

Down Syndrome

What causes Down syndrome?

Down syndrome is caused by a duplication of all or part of chromosome 21, making three copies of the chromosome rather than the usual two copies. Down syndrome is also called trisomy 21. The extra genetic material leads to certain physical changes in persons with Down syndrome.

How common is Down syndrome?

The frequency of Down syndrome is approximately 1 in every 800 births, the rate is increased in older mothers. It is one of the most common chromosomal abnormalities in live-born children. The improved quality of medical care and educational resources have allowed for a more productive life and a longer life expectancy.

Do children with Down syndrome have eye problems?

Individuals with Down syndrome are at increased risk for a variety of eye and vision disorders. Fortunately, many of these eye problems can be treated, especially if discovered at an early age. The quality of life can be further enhanced by the proper assessment and correction of eye problems. The most common eye findings include:

- [Refractive errors](#) - Children with Down syndrome are more likely to need glasses than are other children. This may be due to [myopia](#) (near-sightedness), hyperopia (far-sightedness), and/or [astigmatism](#). Refractive error may develop early in life or later on.
- [Strabismus](#) - Between 20% and 60% of individuals with Down syndrome have eyes that are misaligned (strabismus). [Esotropia](#) (eyes that drift in) is most common while [exotropia](#) (eyes that drift out) occurs less frequently. Strabismus may be treated with glasses, patching and/or eye muscle surgery.
- [Keratoconus](#) - A cone shaped distortion of the cornea (front layer of the eye), occurs in up to 30% of those with Down syndrome. Keratoconus is usually diagnosed around puberty and should be monitored regularly. Blurred vision, corneal thinning, or corneal haze may result from keratoconus. Keratoconus is worsened by eye rubbing; therefore, eye rubbing should be discouraged.
- [Cataracts](#) - There is an increased incidence of congenital cataracts (present at birth) as well as acquired cataracts (develop later). The cataracts may progress slowly and should be monitored regularly, with surgical treatment performed when appropriate.
- [Glaucoma](#) - There is an increased risk of infantile glaucoma (elevated pressure within the eye).



- [Blepharitis](#) - Inflammation of the eyelids with redness at the edge of the lids and crusting around the lashes may occur and cause a feeling of dryness or burning. Treatment is with eyelid hygiene and topical antibiotics.
- Tearing - Excessive tears or watering of the eyes may occur because the drainage channels are blocked or narrow ([nasolacrimal duct obstruction](#)). This may require surgical intervention.
- [Nystagmus](#) - This is an involuntary “back-and-forth” movement or shaking of the eyes. It can affect vision to a mild or severe degree.

When should children with Down syndrome receive an eye exam?

The American Academy of Pediatrics recommends that children with Down syndrome be examined by a pediatric ophthalmologist or an ophthalmologist who is familiar with the examination of infants with disabilities. The initial exam should be done by six months of age with follow up exams once per year or more if needed.

Where can I find more information regarding Down Syndrome?

- [National Down Syndrome Society](#)
- [Down's Syndrome Association](#)