Contents:

b. The infant with whitish pupil.
c. The infant with watery eye.
d. Prematurity and ROP.

OBJECTIVES

- To be aware of specific important disorders related to infancy and early childhood about which the pediatrician can help spread awareness.

- To recognize when to refer the child to the ophthalmologist and what to expect from the pediatric ophthalmologist in the specified situation.
- Visual Development and assessment of vision in infancy and childhood

The human eye undergoes extensive growth in infancy and early childhood. At birth the diameter of the globe is 66% of that of the adult eye. The axial length of the infant's eye is less than that of the adult. From birth to age two years, rapid increase in axial length occurs (6mm). Growth slows down from age of 2-13 years (approximately 2mm). The refractive state of the eye changes as the axial length increases and the cornea and the lens flatten. Eyes are usually hyperopic at birth. This hyperopia corrects as the eye reaches adult dimensions. Pigmentation of the iris increases during the first years of life so that the color of the iris deepens. At birth the fovea is not yet fully developed, however it continues to mature until the age of 3 years.

A newborn baby perceives light and closes the eye whenever there is a bright light. Eye movements are uncoordinated at first and transient deviation of the eyes may be present, this should stabilize by the 4th to 6th month when ocular coordination as well as good fixation and following should be achieved.

**Visual Milestones:**

- 6 – 8 weeks: can make an eye contact and maintain it.
- 3 months: can fix and follow a slow target and converge.
- 6 months: reaches out for objects accurately.
- 2 years: picture matching.
- 5 years: Snellens chart matching and naming.
Assessment of visual development in infants and children:

- **Newborn**: evaluation of the red reflex, pupillary testing.
- **Infants below 1 year**: fixation and following. Fixation preference is checked by observing the child’s response to covering one eye compared to covering the other.
- **6 months to 2 years**: preferential looking (Fig 1).
- **2 to 4 years**: picture tests.
- **4 to 5 years onwards**: adult acuity tests as Snellens chart.

Fig1: Preferential looking; The infant prefers to look at a pattern
As discussed earlier (chapter 5), infants and children can present with a wide array of refractive errors and these can be associated with strabismus and possibly amblyopia. Parents sometimes notice that their infant squints or sits too close to the television screen, but often the errors of refraction are accidentally discovered during routine examination.

Two other problems that parents may notice are: 1) if their infant has a white pupil and 2) if their infant has a watery eye.

- **The Infant with the white pupil (leukokoria) (Fig 3)**

Upon examination of the pupillary area or trying to elicit a red reflex, an abnormal white or yellowish white reflex is noticed. Generally the presence of leukokoria requires prompt referral to an ophthalmologist because the causes are either life or permanent vision threatening. This chapter will discuss the most important causes of leukokoria.
Causes of leukocoria (white pupil)

1. Cataract
2. Retinoblastoma
3. Tractional Retinal detachment due to Retinopathy of prematurity
4. Coats’ disease
5. Endophthalmitis

1-Congenital Cataract

These are opacities in the lens discovered at birth or in the early years of life.

Etiology:

- Idiopathic
- Hereditary.( autosomal dominant)
- Intrauterine infections especially Rubella.
- Metabolic e.g. galactosemia.
- Chromosomal abnormality eg Down’s syndrome
- Traumatic

Cataract Morphology:

- Complete, involving the whole lens
- Incomplete, involving part of the lens

The location and density of lens opacity affect the visual prognosis.

- Anterior polar cataract, usually small (less than 3 mm) involving center of the anterior capsule. It is usually insignificant.
- Nuclear cataract, density and size are variable. It involves the center of the lens. (the nucleus)
- Lamellar cataract, typically large involving layers of the cortex around the nucleus. Patients usually have good prognosis.
- Posterior polar / posterior subcapsular cataract, involving posterior cortex or capsule. It is usually visually significant.

How to differentiate between Cataract and retinoblastoma?
The opacity lies in the lens NOT in the posterior segment

The earlier the onset, the more amblyogenic the cataract will be. All newborns should have a screening eye examination including red reflex evaluation to detect visual axis opacities.
Clinical picture:

Symptoms: Usually given by the mother who may notice:

- A white pupil (leukocoria).
- Defective vision of her baby.
- Roving movement of the baby’s eyes.

Signs:

- Using the retinoscope to assess the red reflex: lens opacity can be detected. Presence of a central opacity more than 3 mm or more is visually significant.
- Presence of nystagmus in bilateral cataract cases indicates that lens opacity is significant.
- Fundus should be carefully examined to exclude retinal anomalies.
- Ultrasonography is done if the fundus is not seen.
- **Complications:**
  Bilateral dense opacities may lead to:
  - Fixation nystagmus

  Unilateral opacity may lead to:
  - Amblyopia.
  - Squint.

**Treatment:**

The treatment of cataract is surgical removal. However, the decision whether to remove a congenital cataract immediately or to observe it conservatively depends on:

1) Clinical judgment to decide if the cataract is visually significant and expected to interfere with the visual development. For example, small anterior polar cataract and snowflake congenital cataract are not known to significantly affect vision. It is better to avoid surgery in order not to deprive the child from accommodation which is in itself amblyogenic.

2) Whether the cataract is unilateral or bilateral. A visually significant unilateral cataract must be operated promptly, even in the first weeks of life, in order to try to avoid deep amblyopia.

3) Age at presentation

   Surgical options include:
   - Timing: the younger the child, the urgency to remove the cataract because of the risk of amblyopia. Cataract should be removed between age of 6 weeks to 2 months.
   - Irrigation-aspiration of the soft lens matter
   - Pars plana lensectomy plus anterior vitrectomy

   **Correction of Aphakia** is done by IOL implantation either in the primary surgery (in unilateral cataract) or as a secondary procedure (in bilateral cases operated early in life). Other options include contact lenses or spectacle correction in appropriate cases.

Prevention and Treatment of amblyopia: see strabismus.
2-Retinoblastoma:

It is the commonest primary malignant intraocular neoplasm in children.

Retinoblastoma occurs in about 1 in 20,000 live births. The average age of diagnosis is 18 months but it may be present at birth or as old as 8 years.

10% Familial 90% Sporadic
30% Bilateral

Clinical features:

Retinoblastoma may present as:

1. Leukocoria: white pupillary reflex, accounts for 65% of cases.
2. Strabismus (squint): if the tumor affects the macular area.
4. Proptosis due to extraocular extension, with occasional red and painful eye.
5. Accidental discovery on routine examination or screening of babies for families with a positive history of retinoblastoma.

Genetic Basis of Retinoblastoma:

Short Arm of Chromosome 13 \( \rightarrow \) Anti-Oncogene (Prevents tumor formation)
Mutation in Chromosome 13 \( \rightarrow \) Loss of Anti-Oncogene (Tumor Develops)

Retinoblastoma is the most serious cause of Leukocoria, in addition to being a tumor that can result in visual loss, if untreated it can lead to infant and child mortality.
6. Spread of the tumor is mainly via the optic nerve and may reach the optic chiasma and subarachnoid space causing malignant deposits in the brain and spinal cord.

![Fig 8: Fundus image of retinoblastoma](image)

**Diagnosis**: is based on:

1. Ophthalmoscopy: with maximal pupillary dilatation, preferably under general anesthesia. It is of extreme importance to examine both eyes due to the relatively high incidence of bilaterality. Fundus examination will reveal a whitish tumor often with prominent vascularity.
2. X-ray may show tumor calcification.
3. Ultrasonography.
4. CT scans

**Treatment:**

1. Enucleation with excision of a long stump of the optic nerve is the treatment of choice for large tumors affecting one eye. The aim is to preserve the child’s life.
2. The second eye must be examined carefully at regular intervals for early detection of tumors in the other eye. In this case, small to medium size tumors can be destroyed while preserving the globe. This may be achieved by laser photocoagulation, transscleral cryotherapy or radiotherapy (according to the tumor size and location in the retina).

Fig 9: Retinoblastoma in an enucleated globe

Fig 10: Retinoblastoma before treatment (Left) and after treatment (Right)

3. Tractional Retinal detachment due to Retinopathy of prematurity (ROP): Retinopathy of prematurity is a disease that affects the retina of infants who are born prematurely or with low birth weight. If progressive and untreated it will lead to tractional retinal detachment that will result in leukokoria by examination. It will be discussed in more detail later.

How to differentiate between ROP and retinoblastoma?

ROP is Tractional RD without a retinal mass,
Retinoblastoma shows a retinal mass with exudative RD

4. Coat's Disease
This is an idiopathic retinal vascular disorder characterized by telangiectasias and
lipid exudation.

Fundus examination will demonstrate exudative retinal detachment with retinal vessel tortuosity and telangiectasia most prominent in the periphery. These may involve the posterior pole and result in visual loss.

5. Endophthalmitis:

Endophthalmitis in an infant presents with a red and painful eye with occasional leukokoria and possibly proptosis. For this reason it is important to remember that Retinoblastoma can also present with a red eye, proptosis and leukokoria. The differentiation between both conditions requires good history taking, establishing if a cause of endophthalmitis is present and a high index of suspicion.

Any Infant presenting with Acute painful proptosis should be referred to an ophthalmologist promptly.
The Infant with the Watery Eye

When approaching the infant with watery eye, we need to remember that any discharge from the eye of a newborn infant is suspicious since tears are not secreted in the first week/ few weeks of life.

The most important causes of a watery eye in an infant or newborn include:

1. Neonatal conjunctivitis and ophthalmia neonatorum
2. Congenital nasolacrimal duct obstruction
3. Congenital glaucoma

1. **Neonatal conjunctivitis and ophthalmia neonatorum**: Ophthalmia neonatorum is any form of conjunctivitis occurring in the first month of life.
   
   **Etiology**:

   1. **Infectious**: ocular contact with contaminated maternal passages or towels.
      - Bacterial: gonococci, staphylococci, streptococci or pneumococci.
      - Viral: herpes simplex and adenoviruses.
      - Chlamydia.
   2. **Chemical**: prolonged postpartum instillation of drops or ointment for prophylaxis against infection.
Diagnosis:

A newly born will not complain but parents may notice:

- Watery eyes
- Sticky discharge that maybe yellowish-whitish or greenish

We depend mainly on the clinical picture. In exceptional cases a conjunctival smear may be taken and sent to the lab for analysis of the causative organism.

Management:

Prevention:

1. Treatment of the mother before labor especially herpetic cervicitis.
2. Washing of the body of the baby from above downward.
3. Penicillin or broad spectrum antibiotic eye drops are instilled in the eyes after birth for 1 week. Prolonged use of topical antibiotics should be avoided as it could result in chemical conjunctivitis.

Treatment: Once suspected, treatment should start immediately:

1. Topical broad spectrum antibiotic eye drops used frequently .
2. Frequent removal of discharge.
3. Antibiotic ointment when the infant is sleeping.
4. Systemic antibiotics in severe cases.
2. Congenital nasolacrimal duct obstruction:

Watery eyes in an infant can occur as a result of obstruction of the nasolacrimal duct which may be due to congenital stenosis, incomplete canalization or an imperforate Hasner's valve (at the end of the nasolacrimal duct).

**Diagnosis:**

Parents may notice:

- Watery eyes
- Sticky discharge
- Rarely, swelling in the region of the nasolacrimal sac if a mucocele is formed which may become a pyocele if it becomes infected.

![Fig 14: Right Mucocele](image)

**Treatment:**

In Congenital nasolacrimal duct obstruction

1. **Conservative treatment** (till the age of 12 months) awaiting spontaneous resolution (in 90% cases). Topical antibiotics are given to wash the discharge and guard against secondary infection and discharge.
2. **Massage**: some physicians ask the parent to apply the thumb or index finger with pressure over the sac area downwards towards the ala of the nose few times daily. It is believed that this may push the secretion towards the lower nasolacrimal duct and may help open a membranous occlusion.

3. **Probing**: this opens the membranous obstruction usually an imperforate Hasner’s valve. The timing of intervention should be around 12 to 15 months. It should not be done before this age in order to give the chance for spontaneous resolution. After 18 months the success rate for probing becomes much less.

4. **Nasolacrimal duct Intubation** using silicone tube if probing fails.

5. **Balloon dilatation**: may be tried.

6. **Dacryocystorhinostomy (DCR)** Persistent symptoms after initial probing can be treated by intubation, balloon catheter dilatation, dacryocystorhinostomy.

### 3. Congenital Glaucoma:

Glaucoma in the first year of life is not uncommon and is usually **primary** due to a developmental defect in the angle of the AC interfering with aqueous drainage. Less commonly, it may be **secondary** to ocular diseases as retinoblastoma, retinopathy of prematurity or neonatal iridocyclitis.

If glaucoma develops in the first 2 years of life before the maturity of corneal and scleral collagen, the external ocular coat expands with the high IOP and the globe enlarges. The normal corneal diameter is 10 mm at birth and about 11.5 mm in the adult whereas in congenital glaucoma, it can reach 13-14 mm.
PRIMARY CONGENITAL GLAUCOMA (BUPHTHALMOS)

Pathogenesis:

A congenital maldevelopment of the angle formerly thought to be with a thin membrane (Barkan’s membrane) overlying the angle structures thus preventing aqueous outflow. The disease may be unilateral or bilateral, commonly autosomal recessive and the earlier the onset in the first year of life the more severe the disease.

Diagnosis:

Early:

Watery eyes (usually not before 2 weeks)
Photophobia (the infant can't tolerate light)
Frequent rubbing of the eyes.

Later stage:

The cornea is noticed to have a large size

In advanced stages:

The cornea turns opaque due to stretch and rupture of Descemet’s membrane resulting in corneal edema.

Corneal haze is caused by elevated intraocular pressure which leads to corneal edema. Breaks in Descemet’s membrane occur with the edema. Corneal enlargement occurs with gradual stretching of the cornea.
Examination is done under general anesthesia and should include measurement of corneal diameter using calipers and measurement of the IOP using handheld applanation tonometer. Normal IOP in infants and young children is 10-12 mmHg.

Ultrasonography is helpful in diagnosis by demonstrating a large axial length of the globe (at birth normally 16mm) provided congenital axial myopia is excluded.
**Treatment: Is essentially surgical:**

Topical antiglaucoma medications as beta blockers and carbonic anhydrase inhibitors can be used to lower IOP before surgery.

**What is the status of the cornea?**

- **Clear**
  - Goniotomy

- **Hazy**
  - Trabeculotomy
    - Trabeculectomy (Late cases/Recurrent cases)

**Early cases** with clear cornea (usually corneal diameter less than 13 mm) are treated by **goniotomy**, a technique in which the membrane overlying the angle structures is incised to allow aqueous outflow.

If the cornea is hazy due to edema, **trabeculotomy** is done ab externo.
Advanced and recurrent cases with large edematous corneas are treated with external fistula as in POAG.

Retinopathy of Prematurity (ROP)

This is a disease that affects infants born prematurely with low birth weight. When the infant is born prematurely, the retina is still not yet fully vascularized. In addition, in utero, the fetus is in a relative hypoxic state in contrast to the situation after birth when the infant is exposed to ambient oxygen. Oxygen is known to have a vasoconstrictor effect. When the immature retina is exposed to ongoing hyperoxia, as in incubators, the vessels will stop growing. Over time, the avascular retina becomes ischemic. This will stimulate VEGF which leads to arterial venous shunts and neovascularization with subsequent scarring and tractional retinal detachment.

Risk factors for developing ROP:

• Low birthweight (less than 1500 grams)
• Gestational age (32 weeks or less)

ROP is a cause of PREVENTABLE blindness, prevention can only be achieved through proper SCREENING. It is the responsibility of the neonatologist and pediatrician to ask for fundus examination for babies in the neonatal ICU and educate the parents about the importance of follow up with an ophthalmologist after discharge.
Screening for Detection of ROP:
Screenings of infants at risk with appropriate timing of exams and follow up is essential in identifying infants in need of treatment. As a rule all premature babies weighing 1500 gm or less OR born at less than 32 weeks of gestation should be screened at 31 weeks or 4 weeks after delivery whichever is later. Older or heavier infants can be examined if other risk factors exist such as respiratory distress syndrome, intraventricular hemorrhage, sepsis and history of high oxygen level in the ICU incubator.

Practical Example:
An infant born at 30 weeks will be examined at age of 34 weeks (4 weeks after birth)
An Infant born at 26 weeks will be examined at age of 31 weeks (5 weeks after birth)

Stages of ROP
There are 5 stages of ROP.
<table>
<thead>
<tr>
<th>Stage</th>
<th>Image</th>
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| Stage 1: Line  
A thin white line that separates the avascular retina anteriorly from the vascularized retina posteriorly | ![Stage 1 Image](image1) |
| Stage 2: Ridge  
The line becomes elevated and wide | ![Stage 2 Image](image2) |
| Stage 3: Ridge with fibrovascular proliferation into the vitreous | ![Stage 3 Image](image3) |
Management:
Early stages of ROP, which show risk criteria for progression, can be managed by either laser treatment to the avascular peripheral retina or injection of Anti-VEGFs intravitreally. Stage 4 and 5 are treated surgically by pars plana vitrectomy with very guarded visual prognosis in most cases.