JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH

How to cite this article:
VISHWANATH B, SHYAM SUDHIR MK.ESCOBAR SYNDROME.Journal of Clinical and Diagnostic Research [serial online] 2010 February [cited: 2010 February 1]; 3:2147-2148.

Available from
http://www.jcdr.net/back_issues.asp?issn=0973-709x&year=2010 &month=February &volume=4&issue=1&page=2147-2148 &id=547

IMAGES IN MEDICINE

Escobar Syndrome

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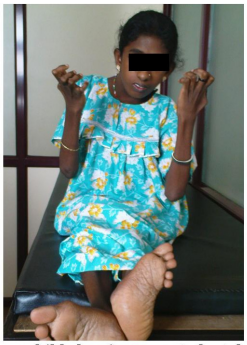
A 14 year old girl was brought with the complaint of inability to extend the limbs at the elbow and knee joints. She was born to second degree consanguineous parents and had a history of delayed gross motor milestones. She weighed 22kgs and her height was 132 cm. (Both <3rd centile). She had an emotionless face with ptosis of eyes, micrognathia,difficulty in opening the mouth widely, down turning of the angles of the mouth, low set ears and pterigia of the neck, axilla and the antecubital areas [Table/Fig 1]. Pterigium of the popliteal area was also seen. Her hands revealed soft tissue syndactyly and camptodactyly and her foot examination showed Rocker bottom feet with valgus deformity [Table/Fig 2]. She also had kyphoscoliosis and the absence of the labia majora on the right side. Her systemic examination was normal. Considering these features, a diagnosis of Escobar syndrome was made.

(Table/Fig 1) Escobar syndrome child showing emotionless face, micrognathia, down turned angles of mouth and pterigia of neck & ante cubital areas



(Table/Fig 2) Same child showing camptodactyly of fingers and rocker bottom feet with valgus deformity

Escobar syndrome was described originally by Bussiere in 1902, but Escobar, in1978, fully delineated it as a distinct entity [1]. It is a very rare disorder and only 50 cases have been reported so far [2]. Other synonyms are Multiple Pterigium Syndrome and Pterigium Universale. It is a genetic disorder and has an autosomal recessive mode of inheritance [3]. It is characterized by multiple pterigia, camptodactyly, syndactyly, short stature and kyphoscoliosis. Affected individuals may also have fusion of vertebrae, dislocation of hip, conductive hearing loss and cardiac defects. Respiratory problems are common due to kyphoscoliosis and are the common causes of death during infancy [3]. Some patients have hypoplasia of the periumbilical skin, leading to umbilical hernia [4]. In the neonatal period, it has to be differentiated from Lethal Multiple Pterigium Syndrome as the latter has 100% mortality.

A majority of affected children have normal intelligence and become ambulatory. Pterigia becomes obvious with time, leading to fixed contractures. Early vigorous physical therapy is suggested to retain joint mobility.

Hearing evaluation is indicated in all individuals.

Recently, a mutation in the foetal or the gamma subunit of the nicotinergic acetylcholine receptor (CHRNG) was identified in most of the individuals who were affected by the Escobar syndrome [5].

References

[1] Escobar V. Bixler D, Gleiser S,Weaver Dd,Gibbs T.Multiple Pterigium Syndrome. Am.J.Dis.Child. 1978; 132:609-11.
[2] Jones KL.Smith's Recognizable patterns of Human Malformations.Sixth Ed. Philadelphia: Elsevier-Saunders; 2006.346-47.
[3] Thompson EM, Donnai D, Baraitser M, Hall CM, Pembrey ME, Fixsen J. Multiple Pterigium Syndrome: Evolution of Phenotype. J.Med.Genet.1987; 24:733-49.
[4] Shalev SA, Spiegel R, Hall JG. A Syndrome Characterised by Contractures and Pterigia of Upper Body Associated with Umbilical Hernia, Short Stature and Distinctive Face in Arabic Family. Am.J.Med.Genet.2005; 138A:236-40.
[5] Hoffmann K, Muller JS, Stricker S, Megarbane A, Rajab A, Lindner JH.et al. Escobar Syndrome is a Prenatal Myasthenia Caused by Disruption of Acetylcholine Receptor Fetal Gamma Subunit. Am.J.Hum.Genet. 2006; 79:303-12.