Eye Problems in Babies

Eye problems in babies may be congenital (usually genetic or through intrauterine insult) or acquired after birth. Early detection and prompt treatment are essential, in order to avoid lifelong visual impairment. The eyes should be examined at the first neonatal check, when babies should be examined for structural abnormalities such as cataract, corneal opacity, ptosis and retinoblastoma, again at the six-week check, and at every well-child screening visit. Any child with an ocular abnormality should be referred to an ophthalmologist specialising in paediatric patients. See also separate Vision Testing and Screening in Young Children article.\[1\]

Failure to detect and appropriately manage disorders of vision in early childhood may have long-term consequences, not only for the development of normal vision but also for self-confidence and the realisation of potential.\[2\]

Congenital problems\[^{3}\]

Congenital problems may either be developmental, mainly secondary to genetic conditions, or to intrauterine effects of drugs, alcohol or maternal blood sugar.

Causes of congenital eye problems

Genetic disorders\[^{4}\]

Genetic disorders are a leading cause of visual impairment in children. They include anophthalmia, aniridia, albinism, anterior segment dysgenesis, Marfan's syndrome, ectopia lentis, neurofibromatosis, retinal haemangioblastomas and familial exudative vitreoretinopathy.

Infection\[^{5, 6}\]

The most characteristic ocular findings associated with congenital infection are chorioretinal scars and chorioretinitis. These are seen in the TORCH group of infections (Toxoplasmosis, 'Other' (syphilis, parvovirus B19), Rubella, Cytomegalovirus and Herpes). There may be extensive eye involvement. Congenital cataracts can also be associated with some congenital infections - eg, rubella, varicella-zoster virus, Epstein-Barr virus.

Alcohol\[^{7}\]

The eye is a primary target for fetal alcohol syndrome, with abnormalities including optic nerve hypoplasia, tortuosity of retinal vessels, coloboma and microphthalmia.

Drugs\[^{8}\]

- Cocaine is associated with vascular disruption in the newborn retina, with superficial and deep haemorrhages. This can resemble haemorrhages related to birth trauma. Severe eyelid oedema can also occur. Other opiates and benzodiazepines have been associated with nystagmus and reduced acuity.
- Some anticonvulsants are associated with a number of abnormalities, including early-onset myopia and strabismus.

Maternal diabetes\[^{8}\]

Optic nerve hypoplasia is the most common ocular malformation in babies born to mothers with diabetes. Tight control of fasting glucose levels reduces the risk.
Types of congenital eye problem

Defects of the globe

- Anophthalmos - complete failure of development of the optic vesicle.
- Congenital cystic eye - failure of development of the globe.
- Coloboma - failure of complete closure of the choroid fissure in utero that can affect the iris, retina or choroid. The hole is present from birth.
- Nanophthalmos - small eye with normal function.
- Microphthalmos - small eye without normal function (eg, cataract, coloboma, congenital cyst).

Defects of the lids

- Congenital ptosis - this is usually due to defective muscles of the upper lid but may also be due to Horner's syndrome and 3rd nerve palsy.
- Eyelid coloboma - this is a full-thickness defect of the eyelid, usually at the junction of the medial and middle third of the upper lid. It is commonly associated with Treacher-Collins' syndrome.
- Cryptophthalmos is a rare congenital anomaly in which the skin is continuous over the eyes, with the absence of eyelids. It is usually associated with other congenital anomalies and the globe is often abnormal. Some syndromes, including Fraser syndrome, are associated with the condition.

Defects of the cornea

- Corneal opacity can be partial or complete and caused by:
  - Congenital glaucoma (most common with an abnormally large eye).
  - Damage from forceps.
  - Endothelial development abnormalities.
  - Persistent attachment of lens.
  - Intrauterine inflammation.
  - Interstitial keratitis.
  - Megalocornea - an X-linked inherited defect associated with an abnormally large but clear cornea.

Defects of the iris and pupil

- Corectopia - this is inappropriately positioned pupils. The condition is relatively common. The pupils are usually positioned upwards and outwards.
- Polycoria - two or more pupils may exist in one iris.
- Coloboma of iris - this is usually seen in the lower part of the iris towards the nose but defects may affect other parts of the eye.
- Aniridia - this is a rare genetic defect with an absent iris, often with secondary glaucoma (in the absence of a family history, it may be associated with Wilms' tumour).
- Albinism - patients may also have poor eyesight and nystagmus.
- Heterochromia - irises of different colours may be associated with normal function or may occur with congenital Horner's syndrome.

Defects of the lens: congenital cataracts

- Congenital cataracts are present at birth but they are not always detected at birth.
- Some (but not all) are progressive.
- Congenital cataracts may be secondary to intrauterine infection (eg, rubella) or be an inherited abnormality.
- Not all congenital cataracts are significant. Small opacities may not cause visual problems but large opacities may cause nystagmus and amblyopia requiring surgery within a few weeks of birth.
- They are generally considered visually significant and requiring removal in the following:
  - The cataract is in the visual axis.
  - Anisometropia - more important than the cataract size is the anisometropia (inequality in refractive power between the eyes) induced by the congenital anterior lens opacities.
- There is a high risk of glaucoma associated with surgery in the first year of life.⁹
- Glaucoma and vitreous haemorrhage show increased incidence in babies with a family history of aphakic glaucoma, or who had nuclear cataract or persistent fetal vasculature syndrome. Patients should be monitored closely postoperatively for the development of these conditions.¹⁰

Other lens and anterior segment defects

- Colobornata (see above)
- Subluxation (eg, in Marfan's syndrome).
- Rarely, incorrect development of the neural crest can cause a number of syndromes which affect the anterior segment. An example is Axenfeld-Rieger's anomaly, which consists of small eyes (microphthalmia), hypoplastic irises, polycoria (iris tears leading to the formation of more than one pupil in the iris) and abnormal patterning of the chamber angle between the cornea and the iris. Glaucoma is often an important complication.¹¹

Vitreous defects

- The remains of the hyaloid artery may appear on the optic disc (Bergmeister's papilla) or of the lens (Mittendorf's dots).
• White pupil (leukocoria) can be caused by:
  • Persistent hyperplastic primary vitreous.
  • Stage V retinopathy of prematurity (retrolental hyperplasia).
  • Severe posterior uveitis/vitritis.

Defects of choroid and retina

• These may be associated with colobomata. A number of rare syndromes can cause them - eg, Aicardi’s syndrome (severe psychomotor impairment, corpus callosum agenesis, chorioretinal lacunae and early-onset infantile spasms).\[^{[12]}\]
  • Scarring can result from congenital toxoplasmosis.

Benign abnormalities are common and include:

• Minor defects of retinal vessels at nerve head.
• Tilted disc from unusual angle of nerve entry.

More severe defects include:

• Central coloboma of the disc - this is also called ‘morning glory syndrome’. The optic nerve head is funnel-shaped with a white dot in the centre, an elevated ring of pigment around the disc and vessels radiating out from the ring like spokes. It thus resembles the morning glory flower.\[^{[13]}\]
• Optic nerve hypoplasia - this may be unilateral or bilateral and is a non-progressive condition. It is now realised to be relatively common with many cases causing only minor visual impairment which may only become apparent later in life. However, in severe cases it can produce a range of visual defects including severe sight impairment. It can be difficult to diagnose and is often associated with congenital defects of the brain and facies. See separate Septo-optic Dysplasia article.

Extra-ocular defects

• Dermoids - most frequently seen supero-laterally.
• Obstruction of nasolacrimal duct - this causes epiphora in up to 30% of neonates. It is thought to be caused by colonisation with bacteria. Most cases resolve spontaneously, although persistent symptoms beyond the age of 12 months may require probing. Associated infection - eg, conjunctivitis and dacryocystitis - may require antibiotics. Dacrocystitis can lead rapidly to generalised sepsis and an aggressive approach with intravenous antibiotics and surgical drainage is sometimes required.\[^{[14, 15]}\]
• Craniofacial anomalies - a number of these can affect vision (eg, craniosynostosis with downward-slanting palpebral fissures).\[^{[16]}\]

Poor vision with no apparent cause

The main causes include:

• Leber’s congenital amaurosis (retinal dystrophy) - see separate Hereditary Retinal Dystrophies article.
• Cone dystrophy.
• Oculomotor apraxia (a difficulty in controlling horizontal eye movement).
• Delayed visual maturation - defined as absence of visual response in a child aged under 3 months, due to gestational immaturity.

Congenital glaucoma

This is often bilateral and associated with other defects. It is the most common non-syndromic glaucoma in infancy; however, it is still rare with a frequency of around 1 in 10,000 children. Early diagnosis is necessary to avoid irreversible severe sight impairment. However, contrary (perhaps) to expectation, it often does not present at birth, although 80% present in the first year of life, typically at 3-6 months.

Symptoms include:

• Tearing
• Photophobia
• Blepharospasm
• Eye rubbing
• Irritability

Signs include:

• Large eyes (buphthalmos)
• Photophobia
• Corneal haze
• Corneal opacity
• Corneal enlargement
• Cupper optic disc

See separate Congenital Primary Glaucoma article.
Acquired problems

Ophthalmia neonatorum

- This refers to any conjunctivitis occurring in the first 28 days of life.
- It is most commonly infective in origin: *Neisseria gonorrhoeae, Chlamydia trachomatis*, bacteria such as staphylococci, streptococci and viruses - notably the herpes simplex virus - but it may also occur as a reaction to chemical irritants. Chlamydial and gonococcal infection can be life-threatening.
- See separate Ophthalmia Neonatorum article.

Retinopathy of prematurity (retrolental fibroplasia)\(^{[14]}\)

- This occurs when there is disruption of the vascularity of the retina.
- 80% of affected babies are less than 1 kg in weight and the condition is associated with prolonged administration of oxygen. Abnormal vessels develop in areas where vascular and avascular tissue meets.
- The condition sometimes resolves spontaneously but may require laser therapy or surgery.
- See separate Retinopathy of Prematurity article.

Strabismus

- Also known as squint, this occurs in fewer than 2% of babies.
- If it persists beyond 3 months of age, referral is indicated to enable normal visual development in the squint. It can be an early presenting feature of retinoblastoma.\(^{[18]}\)
- See separate Strabismus (Squints) article.

Amblyopia

- This is defined as reduced visual function in one or both eyes, not improved by refraction or removal of pathological obstruction to vision.
- Amblyopia is caused by sensory deprivation during the sensitive or critical period of retinal development in the first 2-3 years of life. The longer the period of visual disability, the worse the prognosis in terms of visual acuity.
The most common causes are:
- Strabismus - secondary to suppression of images from the affected eye to avoid double vision.
- Anisometropia - significant difference in refractive power between the two eyes.

See separate Amblyopia article.

Non-accidental head injury (NAHI), or shaken baby syndrome[19, 20, 21, 22]

- The term 'shaken baby syndrome', which consists of a triad of symptoms, has become controversial, both medically and legally.
- In 2011 the Crown Prosecution Service recommended the term be replaced with NAHI. The American Academy of Paediatrics adopted the term 'paediatric abusive head trauma'.
- The condition is often fatal. Where it is not, there is significant risk of permanent impairment to sight, motor function and cognition.
- The original definition of the triad, then entitled 'whiplash shaken infant syndrome', was made in 1973. It drew evidence from research on brain injury in monkeys but commented that the evidence was 'manifestly incomplete and largely circumstantial'.[23, 24]
- The 'triad' of symptoms consists of bilateral subdural haemorrhages, retinal haemorrhage and cerebral swelling/diffuse traumatic axonal injury.
- Some forms of retinal injury are highly characteristic, particularly retinal folds and traumatic retinoschisis. It has been suggested that vitreoretinal traction is the cause of these findings, which tend to correlate well with the intracranial findings.[25]
- For many years consensus of medical opinion was that the 'triad' was pathognomonic of 'forceful' shaking. However, in 2005 the UK Court of Appeals found that the triad was suggestive of NAHI but not 100% diagnostic in the absence of clinical history.
- There has also been uncertainty regarding the level of force needed to produce the damage seen in NAHI. A biomechanical analysis in 2005 concluded that forceful shaking would injure or kill a baby through cervical spine damage rather than through rotational injury but there was criticism of the study's methodology.[26]
- A review in 2009 concluded that child abuse should be suspected in children with retinal haemorrhages and a parental explanation of accidental head injury, particularly if the retinal haemorrhages are bilateral, flame-shaped, or extend through all layers of the retina.[19]
- Conditions supporting a diagnosis of NAHI include retinal bleeding, bone fractures and cervical spine injury.
- Conditions which must be ruled out include hydrocephalus, sudden infant death syndrome, seizure, infectious disease (eg, meningitis), blood dyscrasias and metabolic disorders.

In summary, retinal haemorrhage is a principal finding in inflicted head trauma, with certain ocular injuries showing a particular association. However, inflicted trauma is not the only cause of retinal haemorrhage and, in the absence of supportive clinical history, it is important to consider other diagnoses.[27]

Further reading & references

- Embryology, 3rd Ed, 2005


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