Translation from Romanian

CONGENITAL GLAUCOMA

DEFINITION

→ A form of glaucoma that results from the abnormal development of drainage structures of aqueous humor, characterized by increased intraocular pressure, enlargement of the eyeball (buphthalmos), corneal edema and optic nerve cupping and clinically presenting the characteristic triad: epiphora, photophobia and blepharospasm.
Synonyms: buphthalmos, primary congenital glaucoma

PREVALECE

→ It is the most common form of glaucoma in childhood. In Europe, the prevalence is estimated at about **1 / 27.800 live births.** Males **(65%)** are more frequently affected than females, and the disease is bilateral in **70-80% of cases.**

CLINICAL ASPECTS

→ The diagnosis is established in the first year of life in about **80%** of cases. The age of onset can be postponed until early adulthood.
→ The classic triad includes epiphora, blepharospasm and photophobia.
→ Affected children have red eyes, hyperlacrimation, blurred cornea, opacification and enlargement of the eyeball, caused by the elongation of the immature eyeball due to increased intraocular pressure.
→ Increased intraocular pressure can cause the areas to stretch and rupture with lens subluxation. Trauma can cause hyphema, retinal detachment and rupture of the eyeball.
→ Children older than 3 years develop progressive myopia and insidious visual field loss.
→ Untreated congenital glaucoma invariably leads to blindness.

GENETICAL ASPECTS

→ The etiology is insufficiently understood
→ The obstruction of aqueous humor flow appears to occur at the iridocorneal angle and trabeculae.
→ Gene mapping of affected families identified three chromosomal loci:

- **GLC3A in 2p22.2 →gene CYP1B1 (2p22.2)**
- **GLC3B in 1p36 and**
- **GLC3C in 14q24.3-q31.1**

→ Mutations have also been identified in the genes **LTBP2 (14q24.3) and MYOC (1q23-q24).**
→ ***Genetic counseling:*** Most cases are sporadic, in about 10% of cases autosomal recessive transmission is observed with variable penetrance. Cases with autosomal dominant transmission have also been described.

DIAGNOSIS

→ The diagnosis is established by a complete ophthalmologic examination which reveals: an opacified cornea, large size and the presence of **Haab striae**, increased intraocular pressure (IOP) (more than 20 mm Hg or asymmetry greater than 5 mm Hg is suggestive), deep anterior room, high iris insertion, poorly developed scleral spur (visible by gonioscopy),

increased cup / disc ratio of the optic nerve head, and refractometry revealing myopia and astigmatism.
→ Examination under anesthesia is done if necessary.
→ **Prenatal diagnosis** may determine the risk of the disease in families with known mutations.

TREATMENT
→ Congenital glaucoma is treated primarily surgically, with medical therapy playing only an adjuvant role.
→ Traditionally, **goniotomy** is recommended for children under 2 to 3 years old, if the cornea is clear.
→ **Trabeculotomy** is performed in children whose camber angle has trabecular abnormalities and older children.
→ **Trabeculotomy and drainage devices** are used in refractory cases.
→ Amblyopia, corneal scars and cataracts are late complications.
→ Early visual rehabilitation is important to prevent amblyopia.
→ Patients may need regular monitoring throughout their lives to monitor intraocular pressure.

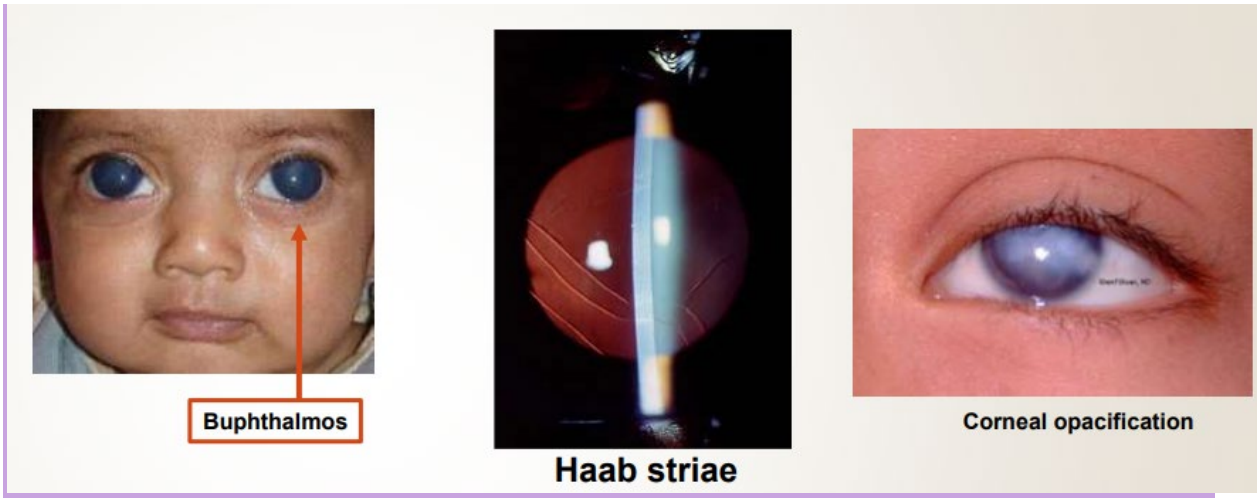
PROGNOSIS
→ The prognosis is largely related to the time of presentation; early diagnosis and prompt surgical treatment significantly influence the visual outcome.

→ Most patients successfully treated from the beginning maintain good control of intraocular pressure, with stable optic nerves and fully functional visual fields in adulthood.

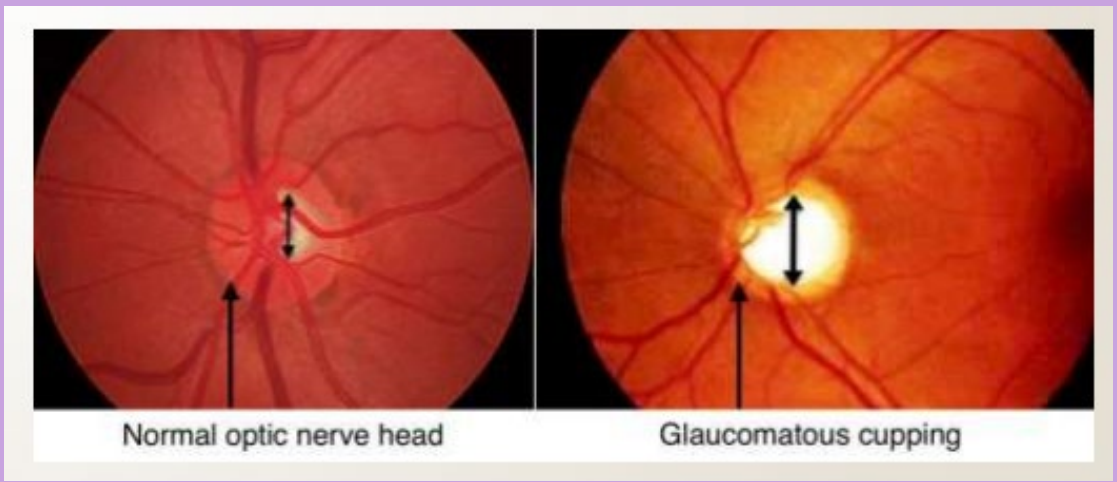
Img.1: Congenital glaucoma - ophthalmia + classic triad: epiphora, photophobia and blepharospasm

Source: https://www.glaucomapatients.org/basic/congenital-glaucoma/

Translation from Romanian



Img. 2: Clinical aspect of buphthalmos disease; Haab striae detectable on biomicroscopic examination; Corneal opacification



Img.3: Optic nerve cupping - can occur quickly, early in the congenital glaucoma

Source : https://www.slideshare.net/SaraMasoudinejad/congenital-glaucoma-71614408

I, BURA TEODORA - NATALIA, certified interpreter and translator for English, by virtue of the authorization no. 35530/20.03.2013, issued by the Ministry of Justice of Romania, hereby certify the accuracy of the translation from Romanian into English.

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