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Popliteal Pterygium Syndrome with Scrotal Agenesis

[1]Sandip Kumar Rahul, [1]Susama Behera, [2]Satyabir Mohapatra, [3]Pallavi Suman
Departments of [1]Surgery and [2]Pediatrics, Nehru Shatabdi Central Hospital, Talcher, Angul, Odisha; [3]Department of Pediatrics, Central Hospital, Barajamda, West Singhbum, Jharkhand, India.

Abstract:

Popliteal pterygium syndrome and agenesis of scrotum are rare. We present a case in which both were present and emphasize on the role of staged surgery and multidisciplinary approach in the management of such cases.

Key words: Cleft Lip, Cleft Palate, Scrotum, Syndactyly, Urogenital Abnormalities.

Introduction

With an incidence of 1:300000, variable expressivity and incomplete penetrance, popliteal pterygium syndrome manifests itself with different combinations of orofacial, skin and genital abnormalities [1,2]. We could find only a few isolated case reports of absent scrotum and none was associated with popliteal pterygium syndrome [3]. Coexistence of both these conditions in the same patient is being described.

Case Report

A 3-week-old male child presented with multiple deformities of face, external genitalia and lower limbs since birth. He had bilateral cleft lip, cleft palate and restricted mouth opening of around 25 mm due to membranous oral synechiae from the margins of the palatal cleft to the floor of the mouth which led to feeding difficulties [Fig.1]. He could not straighten either of his lower limbs due to thick fibrous band over the popliteal region [Fig.2]. Scrotum was not formed and bilateral testes were impalpable [Fig.3,4]. There was bilateral syndactyly involving the third and fourth toe. A fold of skin could be seen over the nail of the right great toe. Rest of the systemic examination was normal.

Child's father had a history of cleft lip surgery in his childhood. Other family members were healthy. Child had a full term normal vaginal delivery with a weight of 2200 grams. Antenatal sonogram had detected the cleft abnormality in the third trimester. Routine hemogram, electrolytes and renal function tests were normal. Abdominal sonogram did not reveal any abnormality. Skiagrams of both lower limbs and chest were done to see for the nature and the extent of the deformities.

Corresponding Author: Dr. Sandip Kumar Rahul
Email: sandeep.rahul65@gmail.com
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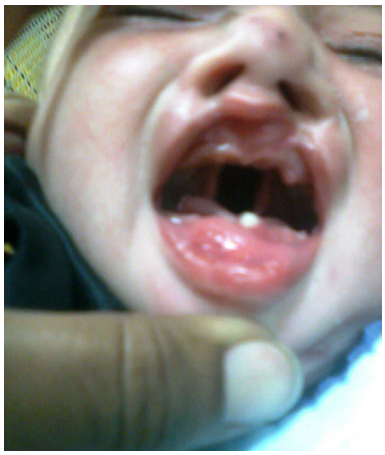

Fig.1: Cleft palate and cleft lip.

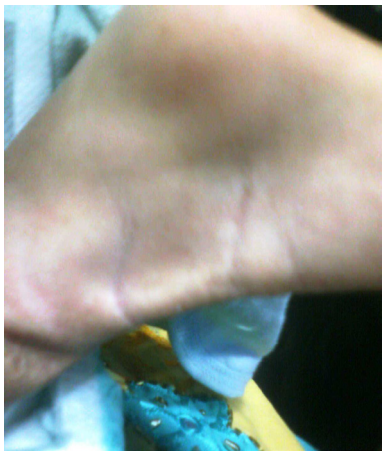

Fig.2: Popliteal webbing.

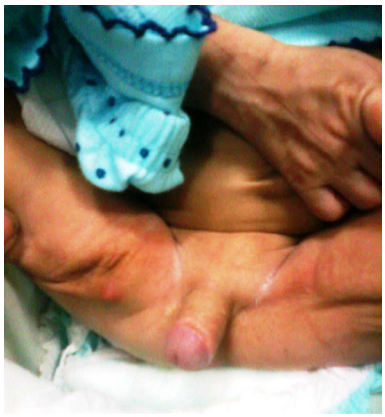

Fig.3: Scrotal agenesis with cryptorchidism.

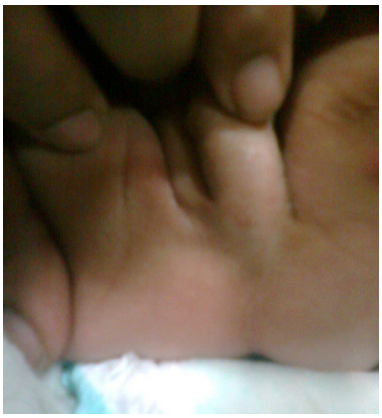

Fig.4: Scrotal agenesis with cryptorchidism (with phallus lifted up).

Parents were told about the syndrome affecting their child and the need for corrective surgeries in different stages by a team of plastic, paediatric, orthopaedic, maxillofacial, otorhinolaryngeal, dental and intensive care specialists.

Discussion

Popliteal pterygium syndrome (PPS) (OMIM #119500) has been described to be an autosomal dominant condition involving mutation in Interferon regulatory factor (IRF)-6 gene on chromosome 1q32 [1]. Abnormalities associated with it include cleft lip/palate, lower lip pits or cysts, syngnathia, congenital ankyloblepharon, scrotal and labial abnormalities, cryptorchidism, popliteal webbing, telipes equinovarus, syndactyly and nail anomalies [1,4]. Antenatal diagnosis by sequence analysis of the IRF6 gene in DNA extracted from amniocentesis or chorionic villus sampling can be done in cases of a positive family history where a disease causing mutation has been detected [2]. Antenatal sonogram can detect the cleft, limb and scrotal anomalies. Agenesis of scrotum is itself rare and we could not find any case in literature with associated PPS [3].

In our patient, the membranous synechiae were divided early at the age of 6 weeks which improved mouth opening and feeding. Following this, the child has been planned for cleft lip surgery when he attains a weight of around 10 pounds. PPS is associated with feeding difficulties, delayed language development, problems with dentition, maxillofacial development and learning difficulties. Speech therapy and appropriate audiological and dental assessment are often needed after a planned but early cleft lip/palate repair.

Scrotal construction in such cases has been variously described. This includes the use of preputial flaps, local perineal skin and tissue expansion techniques [3,5,6]. Silay *et al*. described a case of successful neoscrotal construction using the local perineal skin [3]. Sengezer *et al*. reported scrotal construction by tissue expansion of labia majora in biological female transsexuals [6]. In our patient, surgery for scrotal reconstruction has been planned at around 6 months of age. Following this, at around 1 year of age surgery for cryptorchidism would be done.

Cleft palate repair has been planned at the time of surgery for undescended testes during the same anaesthesia. Popliteal webs present as a formidable orthopaedic challenge with concerns about the sciatic nerve and popliteal vessels in the thick fibrous band with deficient skin. Syndactyly needs additional corrective surgery later by the plastic surgery and orthopaedic team. Complex deformities can present with difficulty in airway management during anaesthesia. Child's overall growth and developmental monitoring is very important throughout the management process. Van der Woude syndrome (OMIM #119300) also shows mutation in the IFR-6 gene and has similar orofacial lesions [7].

The overall prognosis of PPS is good. Growth and intelligence are usually normal but success of surgery done for popliteal web depends on the severity of the web. Cryptorchidism may lead to infertility. Multiple pterygium syndrome with its Escobar (OMIM #265000) [8] and lethal variants (OMIM #253290) [9] and Bartsocas Papas syndrome (OMIM #263650) with similar clinical features and autosomal recessive inheritance have worse prognosis compared to PPS [10].

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