



## Popliteal Pterygium Syndrome, Lethal Type

### Alternative Names

Bartsocas-Papas Syndrome

BPS

Pterygium Popliteal Lethal Type

### WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

### OMIM Number

263650

### Mode of Inheritance

Autosomal Recessive

### Description

The popliteal pterygium syndrome is a severe autosomal recessive disorder also known as the Bartsocas-Papas syndrome. The chief features of the popliteal pterygium syndrome include a popliteal pterygium with a cord containing nerves and vessels, synostosis of hand and foot bones with digital hypoplasia and syndactyly, genital abnormalities, facial clefts with short palpebral fissures, ankyloblepharon, hypoplastic nose, small mouth, and filiform bands between the jaws. In most cases with popliteal pterygium syndrome death occurs at the neonatal or intrauterine stages.

### Epidemiology in the Arab World

#### Qatar

Massoud et al. (1998) described popliteal pterygium syndrome in 4 sibs in an Arab Bedouin family. The phenotypically normal parents were unrelated; the father and mother were from Qatar and United Arab Emirates, respectively. The patients showed oral cleft, filiform bands between the jaws, ankyloblepharon, popliteal pterygium, syndactyly of fingers and toes, phalangeal anomalies with synostosis, clubfeet, nail hypoplasia, and genital anomalies. Additional traits included cutis aplasia, widely spaced nipples, low-set umbilicus, and unilateral renal hypoplasia. One of the sibs was stillborn; the other 3 children lived 10 to 17 months. The absence of parental consanguinity in the family was suggestive for a high gene frequency of this syndrome in the Bedouin community (Massoud et al., 1998).

#### United Arab Emirates

[See also: Qatar > Massoud et al. (1998)].

#### References

Massoud AA, Ammaari AN, Khan AS, ven Katraman B, Teebi AS. Bartsocas-Papas syndrome in an Arab family with four affected sibs: further characterization. *Am J Med Genet.* 1998; 79(1):16-21.

#### Contributors

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