

Reduced Vision

A number of eye conditions can lead to a decrease in the ability to see. This information sheet explains three such conditions that have been reported in individuals with 22q11.2 deletion syndrome (22q11.2DS) and 22q11.2 duplication syndrome (22q11.2DupS). They are coloboma, cataract, and amblyopia.

Coloboma

- The eye is **missing some normal tissue at birth**
- Results in a **loss of vision** and **sensitivity to light**
- Can happen in one or both eyes
- Caused by problems with eye formation when the baby is in the womb – the optic fissures fail to close completely
- Increases the risk of amblyopia (see next page), cataracts (if coloboma is in the lens; see below), as well as abnormal blood vessel growth and retinal detachment (if coloboma is in the retina)



An iris coloboma – In this case, the pupil is shaped like a keyhole instead of a circle

Coloboma Type (Missing Tissue)	Visible Signs	Vision	Accommodation or Treatment
Eyelid	<ul style="list-style-type: none"> • Incomplete upper or lower eyelid 	<ul style="list-style-type: none"> • May be reduced 	<ul style="list-style-type: none"> • Bandages, special contact lenses, or artificial tears to keep the eye moist • Surgery to repair eyelid in severe cases
Lens	<ul style="list-style-type: none"> • The lens that is missing tissue cannot be seen from outside 	<ul style="list-style-type: none"> • Blurry vision 	<ul style="list-style-type: none"> • Eyeglasses or contacts • Surgery to replace the affected lens
Optic nerve (or optic disc)	<ul style="list-style-type: none"> • The optic nerve that is missing tissue cannot be seen from outside 	<ul style="list-style-type: none"> • Blurry vision • Reduced vision • Blind spots 	<ul style="list-style-type: none"> • Low vision aids if needed • Not possible to repair
Iris	<ul style="list-style-type: none"> • The pupil appears like an oval or a keyhole instead of a circle; • A hole in the iris makes a second pupil 	<ul style="list-style-type: none"> • Light sensitivity • Blurry vision 	<ul style="list-style-type: none"> • Wear colored contact lenses to make the iris appear round and to reduce light sensitivity • Surgery to repair the iris
Chorio-retinal (retina)	<ul style="list-style-type: none"> • The gap in the back of the eye cannot be seen from outside 	<ul style="list-style-type: none"> • Distorted • Blind spots 	<ul style="list-style-type: none"> • May or may not be necessary • Surgery if complications arise

Cataract

- The **lens inside the eye is cloudy instead of clear**, so it blocks the light and cannot transmit it properly
- Results in hazy, blurry, and less colorful vision
- Present at birth or appears at any age to one or both eyes
- Symptoms include cloudiness in the eye, reduced vision, light sensitivity, lack of interest in colors etc.
- A photo of an eye with a cataract has a dim or white (not red) reflex. ([Photo requirements](#): The child looks directly at the camera lens, the camera flash is on, the background is dimly lit, and red-eye reduction is off.)
- Caused by genetic factors, certain health conditions (e.g. diabetes), eye injuries, infection during pregnancy etc.
- Surgery may be needed to remove the cloudy lens or replace the lens with an artificial one (intraocular lens, IOL)



A child young with a cataract.

Treat Children's Cataracts Early!

If only 1 eye is affected by a cataract, the child's brain learns to ignore images from that eye, leading to **permanent vision loss**. See **Amblyopia** on the next page.

