

## Ptosis (Droopy Eyelid)

**Ptosis** (pronounced “toe-sis”) refers to a droopy upper eyelid that sags and covers part of the eye. The abnormally low position of the eyelid is also called **blepharoptosis**, upper eyelid ptosis, or droopy eyelid. The condition can affect one or both eyes. An eyelid that sags severely will prevent the eye from seeing properly.

### Symptoms

- In addition to the visible drooping of the eyelid, the patient may also:
  - Feel tiredness and achiness around the eye(s)
  - Tear up frequently
  - Have decreased vision
  - Tilt the head back to see

### How Ptosis Happens

- Our eyes can open and close because of the movement of the eyelids, which depends on the contraction and relaxation of the levator muscles.
- If there are issues in the levator muscles or the connecting nerves, the upper eyelid will sag over the eyes, resulting in ptosis.
- Ptosis can happen from birth. It can also develop as a result of age, nerve damage, other medical conditions, or eye surgeries.

### Diagnosis and Treatment

- The healthcare provider can diagnose ptosis based on the sagging of the eyelid. He/she may also test the vision and eye movement.
- Ptosis that does not affect vision may not need to be treated.
- Severe ptosis can be corrected by surgery to lift the eyelid.
- **In children, ptosis should be assessed and treated as early as possible to minimize the impact on their vision development.**
  - Untreated severe ptosis may lead to [astigmatism](#), [amblyopia](#), and neck problems (from tilting the head in order to see)



Ptosis

### Ptosis and 22q Differences

Ptosis has been reported in ~4% of children with 22q11.2 deletion syndrome. The frequency of ptosis in children with 22q11.2 duplication syndrome is unclear due to the small number of patients in each study.

### Conditions That May Be Mistaken as Ptosis

**Hypotropia** is a type of strabismus in which the eye is not aligned well and turns downward. See [Strabismus \(Misalignment of the Eyes\)](#).

**Hooded eyelid** is a normal trait caused by excess skin folding, and there is nothing wrong with the levator muscle or the nerves that lift the eyelid. Unlike ptosis, hooded eyelids are a cosmetic problem but not a medical one.



Hooded eyelid

It is recommended that individuals with 22q11.2 deletion or duplication have a **comprehensive eye examination** at diagnosis, with follow-up as indicated by findings. Typically, this means having a comprehensive eye exam **every few years for kids**, and **every 1-2 years for adults**.

### Resources

- [Ptosis \(Droopy Eyelid\)](#) – Cleveland Clinic
- [What Is Ptosis?](#) – American Academy of Ophthalmology
- [Ocular findings in the chromosome 22q11.2 deletion syndrome](#) (2007)
- [Ocular findings in 22q11.2 deletion syndrome: A systematic literature review and results of a Dutch multicenter study](#) (2022)
- [Ocular findings associated with chromosome 22q11.2 duplication](#) (2016)
- Updated clinical practice recommendations for managing [\[children | adults\]](#) with 22q11.2 deletion syndrome – 2023 [These documents can also help guide the care of individuals with **22q11.2 duplication syndrome**, as the associated features are quite similar, just with lower frequency.]
- ❖ Images of Ptosis and Hooded Eyelids – Dr. Brian Forbes, Children’s Hospital of Philadelphia