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Chromosome  
Disorder Outreach

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## ABOUT US

Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

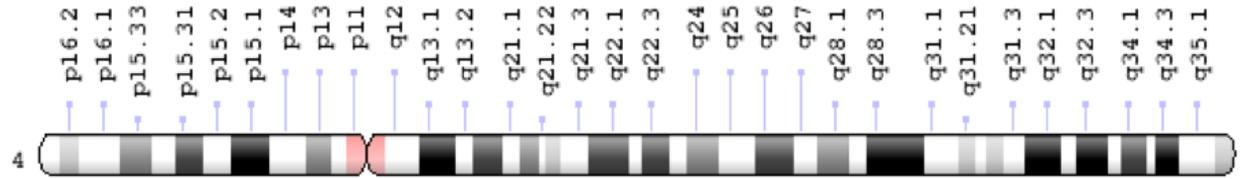
### Wolf-Hirschhorn Syndrome

### 4p16.3 deletion

# Wolf-Hirschhorn Syndrome

## 4p16.3 deletion

Wolf-Hirschhorn Syndrome (WHS) – caused by a deletion in 4p16.3, the most distal area of the short arm of chromosome 4 – was first reported as a genetic disorder in 1961. Clinical features include prenatal hypoplasia (low birth weight), severe growth delay, hypotonia (low muscle tone), microcephaly, and seizures within the first three years of life. More than 1,000 patients with WHS have been reported to date.



Other clinical features of WHS are dependent on the size of the deletion. WHS can be divided into 3 groups: patients with small deletions (less than 3.5 Mb), average deletions (4-18 Mb), or large deletions (over 20 Mb). Typically the patients with larger deletions have more severe manifestations, including developmental delay, cleft palate, eye defects, congenital heart defects, and kidney defects. Characteristic facial dysmorphism, hearing impairment and abnormal tooth development are also common.

Of the abnormalities of the eyes the most common defect is coloboma of the iris and/or the choroid. Other defects can include microphthalmia, cataracts, glaucoma, microcornea, and corneal opacity. The congenital heart defects occur relatively frequently, but are usually not life-threatening. The most common heart defects in these patients are atrial septal defects, ventricular septal defects, and stenosis of the pulmonary artery.

Kidney defects are less common than heart defects. These defects include hypoplastic kidneys (sometimes with cystic lesions), hydronephrosis, horseshoe kidney, and malposition of kidneys. Many affected boys have hypospadias.

The distinct facial abnormalities – typically found in individuals with larger deletions – include a prominent glabella, protruding eyes, epicanthus, micrognathia, low set ears, preauricular tags and pits. The distinct shape of the nose defects with the high forehead have often been compared to the shape of a Greek warrior helmet.