Case Report

# Escobar syndrome in three male patients of same family

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We describe three male individuals from a consanguineous south Indian family affected with the multiple pterygium syndrome (Escobar syndrome). Common clinical features included short stature, multiple pterygium, skeletal anomalies, and normal intelligence. The first report of this condition was made in 1902 from this same place (Pondicherry) and the disease received its present popular name Escobar syndrome in 1982. The genetic defect for this condition was identified in 2006 as mutation in the fetal acetylcholine receptor.

**Key words:** Escobar syndrome, familial, multiple pterygium syndrome.

## Introduction

Escobar syndrome or the multiple pterygium syndrome is an autosomal recessive disorder, though other modes of inheritance have also been suggested. The central manifestation of this disorder is the presence of multiple pterygia or cutaneous contractures with normal intelligence. It was first described by Bussiere [1] in 1902 from Pondicherry. It however got its name Escobar syndrome in 1982 after Escobar[2] who along with his associates prepared an extensive report on this disease in 1978. The genetic defect of this condition has been identified as the gamma subunit of the nicotinic acetylcholinergic receptor.

Till date, there is only one more single case report from India. This is the first report of three such cases in a south Indian family from Pondicherry, two of whom we could document clinically, while the third is historical.

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## Case Report

The proband, a 21-year-old young adult, was admitted with the complaints of fever, cough with mucopurulent expectoration, and shortness of breath. He was diagnosed as a case of severe pneumonia, and hospitalized.

On examination, he was also found to have multiple pterygium involving the neck, elbow [Figure 1], fingers [Figure 2] and popliteal area, receding chin, and severe scoliosis [Figure 3]. Radiological survey showed fusion of C3, C4, and C5 vertebrae [Figure 4]. His height was 125 cm (<3rd percentile). His intelligence on formal bedside testing was normal.

The above patient was diagnosed as severe pneumonia with multiple pterygium syndrome. The pneumonia was treated with intravenous antibiotics. He recovered fully from the pneumonia. Before discharge from hospital, spirometry was done to assess the lung function in view of severe scoliosis. It showed severe restrictive pattern of lung disease. Karyotyping of the proband was normal.

After the diagnosis of multiple pterygium syndrome, the family history was analyzed retrospectively. The family consisted of consanguineous parents with five children [Figure 5]. The above described proband is the third child. The second male child also had contractures involving finger joints and died at two years of age due to unknown reasons. Further details of second child were

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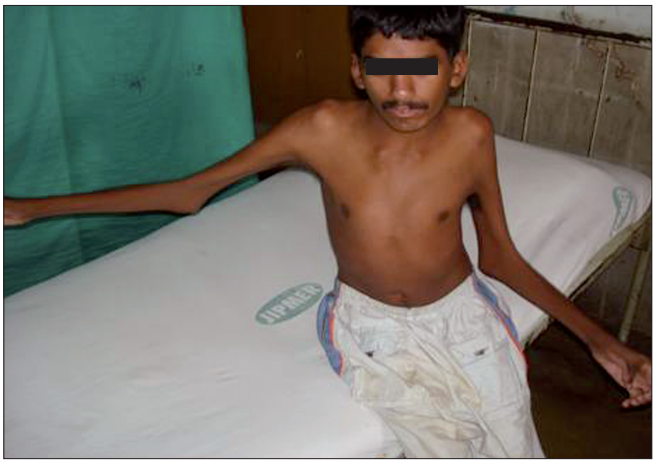

Figure 1: Pterygium of elbows.

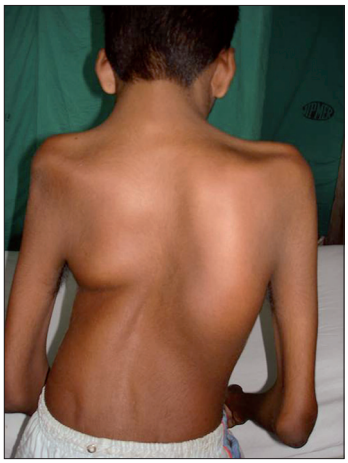

Figure 2: Pterygium of the neck and scoliosis.

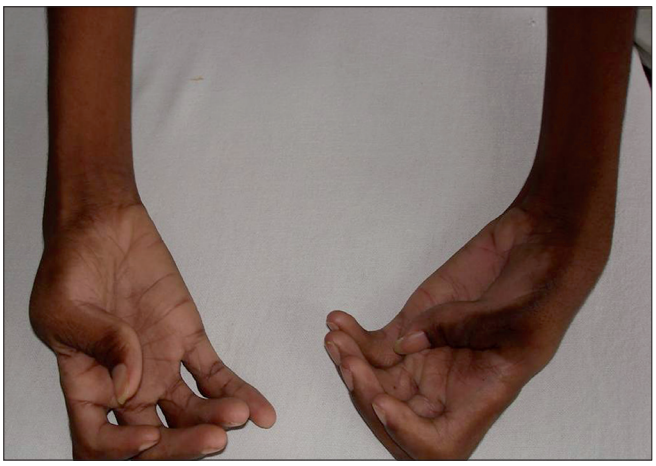

Figure 3: Pterygium of the fingers.

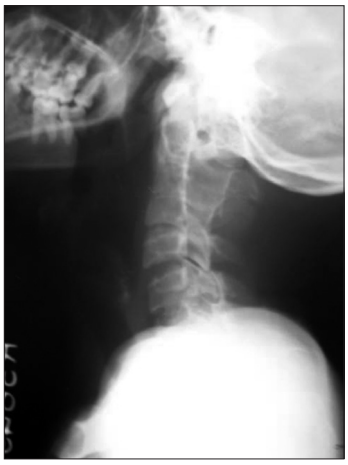

Figure 4: Fusion of C3, C4, and C5 vertebrae.

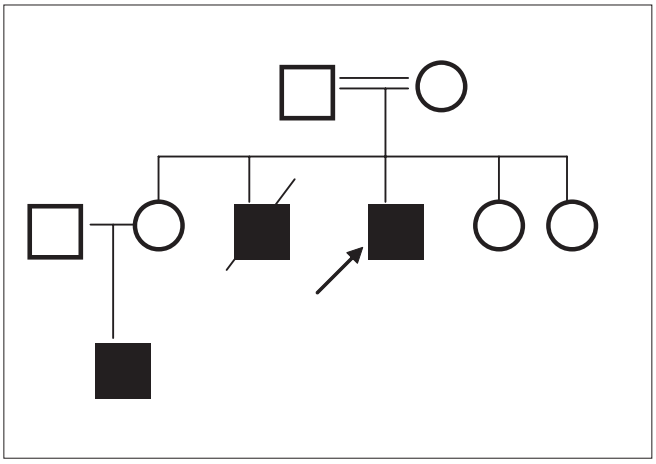

Figure 5: Pedigree chart.

not available.

From the family history of the proband, we found that his nephew (the second son of the first female sibling of above family) also was having similar features. This five-year-old male child [Figures 6-8] was found to have all the above mentioned features [Table 1] and also ptosis, epicanthic fold, long philtrum, low set ears, antimongoloid slant, and rocker bottom feet.

Plastic surgery opinion was sought for both patients. The expert opinion was that in the proband, correction is difficult, whereas in the child, correction of the defects is planned.

## Discussion

The clinical manifestations of the two patients we described above were consistent with Escobar syndrome. Escobar syndrome is a subtype of arthrogryposis multiplex congenita.

This condition was first described by J.A. Bussiere in 1902 in Annales d`hygiene et de medecine from

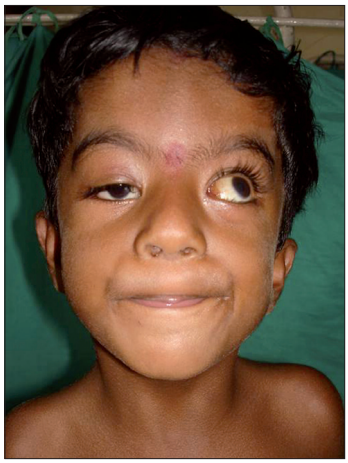

Figure 6: Right ptosis, epicanthic fold, anti mongoloid slant, and receding chin.

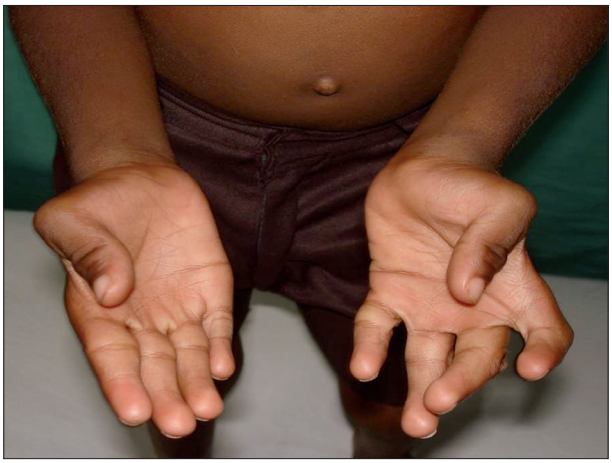

Figure 7: Pterygium of the fingers.

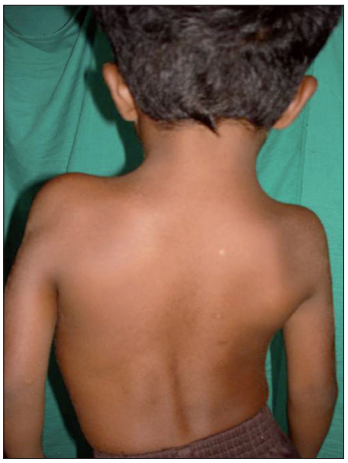

Figure 8: Scoliosis.

**Table 1: Clinical features of both patients**

| | Patient No. 1 (proband) | Patient No. 2 |
|---|---|---|
| Pterygium of neck, axilla, fingers, cubital and popliteal fossae | + | + |
| Receding chin | + | + |
| Ptosis, low set ears | - | + |
| Short stature | + | + |
| Scoliosis | + | + |
| Fusion of cervical vertebrae | + | + |
| Rocker bottom feet | + | + |

Pondicherry. He described a patient with multiple pterygia with resemblance to a cobra (cobra man/l'homme cobra), possibly because of the webbing of neck and microcephaly. In 1978, Victor Escobar and his associates compiled the various clinical manifestations and the disease was termed as Escobar syndrome in 1982.

It is a very rare genetic disorder with multiple congenital anomalies as described; however, noteworthy is the preserved intelligence. Other features described in literature and not present in our patients include genital anomalies, lordosis, vertical talus, cleft lip, cleft palate, down turned angles of mouth, furrowed tongue, peculiar spoon shape tongue (lingua cochlearis), hemangiomas of forehead, cutaneous dents, pulmonary hypoplasia, cryptorchism, small penis, small clitoris, and hypoplastic/absent labia majora.

The mode of inheritance is usually autosomal recessive and rarely autosomal dominant. The pattern of inheritance in our report appears to be autosomal recessive, though X-linked cannot be ruled out since all affected were males.

The gene responsible has been localized to 2q36-q37 region according to Hoffman *et al.*,[3] and Morgan *et al.*[4] in 2006. This gene codes for the gamma subunit of the acetylcholine receptor. The acetylcholine receptor has five subunits (2 alpha, 1 beta, 1 delta, and 1 gamma/epsilon). The gamma subunit is replaced by the epsilon in later fetal or perinatal life. Loss of gamma subunit in fetal life causes reduced fetal movement which is responsible for the contractures. The formation of normal epsilon unit later prevents the development of myasthenia in adults. Thus, Escobar syndrome is an example of a devastating dysmorphology due to a transient neuromuscular end-plate disturbance, but whether all the features are due to this defect alone is debatable.

Till date, there is only one more single case report from Vellore, South India by Madhuri *et al*,[5] where they described a 5-year-old girl with multiple pterygia, genital anomalies, facial anomalies, but no spine deformities

and translocation involving chromosomes 6 and 7, which was not seen in our patient. This is the first report of a family with three affected individuals and all being male members.

## Conclusion

We describe three male patients with Escobar syndrome from the same family.

There is no specific treatment for Escobar syndrome. At present, genetic counselling and *in utero* detection, wherever possible, remains the mainstay of treatment.

We could not confirm whether this family were in any way descendents of the first patient of Escobar syndrome described by J.A. Bussiere in 1902.

## Acknowledgements

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