Association of type II Waardenburg syndrome with hypermetropic amblyopia

ABSTRACT

We present a case of hypermetropic amblyopia in type II Waardenburg syndrome (WS) to highlight the association. WS is an "oculo-dermato-auditif" dysplasia described in 1947 by Waardenburg and by Klein in 1950. It is distributed worldwide, with no predilection for race or gender. The prevalence is estimated to be 1:42 000 live births in the general population. WS is a genetic disease with autosomal dominant transmission with incomplete penetrance and variable expressivity. Complex network of interaction between six genes have been identified to date. They are PAX3 gene, primarily responsible for type I and III WS; MITF, SOX10, and SNAI2 genes in type II WS; EDN3 and EDNRB genes in type IV WS.