Why is a regular ophthalmological exam critical in VHL?

Brian Madow, MD
Department of Ophthalmology, University of South Florida
VHLA Family Meeting in Tampa 2017

Screening

Screening is the testing of individuals at risk for, or who have von Hippel-Lindau disease (VHL) who do not yet have visual symptoms.

Eye exams:

Screening for the occurrence of VHL disease
Monitoring the known VHL disease affecting the eye

When is eye screening performed
Why is a Regular Ophthalmological Exam Critical in VHL

Ages 1-4
- Annually
  - Eye/retinal examination with indirect ophthalmoscope by an ophthalmologist skilled in diagnosis and management of retinal disease, especially for children known to carry the VHL mutation.

Ages 5-15
- Annually
  - Dilated eye/retinal examination with indirect ophthalmoscope by ophthalmologist informed about VHL.

Age 16+
- Annually
  - Dilated eye/retinal examination with indirect ophthalmoscope by ophthalmologist informed about VHL.

During Pregnancy (for women with VHL)
- Annually
  - Regular retinal checkup to anticipate potentially more rapid progression of lesions.
What are we looking for inside of the eye?

In 1904, Eugen von Hippel described a rare disorder of the eye.

- Old term: angiomatosus
- New term: capillary hemangioblastoma

Ball of small pipes (small blood vessels known as capillaries)

Benign vascular neoplastic tissue

- Non-malignant tumors of vascular origin
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Occurrence of retinal capillary hemangiomas in VHL disease has been reported to vary from 50% to 80%.

In every 10 people with VHL 5-8 people will have the “ball”
At what age is most likely to find the “ball”

- The mean age at diagnosis of retinal capillary hemangiomas in VHL disease is approximately 25 years.
- Most patients present between the ages of 10 and 40 years.
- The probability of developing a retinal capillary hemangioma increases progressively with age.

Most common locations in the retina
Photography begins with a complete explanation of the procedure by the examiner.

It is important to reassure that no retinal damage is caused by this procedure.

The camera flash is bright and the patient should know when to expect a flash. Since pictures will include the macula (area of central vision), it is normal to experience a blue or red tint to vision immediately following the flash.

This totally disappear within two to five minutes.
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Red-free photography

Fluorescein angiography
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1. Blue excitation filter
2. Yellow-green filter

How do we screen
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VISUAL ACUITY OF THE BETTER EYE IN PATIENTS AFFECTED WITH VON HIPPEL-LINDAU DISEASE, WITH OR WITHOUT OCULAR INVOLVEMENT

<table>
<thead>
<tr>
<th>BEST-CORRECTED VISUAL ACUITY</th>
<th>NO. (%)</th>
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<tbody>
<tr>
<td>20/20 or better</td>
<td>357 (88.1)</td>
</tr>
<tr>
<td>20/25 to 20/40</td>
<td>34 (8.4)</td>
</tr>
<tr>
<td>20/50 to 20/160</td>
<td>6 (1.5)</td>
</tr>
<tr>
<td>20/200 or worse</td>
<td>8 (2.0)</td>
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Who performs the screening?

How frequent should retina be examined?

Why is important to check the retina periodically?
Ball of small pipes (small blood vessels know as capillary)

Capillary leakage
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Puling (traction)

Bleeding

Cataract
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Ultrasound testing

Prevention of vision loss

Early, timely and continuous periodic retinal examination by retinal specialist with knowledge of VHL

Can Prevent severe vision LOSS