

Case Reports

Bartsocas-Papas Syndrome in a Pakistani Family from Kuwait

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We report a rare case of Bartsocas Papas Syndrome, a lethal autosomal recessive type of Popliteal Pterigium syndrome, from a consanguineous Pakistani family who had typical anomalies of face, limbs and genitalia with additional peripheral pulmonary stenosis. Antenatal diagnosis and option for termination of pregnancy is advised.

Keywords: Ankyloblepharon, Facial cleft, Popliteal Pterigium

The Popliteal pterigium syndrome in its lethal autosomal recessive form, the Bartsocas Papas syndrome (BPS), was first described in 1972(1). The chief features include a popliteal pterigium with a cord containing nerves and vessels, synostosis of hand and foot bones with digital hypoplasia and syndactyly, facial clefts, ankylo-blepharon and filiform bands between the jaws(2). More than 20 cases have been reported in the literature, mostly from mediterranean ancestry(Greek, Italian, Spanish) but

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*Manuscript received: June 25, 2003;
Initial review completed: August 6, 2003;
Revision accepted: December 11, 2003.*

occasionally from Dutch, Arabs, Turkish and Iranian(2-7).

We report, for the first time, patient with BPS from a Pakistani family living in Kuwait along with review of literature.

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This boy was born to an 18-year-old Pakistani primi mother at full-term by vaginal delivery assisted by forceps. Parents were first-degree cousins and were healthy. There was no family history of any congenital anomalies. The mother had one abortion at 3 months' gestation one-year before, the cause was unknown. The antenatal period was uneventful. Birth weight of the neonate was 2.7 kg, head circumference 34 cm, length 47 cm and chest 32.5 cm. Apgar scores were 5 and 7 at 1 and 5 minutes respectively. The baby had striking anomalies of face, hands, legs and genitalia (*Fig. 1*). There were absent hair on scalp, eyebrows, and eye lashes with bilateral ectropion and corneal opacities. Hypoplastic deformed nose, bilateral cleft palate and lips and signathia (fusion of lips) were also noted (*Fig. 1*). Saliva bubbles were coming out at both corners of mouth from salivary pits. There were absent thumbs and fusion of other fingers (mitten hands) with hypoplasia of nails. Marked shortening of both lower limbs with thick pterigium extending from ischea to heels was noted in addition to syndactyly of toes (all on left, and second to fourth toes on right). The genitalia were ambiguous with small phallus and pigmented folds of skin of scrotum. Skeletal survey showed absent first phalanx and shortening of tibia and fibula. Ultrasound of head and abdomen were normal. An asymptomatic soft systolic murmur was



Fig. 1. Neonate with alopecia, facial defects, thick popliteal pterygium, mitten hands with absent thumbs, syndactyly of toes and hypoplastic genitalia.

diagnosed as peripheral pulmonary stenosis by echocardiography. Karyotype was normal. The soft tissue fusion of lips was incised by plastic surgeon, but mouth opening remained restricted and feeds were given by an orogastric tube. The eyelids were stitched to prevent exposure keratitis. The baby was discharged home at 2 months of age on gavage feeding, but suddenly died at 4 months of age at home. The exact cause of death could not be known as autopsy was refused.

Discussion

Bartsocas and Papas reported a family, in which the parents were third cousins, with 4 siblings (3 female, 1 male stillborn) severely

affected by BPS(1). Clinical features include popliteal pterygium with a cord containing nerves and vessels, low birth weight, synostosis of hand and foot bones with digital hypoplasia and syndactyly; facial clefts, microcephaly, ankyloblepharon, hypoplastic nose, filiform bands between the jaws, lower lip salivary pits and hypoplastic genitalia(2). Our case had most of the typical features of BPS along with the additional finding of peripheral pulmonary stenosis, which has not been reported in literature. Various anomalies associated with BPS include renal agenesis, esophageal atresia, hypo-plastic diaphragm, agenesis of the shaft of the penis and anal atresia(4); and super-numerary nipple(7); none was present in our case.

BPS should be differentiated from the lethal multiple pterygium syndrome having generalized pterygia and which has 3 distinct types based on bony fusion and modeling errors of bones(8); the pterygium in BPS is localized to lower limbs.

Most cases described are among the Mediterranean ancestry(2). Hennekam, *et al.* described 2 Dutch sibs(4), and Massoud, *et al.* described 4 sibs in a non-consanguineous Arab-Asian family, one was stillborn; the other 3 children lived 10 to 17 months(5). Mavilli, *et al.* described an 8-month-old Turkish girl(6), and Shafeghati, *et al.* an Irani infant(7) with BPS. Our case is from south-eastern-Asian ancestry (Pakistani) which has not been described previously to our knowledge.

The exact pathogenesis of BPS is unknown. The potential teratogenic effect of transient or persistent edema during embryonic and fetal development was thought but absence of edema in BPS points towards other etiologies(8). A unique and generalized vascular problem is suggested as the disruptive process is not located in an anatomic site(4). An

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alternative explanation proposed is a pleiotropic autosomal recessive dysplasia sequence of the “ectoderm ring” of Blechsmidt, based on the characteristic malformations, which are “atypical, bizarre, and ragged-at-the-edges”, indicating tissue necrosis(2). Despite the lack of apparent consanguinity in their report the autosomal recessive pattern can not be ruled out because of relatively small Arab community as reported by Massoud, *et al.*(5). The presence of four affected siblings and first degree consanguinity among healthy parents in our case indicates the presence of autosomal recessive inheritance.

Most cases die in early weeks of life, the oldest being 8-year-old(9). Our case died at 4 months due to sudden death. The reported causes of death are broncho-pneumonia, respiratory distress and sepsis(2,4). Antenatal diagnosis is possible as early as first trimester using transvaginal ultrasound(10). Due to its lethal nature, early antenatal diagnosis and option for termination of pregnancy should be discussed with parents, along with awareness that occasional cases may have a good prognosis.

Contributors: RMZ was involved in patient care, supervised this manuscript and is guarantor of the study. ALS collected the data, reviewed the literature and prepared this manuscript. EMA helped in reviewing the literature. RKG operated on the case and reviewed literature.

Funding: None.

Competing interests: None stated.

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