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Congenital Primary Glaucoma



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Synonym: trabeculodysgenesis

Introduction

There are various forms of congenital glaucoma. Some arise in the context of other abnormalities (eg, Reiger's syndrome, [Sturge-Weber syndrome](#) and [neurofibromatosis](#)). Childhood glaucoma can develop secondary to ocular problems such as tumours, trauma (accidental or surgical), inflammation or infection.

Early recognition of this condition is very important, as prompt treatment can significantly improve the child's vision.

If you want more information about later-onset glaucoma, see separate articles: [Glaucoma and Ocular Hypertension](#), [Primary Open Angle Glaucoma](#), and [Angle-Closure Glaucoma](#).

You may also find the following separate articles relevant: [Eye Problems in Babies](#), [Ophthalmia Neonatorum](#), [Vision Testing and Screening in Young Children](#), [Amblyopia](#), [Coloboma](#) and [Hereditary Retinal Dystrophies](#).

Epidemiology

- The most common type of infantile glaucoma is primary congenital glaucoma. It is very rare, of the order of 1:10,000 births.
- In 90% of cases, this is sporadic and in the remaining 10% of cases, there is an autosomal recessive pattern of inheritance.^[1]
- It may also be associated with abnormalities of the CYP1B1 gene (which encodes for cytochrome P4501B1)^[2] as well as others such as the LTBP2 gene.^[1]
- The problem lies in defective aqueous outflow from the anterior chamber due to malformation of the trabecular meshwork, the main drainage passage for aqueous. This results in a rise in intraocular pressure (IOP): in true congenital glaucoma (40% of cases), this occurs during intrauterine life and in infantile glaucoma (55% of cases), the rise is evident before the child's third birthday. The remainder of cases fall into the juvenile glaucoma group whose problem manifests itself after the third birthday but before the age of 16.
- Most cases (75%) are bilateral although involvement is usually asymmetrical.^[3]
- 65% of patients are boys.
- Its impact, if unrecognised, can be significant on the child's vision: it accounts for about 2-15% of patients being treated/cared for by institutions for the blind.^[3]

Presentation

- This depends on the age of onset of IOP rise but general trends are common in all children.
- Although it is present at birth, this condition may not be diagnosed until infancy or early childhood.
- The majority of cases are diagnosed in the first year.
- Epiphora, photophobia and blepharospasm are the three most common manifestations.

Symptoms

- Corneal changes including oedema which causes haziness. This may be the first abnormality noticed by the parents.
- Globe enlargement (if the IOP rises before the age of 2), known as buphthalmos. This is not generally noticed by parents unless it is unilateral or markedly asymmetrical, when they may report that their child has a 'large eye'.

Signs

The ophthalmologist will make a note of the following:

- Presence of buphthalmos.
- Corneal diameter: a horizontal diameter of >12 mm before the first year of life is highly suggestive.^[3]
- Abnormalities of one of the layers of the cornea (Descemet's membrane) which can split, so causing corneal oedema.
- A rise in IOP.
- Abnormalities on examination of the trabecular meshwork.
- Optic disc cupping.

- Refractive changes - particularly myopia (short-sightedness).

Differential diagnosis^[4]

- **Cloudy cornea at birth** - birth trauma, intrauterine rubella, some metabolic disorders (eg, mucopolysaccharidoses), congenital corneal dystrophies.
- **Large cornea** - very high myopia, megalocornea.
- **Epiphora** - delayed canalisation of the nasolacrimal duct (very common).
- **Raised IOP** - tumours (eg, retinoblastoma), retinopathy of prematurity, intraocular inflammation, trauma.

Investigations

This depends on the age of the child. The ophthalmologist will need to ascertain the visual acuity, and the IOP and have a good look at the trabecular meshwork as well as the fundus via dilated fundoscopy. Visual acuity can be determined to a certain extent when the child is awake but a full examination of the globe usually warrants a general anaesthetic.

Associated diseases

By definition, this condition occurs in an otherwise healthy eye with no associated systemic disease.

Management

- **Primary care** - any child presenting with symptoms or signs suggestive of glaucoma should be referred to an ophthalmologist.
- **Medical** - this is usually temporary and initiated on diagnosis whilst awaiting surgery. It may involve the use of topical beta-blockers ± systemic carbonic anhydrase inhibitors.
- **Surgical** - surgery is the definitive treatment and can involve one or more of the following:
 - *Goniotomy* - this is usually performed at the initial examination under anaesthetic once the diagnosis is confirmed. It involves opening Schlemm's canal (which drains the aqueous) via an incision into the trabecular meshwork. This procedure may need to be repeated more than once but it is generally successful.
 - *Trabeculotomy* - this is similar to the above procedure but uses a different technique; it is used when the cornea is too cloudy to allow for a goniotomy.
 - *Trabeculectomy* - this involves removal of part of the trabecular meshwork (or where it would be if it were correctly formed) in order to allow aqueous to drain into Schlemm's canal.
 - *Pediatric glaucoma surgery* - this is challenging because of the differences in anatomy from the adult, differences in the behaviour of the tissues of a child's glaucomatous eye, the variety in causes of the disease, and difficulties with postoperative management^[5]
- **Intractable glaucoma** - this may be treated with laser therapy or cryotherapy but this is very much a last resort.
- **Optical** - any refractive errors will also be addressed in order to minimise the possibility of amblyopia.

Complications^[3]

Undiagnosed glaucoma can lead to lifelong visual impairment. Diagnosis can be missed due to the rare and insidious nature of the problem. The most serious complications are unusual but tend to relate to having a general anaesthetic or to the surgery itself (hyphaema, infection, lens damage and uveitis).

Prognosis

- The corneal oedema may persist for several weeks but often resolves, as does the optic disc cupping (unlike adults).^[6]
- Apparently stable congenital glaucoma may progress with sight-threatening complications after many years of IOP stability. Monitoring of these patients is therefore usually indicated throughout life.^[7]
- About 50% of patients are left with a degree of visual loss which may be due to persistent oedema, persistent optic disc cupping, nystagmus, large refractive errors or amblyopia. An enlarged globe will also be more susceptible to damage in the future.

Further reading & references

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