The Abnormal Red Reflex: Congenital Cataracts

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Good Red Reflex

- Bright red
- Symmetric
- How to acquire the reflex?
  - Dim room
  - Direct ophthalmoscope
  - Simultaneous viewing of both eyes at arm's length

Abnormal Red Reflex

Unequal:
- Color
- Intensity
- Clarity
- Semi-urgent
- Within 1 week

Presence of:
- Opacities
- Dark spots
- White reflex

Sight-threatening (Cataract)

Life threatening (Retinoblastoma)

 Causes of Abnormal Red Reflex

Asymmetrical reflex:
- Refractive error
- Strabismus
- Cataract
- Retinal pathology
- Rb
- Corneal opacity

Blunted reflex:
- Cataract
- Retinal pathology
- Rb
- Corneal opacity
- Glaucoma

Congenital Cataracts

- 1:4000-1:10000 per year
- Congenital / Infantile
- Significant cause of visual morbidity
- Genetic basis in 50%
  - >40 genetic loci, >17 genes
- Associated with systemic disease
- Timing of surgical intervention is critical

Clinical Presentation

- Abnormal red reflex
- Not following objects
- Sensory strabismus
- Unilateral nystagmus
Cataract Morphology

Laterality

<table>
<thead>
<tr>
<th>Bilateral Cataracts</th>
<th>Unilateral Cataracts (local dysgenesis)</th>
</tr>
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<tbody>
<tr>
<td>Idiopathic (60%)</td>
<td>Idiopathic (80%)</td>
</tr>
<tr>
<td>Familial (10%)</td>
<td>AD, X-linked</td>
</tr>
<tr>
<td>Chromosomal (&lt;5%)</td>
<td>Trisomy 21, 13, 18</td>
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<td>Metabolic (&lt;5%)</td>
<td>Galactosemia, diabetes mellitus, Fabry, Wilson</td>
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<tr>
<td>Craniofacial syndromes (&lt;5%)</td>
<td>Hallermann-Streiff, Rubinstein-Taybi</td>
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<tr>
<td>Renal syndromes (&lt;5%)</td>
<td>Lowe, Alport</td>
</tr>
<tr>
<td>Intrauterine infection (3%)</td>
<td>Toxoplasmosis, rubella, CMV, varicella, syphilis</td>
</tr>
<tr>
<td>Ocular anomalies (2%)</td>
<td>Aniridia, anterior segment dysgenesis</td>
</tr>
<tr>
<td></td>
<td>masked bilateral</td>
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</tbody>
</table>

Course of Action

Abnormal Red Reflex

- Dilate Pupil
- Refer

Dilate Pupil
- Tropicamide 0.5%
- Cyclopentolate 0.5%-Phenylephrine 2.5%
- Wait for 15-20 min
- Repeat Red Reflex Test

Within 1 week

Pediatric Ophthalmologist
Emphasize “Abnormal Red Reflex”

Management of Congenital Cataracts

• Visually significant opacities
  - Cataract extraction ± IOL implantation
  - Refractive correction (i.e. glasses)
  - Occlusion (to reverse amblyopia)

• Non-significant opacities
  - Close monitoring
  - Tendency to progress
  - Correct refractive errors ± patching

Prognosis

• Congenital vs. acquired
• Early onset opacity
• Longer duration of deprivation
• Unilateral onset congenital cataracts

<table>
<thead>
<tr>
<th>Timing of Surgery</th>
<th>Visual Outcome</th>
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<tr>
<td>&lt; 8 weeks</td>
<td>20/800-20/30</td>
</tr>
<tr>
<td>&gt; 8 weeks</td>
<td>HM-20/160</td>
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* Early surgery by itself does not ensure a good outcome!


Conclusions

• All infants should have a red reflex evaluation during the first 2 months of life.
• Abnormal red reflex requires a semi-urgent referral
• Underlying reason may be sight or life threatening
• Children will not complain!
• Have a low threshold for referral
  - If truly abnormal → Patient receives proper care
  - If normal → Everyone’s relieved & happy!

1. Asymmetry
2. Dark spots
3. Diminished reflex
4. White reflex