

*The AAPOS Genetic Eye Disorders Committee
proudly presents...*

Genetic eye disorders in children: Five practice pearls

Presenters: Virginia Miraldi Utz, Arif Khan, Emily McCourt,
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Board Liaison: Sean Donahue

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AAPOS 2026

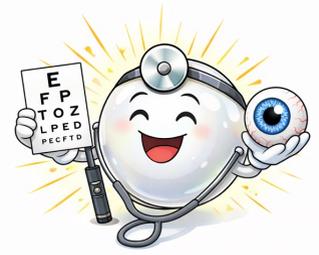
51st **Annual Conference**

Westin Seaport District

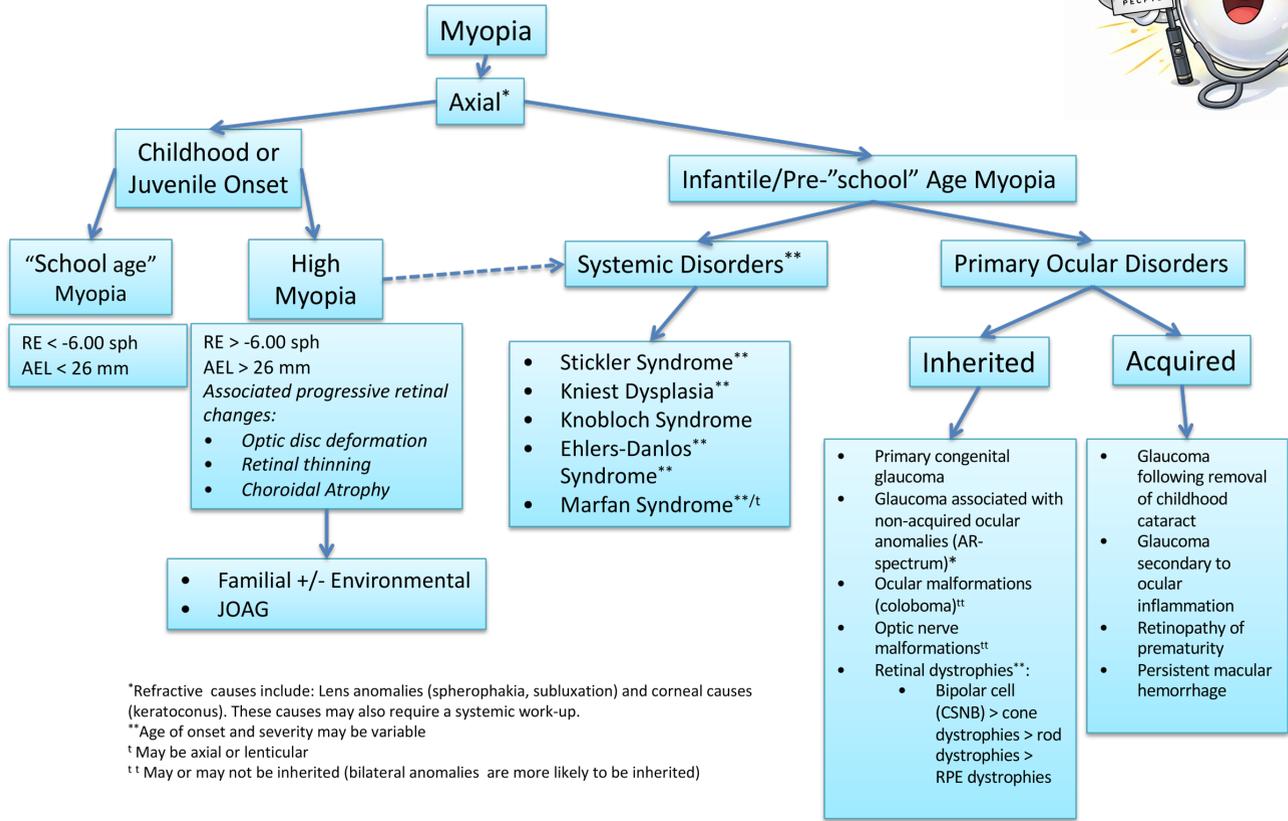
March 18–22, 2026

Boston, Massachusetts

1. Recognize systemic clues from the ocular examination



Example: High, pre-school aged or congenital myopia



*Refractive causes include: Lens anomalies (spherophakia, subluxation) and corneal causes (keratoconus). These causes may also require a systemic work-up.

**Age of onset and severity may be variable

† May be axial or lenticular

†† May or may not be inherited (bilateral anomalies are more likely to be inherited)

Select Syndromic Causes of Myopia:

Syndrome	Molecular Basis	Ocular Features	Systemic Features	Referral
Stickler Syndrome	<i>COL2A1</i> (Type I), <i>COL11A1</i> (Type II), <i>COL11A2</i> (Type III)	High myopia (often congenital), vitreous abnormalities (membranous or beaded type), retinal detachment risk, early cataract, glaucoma	Midface hypoplasia, cleft palate (Pierre Robin sequence), sensorineural hearing loss, early-onset osteoarthritis, skeletal dysplasia	Genetics, Ophthalmology, ENT, Cardiology (mitral valve prolapse), Rheumatology
Marfan Syndrome	<i>FBN1</i> (fibrillin-1)	High myopia, ectopia lentis (classic: superotemporal displacement, 30-60%), retinal detachment risk, flat cornea	Aortic root dilatation/dissection, tall stature, arachnodactyly, pectus deformity, scoliosis, joint hypermobility, dural ectasia	Cardiology (urgent for aortic monitoring), Genetics, Ophthalmology, Orthopedics
Knobloch Syndrome	<i>COL18A1</i> (biallelic pathogenic variants)	High myopia (-10 to -20 D), smooth (cryptless) irides, temporal ectopia lentis, distinctive vitreoretinal degeneration (severe RPE atrophy, macular atrophic lesions, white fibrillar vitreous), retinal detachment (50% of eyes; median onset 2.5 years), posterior perinuclear lens opacity, cone-rod dysfunction on ERG	Occipital encephalocele/meningocele (variable; may be absent), occipital skull defects, cutaneous scalp changes, learning difficulties, epilepsy, congenital renal abnormalities, developmental delay; most patients have normal intelligence	Ophthalmology (urgent for RD risk), Genetics, Neurosurgery (if encephalocele present), Neurology (if seizures/developmental delay), Nephrology (if renal abnormalities)

Dr. Traboulsi and Panel:

- What would you do next?
- Differential diagnosis?
- Clues to identifying systemic disease

2. Just diagnosed my patient with an inherited retinal disorder.



Case presentation: Dr. Del Valle

Counseling with hope and realistic expectations (Dr. Levin and Panel Discussion)

- Will I go blind?
- Will I be able to drive?

Supportive resources to consider (low vision, educational support), registries
(Compiled by Jen Rossen, MD)

a. Low Vision/Visual Rehabilitation: AAPOS Low Vision Resource Page:

- <https://aapos.org/syndicated/pediatric-low-vision>
- <https://higherlogicdownload.s3.amazonaws.com/AAPOS/159c8d7c-f577-4c85-bf77-ac8e4f0865bd/UploadedImages/Documents/AAPOS-Patient-Resources.pdf>

The screenshot shows the AAPOS website page for Pediatric Low Vision Rehabilitation Resources. The page features a navigation menu with links for Job Opportunities, Store, Join, Contact Us, and LOGIN. The main content area is titled "Pediatric Low Vision Rehabilitation Resources" and includes three columns of resources:

- LOW VISION RESOURCES**
 - [Low Vision Patient and Family Resource Information Sheet](#)
 - The Importance of Vision:**
Why is the vision so important? What special role does it play in learning and guiding development from the moment of birth?
- BRAIN BASED VISION IMPAIRMENT: CVI RESOURCES**
 - [AAPOS CVI Webinar: Expert Panel from Pediatric Ophthalmology and Neuro-Ophthalmology and Parent Perspective](#)
 - [AAPOS Pediatric Low Vision: Helping You Help Your Patients](#)
- REFERENCES**
 - [List of High Yield Journal Articles and References](#)
- POWERPOINTS**
 - [Cortical/Cerebral Visual Impairment](#)
 - [Cortical Visual Impairment](#)
 - [Retinopathy of Prematurity Medical](#)

b. Early Intervention services

Support for 504 and IEP accommodations

- At time of diagnosis
- Or in preparation for Kindergarten for congenital conditions

But what's the difference between a 504 Plan and an IEP?

- **504 Plans** describe adjustments in the school setting to remove barriers to the regular education in school
- **IEPs** remove barriers *and also provide specialized instruction and services* to help a child develop the skills that their disability makes harder to learn

- AAP 504 Plan v. IEP (Healthychildren.org)



c. Foundation Fighting Blindness

MyRetina Tracker

Parents can sign up their child with clinical and genetic information to be contacted for studies

A screenshot of the website fightingblindness.org/my-retina-tracker-registry. The browser address bar is highlighted in red. The page features the Foundation Fighting Blindness logo and the 'My Retina Tracker Registry' title. Below the title are two green buttons: 'Member Login' and 'Register', with the 'Register' button highlighted in red. A 'Clinician Portal' link is also visible. On the right, there are links for 'MY RETINA TRACKER PROGRAM', 'Why Genetic Testing is Important', and 'Open Access Genetic Testing Program'. An accessibility menu is open on the right side, showing options for 'Text Size' (set to 100%) and 'Theme' (with three color options: A, A, A).

Sign up

Welcome to My Retina Tracker Registry

Any person who has an inherited retinal degeneration or has a genetic relationship (often called a blood relative) to a person with the disease – such as a parent, child, uncle, grandparent, may join the registry.

The registration process consists of three steps:

First, sign up by entering a few account credentials. If you are a parent or guardian creating an account for your affected child, we will also ask you to provide your contact information.

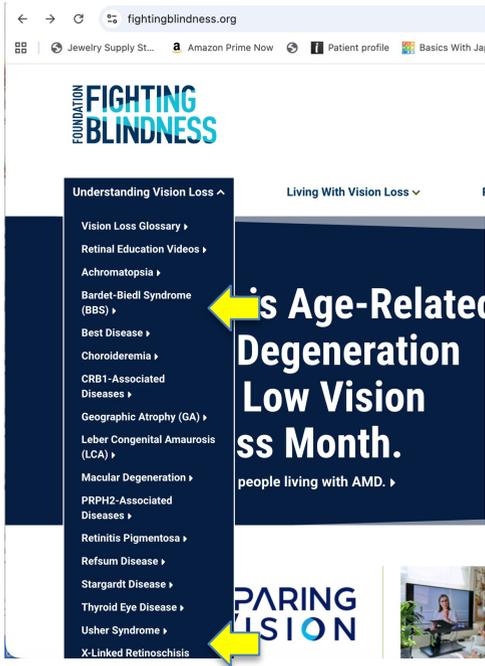
Second, provide some additional information about yourself as a new member. If you are entering this on behalf of someone else at their request, the words "you", "yourself", and "member" refer to the person you are entering information for.

Third, complete an informed consent. This consent form provides important information about what it means to be involved in this registry. People who want to take part in this registry must read this consent form and agree to participate by checking an "I agree" at the end.

After these steps, you will be guided through a series of questions that will help you build your retinal health profile.

FFB Patient Handouts (Infographics)

- Support and Resources: <https://www.fightingblindness.org/>
- Check out the infographics: (example: Bardet Biedl Syndrome)



Scroll to bottom of BBS page for infographics

RESOURCE DOWNLOAD

What You Should Know About Bardet-Biedl Syndrome Infographic - 11X17

Download (PDF, 553 KB)

RESOURCE DOWNLOAD

What You Should Know About Bardet-Biedl Syndrome Infographic - 8.5X11

Download (PDF, 496 KB)

RESOURCE DOWNLOAD

What You Should Know About Bardet-Biedl Syndrome Infographic - 8.5X11 (Spanish Version)

Download (PDF, 8.84 MB)

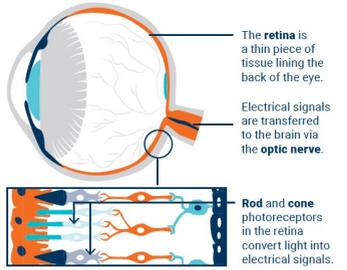
Spanish Infographics

LO QUE DEBE SABER ACERCA DE LA RETINOSQUISIS LIGADA AL CROMOSOMA X

DISEASE INFORMATION

Bardet-Biedl Syndrome

Often diagnosed in childhood or adolescence, Bardet-Biedl syndrome (BBS) is an inherited disease causing progressive loss of night and peripheral vision from retinitis pigmentosa (RP).



HOW IS BBS INHERITED?
Bardet-Biedl syndrome is autosomal recessive. That means it is **genetically passed** when both parents have one gene for the syndrome paired with one normal gene. There is a **25% chance** of their child inheriting the two BBS genes. **18 BBS causing genes** have been identified.



ABOUT BBS
BBS is often diagnosed at birth or in childhood when problems due to retinitis pigmentosa or other problems related to the syndrome are discovered.

People with BBS-related RP experience a gradual decline in their vision, because photoreceptors degenerate.

GENETIC TESTING
Genetic testing helps with attaining an accurate diagnosis. It also helps assess the risk of passing the disorder to offspring. A patient with an accurate diagnosis is in a better position to understand which emerging treatment approaches and clinical trials are most appropriate for them.

WHAT TO EXPECT WITH BARDET-BIEDL SYNDROME

Vision-related symptoms



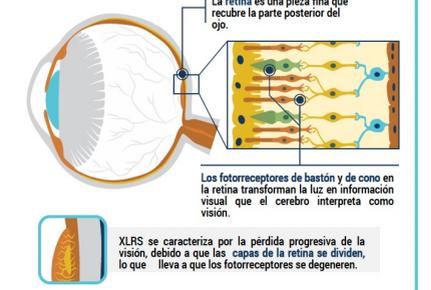
- Other related symptoms** (Symptoms vary from person to person)
- Polydactyly**: Extra fingers and toes are usually removed in infancy or early childhood.
 - Obesity**: May be present by childhood and is usually limited to the trunk of the body.
 - Renal (Kidney) Disease**: Renal abnormalities can affect the structure and the function of the kidneys.

Spanish Infographics

LO QUE DEBE SABER ACERCA DE LA RETINOSQUISIS LIGADA AL CROMOSOMA X

¿QUÉ ES LA RETINOSQUISIS LIGADA AL CROMOSOMA X?

La Retinosquisis Ligada al Cromosoma X (XLRS) es una enfermedad hereditaria en la retina, que provoca la pérdida de la visión central y periférica debido a la degeneración de la retina. Cerca de 35,000 personas en los Estados Unidos sufren de esta condición.

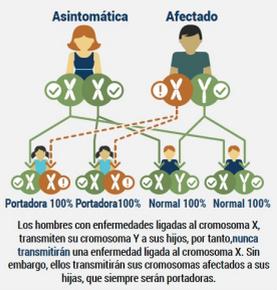
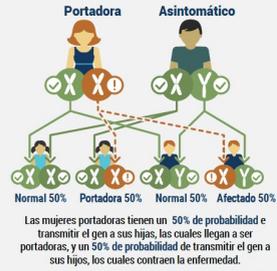


SÍNTOMAS DE LA RETINOSQUISIS LIGADA AL CROMOSOMA X

- Uno de los primeros síntomas de la XLRS es la agudeza visual reducida.
- La lesión muscular quística (como ampollas) en la retina es el rasgo común de la XLRS, y agrava la falta de la visión.
- La lesión muscular quística puede tratarse con medicamentos que lleven inhibidores de la anhidrasa carbónica orales o tópicos.

¿CÓMO SE HEREDA LA RETINOSQUISIS LIGADA AL CROMOSOMA X?

La XLRS es causada por las mutaciones del gen RS1. La enfermedad se contrae en las familias por el patrón de herencia ligado al cromosoma X.



- Afecta principalmente a los hombres.
- Generalmente se diagnostica en la niñez.
- La pérdida de la visión es a menudo progresiva.
- Casi siempre conlleva a la ceguera total.

- Navigating Clinical Research: Referral to Treatment Trials
 - What steps does the general pediatric ophthalmologist need to take to refer? : Discussion led by Dr. Alina Dumitrescu
 - Before referral:
 - Obtain molecular confirmation whenever possible (CLIA-certified testing if available)
 - Clarify phenotype: onset, progression, imaging features, electrophysiology
 - Ensure the genotype matches the trial’s inclusion criteria (e.g., biallelic pathogenic variants, specific mutation types)
 - Counseling Considerations / Contacting Sponsor
 - Protecting the relationship with the Family

- Navigating Clinical Research: Referral to Natural History Studies
 - What are natural history research studies and how to discuss with patients: Discussion led by Dr. Laryssa Huryn
 - What are natural history studies?
 - Before referral (follow as in the section above) – genetic testing, clarifying phenotype, and ensure patient’s diagnosis matches trial’s inclusion criteria.
 - What does participation entail?
 - What are the benefits of participation?



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Current Gene Therapy Trials



CLINICAL TRIALS FOR GENETIC EYE DISORDERS

AAPOS has no financial interest in any clinical trials. AAPOS does not promote any particular study, and does not endorse any study. Clinical trials by definition offer treatments that have not been tested and may or may not benefit patients who enroll.

Partial list of treatment trials for children with genetic eye disorders.

Please refer to www.ClinicalTrials.gov for more information.

There are many more gene therapy trials for patients 18 years and older.

There is also a registry and natural history observational study for many retinal degenerations. For more information contact Coordinator@MyRetinaTracker.org 1-800-683-555. Free molecular genetic testing is also available for a large number of retinal genes through MyRetinaTracker; go to <https://www.fightingblindness.org/my-retina-tracker-registry>.

[Clinical Treatment Trials Recruiting Children with Genetic Eye Disorders](#)

Updated 11/2025

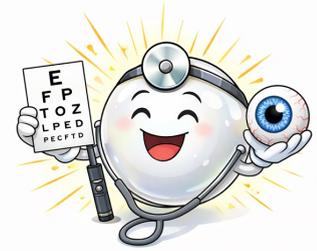
GENETIC EYE DISORDERS
RESOURCES

Case summary and discussion (Led by Dr. Alex Levin)

3. Ocular Malformation: Consult in the NICU or outpatient setting with an anterior segment malformation – how to approach and what to do next.

Case Presentation by Dr. Gerald Zaidman:

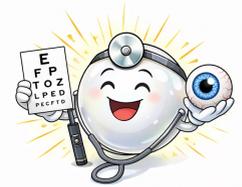
- Infant with bilateral anterior segment anomalies



Case Discussion led by Dr. Arif Khan.

- Malformation complex: clues to systemic association
- Resources to use to look for association
- Diagnostic approach to genetic testing with counseling
 - Microarray v. Panel v. WES/WGS
- Negative testing: What next?
 - Counseling families when results with results are negative or inconclusive.

4. Patient referred for eye exam with a known genetic condition that I have never heard of... Steps to an efficient and thorough evaluation in a busy pediatric ophthalmology clinic.



Case Presentation by Dr. Jose Puthussery

Resources to use in real-time by Drs. Jen Rossen, Emily McCourt and Jose Puthussery

- GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK24676/>
- Free, Gold standard for genetic references and guidelines
- Control + F – search “ophthal” and scroll



National Library of Medicine
National Center for Biotechnology Information

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Dyskeratosis Congenita and Related Telomere Biology Disorders
Synonym: Zinsser-Cole-Engman Syndrome

Sharon A Savage, MD and Marena R Niewisch, MD.
Author Information and Affiliations
Initial Posting: November 12, 2009; Last Revision: January 19, 2023.
Estimated reading time: 46 minutes

Summary

fibrosis	diagnosis	ophthal
Pulmonary arteriovenous malformations	Bubble echocardiogram	In case of pulmonary symptoms in absence of pulmonary fibrosis
Oral & dental issues	<ul style="list-style-type: none"> Exam by dentist & dental hygienist Monitoring of leukoplakia & biopsy of suspicious lesions 	Every 6 mos
Immunologic issues	Follow up based on findings at initial eval	At immunologist's discretion
Cutaneous manifestations	Full-body skin exam by dermatologist	Annually
Developmental delay	Clinical eval & referral to specialist if needed	At each visit in childhood
Ophthalmologic problems	Ophthalmologic exam (vision problems, abnormally growing eyelashes, lacrimal duct stenosis, retinal changes, bleeding, cataracts, & glaucoma)	Annually
Avascular osteonecrosis	Clinical eval based on symptoms	At each visit
Liver disease	Liver function tests (irrespective of androgen therapy)	Annually

BMF = bone marrow failure; CBC = complete blood count; PFT = pulmonary function test
1. Including 5q-, 7q-, monosomy 7, trisomy 8, 20q-, 11q23 translocation, 3q abnormalities

Real-time Resources Drs. Jen Rossen, Emily McCourt and Jose Puthussery

- An Online Catalog of Human Genes and Genetic Disorders

(<https://www.omim.org/>)



CHARGE syndrome

#214800
Table of Contents

CHARGE SYNDROME

Alternative titles; symbols

CHARGE ASSOCIATION--COLOBOMA, HEART ANOMALY, CHOANAL ATRESIA, RETARDATION, GENITAL AND EAR ANOMALIES
HALL-HITTNER SYNDROME; HHS

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
Ra12.2	CHARGE syndrome	214800	AD	3	CHD7	608892

External Links

- Clinical Trials
- EuroGentest
- Gene Reviews
- Genetic Alliance
- MedlinePlus Genetics
- OrphaNet

Animal Models

INHERITANCE
- Autosomal dominant

GROWTH
Other
- Postnatal growth retardation

HEAD & NECK
Head
- Microcephaly

Face
- Square face
- Malar flattening
- Micrognathia
- Facial asymmetry

Ears
- Small ears
- Lop ears
- Cup-shaped ears
- Deafness (sensorineural or mixed sensorineural and conductive)
- Mondini defect
- Hypoplastic semicircular canals

Eyes
- Colobomas (iris, choroid, retina, disc, and optic nerve)
- Anophthalmia
- Microphthalmia
- Proptosis
- Hypertropism
- Downsloping palpebral fissures

Nose
- Posterior choanal atresia (membranous and/or bony)
- Anomia

- Uptodate.com [requires institution subscription]



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Telomere biology disorders, including Dyskeratosis congenita

Outline

SUMMARY AND RECOMMENDATIONS

INTRODUCTION

PATHOPHYSIOLOGY

Role of telomeres

Genetics

Inheritance patterns

EPIDEMIOLOGY

CLINICAL FEATURES

Classic DC presentation

Bone marrow failure

Additional somatic features

AUTHORS: Timothy S Olson, MD, PhD, Kasiani C Myers, MD
SECTION EDITOR: Peter Newburger, MD
DEPUTY EDITOR: Alan G Rosenblatt, MD

Literature review current through: Jan 2026.
This topic last updated: Jan 17, 2025.

INTRODUCTION

Telomere biology disorders (TBDs), also called telomeropathies, are inherited disorders characterized by bone marrow failure (BMF), cancer predisposition, and somatic (ie, nonhematologic) abnormalities. TBDs are caused by mutations that interfere with the normal maintenance of telomeres, the regions at the ends of the chromosomes that protect nucleated cells from the loss or gain of genetic material. Dyskeratosis congenita (DC) and other TBDs are rare syndromes associated with short telomeres.

- Open Evidence AI (openevidence.com)

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OpenEvidence

Dosing adjustment for ceftriaxone in patients with severe renal impairment?

Write a Prior Auth Letter Write Home Care Instructions Prepare For MOC Exams

Explore More Capabilities

What are the eye manifestations associated with Leigh Syndrome?

Finished thinking

Leigh syndrome presents with a **wide range of ocular manifestations**, with strabismus being the most frequently observed finding, occurring in approximately 40% of patients. Other common ophthalmologic features include pigmentary retinopathy, optic atrophy, ptosis, and nystagmus.

Strabismus is the predominant finding, with exotropia more common than esotropia (mean exotropiation 29.6±12.5 prism diopters; mean esotropiation 24.0±8.9 prism diopters). Notably, all patients with esotropia in one series had disease onset before 1 year of age.

Pigmentary retinopathy occurs in approximately 22-23% of patients. In patients with the MT-ATP6 m.5993T>G mutation specifically, pigmentary retinopathy was reported in 12 of 43 individuals. Bull's-eye maculopathy has also been documented in infants with this mutation.

Optic atrophy is present in approximately 22% of patients. Histopathologic examination has demonstrated thinning of the nerve fiber and ganglion cell layers in the macula, along with mild atrophy of the optic nerve.

Ptosis occurs in approximately 16% of patients and can be a presenting sign, sometimes leading to midline ptosis as juvenile myasthenia gravis. Ptosis may be particularly associated with the m.1351G>A mutation in the MT-ND5 gene.

Nystagmus and abnormal eye movements are common, occurring in 14% of patients. Nystagmus or roving eye movements were the most frequent ophthalmologic presenting symptoms in one series (13/20 patients).

Additional findings include **ophthalmoplegia** (weakness or paralysis of extraocular muscles), refractive errors (particularly astigmatism), reduced visual acuity, and retinal vessel attenuation.

Ophthalmologic manifestations often appear or progress over time, making regular ophthalmologic follow-up essential for all children diagnosed with Leigh syndrome.

OpenEvidence is designed to support healthcare professionals by providing evidence-based answers to medical queries.

Can also create a patient hand-out in real-time (See next page)

- **Open Evidence AI (openevidence.com) continued/**

 Eye Problems in Leigh Syndrome: A Guide for Families



What is Leigh Syndrome?

Leigh syndrome is a rare disease that affects the brain and nervous system. It usually starts in babies or young children. The disease happens when the body's cells cannot make enough energy to work properly. This affects many parts of the body, especially the brain, muscles, and eyes.

How Does Leigh Syndrome Affect the Eyes?

Most children with Leigh syndrome (about 8 out of 10) will have some kind of eye problem. These problems can happen when the disease first starts, or they may develop over time. This is why regular eye doctor visits are very important.

Common Eye Problems in Leigh Syndrome

Crossed or Wandering Eyes (Strabismus)

This is the most common eye problem, happening in about 4 out of 10 children with Leigh syndrome. The eyes may turn inward, outward, or in different directions. This can make it hard to see clearly.

Eye Movement Problems

Some children have eyes that move back and forth on their own (called nystagmus) or have trouble moving their eyes in certain directions. About 1 or 2 out of 10 children have these problems.

Droopy Eyelids (Ptosis)

The upper eyelid may droop down and cover part of the eye. This happens in about 1 or 2 out of 10 children.

Problems with the Back of the Eye

- **Retina changes:** The retina is the part of the eye that helps you see. Some children develop

- **Chat GPT / Gemini / MS Pilot**
 - **Free and subscription options**
 - **Need to verify references / content**

Sample Prompts:

Initial Orientation

“What are the ophthalmic manifestations of Usher Syndrome that a **general pediatric ophthalmologist** should screen for?”

Exam-Focused

“Which eye findings in **1q21.1 deletion syndrome** would most likely present in a pediatric ophthalmology clinic?”

Variant Context (Non-interpretive)

“This child has a **pathogenic variant in COL2A1**. What ocular findings are associated with this gene?”

AI Key Points:

- “AI is best thought of as a **clinical assistant**, not a diagnostic tool.”
- “These tools help us **ask better questions** and perform more focused exams.”
- “AI can be especially helpful for **rare conditions we may see once or twice in a career.**”
- “We still rely on **genetic counselors and subspecialists** for interpretation and management.”
- “The goal is improved care and confidence — not replacing expertise.”



MS Pilot



Chat GPT



What are the ophthalmic manifestations of NF-2



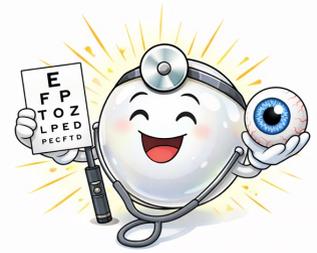
Neurofibromatosis type 2 (NF2), also known as NF2-related schwannomatosis, has distinct ophthalmic manifestations that are often the first clinical signs of the disease, appearing well before the hallmark hearing loss.

Unlike NF1, which is famous for **Lisch nodules** (iris hamartomas), NF2 primarily affects the **lens** and the **retina**.

Primary Ocular Findings

- **Juvenile Cataracts (60–80% of patients):** This is the most common ocular feature. These are typically: 
 - **Posterior Subcapsular (PSC):** Small opacities at the very back of the lens. 
 - **Cortical Wedge-Shaped:** Peripheral opacities that may only be visible when the eye is fully dilated. 
 - These often appear in childhood or early adulthood and are a key diagnostic criterion. 
- **Epiretinal Membranes (ERM):** Translucent, grayish membranes that grow on the surface of the retina. They occur in about **12–40%** of patients and can lead to distorted or blurred vision. 
- **Retinal Hamartomas:** Benign tumors of the retina or retinal pigment epithelium. Specifically, **Combined Hamartomas of the Retina and RPE (CHRRPE)** are classic for NF2.  

- 5. Everyday Cases: *GENETIC TESTING PITFALLS AND HOW TO AVOID*

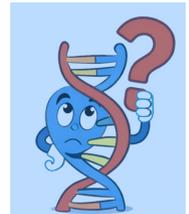


Example Cases:

- Case 1: early-onset retinitis pigmentosa - Dr. Arif Khan
- Case 2: a child with infantile-onset nystagmus – Dr. Emily McCourt
- Case 3: child with ocular albinism and fhx – Dr. Jen Rossen

Key Points and Panel Discussion led by Dr. Laryssa Huryn

1. Testing is complex — important considerations before, during, and after results
2. Psychosocial impact on families: anxiety, privacy concerns, guilt, implications for siblings
3. Test selection matters: inappropriate or incomplete testing may miss the diagnosis
4. Results can be challenging: VUS, unexpected findings, syndromic implications
-Variant of unknown significance: A VUS is a “we’re not sure yet” result — not a diagnosis – more work-up to be done
5. Genetic counseling is essential to ensure appropriate interpretation and follow-up



- If you have a patient who you think might qualify for a clinical trial, we are happy to help.
- Did you know that a Mystery Case of the Month is provided by the Eye Genetics Committee in the monthly eBlast? Great for docs in practice or trainees who want some extra cases in genetics...
- Need a specific resource? Suggest it to our committee!

aapos@aao.org

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Boston, Massachusetts

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and Strabismus