

HYPERTELORISM-HYPOSPADIAS SYNDROME* (OPITZ SYNDROME)

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Two or more morphologic anomalies occurring in the same patient, usually characterize a malformation syndrome. Although with genetic etiology, the majority of the syndromes does not show a clear heredity pattern. This attractive aspect of dysmorphic syndrome studies impel us to report all the syndromic association at this level of knowledge. The aim of this communication is to report a familial example of the Opitz syndrome. This nosologic entity is characterized by telecanthus or apparent hypertelorism and hypospadias associated or not to mental retardation. Other morphologic abnormalities are strabismus, cryptorchidism, cleft lip and palate, and urinary tract defects. The prevalence of the Opitz syndrome is not known but it probably is much more frequent than it may be supposed. The most frequent characteristics of the syndrome, e.g., the telecanthus and hypospadias can be missing at the examination because of the wide variation of the telecanthus morphologic importance and, on the other hand, hypospadias is restricted to the male. Fig. 1 shows the mother with her first child; fig. 2 shows her second and last child (the propositus) at three years old, a more severe affected example of the syndrome. This syndrome presents some interesting aspects: Female have been reported to have telecanthus and no other findings, and no affected female are known to have died from the syndrome. These females, however, always link the generations in the case of familial Opitz syndrome, that is, it is admitted a dominant trait with no documented case of male-to-male transmission. Another important aspect to be faced in the study of the pathogenesis of this syndrome is a probable relationship between the process

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Fig. 1 — Mother and her first son. Note telecanthus.



Fig. 2 — The propositus showing a complete Opitz syndrome.

of twinning and the syndrome. The frequency of twins among affected individuals is significantly higher than the general population. Because of these pathogenetic implications and the primary prevention performed through genetic counseling, attention should be given to male patient showing hypospadias or an isolated female with telecanthus.

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