



Nablus Mask-Like Facial Syndrome

WHO International Classification of Diseases

Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified

OMIM Number

608156

Mode of Inheritance

Autosomal recessive?

Description

Principal features of the Nablus Mask-Like Facial syndrome include, striking, expressionless, facial appearance with a long face; upswept frontal hairline with normal hair; tight glistening facial skin with small palpebral fissures; apparent hypertelorism; sparse almost absent eyebrows, bulbous nose with prominent columella; long upper and everted lower lip; maxillary hypoplasia; longitudinal cheek dimples; and abnormal posteriorly angulated ears without lobules. Other parts of the body including skin, nails, and genitalia are unremarkable. A Palestinian boy and a Sicilian girl are the two cases reported in the literature with this syndrome to date.

Epidemiology in the Arab World

Palestine

Teebi (2000) described a 4-year-old boy from the city of Nablus with strikingly unusual facial mask-like appearance. The parents were phenotypically normal and non-consanguineous. The patient's twin sister was phenotypically normal. Another 7-year-old brother was also normal. The affected child appeared unusual at birth, whereas his twin sister appeared normal. His psychomotor development was normal. On examination he was interactive and apparently intelligent boy. Teebi (2000) concluded that the appearance was apparently unique and different from that in urofacial syndrome.

References

Teebi AS. Nablus mask-like facial syndrome. *Am J Med Genet.* 2000; 95(4):407-8.

Contributors

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