Escobar Syndrome

Pages with reference to book, From 140 To 141

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Originally described by Bussiere in 1902, this disorder was fully delineated by Escobar et alas a distinct entity in 1978[1,2]. About 50 cases have been reported[2]. The disorder mainly appears to affect muscles, with muscle degeneration and disorganization of myofibrils having been noted in biopsies. The inheritarice is autosomal recessive. The classical syndrome also known as multiple pterygium syndrome has been found to have the following features[1,2]: small stature, ptosis of eyelids, inner canthal folds, micrognathiawith down- turning of mouth, pterygia of neck, axillae, elbows and knees (pterygium means shaped like a wing), pterygia plus camptodactyly (permanent and irreducible flexion of one or more fingers), syndactyly, equinovarus and/or rocker-bottom feet, numerous flexion contractures of joints, cryptorchidism, absence of labia majora and vertebral and/or rib anomalies. Occasional abnormalities include scoliosis, dislocation of the hip, hypoplastic nipples and cleft palate.

Case Report

A four years old girl from Dera Ismail than was admitted with complaints of "multiple limb deformities". She was the product of a full term, uncomplicated pregnancy and vaginal delivery. Her parents were first cousins, with three other normal children. Father was 24 and mother was 19 years old at the time of birth. The family history was negative for congenital abnormalities. At birth she had flexion deformity of both the elbow and ankle joints. Birth weight was not done. Motor development was delayed, she held neck at 10 months, sat at 1 year and started walking at three and a half years of age. Intellectually she developed normally. On examination she had small stature, her weight and height were both below 2 standard deviations for her age. She had a flat, emotionless face with downward turning of the mouth and low set ears. She did not have micrognathia. She had pterygia of neck, axillae, antecubital and popliteal areas. She had scoliosis and widely spaced hypoplastic nipples. Camptodactyly with partial syndactyly of the fingers was present. Both feet had a rocker bottom appearance. All her nails were normal. On genital examination her labia majora were absent, normal labia minora and introitus were present. Pelvic ultrasound revealed a hypoplastic uterus, but ovaries could not be clearly identified. No abnormality could be detected in cardiovascular, respiratory, gastrointestinal or central nervous system examination. She had difficulty in standing straight and walking, probably due to limb deformities and spinal defect. Upper limb and hand functions were quite reasonable. Chromosomal analysis showed normal karyotyping with 46XX chromosomes. Echocardiography did not reveal any congenital defect. Muscle biopsy could not be done.

Discussion

The most common recessively inherited disorder with limb pterygia is multiple pterygium syndrome, which must be differentiated from popliteal pterygium syndrome which has dominant inheritance[3]. The most distinctive features in the popliteal pterygium syndrome include: pterygia of popliteal areas, lip pits, ankyloblepharon fihiforme adnatum (congenital adhesion of the upper and lower eyelids by filamentous bands), oral frenula (mucosal folds), reduction defects of the digits and the characteristic nail anomalies (a pyramidal skin fold extending into the base of the nail and dermal skin ridges over toes, particularly the hallux) [3]. Some of these cases are confounded with other entities primarily

because of incompleteness and inconsistency in reporting of clinical findings. It might be mistaken for the 'Turner' phenotype with arthro-gryposis[4], Marfan syndrome[5], Pterygoarthromyo-dysplasia Congenita and Bonnevie-Ulrich syndrome[3]. Escobar[1] and Hall[3] in their excellent reviews of 20 and 48 cases respectively differ in the frequency of different findings present in the reported cases of multiple pterygium syndrome (Table).

Table. Comparison of frequency of most consistent findings.

Clinical findings	Escobar et al[1]	Hall et al[3]
Pterygia of neck	100%	85%
Antecubital pterygia	90%	54%
Popliteal pterygia	90%	60%
Syndactyly	74%	48%
Camptodactyly	84%	71%
Foot deformities	74%	63%
Scoliosis	35%	60%

The pattern of malformations seen in this case clearly represents the multiple pterygium syndrome. Since these patients have normal I.Q. the long term consequences depend upon the extent of management in early life. If adequately managed by appropriate physiotherapy and surgical correction in time, they can lead an almost normal life. Sexual development though late, is usually complete. A case has been reported where a female patient has given birth to a completely normal child[6].

References

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