

A Family from Turkey with Bartsocas-Papas Syndrome

Türkiye'den Bartsocas Papas Sendromlu Aile

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A 34-year-old pregnant Turkish woman was admitted to the emergency service because of contractions at 33 weeks. Her vaginal examination revealed 8-9 cm cervical dilatation and 80-90% effacement. Fetal membranes were intact, and the fetus had a vertex presentation. Shortly after, she had a vaginal delivery. The newborn had mid-facial cleft, low-set ears, cleft lip, bilateral pterygia, ambiguous genitalia, syndactyly, and hypoplastic toes. There were thick skin bands between the soles of both feet and the upper part of the genitalia (Figure 1, 2). The newborn died of obstructive apnea. In the postmortem ultrasonography and X-ray image of the newborn, the visceral organs and skeleton were observed to be normal. The family did not allow genetic tests and autopsy to be performed. They were a case of cross-cousin marriage family. Her husband and she and their brothers had children with Bartsocas-Papas syndrome (BPS) who could not take their first breath. Six newborns died in the neonatal period. One child is 4 years old, and his treatment is in progress. The patient's permission was received for publishing this case.

The first case of BPS was recorded in 1600, but Bartsocas and Papas was described in 1972 in terms of heredity pattern and other anomalies. The frequency of BPS is 1 in 650000 cases. BPS is a lethal form of popliteal pterygium syndrome (PPS), but BPS is more frequently seen in the hands and feet and results in more facial abnormalities than PPS. Inheritance is autosomal recessive. Various abnormalities are seen such as atypical face,



Figure 1. The face of the newborn had mid-facial cleft, low-set ears, cleft lip, and ectropion of the eyelids.



Figure 2. The newborn had bilateral pterygia, bilateral popliteal and thick skin bands connecting the upper part of the genitalia and soles of both feet, talipes equinovarus with hypoplastic toes, and bilateral syndactyly.

hypoplastic nose, short palpebral fissures, microcephaly, syndactyly, popliteal webbing, genital abnormalities, and ectodermal anomalies. On performing ultrasound in the first trimester, limb abnormalities can be diagnosed and identified [1]. Visceral organs may be affected, but usually, they are reported to be normal and functioning. Abnormalities in the urinary system such as dysplastic kidney and absence of the urethra can be seen in BPS. BPS is fatal in the newborn period in most cases. However, cases of patients who lived until childhood have been reported [2]. BPS is seen more often in Mediterranean countries, and it is seen that this form, which is deadly and hereditary transitive, is repeated for generations [1, 2]. As in our case, due to reasons such as the continuation of lineage and ostra-

cisation, deadly incidents and sick children are not revealed and marriages among relatives are realized. Providing genetic consultations to families with low socioeconomic levels may cause a considerable decline in the incidence rate of the disease.

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