

Ohdo blepharophimosis syndrome

Ohdo syndrome is a multiple congenital malformation syndrome characterized by:

- Blepharophimosis
- Ptosis
- Dental hypoplasia
- Hearing impairment
- Intellectual disability
- Male patients may show cryptorchidism and scrotal hypoplasia.
- Abnormal ears, microcephaly, and growth retardation have been reported occasionally.

So far, less than 30 patients have been reported worldwide.

Most reported cases are *sporadic*, except the original cases of Ohdo who described two affected sisters and a first cousin, favoring *autosomal recessive* inheritance. *Autosomal dominant*, *X-linked*- and *mitochondrial inheritance* have also been suggested.

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