Monitoring the Patient with von Hippel-Lindau

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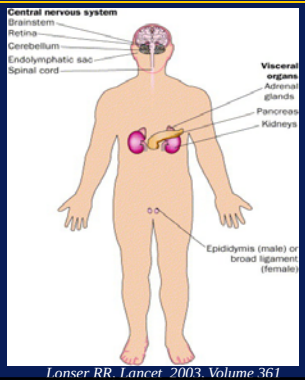
von Hippel-Lindau Disease

- **Genetic Mutation in *VHL***
  - Tumor suppressor gene
- **Dysregulation of angiogenesis**
  - growth of clusters of blood vessels and tumors/masses in various organs

Tumors in VHL

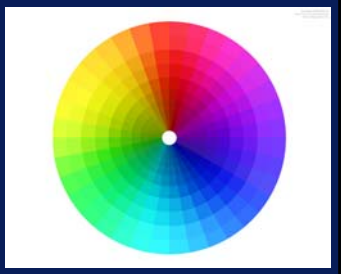


	Frequency
Hemangioblastoma	
retina	**25-60%**
cerebellum	**44-72%**
brainstem	**10-25%**
spinal cord	**13-50%**
Renal cell carcinoma	**25-60%**
Pancreas cysts	**17-56%**
neuroendocrine tumors	**8-17%**
Pheochromocytoma/PGL	**10-20%**
Endolymphatic sac (ear)	**11-16%**

Lonser RR. Lancet. 2003. Volume 361

Variation in Spectrum of Disease

- **Type of VHL mutation (Type 1, 2A, 2B, 2C)**
- **Other modifiers**
  - Genetic
  - Environmental
  - Lifestyle (diet, exercise, smoking)



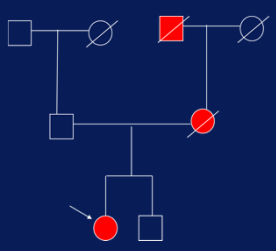
The Case for Screening in VHL

- "A stitch in time saves nine…."

Lesions are easier to treat when they are small

Genetic Testing for VHL

- Confirms the diagnosis
- Facilitates risk stratification for family members



Where to start?

VHL Alliance as a Resource

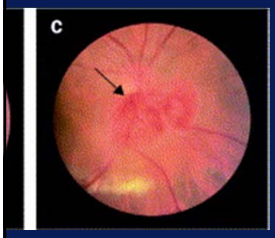
www.vhl.org

Guidelines for VHL Screening/Monitoring

Organ	Test	Age to Start	Frequency
Eyes	Exam by retinal specialist	1-4	annually
Ears/hearing	Audiology testing	1	Every 2-3 years
Adrenals	Plasma metanephrines and normetanephrines	Age 5	annually
Kidneys, Adrenals, and Pancreas	Ultrasound of abdomen	Age 8	Annually
	MRI abdomen	Age 16	At least every other year
Brain and cervical spine	MRI	Age 16	Every 2 years

VHL Handbook, 2012

Eyes

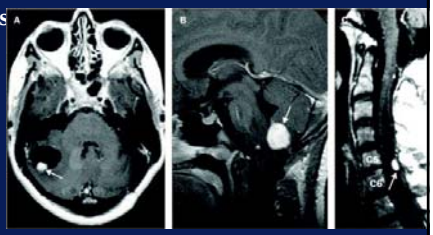


- **Hemangioblastomas**
- **Annual exams starting in infancy**
- **More frequent exams during pregnancy**

Image from Lonser RR, Lancet 2003, Volume 361

Brain and Spine

- **Hemangioblastomas**
- **MRI brain and c-spine every other year starting in teens**



Images from Lonser RR, Lancet 2003, Volume 361

Abdomen and Pelvis

- **Kidneys (renal cell carcinoma)**
- **Adrenals (pheochromocytoma)**
- **Pancreas (cysts, neuroendocrine tumors)**
- **Imaging at least every other year starting in teens**

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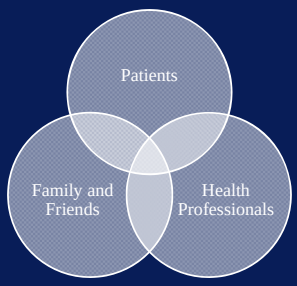
VHL Handbook, 2012

Special Considerations

- Symptoms (headache, dizziness, vision/hearing changes)
- What findings require intervention?
  - Threshold is often different in setting of VHL
- Timing and type of surgery depend on symptoms, rate of growth
- Pregnancy=close monitoring
  - Retinal exams, testing for pheochromocytoma, CNS lesions

Living with VHL: A Team Approach



Med/Surgical Subspecialists
- Neurology/Neurosurgery
- Ophthalmology
- Surgery (GI/Urology/ENT)
- Endocrinology
- OB/GYN, pediatrics

- **Genetic counselors**
- **Physical Therapists**

Care Coordination

- **Talk with your doctors**
  - VHL-specific screening and monitoring
  - Symptoms
  - Genetic testing
- **Seek opinions from experts, as needed**
  - VHL Alliance designated Clinical Care Centers
  - Genetics Clinics
  - NIH
- **Research/Clinical trials**

VHL Clinical Care Centers

www.vhl.org

Thank you
and
Welcome to Ann Arbor

www.uofmhealth.org