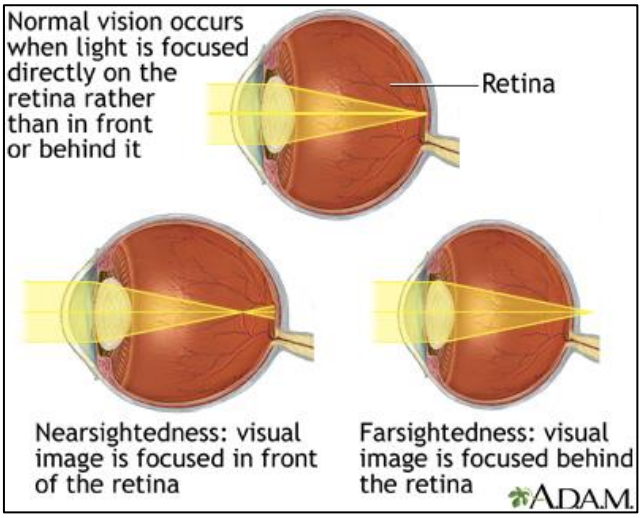
Refractive Errors

In a person with normal vision, the light that enters the eye is refracted (bent) slightly at multiple spots. Then the light is able to focus correctly on the retina (back of the eyeball), and the person gets a clear image of the object. [See our info sheet "Introduction to the Eye"].

Refractive errors happen when the shape of the eye (or its parts) does not allow incoming light to bend properly. The light does not focus properly on the retina, leading to blurry vision. **Nearsightedness**, **farsightedness**, and **astigmatism** are the main types of refractive errors. These errors can be accommodated using eyeglasses or contact lenses. Less commonly, surgeries are used to fix the problem.



Types of Refractive Errors

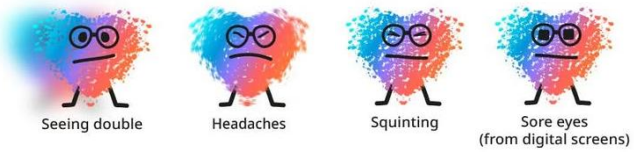
	Nearsightedness (Myopia)	Farsightedness (Hyperopia)	Astigmatism
Problem	• Far away things look blurry	• Near things look blurry	• Things at any distance look blurry
Light	• Light entering the eye focusses in front of the retina instead of on it	• Light entering the eye focusses behind the retina instead of on it	• Light entering the eye bends unevenly and cannot focus properly [see diagram]
Causes	• Eye ball is too long from front to back; OR • Cornea or lens is more curved than it should be	• Eye ball is too short from front to back; OR • Cornea or lens is not curved enough	• Eye ball is oval-shaped rather than round; OR • The cornea or lens is more curved than it should be
Risk Factors	• Genetics • Ethnicity • Frequent reading and use of computer/phone screens	• Genetics • Certain genetic disorders	• Genetics • Eye injuries • Eye disease • Eye surgery
Start time	• Starts and worsens in childhood • Worsening will likely slow down or stop by the end of the teenage years	• Present at birth or starts at an early age; normal growth may correct the problem • ~40 years old for the type of farsightedness (**presbyopia**) that arises due to age related changes to the lens	• Present at birth or starts at any point in life • Can worsen, improve, or stay the same
Prescription for eyeglasses OD = right OS = left	• A negative number (such as -3.00) in **SPHERE** (1st number on prescription) means nearsightedness. The higher the number, the stronger the glasses you need.	• A positive number (such as +3.00) in **SPHERE** (1st number on prescription) means farsightedness. The higher the number, the stronger the glasses you need	• A higher degree in **CYL** (**Cylinder**; 2nd number on prescription) means worse astigmatism, and the eye is more oval than round • The degree in **AXIS** (3rd number) shows the location of the astigmatism on the cornea

Eye Series for Individuals with 22q11.2 Differences

Refractive Errors (continued)

Symptoms of Refractive Errors

- Blurry vision (up close, far away, or both)
- Seeing double
- Headache
- Squinting
- Eye strain
- Eye pain



⚠️ Individuals who are young and/or have intellectual disabilities may not be able to say that their vision is blurry. Parents and caregivers are advised to watch out for signs that may be related vision problems, e.g. squinting, school grades getting worse

Comprehensive Eye Exams

- During a comprehensive eye exam, eye care professionals check:
  - How well can the person see? (see Snellen Chart on the right)
  - How healthy are the eyes?
  - Do both eyes work together?
  - Are the inside and the surface of the eyes normal?
  - Are there early warning signs for diabetes, high blood pressure, autoimmune diseases, cancers, or other serious conditions?
- Eye care professionals can:
  - Help correct a person's vision if needed
  - Help prevent the problems from getting worse.
  - Make referrals for additional medical attention

For individuals with 22q11.2 deletion or duplication syndrome, a **comprehensive eye examination** is recommended 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**.

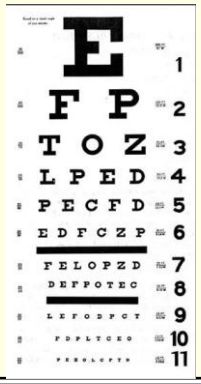
Refractive Errors and 22q Differences

The most common refractive errors in people with 22q11.2 deletion syndrome are hyperopia (6–48%) and astigmatism (3–23%).

The most common refractive errors in people with 22q11.2 duplication syndrome are hyperopia (22–26%) and astigmatism (~11%).

The Snellen Chart

The Snellen Chart is used to check how well people see. Children who cannot read yet may use an eye chart with figures instead of letters.



Potential Complications

People with refractive errors are at a higher risk of developing:

- Glaucoma – An increase in eye pressure that may lead to blindness
- Amblyopia (lazy eye) – vision reduced in one eye
- Strabismus – misaligned eyes
- Low vision

Resources

- Refractive errors | Nearsightedness (Myopia) | Farsightedness (Hyperopia) | Age-related Farsightedness (Presbyopia) | Astigmatism – Cleveland Clinic
- Farsightedness (Hyperopia) – Government of Alberta, Canada
- What does 20/20 Vision Mean? | Astigmatism – American Academy of Ophthalmology
- World Sight Day 2022: #LOVEYOUR EYES – The International Optician's Association
- See the Full Picture of Your Health with an Annual Comprehensive Eye Exam | Comprehensive Pediatric Eye and Vision Examination – 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)
- 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 Normal, nearsightedness, and farsightedness – Medline Plus Encyclopedia, National Library of Medicine
- ❖ Images of eye problems from World Sight Day Activities – Preventblindness.org
- ❖ Image of the Snellen Chart – Encyclopaedia Britannica

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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