The baby that can’t see – Tips and tricks

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FRCOphth

Teaching points

- Prevalence and causes of VI in children
- UK vision screening
- What level of vision should you and parents expect?
- What are the important features in the history
- How to examine an infant’s eyes
- Decision flow chart
- Electrophysiology
- Common causes of visual impairment in babies

Causes of Childhood VI in UK: 2:1000

27% AVOIDABLE / TREATABLE

73% UNAVOIDABLE

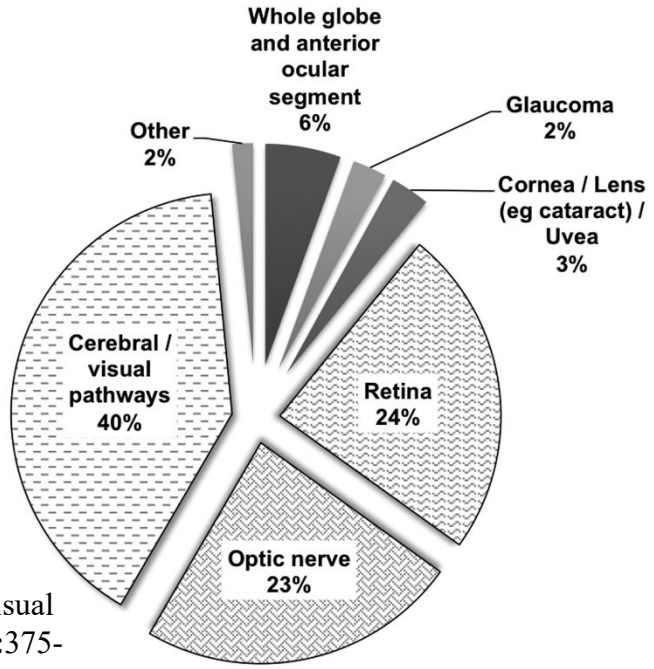
Causes of Childhood VI in UK: 2:1000

27% AVOIDABLE / TREATABLE

- Cataract (5%)
- Glaucoma (3%)
- Uveitis (2%)
- Refractive error (2%)
- Raised ICP (4%)
- Retinopathy of Prematurity (3%)
- Non-accidental injury (3%)
- Retinoblastoma (1%)
- Diabetes (1%)

73% UNAVOIDABLE

- Cerebral visual impairment (50%)
- Retinal dystrophies (10%)
- Optic nerve anomalies (5%)



Solebo AL, Rahi J Epidemiology, aetiology and management of visual impairment in children *Archives of Disease in Childhood* 2014;**99:**375-379.

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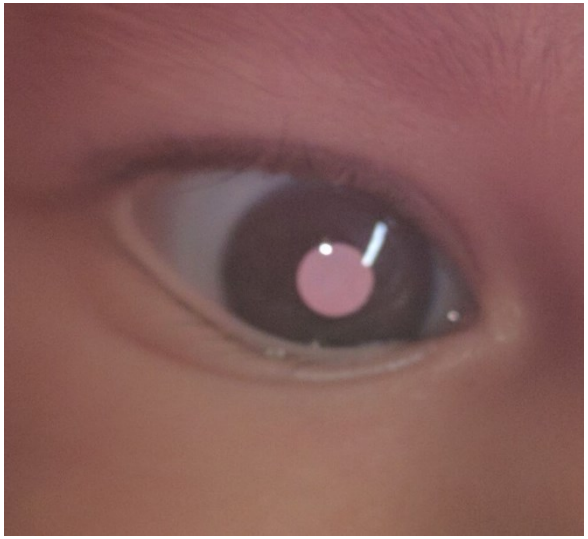
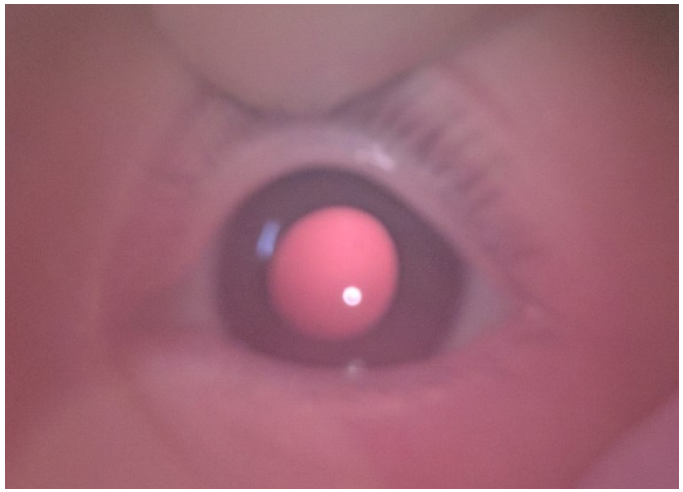
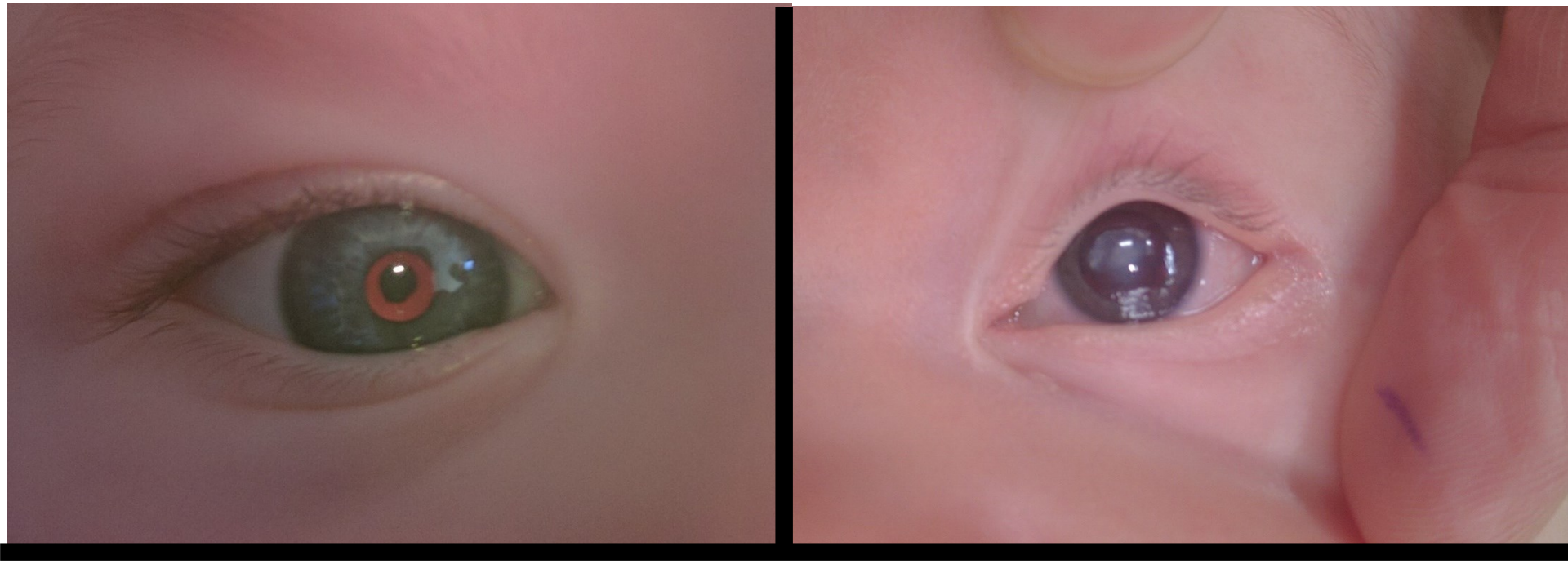
UK vision screening

- NIPE (newborn and infant physical examination) designed primary to pick up cataract
  - Within 72 hours of birth: red reflex
  - 6-8 week check: red reflex
  - Approx 50% undetected
  - Approx 80% referred cases are false positive, esp ethnic minority





Rahi et al. National surveillance study, BMJ Feb 1990. Vol 318 362-5
47% detected at NIPE screening
43% diagnosed before 12 weeks of age



What are the differences between infant and adult cataract in morphology and aetiology?

How are unilateral and bilateral congenital cataracts different?

UK vision screening – how and why?

UK vision screening

- Aiming to detect amblyopia (5% of children) before visual maturity
- Vision screening in schools at 5 years of age
- What about earlier photoscreening – what is it testing / pros and cons?
- Use Keeler letter screening cards
- 40% of referrals are false positives

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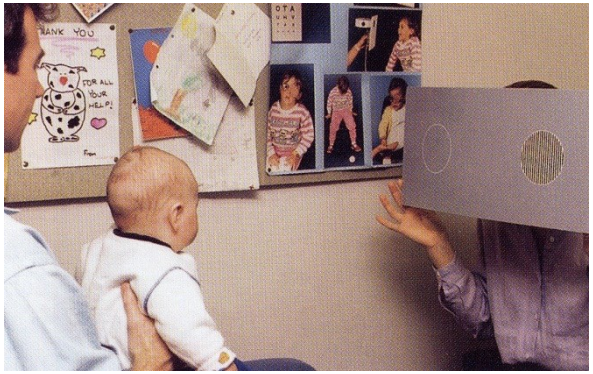
What can an infant see?

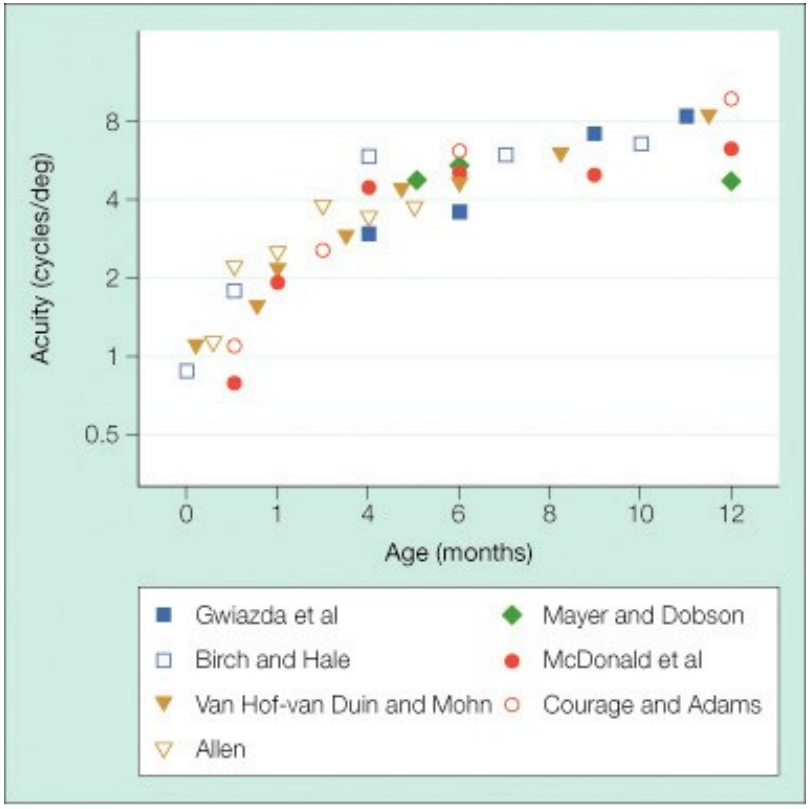
- VA about 6/36-6/60
- Preferential looking techniques (resolution rather than recognition)
- Fix and follow a large toy target
- Central, steady , maintained (CSM)



Visual development

- When does a normal neonate begin to see?
  - CF at birth
  - 4-6 weeks central foveal fixation/saccades
  - 6 weeks smooth pursuit movements
  - Critical period first 2-3 months
  - Approx 6/9 at 3yo

  - Resolution vs recognition testing?



Teaching points

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Visual behaviour

Visual behaviour

- Does the baby lock on eyes with parent when feeding?
- Do they feel that the baby's visual interest fluctuates?
- Do they ever notice nystagmus?
- Does the baby stare at lights or startle to touch?

Birth history

Birth history

- Prematurity
- Maternal rashes / fevers
- Maternal drug use
- Consanguinity
- Neonatal course
- Subsequent development
- Family history

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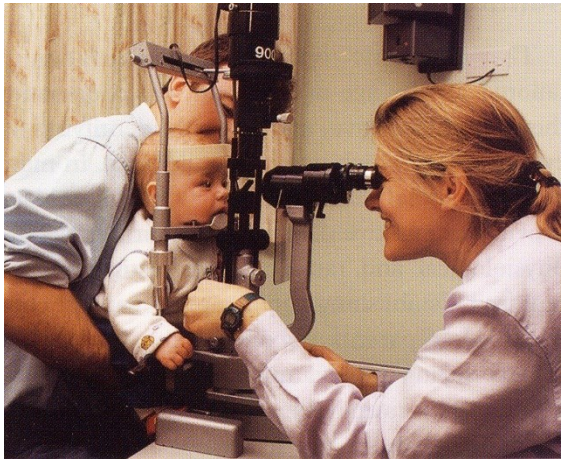
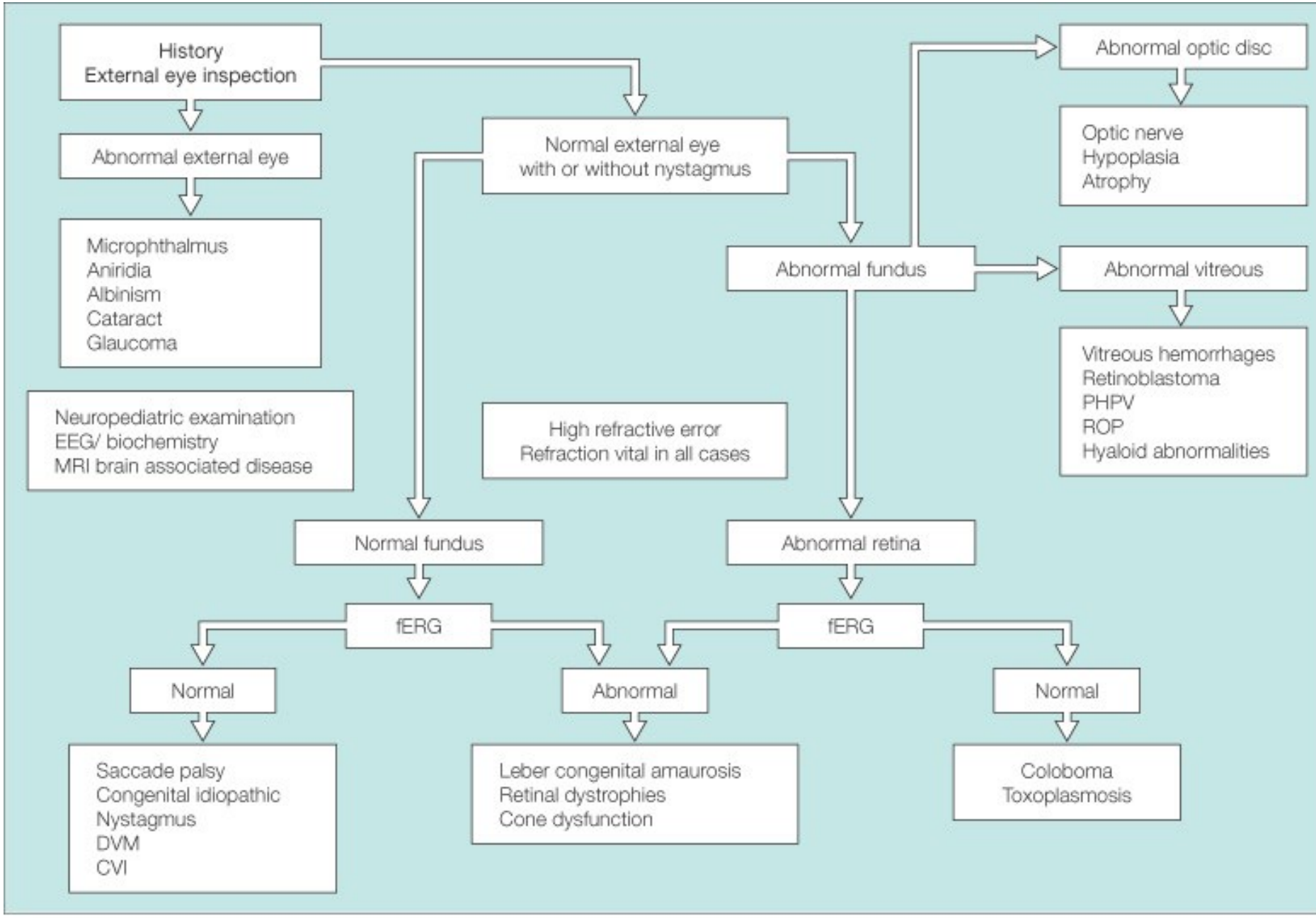
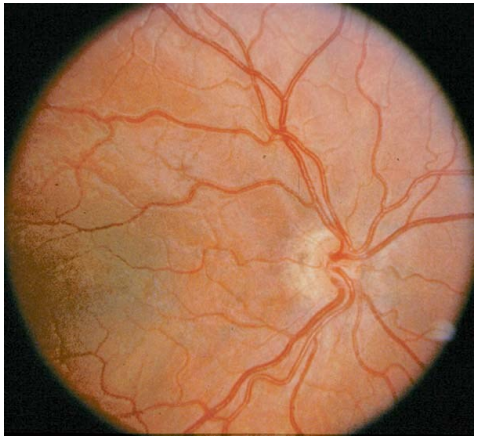
Eye examination in infancy

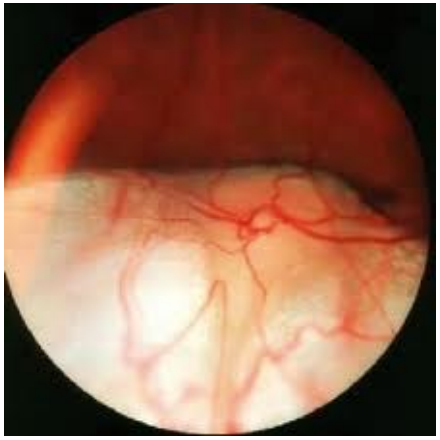
- Check globe for congenital abnormality
  - Aniridia, microphthalmia, cataract, glaucoma
- Pupil responses
- Eye movements to toy /face, corneal light reflex symmetry
- Look for nystagmus
- Cycloplegic refraction (0.5% cyclo)
- Fundoscopy with indirect and 28D
- Retcam / Optos
- Family examination

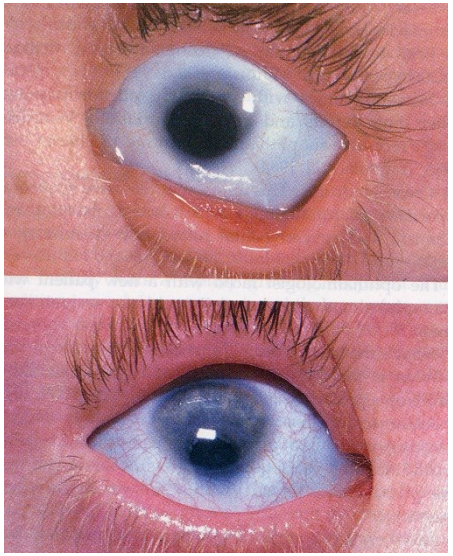


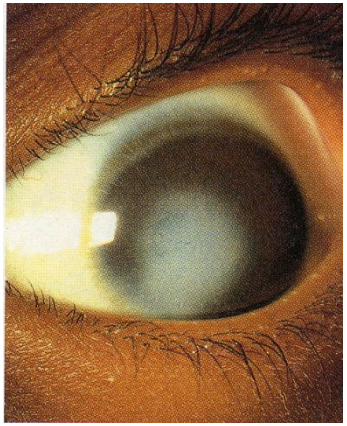


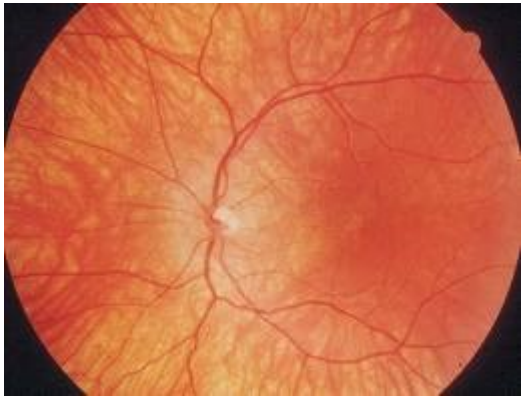
Abnormal examination



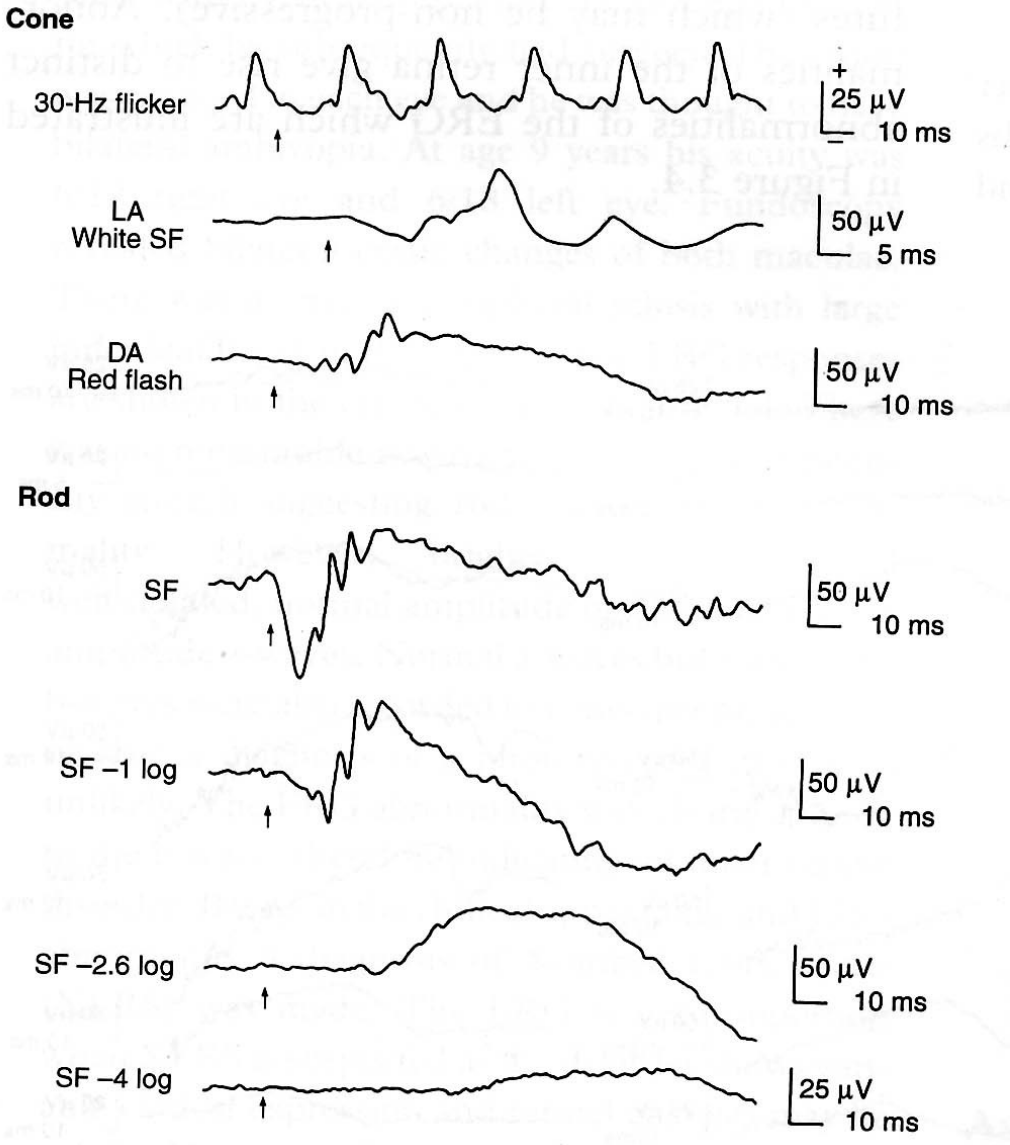






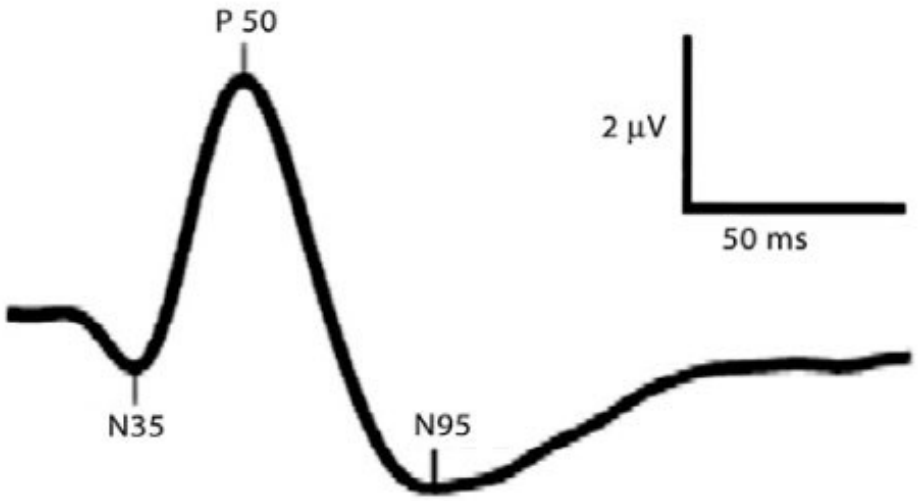


The Full field ERG



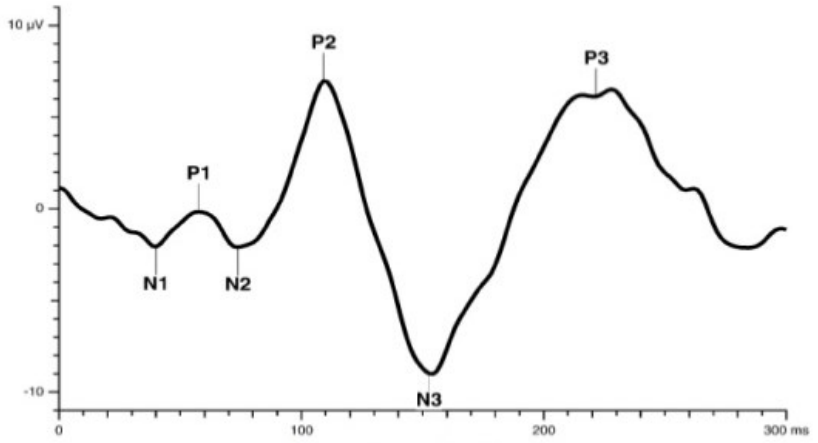
pERG: macular pathway

- Pattern reversal chequerboard stimulus
- Usually not possible in infants
- P50 component is derived from macular retina
- N95 component is derived from optic nerve



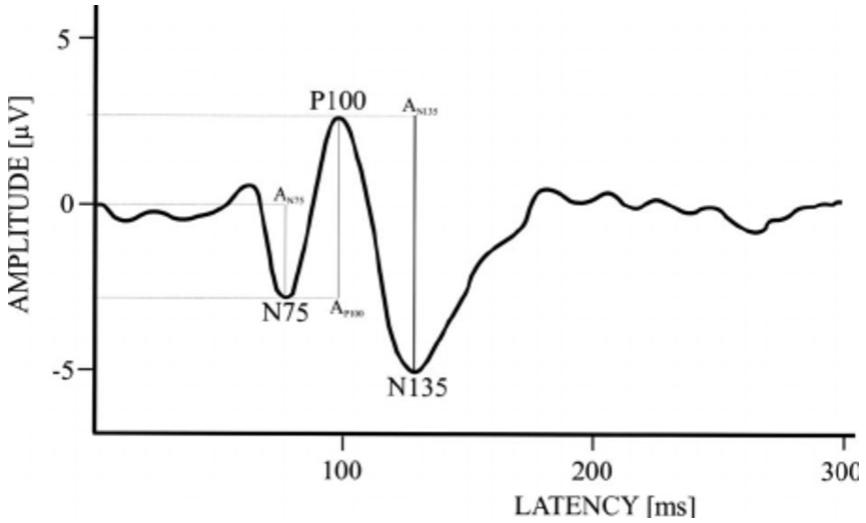
Flash VEP

- Useful in infants or if VA is poor
- No visual fixation required
- Allows assessment of visual acuity in each eye and hemianopias
- Tests the visual pathway as a whole: doesn't identify location of problem although P100 latency changes are seen in demyelination
- Helpful if used with pERG



Pattern VEP

- Useful in children with good fixation
- Helpful for functional cases
- Allows comparison between each eye / between hemispheres
- Tests the visual pathway as a whole: doesn't identify location of problem eg poor in amblyopia, inattention

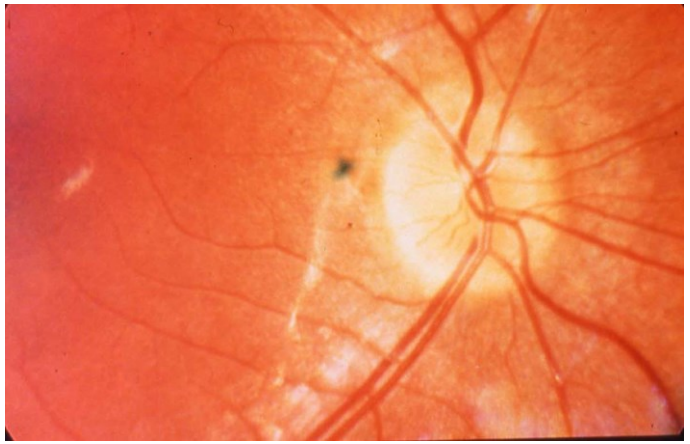


Case 1: Theo

- 4/12 old caucasian baby boy
- Poor visual attention
- Roving nystagmus since 1/12
- Born full term, no problems during pregnancy or in neonatal period
- Otherwise thriving
- No FH, no parental consanguinity

Theo - examination

- Sl photophobic
- Eye rubbing
- Not fixing and following
- Large amplitude, roving horizontal nystagmus
- Sluggish pupillary reaction to light

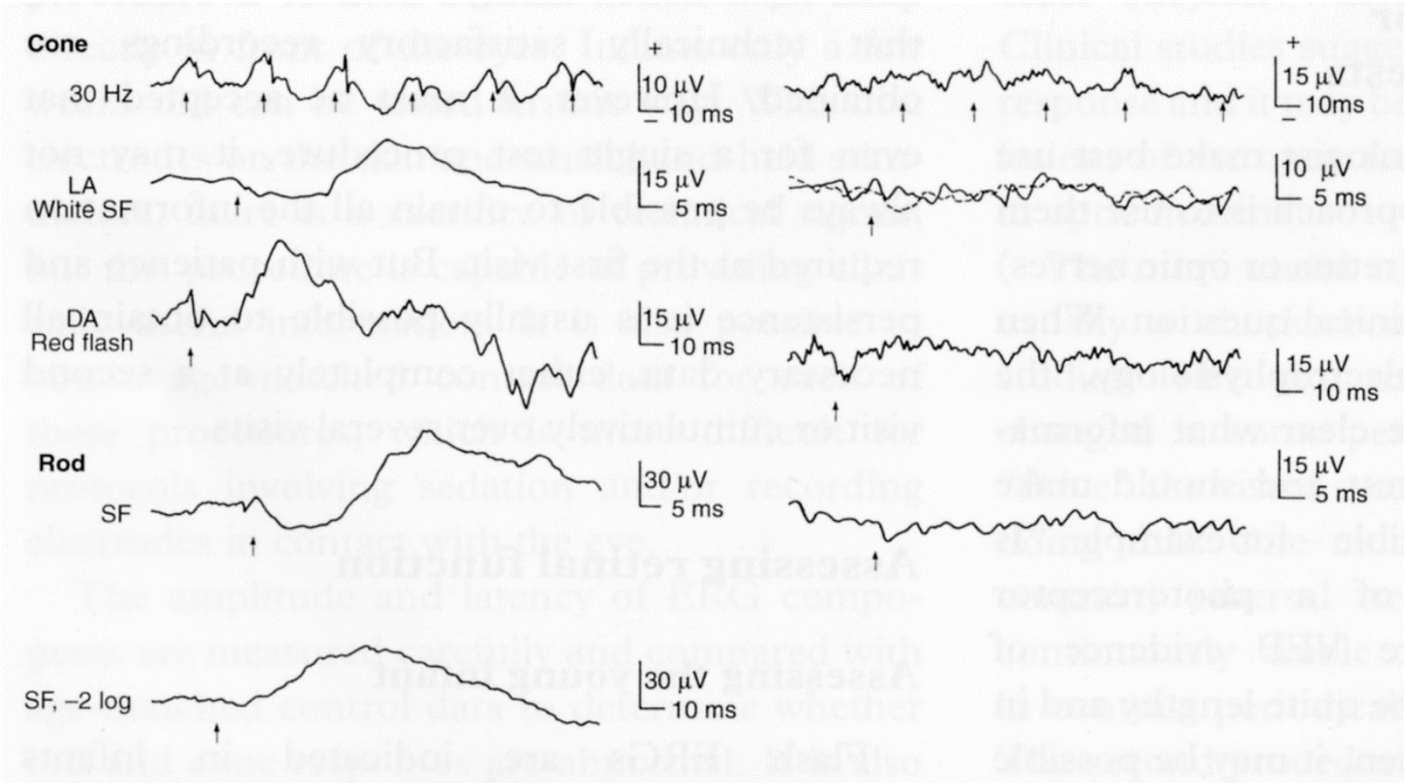


Theo-examination

- Anterior segment normal
- Cycloplegic refraction: +6DS each eye
- Fundoscopy was normal

- Possible diagnoses?
- What Ix would you want?

Theo's infant protocol ERG



Normal infant

Child with EORD

Theo-diagnosis

- Early onset severe rod/cone dystrophy (Leber congenital amaurosis)
- Numerous genotypes – main ones
  - *RPE65* (vitamin A metabolism)
  - *GUCY2D* (involved in transduction pathway)
  - *CRX* (involved in photoreceptor development)
  - ***CEP120***
- 266 retinal dystrophy gene panel – about 60% hit rate
- *RPE65* phenotype is less severe, photophilic with later onset – Luxterna

What support can we offer?

- Check hearing, think syndrome
- Genetic review / counselling
- Child Psychology service
- Visual impairment teacher support
  - Developmental milestones delayed
- Child will be able to maintain mainstream school with support – need educational statement
- Photochromic glasses may be helpful
- Red tinted glasses for achromatopsia
- Dietary advice
- Future gene therapy for some forms

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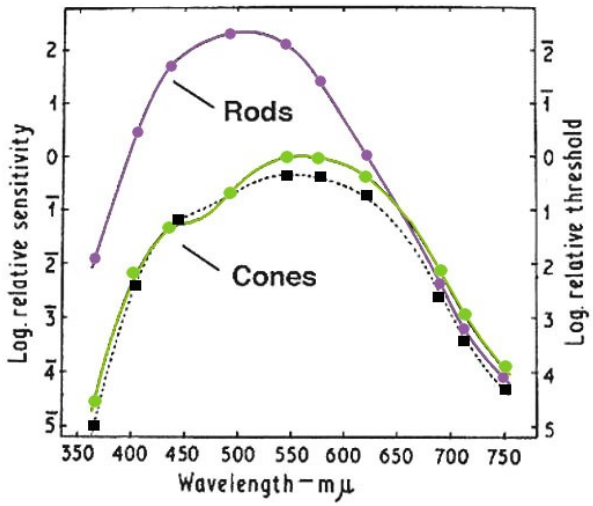
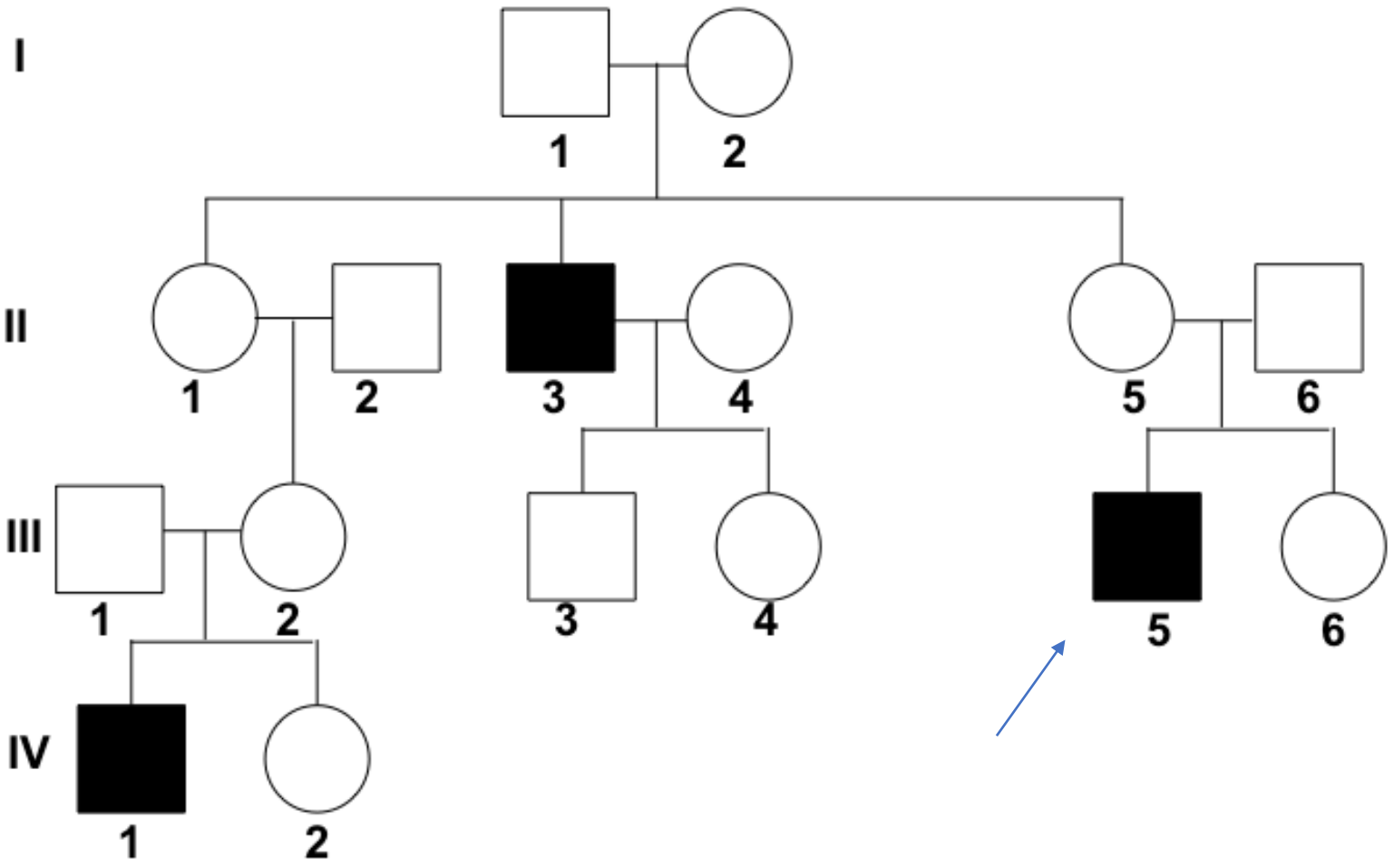


Case 2 - Alfie

- 3/12 old caucasian boy
- Parents worried that he can’t see
- Noticed roving nystagmus since 1/12 old
- No problems during pregnancy/delivery, born full term
- FH: maternal uncle has wobbly eyes


I
1
2
II
1
2
3
4
5
6
III
1
2
3
4
5
6
IV
1
2

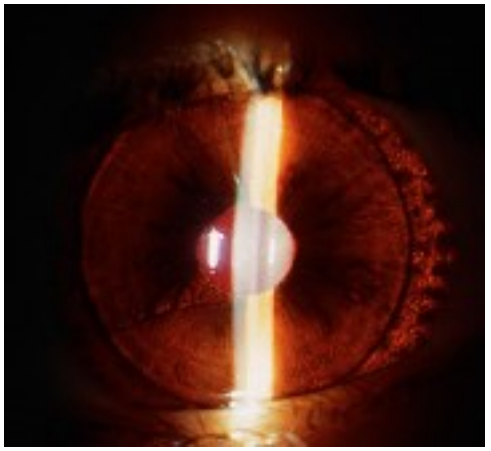
Alfie - exam

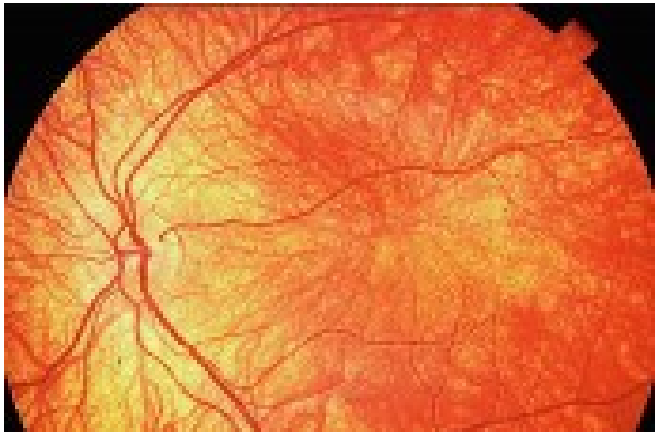
- Horizontal pendular nystagmus
- Normal pupillary reactions
- Not fixing and following

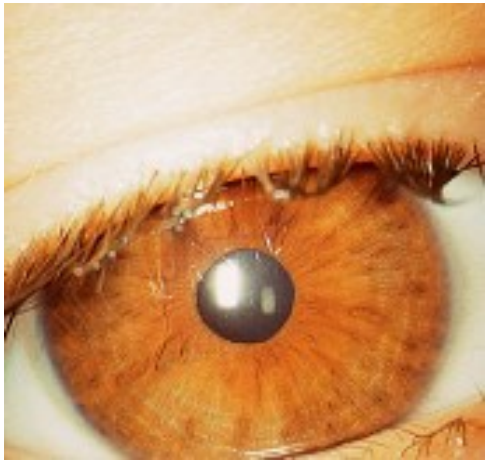
- What next?

Alfie - exam

- Blond boy but not white haired
- Horizontal pendular nystagmus
- Variable angle right esotropia
- Refraction: +4/-2x180



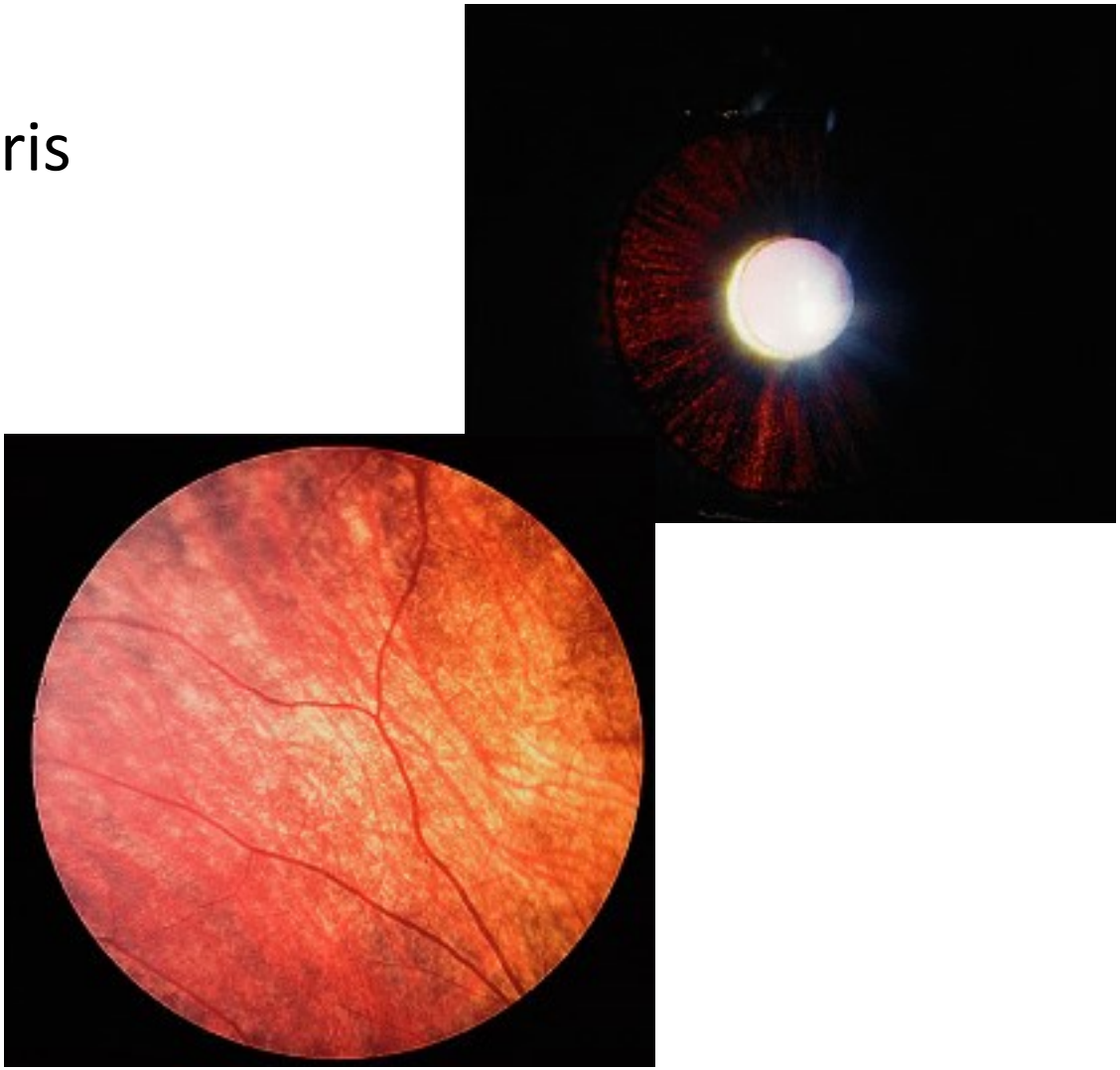




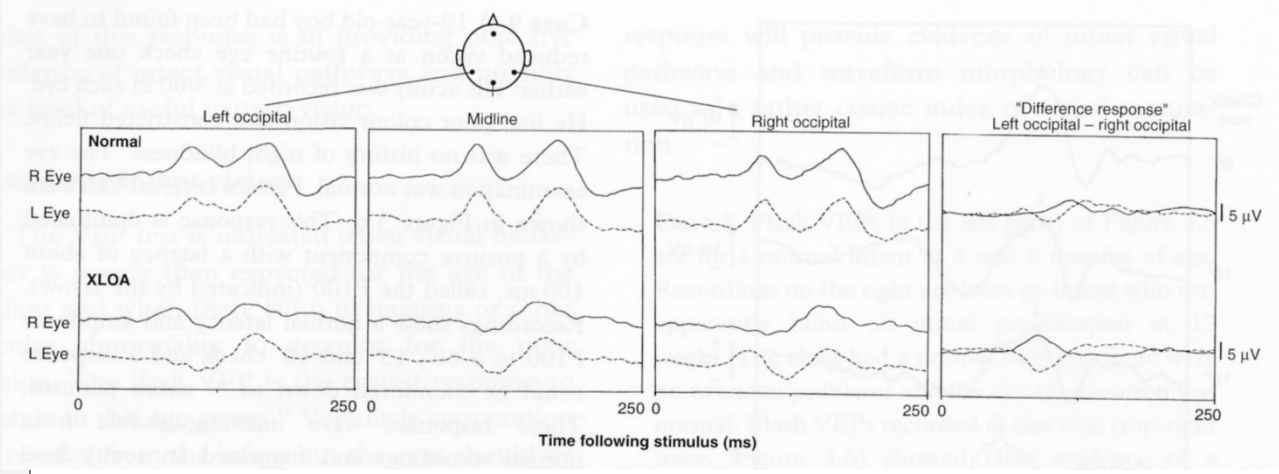
Maternal examination

- Mild peripheral iris translucency
- Mud-splatter appearance on fundoscopy

- Diagnosis?



Albino protocol VEP



Diagnosis: ocular albinism (XL)

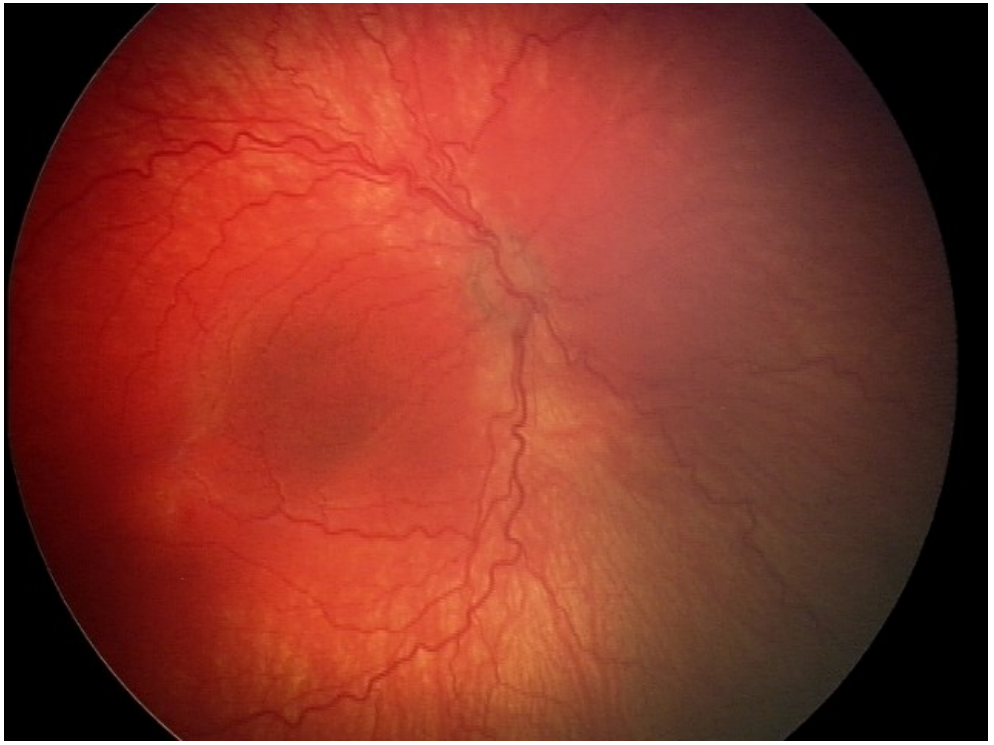
- Associated delayed visual maturation
- OA1 Caused by mutations in *GPR143* protein is expressed in RPE and melanocytes
- Problem with melanin trafficking rather than production
- Prognosis: probably 6/24 or so, better for near
- Likely to develop a head posture

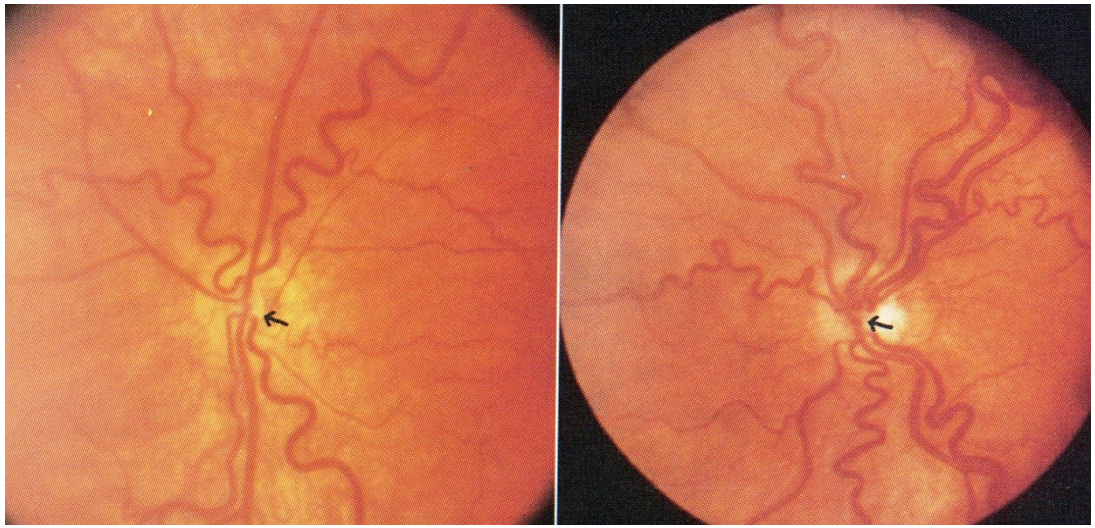
Case 3- Max

- 3/12 old caucasian boy
- Parents concerned about vision
- Noticed roving nystagmus since soon after birth
- Maternal IDDM
- Full term, prolonged jaundice
- No FH of eye problems

Max -exam

- Not fixing and following
- Roving horizontal nystagmus
- Normal pupil reactions
- Refraction: +2.50BE

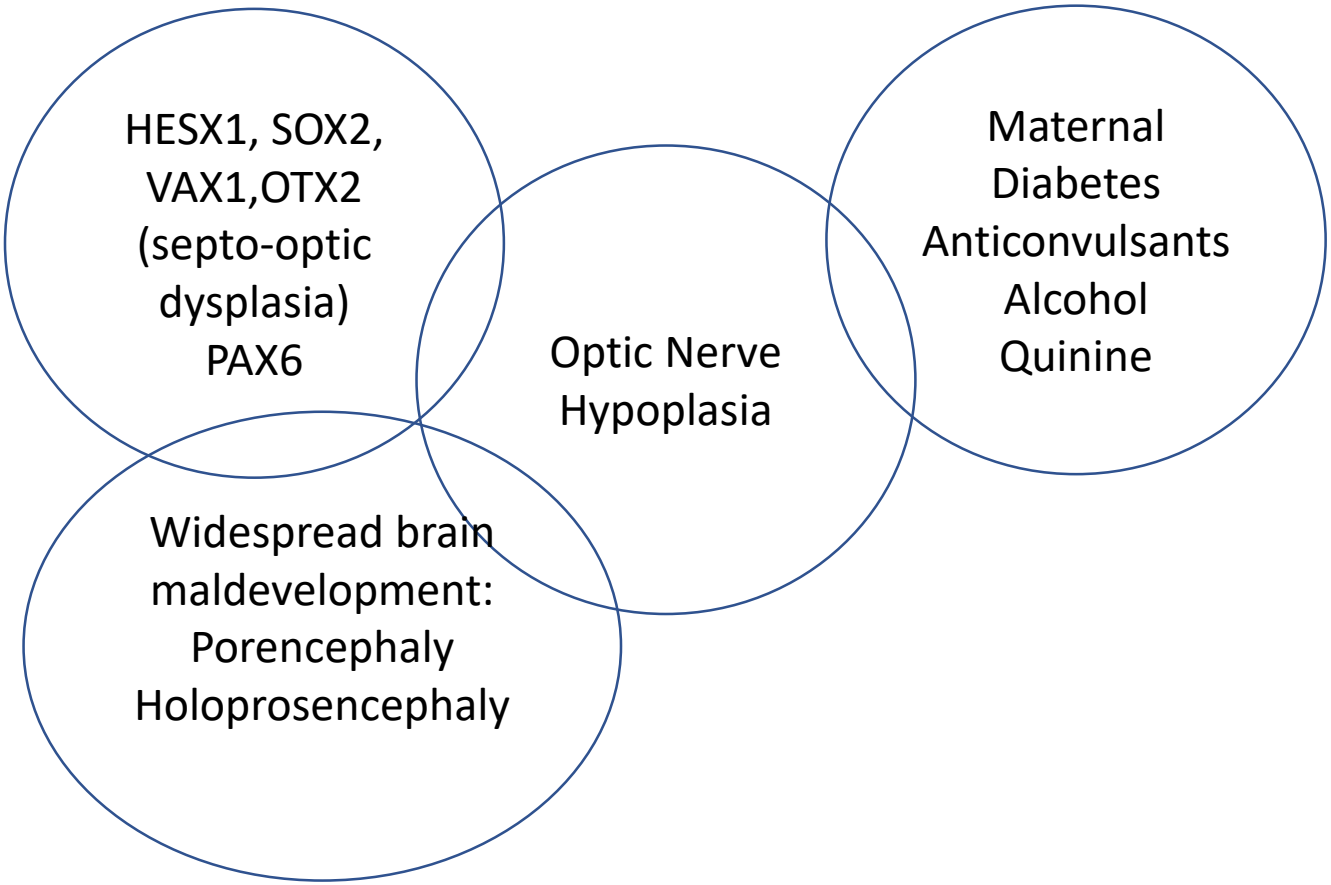




Optic nerve hypoplasia 1:10,000

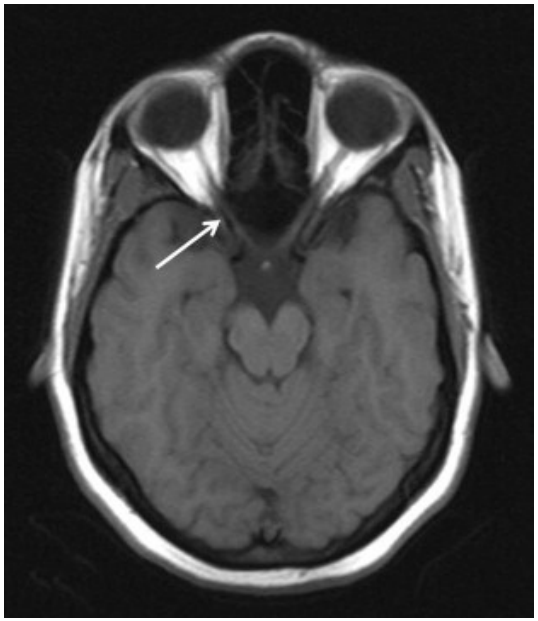
- Unilateral / bilateral
- Isolated / syndromic
- Associated with CNS and endocrine abnormalities
- Commonest congenital optic disc anomaly seen
- Often missed
- Insult early in gestation causes reduced number of axons
- Visual acuity can not be predicted by size of disc
- Non-progressive visual problems

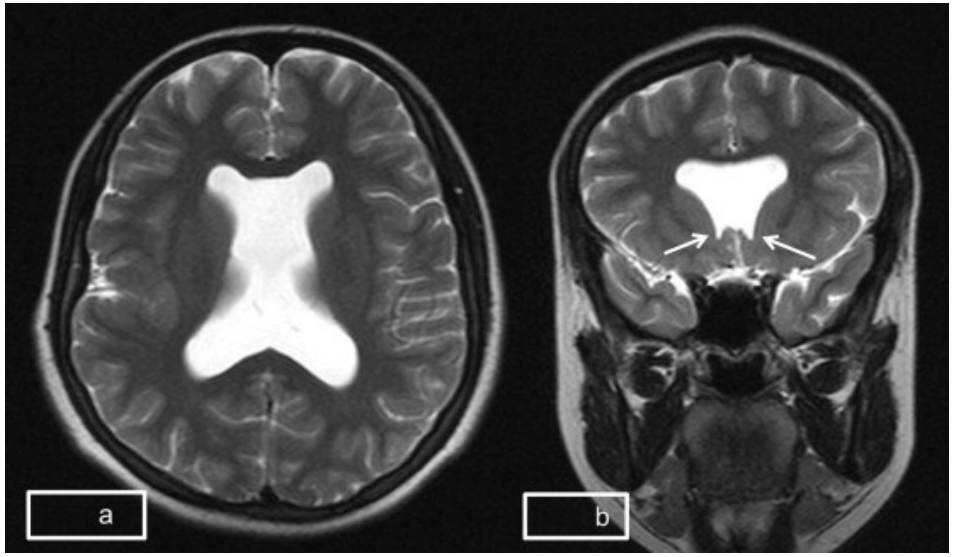
Associations of optic nerve hypoplasia



MRI

- Thinning of ON/chiasm
- Hemispheric migration abnormalities
- Absence of septum pallucidum
- Thinning / agenesis of corpus callosum
- Pituitary ectopia





Diagnosis: septo-optic dysplasia

- Refer to paed neurologists / geneticists
- Refer to paed endocrinologists
  - Low GH started on hormone replacement
  - hypothyroidism

- Refer to visual impairment teachers
- Can’t titrate nerve size to future vision
- VEPs can be useful
- Element of delayed visual maturation

Delayed visual maturation

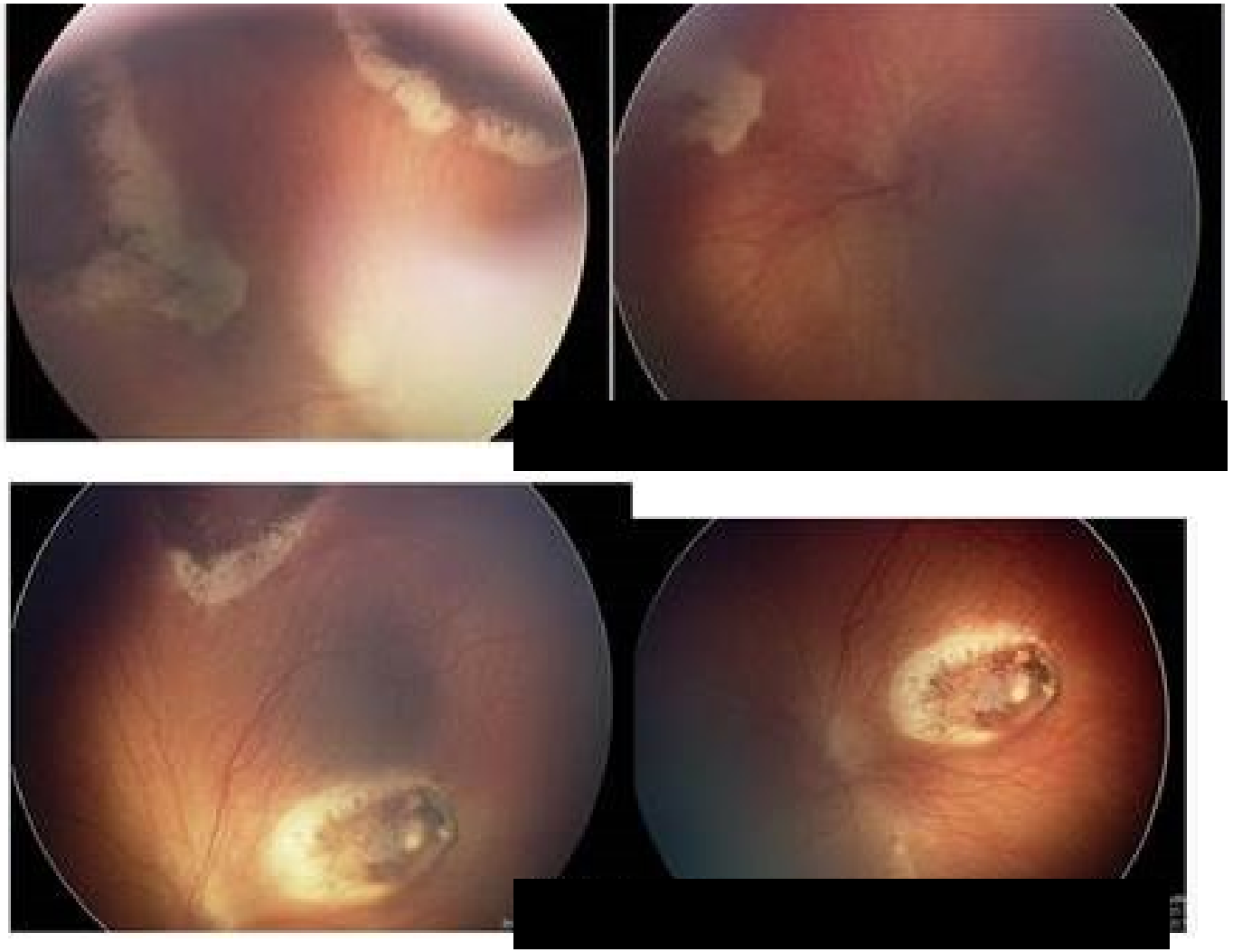
- Type I-isolated
  - Presents at 3-4 months of age
  - Improve by 6 months
  - May be associated with slowness to speak
- Type 2 – systemic disease / global delay
  - Ex-prems, infantile spasms
  - Takes years to improve
- Type 3 – associated with early onset ocular disease
  - Albinism, cataracts, ON hypoplasia
  - Takes years to improve

Case 4: Jude

- 3 month old boy
- Parents concerned about lack of visual responses and nystagmus
- Full term birth
- No prenatal problems
- No family history
- Neonatal gastroenteritis and prolonged jaundice, liver function tests remain abnormal

- Nystagmus and poor visual responses confirmed on examination
- Refraction normal

Fundus examination

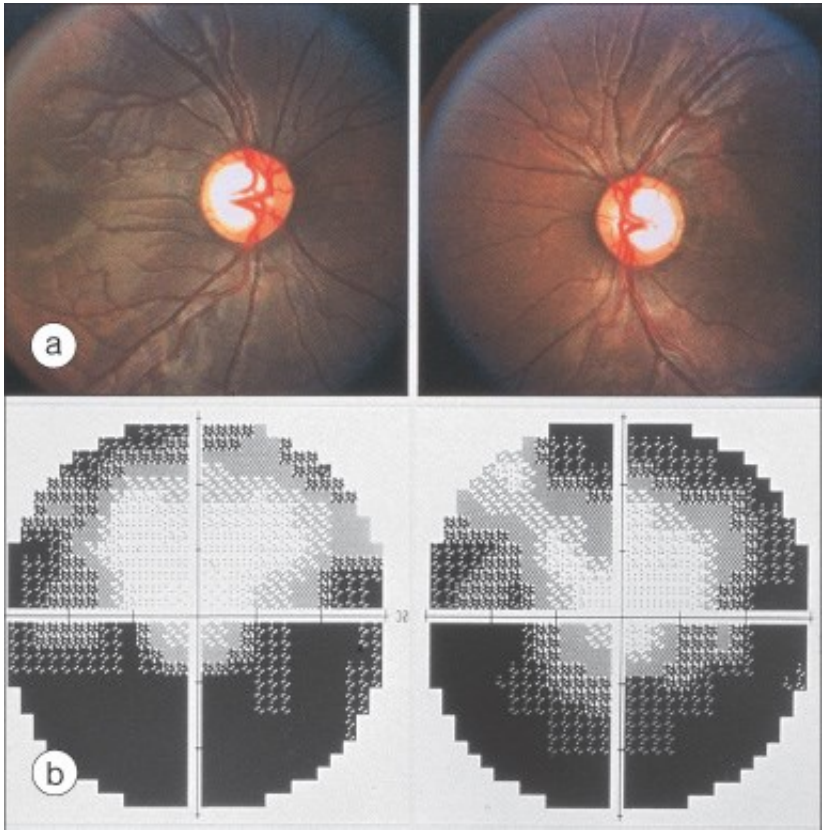


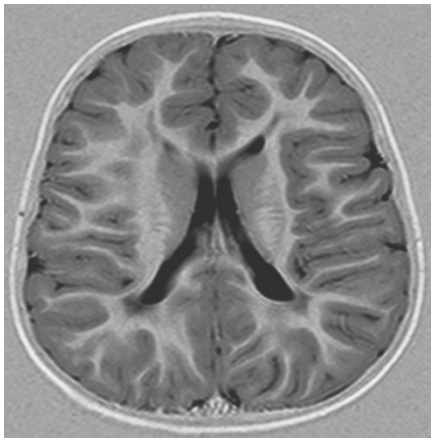
Case 5: Charlie

- 4/12 old child ex –prem 25 week gestation
- Parents concerned that child can not see
- Previous ROP Stage 2 spontaneously regressed
- Rocky neonatal course
  - Perventricular leukomalacia
  - Previous intra-ventricular haemorrhage
  - Chronic lung disease
- No FH or parental consanguinity

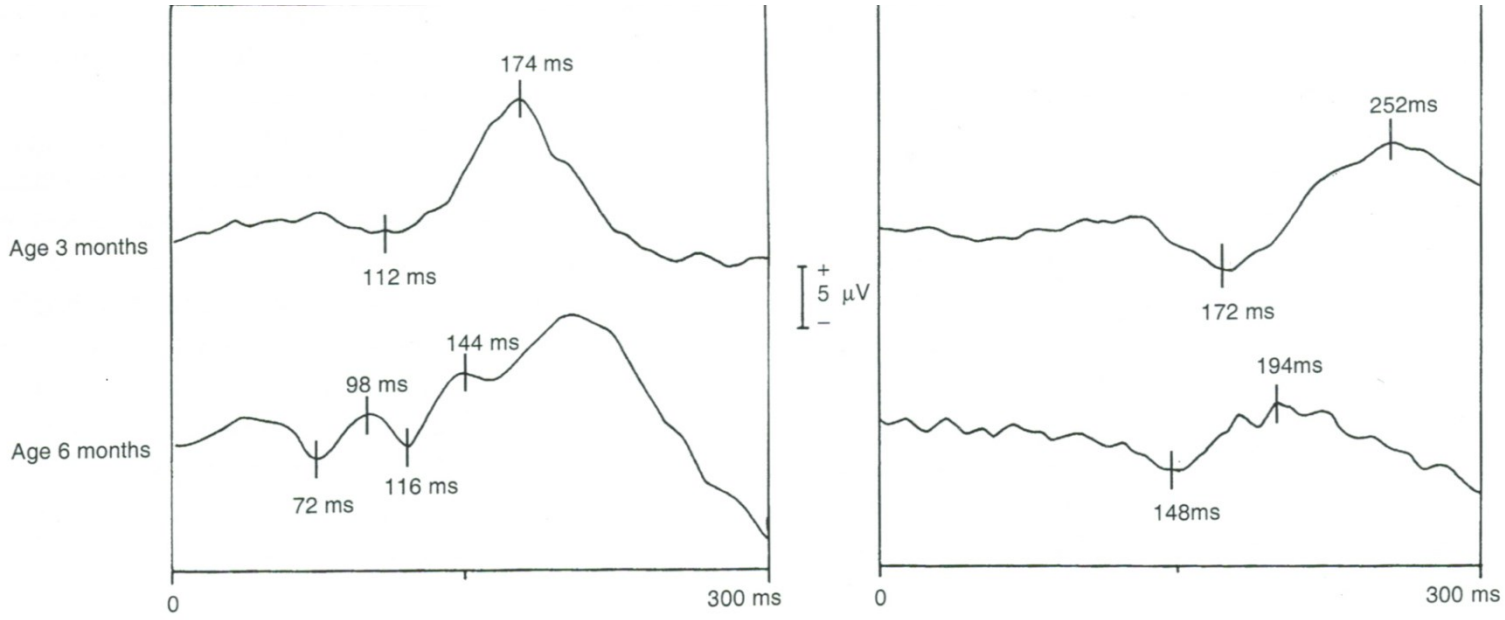
Charlie

- No nystagmus
- Will not fix or follow
- Pupil reactions normal
- Refraction: +2.00DS
- Fundoscopy: slightly pale, 0.8c/d ratio





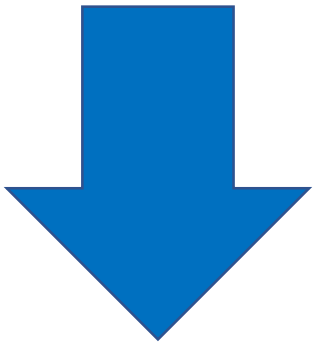
Infant protocol flash VEP



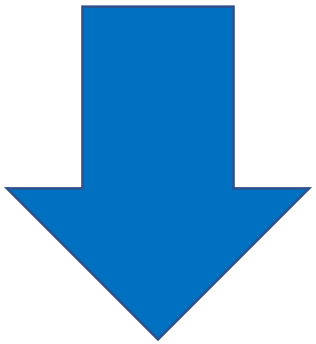
Normal child

Infant with CVI

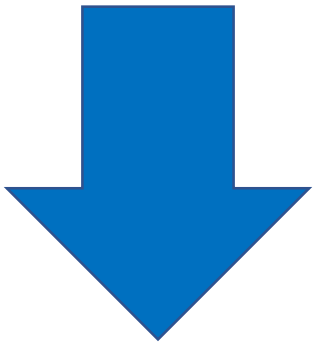
Three main groups



Profound visual impairment



Some useful functional vision alongside cognitive and other disabilities



Useful vision and the ability to use it

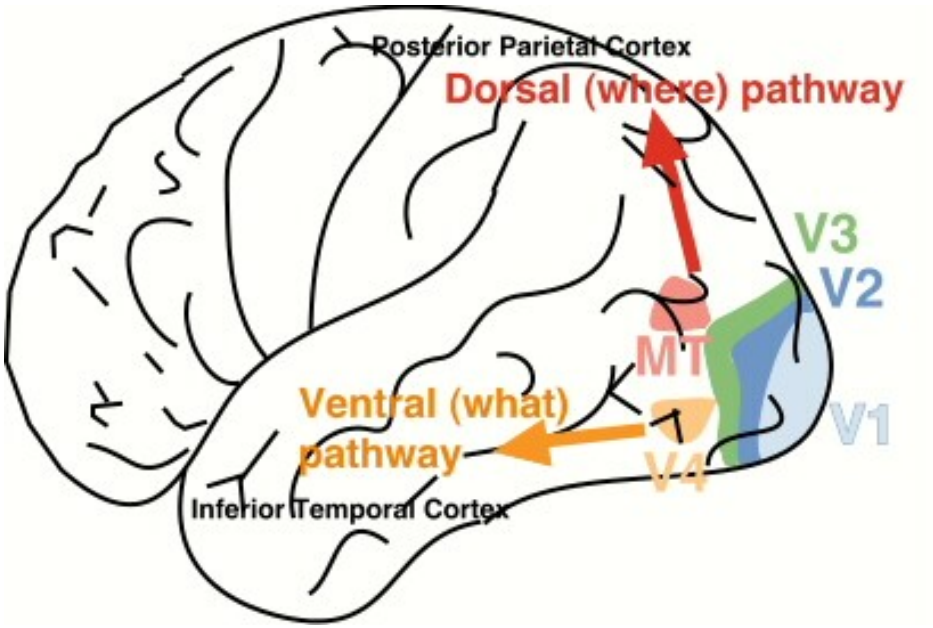
All can be associated with cerebral palsy / prematurity
+/- movement disorder (dyskinesia) +/- accommodative disorders

Dutton and Lueck, Vision and the Brain understanding CVI in children, AFB press

CVI Screening questions:

- Does your child have difficulty walking downstairs?
- Does your child have difficulty seeing things which are moving quickly, such as small animals?
- Does your child have difficulty seeing something which is pointed out in the distance?
- Does your child have difficulty locating an item of clothing in a pile of clothes?
- Does your child find copying words or drawings time-consuming and difficult?

Higher visual processing



- Dorsal stream “where?”
  - Posterior parietal lobes appraise overall scene
  - Temporal lobe input (recognition)
  - Frontal lobe input (choice)
  - Motor response
  - Seen in PVL
- Ventral stream “what?”
  - Medial temporal lobes (recognition)

Higher visual processing issues

Dorsal stream dysfunction

- Impaired simultaneous perception
  - Can't recognise mother in crowd
  - Difficulty with complex visual environments
- Impaired movement in 3D space (optic ataxia) eg kerbs, floor boundaries
- Impaired perception of movement
  - Avoid situations with fast motion
  - Aware of fast targets only when they stop

Ventral stream dysfunction

- Problems with visual recognition
- Problems with recognising facial expression
- Impaired face recognition (right side dominant)
- Impaired object and shape recognition with reading problems (left dominant)

Approaches to help with CVI

- Check accommodation is sufficient
- Use high contrast images
- Simplify backgrounds, remove clutter
- Avoid noisy environments
- Try masking text surrounds
- Mobility training for field defect
- Try child using wheeled toy or pram if problems with inferior field

Summary

- Take parents concerns seriously
- Nystagmus is an important diagnostic feature
- Full clinical exam including refraction
- Ix as necessary, remember MRI requires GA
- Do not tell the parent the child is blind – most conditions have associated with DVM and vision is likely to improve (although possibly not to normal levels)
- Involve the VI team early even when uncertain of diagnosis – early stimulation speeds improvement and may influence overall development
- Certification and help with Disability Living Allowance application
- Support groups