Eye Issues in 22q Differences

Our **eyes** are complex organs that process the incoming light to help us see. When parts of the eyes do not work well or did not develop fully, we may not be able to see properly. The "Eye Series" provides explanations on the common and/or significant eye problems in individuals with 22q11.2 deletion and duplications. The **Health Conditions Explained** page on our website contains all the info sheets listed below.

Issues reported in Individuals with 22q11.2DS	See our info sheet on:
• Nearsightedness (**Myopia**) • Farsightedness (**Hyperopia**) • **Astigmatism**	• Refractive Errors
• Missing tissues in a part of the eye (**Coloboma**)	• Reduced Vision
• **Cataract**	
• Lazy Eye (**Amblyopia**)	
• **Exotropia, Exophoria, Esotropia, Esophoria**, or any form of eye misalignment (**Strabismus**)	• Strabismus (Misalignment of the Eyes)
• Cloudiness of the Cornea (**Sclerocornea**)	• Sclerocornea
• Droopy Eyelid (**Ptosis**)	• Ptosis (Droopy Eyelid)

Eye Issues in 22q11.2DupS
Strabismus, hyperopia and astigmatism are the most common eye issues in 22q11.2DupS. The other issues are rare.

Conditions that are common in 22q11.2DS but don't affect vision

Tortuous retinal vessels – Blood vessels on the retina are twisted. This condition may be related to increased blood pressure.

Posterior embryotoxon – An opaque ring is visible in the cornea (which is normally transparent). This ring is present since birth and may happen together with other genetic conditions.

Hooded eyelid – The upper eyelid has excess skin folding but can still open and close. Hooded eyelid is not the same as Ptosis (Droopy Eyelid), which is due issues with the levator muscles or related nerves.

Comprehensive Eye Exams

A comprehensive eye exam checks these aspects:

- Personal and family history of health, work, and environment
- Visual acuity – How clearly the person sees (on the eye chart)
- Refraction / lenses needed
- Depth perception
- Color vision
- Side (peripheral) vision
- Eye muscle movements
- How well the eyes work together
- Pupils' response to light
- Curvature of the cornea
- Any cataracts
- Eyelids, iris, and lens
- Eye pressure
- Retina and optic nerves (pupils dilated)

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

- Associations of Retinal Microvascular Diameters and Tortuosity With Blood Pressure and Arterial Stiffness: United Kingdom Biobank – 2019
- Posterior embryotoxon (PE) – Children's Health
- Eye Exam and Vision Testing Basics – American Academic of Ophthalmology
- Comprehensive eye exams – American Optometric Association
- 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)
- Updated clinical practice recommendations for managing [**children** | **adults**] with 22q11.2 deletion syndrome – 2023

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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