Pediatric Retina

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Hey, Will you Peek at this Kid?
4 yo w/ Fundus Abnormality
Closer look
What’s your Dx?

Retinoblastoma
Retinoma
Toxoplasmosis
Toxocara canis
Astrocytic Hamartoma
Other
Case #2

Courtesy of G. Baker Hubbard MD
Case #1
Courtesy of G. Baker Hubbard MD

Right Eye  8L  90dB

Right Eye  8E  60dB
What’s your Dx?

Retinoblastoma

Coats Disease

Juvenile Idiopathic Arthritis related Uveitis

Intermediate Uveitis

Infectious Retinitis (bacterial or fungal)

Other
Case #1

Courtesy of Hans Grossniklaus MD
Case #1

Courtesy of G. Baker Hubbard MD
Case #1

 Courtesy of G. Baker Hubbard MD
Case #2

Courtesy of G. Baker Hubbard MD
Right Eye  HAX  90dB
MRI with Contrast
Case #2
Courtesy of Hans Grossniklaus MD
Case #2

Courtesy of Hans Grossniklaus MD
Case #2

Courtesy of Hans Grossniklaus MD
Statistics

Incidence
1:15,000
300 cases/yr in US

Bilateral
1/3 of cases

Unilateral
2/3 of cases
15% will have germline mutation
Genetics

Heritable (i.e. Germline Mutation)
- Usually Bilateral
- Younger Age of Onset
- (+) Family History

Non-Heritable (i.e. Somatic Mutation)
- Usually Unilateral
- Solitary tumors
- Slightly older age of onset
- (-) Family History
# International Classification of Retinoblastoma: ICRB

<table>
<thead>
<tr>
<th>Group</th>
<th>Tumor Features</th>
<th>% globe Survival</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Small Tumors $&lt; 3$ mm</td>
<td>100%</td>
</tr>
<tr>
<td>B</td>
<td>Large Tumor $&gt; 3$ mm, Macular, Juxtapapillary, subretinal fluid $&lt; 3$ mm from disc margin</td>
<td>93%</td>
</tr>
<tr>
<td>C</td>
<td>Focal Subretinal Seeds $&lt; 3$ mm from RB Vitreous Seeds $&lt; 3$ mm from RB</td>
<td>90%</td>
</tr>
<tr>
<td>D</td>
<td>Diffuse Seeds $&gt; 3$ mm from RB Vitreous Seeds $&gt; 3$ mm from RB</td>
<td>50%</td>
</tr>
<tr>
<td>E</td>
<td>Extensive Tumor $&gt; 50%$ of globe NVG, or opaque media, ON invasion, $&gt;2$ mm choroidal invasion, orbit involv.</td>
<td>0%</td>
</tr>
</tbody>
</table>

IAC Intra-Arterial Chemotherapy

### Intra-arterial Chemotherapy for Retinoblastoma in 70 Eyes

**Outcomes Based on the International Classification of Retinoblastoma**

<table>
<thead>
<tr>
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<th>Pre-IAC</th>
<th>Current</th>
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</thead>
<tbody>
<tr>
<td>A</td>
<td>100%</td>
<td>100%</td>
</tr>
<tr>
<td>B</td>
<td>93%</td>
<td>nearly 100%</td>
</tr>
<tr>
<td>C</td>
<td>90%</td>
<td>nearly 100%</td>
</tr>
<tr>
<td>D</td>
<td>50%</td>
<td>95%</td>
</tr>
<tr>
<td>E</td>
<td>0%</td>
<td>36%</td>
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</table>

ICRB Classification

Images Courtesy of Jill Wells

Group A

Group B
ICRB Classification

Group C

Group C late
Group C: IAC
Shields et al. 2014
ICRB Classification

Images Courtesy of Jill Wells

Group D  Group E
Group D
Shields et al. 2014
Complications
Shields et al. 2014

Supra-orbital branch
LUL Ptosis
Complications
Shields et al. 2014
What’s your Dx?

Retinoblastoma
Coats Disease
Persistent Fetal Vasculature aka PHPV
Toxoplasmosis
Toxocara canis
Other
Retina at the Lens
Diagnosis?
MRI
CT
What’s your Dx?

Retinoblastoma

Coats Disease

Persistent Fetal Vasculature aka PHPV

Toxoplasmosis

Toxocara canis

Other
Histopathology
18 yo Adopted Asian M

Referred October 2006; 2nd Opinion
Exudative Maculopathy OS
Decreased vision OS x since June ’05
Dense Central Scotoma
Prior Tx

- Laser Tx Outside
  - x2
- Anti-VEGF
  - x3
PMH

Wisdom Teeth Extraction
Ocular Exam

OD
20/20
12
FTFC

OS
20/400
12
Constricted VF’s
What do you Recommend?

- More Cryotherapy
- More Anti-VEGF
- Both #1 and #2
- PPV w submacular extraction
- Other
PPV ?
<table>
<thead>
<tr>
<th></th>
<th>OD</th>
<th></th>
<th>OS</th>
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<tbody>
<tr>
<td></td>
<td>20/20</td>
<td></td>
<td>20/200</td>
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<tr>
<td></td>
<td>8</td>
<td></td>
<td>10</td>
</tr>
<tr>
<td></td>
<td>FTFC</td>
<td></td>
<td>Improved VF’s</td>
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Before

After
Any Other Questions?
Diagnosis?

Bardet Biedle

- BBS1 Mutation
- Auto Recessive
- Obesity, RP, polydactyly, Developmental delay, hypogonadism

Lawrence Moon

- no polydactyly
- no obesity
- spastic paresis

Bardet Biedle Lawrence Moon
Model Release Signed; Parental Permission Given
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Stickler’s Syndrome
9 yo WM w/ Seizure
2 yo Asian Female
Gliosis of Macula

Pigment changes
FA
Skin Lesions
Skin Lesions
Early Skin Lesions
Skin Changes
Adolescent Skin Changes
Adolescent Skin Changes
Dental Changes

Peg-Shaped Teeth
Caucasian Skin
Parental Model Release Signed
What’s your Dx?

Sjogren Larson Syndrome
Bloch-Sulzberger Syndrome
Incontinentia Pigmenti
Both #2 and #3
Bardet-Biedel Syndrome
What’s the Inheritance pattern?

- Autosomal Dominant
- Autosomal Recessive
- X-linked Dominant
- Mitochondrial
- Not Inherited
Incontinentia Pigmenti

Bloch-Sulzberger Syndrome
X-Dominant: Lethal in Males

Always?? . . .
Summary

Don’t Just Peek at a Kid!
Listen and Study Carefully
Family History is Important
Low Threshold for an EUA
Thank you for your attention!