

Sclerocornea

The **cornea** is the front outer part of the eye and is usually clear. It lets light enter the eye and helps focus it. **Sclerocornea** is a rare birth defect of the eye in which the cornea is cloudy, and vision may be reduced. It has been reported in a small number of children with 22q11.2 deletion syndrome (22q11.2DS).

What is Sclerocornea

- The cornea is cloudy, like the **sclera** (white part) around it.
- The cloudiness can happen to the whole cornea or part of it.
- The **limbus** (border between the cornea and the sclera) is not obvious.
- The cloudiness may decrease the amount of light that enters the eye and therefore reduce vision, but some patients have near-normal eyesight.
- Sclerocornea usually affects both eyes. It does not get better or worse.
- Sclerocornea is not associated with inflammation.

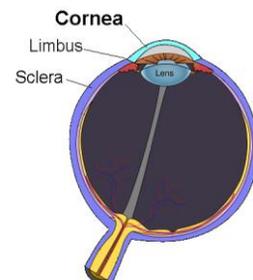
How does Sclerocornea Happen

- Sclerocornea can happen with or without family history and may be passed onto the next generation.
- It can happen with additional eye problems.
- It can happen with or without intellectual disabilities, deafness, abnormalities of the head and face etc., and other health problems.
- It happens due to the abnormal migration and development of neural crest cells when the fetus is between 7-10 weeks old in the womb.

Diagnosis and Treatment

- An optometrist or ophthalmologist can diagnose sclerocornea and check for other (potentially serious) conditions by performing a **comprehensive eye exam**.
- If sclerocornea is the only problem, a corneal transplant (**keratoplasty**) may be recommended. However, the procedure is complex, with a high rejection rate and poor long-term outcome.
- The patient may need to be monitored and treated throughout his/her life by multiple eye-care specialists.

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**.



Other conditions that can cause cloudiness in the cornea:

- Glaucoma (optic nerve damage)
- Peter's Anomaly
- Trauma and injuries to the eye
- Infections
- And many others

Resources

- [Sclerocornea](#) – Eyewiki.org
- [Keratoplasty](#) – Mayo Clinic
- [Overview of sclerocornea](#) – 2023
- [Sclerocornea Associated With the Chromosome 22q11.2 Deletion Syndrome](#) – 2008
- 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.]
- ❖ [Image of Sclerocornea](#) – Eyewiki.org
- ❖ [Schematic diagram of the human eye](#) – Wikimedia / Mikael Häggström