table 1: Differentiation table of seemingly similar conditions |
|---------------------------------------------------------------|
| Escobar syndrome (MPS)                                      | Arthrogryposis multiplex congenital | Popliteal pterygium syndrome |
| Camptodactyly syndactyly, short stature, rock-bottom or club feet, joint contractures, webbing of the neck, micrognathia, cleft palate down-slanting eyes, drooping eyelids, low-set ears, kyphoscoliosis Spinal fusion abnormalities Undescended testes and an abnormally small penis Underdeveloped or missing labia majora MPS: Multiple pterygium syndrome | Reduced mobility of multiple joints, the shoulders are bent inward and internally rotated The elbows are extended and the wrists and fingers are bent, the hips may be dislocated and are usually slightly bent, clubfoot | Cleft lip and palate, lower lip pits, popliteal and intercral pterygium, underdevelopment of the genitals, syndactyly, adhesion of upper and lower jaw and adhesion of upper and lower eyelids A cone-shaped fold of skin on the nail of the big toe |
Ours is the first case report of an isolated case, showing clinical manifestations of Escobar syndrome, from a north Indian family. This case shows the clinical manifestation of Escobar syndrome such as pterygium, characteristic facial features, ophthalmological findings, and muscle atrophy and it additionally shows hand anomaly, i.e., monodactyly as a newer association.

A multidisciplinary approach involving a physician, ophthalmologist, orthopedic surgeon, physiotherapist and plastic surgeon is the ideal way to manage this condition.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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