



Ophthalmologic Features in Homocystinuria



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Ophthalmic Case Scenarios-Case 1

Case 1: Known Case of Homocystinuria

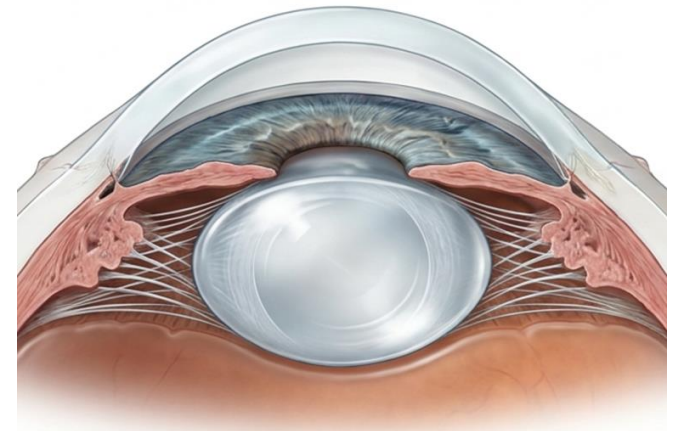
Patient Snapshot

SEX / AGE	5 years old, Boy
MEDICAL HISTORY	Known case of homocystinuria
CURRENT CORRECTION	Sphere -2.00, Cylinder -1.00
CHIEF COMPLAINT	Routine ophthalmic follow-up in a known case



Ocular Examination

Parameter	Right Eye	Left Eye
VA with current spectacles	Reasonably adequate	Reasonably adequate
Cycloplegic refraction	Close to current correction (-2.00 / -1.00)	Close to current correction (-2.00 / -1.00)
Slit lamp — lens	No obvious displacement, or very mild tilt	No obvious displacement, or very mild tilt
Intraocular pressure	Normal	Normal
Fundus	Normal	Normal





Ophthalmic Case Scenarios-Case 2

Case 2: High Astigmatism and Uncorrectable Vision

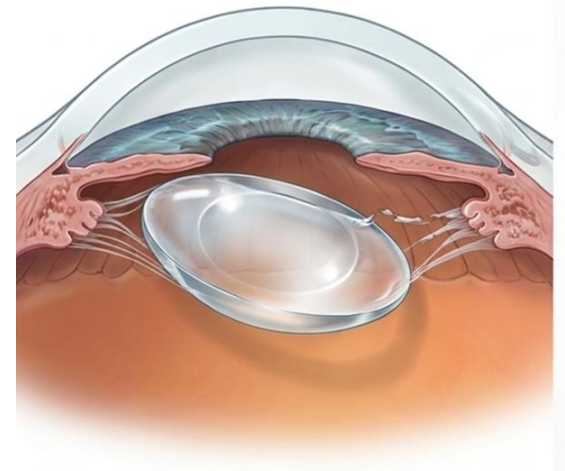
Patient Snapshot

CHIEF COMPLAINT: Established spectacle wearer with high astigmatism and vision not correctable to normal with glasses

MEDICAL HISTORY: May have a prior diagnosis of homocystinuria, or this visit may be the first time it is suspected

Ocular Examination

Parameter	Right Eye	Left Eye
VA with spectacles	Not correctable to normal	Not correctable to normal
Cycloplegic refraction	High, irregular astigmatism	High, irregular astigmatism
Slit lamp – lens	Ectopia lentis (clear lens displacement), Iridodonesis may be present	Assess for concurrent involvement, Iridodonesis may be present
Intraocular pressure	Normal	Normal
Fundus	Myopic change	Myopic change





Ophthalmic Case Scenarios-Case 3

Case 3: Sudden Vision Loss After Minor Trauma

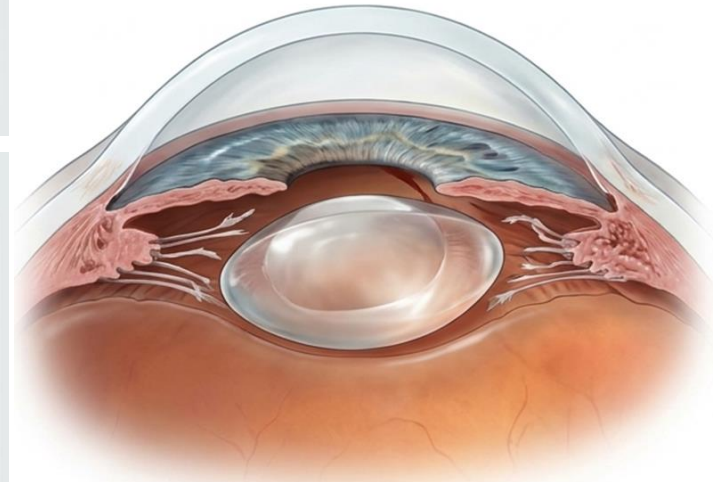
Patient Snapshot

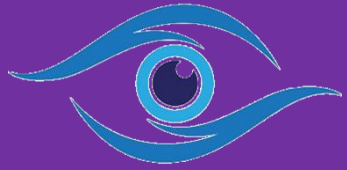
CHIEF COMPLAINT: Sudden vision loss following a minor trauma (e.g., a ball impact or a mild childhood play-related knock)

MEDICAL HISTORY: Diagnosed homocystinuria — either previously known or newly discovered during this visit

Ocular Examination

	Parameter	Affected Eye (Trauma)	Fellow Eye
1	Visual acuity	Sudden decline	May be normal or mildly myopic
2	Intraocular pressure	May be elevated if pupillary block occurs	Normal
3	Slit lamp — lens	Partial or complete dislocation following trauma	Careful exam for occult zonulolysis
4	Fundus	Assess for vitreous prolapse or hemorrhage	Assess underlying status



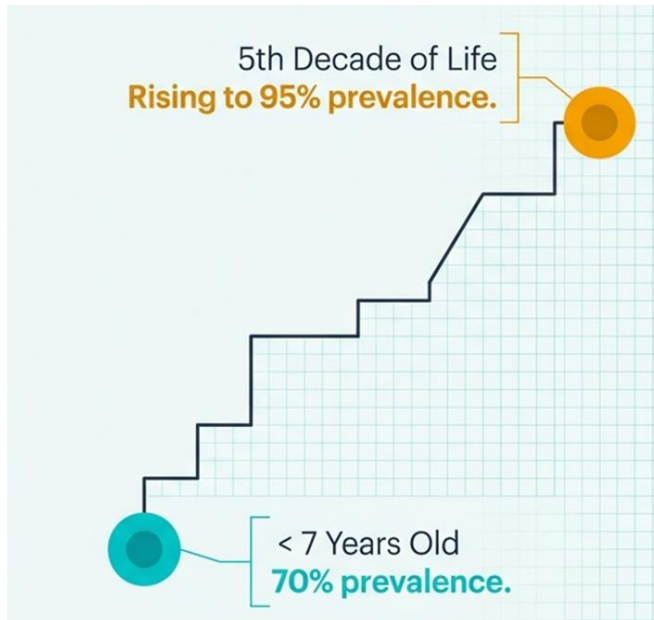


Ectopia lentis

May be the **first** and **only** sign of disease

Most common ophthalmic complication (**90%** of patients).

- ❑ In patients **<7 years** old = **70%**
- ❑ Rising to **95%** in the **fifth decade**.

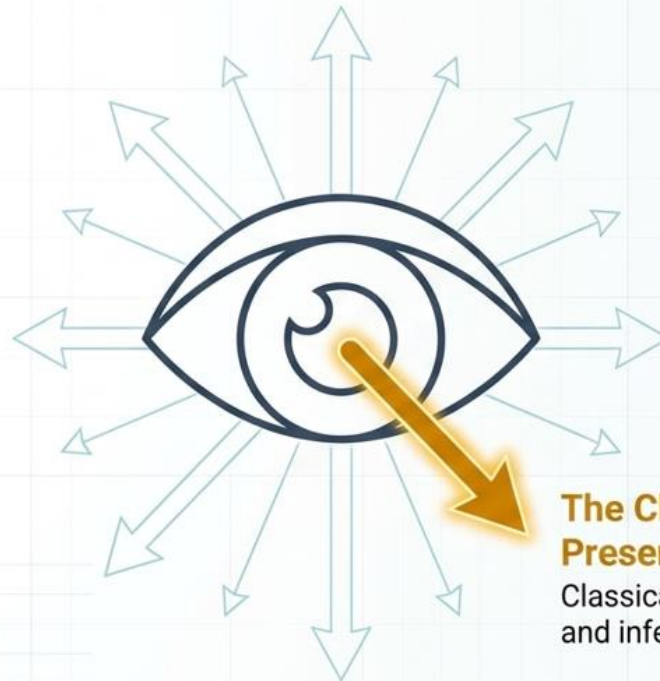




Ectopia lentis

Classically, lens dislocation is bilateral and inferonasal.

- ❑ Can happen in any direction.
- ❑ Direction of lens dislocation should not be considered pathognomonic for homocystinuria.



The Classic Presentation:
Classically bilateral and inferonasal

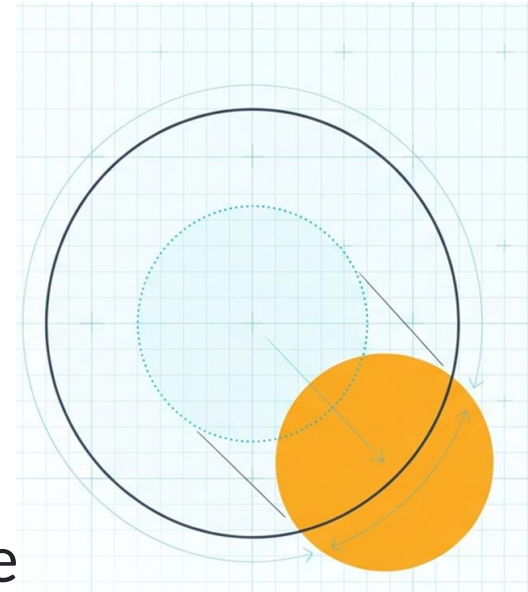


Ectopia lentis

Age related increased lens mobility and zonular disruption

Individuals with complete anterior lens dislocation are often older (10 years old).

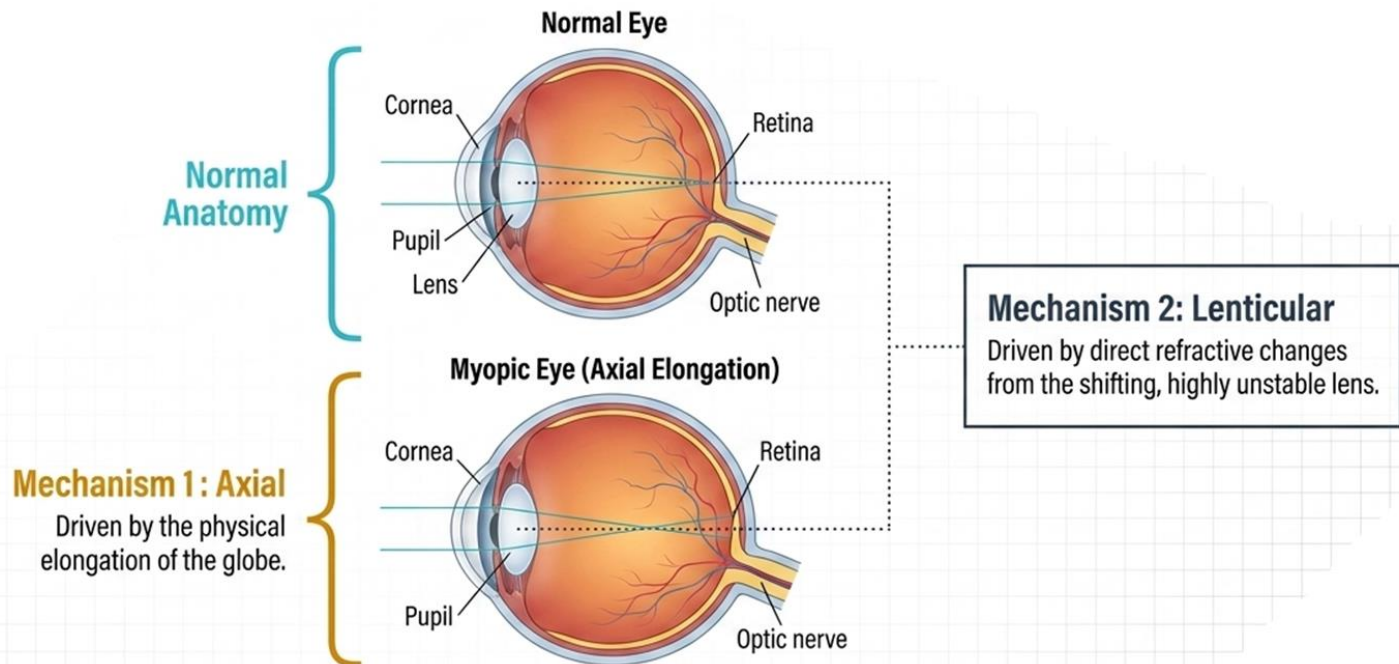
Lens dislocation may also be due to complete absence of zonules which can also lead to vitreous prolapse.





High myopia

Second-most common ocular manifestation
Both **axial** and **lenticular**

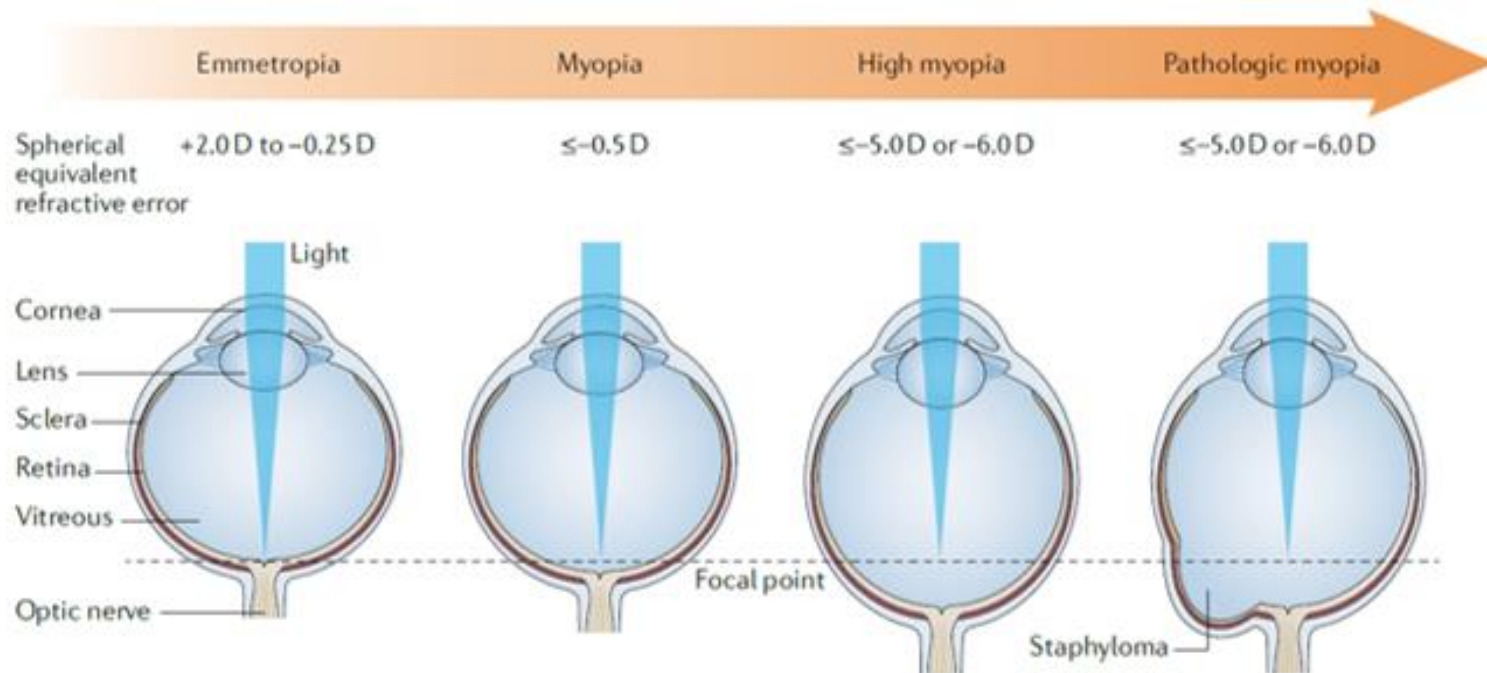




High myopia

The severity of myopia varies depending on:

- ❑ Age of diagnosis
- ❑ Level of control.





Timing

The Critical Intercept: Timing Dictates Visual Outcomes

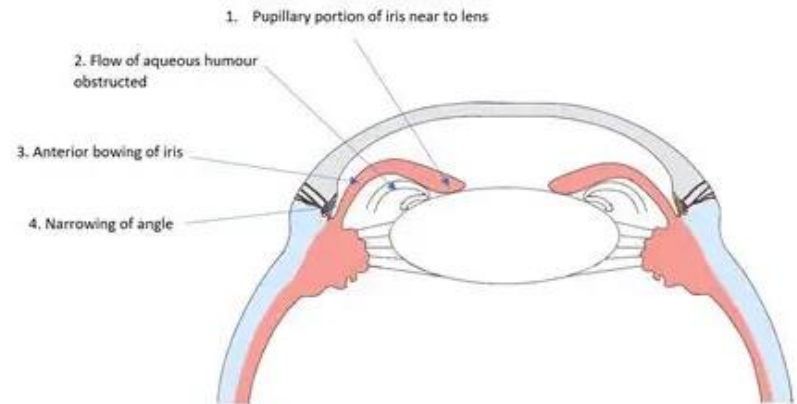
Patient Trajectory & Variables	Refractive Outcome	Visual Acuity
The Optimal Trajectory Diagnosed at birth + Treatment <6 weeks + Good control.	Emmetropic (or mild/moderate hyperopia/myopia).	100% achieve 20/40 or better (Maintained even if poorly controlled later in life)
The Progressive Trajectory Diagnosed <6 weeks + Failure to maintain good homocysteine control.	Usually highly myopic (>-5D) with progressively worsening myopia.	Progressive deterioration.
The Late Trajectory Diagnosed later in life (usually presenting first to an ophthalmologist).	Severe structural complications established.	<30% achieve visual acuity of 20/40 or better.



Pupillary block glaucoma

Pupillary block glaucoma

- ❑ Anterior dislocation
- ❑ Traumatize the corneal endothelium,
- ❑ Corneal edema
- ❑ Stromal opacification
- ❑ Bullous keratopathy.





Open angle glaucoma

Homocysteine can increase **arterial pressure** and can contribute to **increased ocular perfusion pressure** and **IOP**, contributing to the development of open-angle glaucoma.



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Homocysteine and Glaucoma

by Joshua Washington ¹ , Robert Ritch ²  and Yutao Liu ^{1,3,*} 

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Ocular complications

Retinal detachments

Non-arteritic anterior ischemic optic neuropathy (NAION),

- Local atherogenesis caused by **hyperhomocysteinemia**.

Cataracts

- Elevated exposure to homocysteine increases **PSC cataracts**

Strabismus

Amblyopia



Differential diagnosis

Several other diseases may present with ectopia lentis but will vary with other clinical features.

Ectopia Lentis et pupillae

- Ectopic pupil
- Flat-appearing iris
- Often presents with cataracts.

Marfan syndrome

- Marfanoid habitus,
- Significant cardiac concerns.
- Will not have intellectual disability or seizures.

Weill-Marchesani syndrome

- Microspherophakia
- Brachydactyly
- Joint stiffness



Systemic treatment

- Homocysteine concentrations should be kept
 - ❑ below **100** $\mu\text{mol/L}$ in **adults**,
 - ❑ below **50** $\mu\text{mol/L}$ in **children**

Halting lens subluxation and allowing standard bone growth to resume.



Guideline | [Open Access](#) |

Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency

[Andrew A. M. Morris](#) ✉, [Viktor Kožich](#), [Saikat Santra](#), [Generoso Andria](#), [Tawfeg I. M. Ben-Omran](#), [Anupam B. Chakrapani](#), [Ellen Crushell](#), [Mick J. Henderson](#), [Michel Hochuli](#) ... See all authors ▾

First published: 24 October 2016 | <https://doi.org/10.1007/s10545-016-9979-0> | [VIEW METRICS](#)

Communicated by: Avihu Boneh

This article makes recommendations for the diagnosis and management of CBS deficiency, based on a



Management

Ophthalmologic treatment

The Structural Problem

The Surgical Solution

Unstable / Dislocated Lens

Lensectomy
The most common surgical procedure.

Zonular Instability +
Associated Vitreous Prolapse

Vitrectomy

Resulting Aphakia

Implantable Intraocular Lenses (IOLs)

- Iris-fixated
- Scleral fixated
- Anterior chamber IOLs



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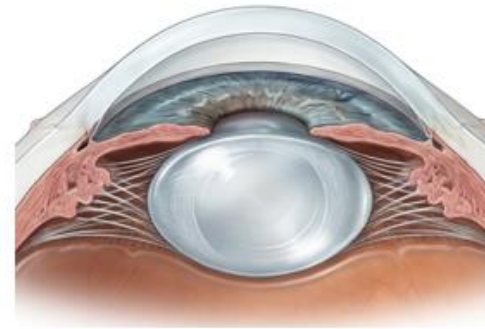
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KEY TEACHING POINT

At this age, the refractive error may still be mild and the lens may not yet show significant displacement. This case illustrates routine follow-up in an early disease stage – regular exams matter even without a dramatic finding, since zonular instability can progress over time.

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Case 2: High Astigmatism and Uncorrectable Vision

➤ Click

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KEY TEACHING POINT

High, irregular astigmatism that glasses cannot adequately correct should always raise suspicion for lens displacement rather than a purely corneal or axial refractive problem.

Differential Diagnosis

- ☒ Marfan syndrome
- ☒ Weill-Marchesani syndrome
- ☒ Isolated familial ectopia lentis

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KEY TEACHING POINT

The mismatch between a trivial trauma history and a severe ocular finding — complete lens dislocation — is itself a key diagnostic clue, since the zonules are already compromised by the underlying systemic disease.

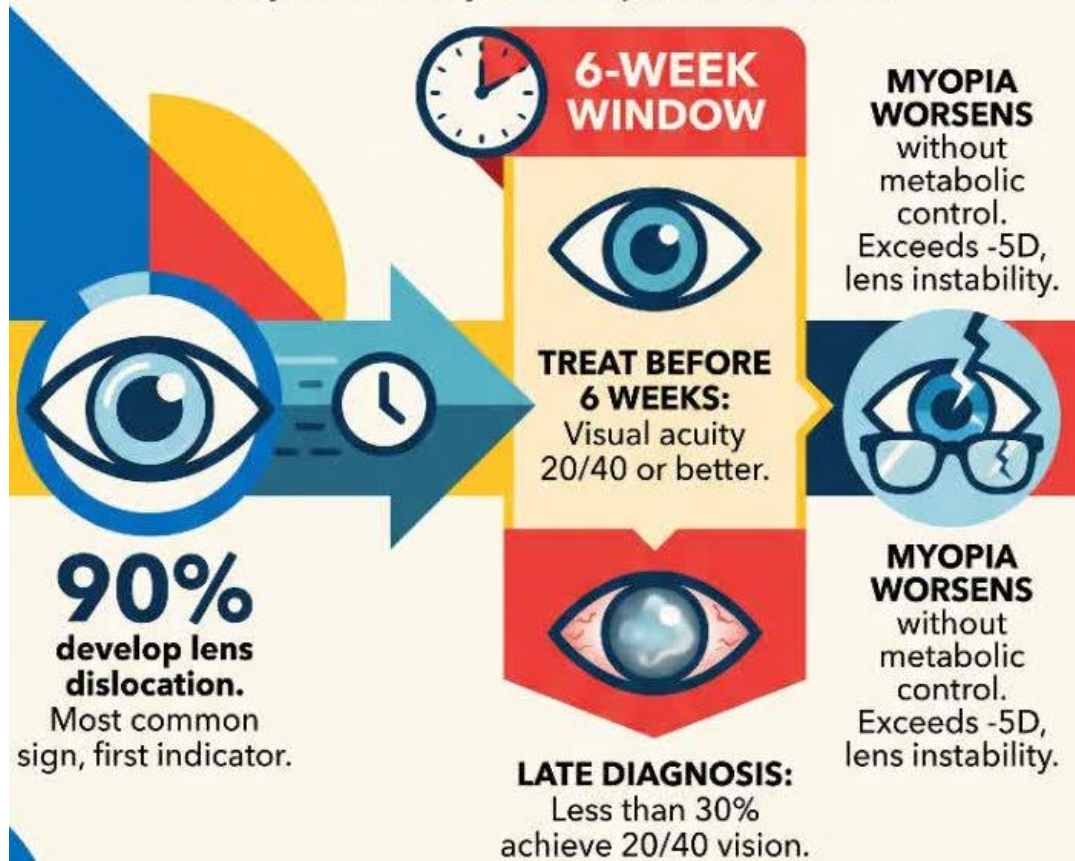
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Take-home message

Ectopia Lentis: The 6-Week Window

Nearly all homocystinuria patients affected.





Thank you for your attention