Q: Picture of leukocoria.
- What is the differential dx
- What are the features of each condition
- How do you work the patient up?

Ddx of **Leukocoria**
1) RB
2) PHPV
3) toxocara
4) Coat's
5) cataract
6) ROP
7) coloboma
8) RD
9) retinal dysplasia
10) uveitis

**RB**
Vitreous seeding, uveitis simulating, NVI, pseudohypopion, usually normal size.

<table>
<thead>
<tr>
<th>Differential Diagnosis of Retinoblastoma</th>
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<tbody>
<tr>
<td><strong>Differential diagnosis of leukocoria</strong></td>
</tr>
<tr>
<td>Coats' disease</td>
</tr>
<tr>
<td>Persistent hyperplastic primary vitreous</td>
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<tr>
<td>Ocular toxocariasis</td>
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<tr>
<td>Cicatricial retinopathy of prematurity</td>
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<tr>
<td>Familial exudative vitreoretinopathy</td>
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<tr>
<td>Incontinentia pigmenti retinopathy</td>
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<tr>
<td>Nerrie's disease</td>
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<tr>
<td><strong>Differential diagnosis of vitreous seeds</strong></td>
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<tr>
<td>Pers planitis (intermediate uveitis)</td>
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<tr>
<td>Microbial endophthalmitis or retinitis</td>
</tr>
<tr>
<td>Leukemic infiltration (rare)</td>
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<tr>
<td><strong>Differential diagnosis of discrete retinal tumors</strong></td>
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<tr>
<td>Astrocytoma of retina</td>
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<td>Retinal capillary hemangioma (usually appears red, but may appear white if associated with retinal fibrosis or accumulation of intraretinal exudates)</td>
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<tr>
<td>Focal patches of myelinated retinal nerve fibers</td>
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</table>
cataract rare, uni and bilateral and may be multifocal, Ca++

- **Coat’s Disease** (Leber’s Miliary Aneurysms) is characterized by the following:
  Males 3:1, 10-15% bilateral
  **Ocular findings**: retinal telangiectasia, sheathed vessels, retinal exudates, subretinal fluid, exudative RD, capillary non-perfusion, usually superotemporal
  **FA**: telangiectasia, micro & macroaneurysm, cap non perfusion, leakage

  **Treatment**
  Mild-Modrate
  Laser photocoagulation (AG, diode)
  Cryo
  Severe
  Scleral buckle with drainage of SRF
  Vit with MP. Drain SRF, Laser

- **PHPV**
  (retrol. mass of fat, SM, collagen, cartil.)
  **Ocular findings**: microphthalmos, microcornea, cataract, angle closure glaucoma, usually unilateral, elongated ciliary processes

Retinoblastoma Genetics

Unilateral RB’s: Risk of subsequent children having RB

<table>
<thead>
<tr>
<th>Parent Status</th>
<th>Child Status</th>
<th>Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Affected parent</td>
<td>Child not affected</td>
<td>3%</td>
</tr>
<tr>
<td>Normal parent</td>
<td>Affected child</td>
<td>3%</td>
</tr>
<tr>
<td>Affected parent</td>
<td>Affected child</td>
<td>30%</td>
</tr>
</tbody>
</table>

Bilateral RB’s: Risk of subsequent children having RB

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<th>Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Affected parent</td>
<td>Child not affected</td>
<td>40%</td>
</tr>
<tr>
<td>Normal parent</td>
<td>Affected child</td>
<td>10%</td>
</tr>
<tr>
<td>Affected parent</td>
<td>Affected child</td>
<td>50%</td>
</tr>
</tbody>
</table>
Work up for leukocoria
1. Complete history and physical exam
2. Examine family members
3. U/S (Ca in RB, ERD in Coats’, Retrolental mass in PHPV)
4. CT scan and MRI
5. F/A if Coats’ in question
6. Serology if toxocariasis suspected

Tid bits about RB

Treatment of RB Traditional
1) unilateral RB with poor visual potential (macular tumors and large tumors - i.e. half the vitreous) à enucleate (still true today)
2) bilateral tumors: enucleate the poorer eye (if advanced)
3) enucleate for: rubeosis, glaucoma, vitreous seeding

Prognosis of Death if ON invasion in RB
1) 8% - no invasion
2) 15% - prelaminar
3) 44% - retrolaminar and before resected end
4) 64% - retrolaminar and past resected end

Prognosis of RB
1) optic nerve not involved: 90% survive
2) tumor into lamina cribrosa: 40%
3) tumor past cut end of nerve: 20%

Baseline Systemic Evaluation in Retinoblastoma

| Complete pediatric history and physical examination |
| Blood for complete blood count |
| Magnetic resonance imaging or computed tomography of brain, especially in bilateral or familial cases, to look for pineoblastoma or ectopic intracranial retinoblastoma |
| Lumbar puncture for cerebrospinal fluid analysis |
| Bone marrow aspiration or biopsy |
| Bone scan |
| Genetic testing of affected child and family members |

a) Currently advocated only for children who have advanced intracocular disease or clinically extracocular disease at baseline.

b) Currently available only as part of a research study in most centers.

Treatment Options for Intraocular Retinoblastoma

- Chemotherapy
- Enucleation
- Radiation therapy
  - external beam radiation therapy proton or gamma ray
  - plaque radiotherapy
- Photocoagulation and laser therapy
- Cryotherapy
- Observation (for spontaneously arrested retinoblastoma, 'retinoma')
Follow up after enucleation: every 4 months for EUA until age 4, then Q 6 months

Sites for mets for RB
1) CNS
2) skull
3) long bones

DDX of Homer Wright rosettes
(fleurette specific for RB)
1) RB
2) medulloepithelioma
3) neuroblastoma

DDx of Flexner-Wintersteiner rosettes
1) retinoblastoma
2) pinealoblastoma
3) ectopic intracranial retinoblastoma
Maturity:
fleurette>FW>HW

DDx of small to medium size retinoblastoma
1) astrocytic hamartoma
2) retinal capillary hemangioma
4) posterior pole granuloma

Workup for RB
1) CSF (LP) - during EUA
2) bone marrow - during EUA
3) bone scan
4) CXR

Treated RB appearance
1) tumor disappeared
2) cottage cheese
3) fish flesh - looks like untreated tumor

Genetic counselling for RB
Risk of next child to have inheritable RB
1) parent had bilateral RB: 45%
2) “normal parents with 2 children with RB: 45%
3) parent has unil. RB : 7-15%
4) “normal” parents with 1 child with bil. RB: 5%
5) “normal” parents with child with unil. RB: <1%
- 15% of sporadic unilateral RB patients are carriers of RB gene and therefore are transmittable (see 2)
- 6% of RB patients have FH of RB

Genetic work up for RB
1. Karyotyping detect 3-5%
2. Esterase D 10%
3. Southern blot (RFLP) 20%
4. DNA polymorphisms PCR/SSCP 100% eventually