**10th International
VHL Medical Conference**

Day 2: Clinical Progress

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Director of Wellness
VHL Family Alliance

Effects and Treatments

- See new draft Handbook on USB drive
- Better ways to diagnose
- Better surgical approaches
- Not all tumors seen require treatment
- Not all "newer" treatments are "better" for VHL. Examine benefits **and limitations** in your situation.
  - Stereotactic radiosurgery
  - Radiofrequency ablation

Endocrine Diseases:
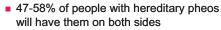## Dr. Pacak re finding pheos

- 50% are incorrectly diagnosed
- 35% are hereditary, 10 known genes
- Octreoscan not recommended (<50%)
- MIBG finds about 85%
- ^{18}F-FDOPA PET is best
- Dr. Pacak has an open invitation to get pheo testing at NIH – travel and lodging at your own expense; testing is free call 301-402-7785 Karen Adams

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Endocrine Diseases:
Adrenal Sparing Surgery

- Gennady Bratslavsky, Syracuse
- 47-58% of people with hereditary pheos will have them on both sides
- Preserve every salvageable scrap of adrenal you can
- Evolution of surgical practice –
- Standard of care is laparoscopic or robotic partial adrenalectomy

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Endocrine diseases:
Treatments for Pheos

- Camilo Jimenez, MDACC
- Not always in or near adrenals
- 5% abdomen, 2% chest, <1% HN
- Risk of metastasis under 3%
- Greater in larger tumors, especially paragangliomas
- Drugs, but no magic bullet
- Moral is: find them early

Endocrine diseases:
Pancreatic islet cell tumors

- Hartmut Neumann, Freiburg
- pNETs do not produce hormones
- Watch growth, risk rises with size
- MRI better than CT
- ^{18}F-FDOPA PET is best
- Working with Dr.Martin Walz in Essen to perfect endoscopic resection of tumors in the tail

CNS and Eye: Ophthalmic Management

- Dan Gombos, MDACC
- Laser is best; Larger lesions may require cryo
- <u>Early detection and regular management maintains control</u>
- Justapapillary tumors hardest
- Some drugs, no magic bullet
- Saving the eye vs. the vision

CNS and Eye: VHL and the nervous system

- Alexander Vortmeyer, Yale Univ
- History of research on eye
- Looked at the cell structure
- Same cell structure in the eye and brain and spinal cord
- Sven Gläsker: vasculogenesis
- Dr. Takayanagi re VHL vs sporadic spinal hemangioblastomas
- Dr. Kanno re occurrence in Japan

CNS and eye: Difficult neurosurgical issues

- Ian McCutcheon, MDACC
- Weigh risks of surgery or not
- Focused (stereotactic) radiation not recommended
- Hemangioblastomas represent only 2% (two percent) of all CNS tumors, therefore experience is hard to find.
- Syrinx in spinal cord will deflate once you remove the tumor feeding it.

CNS and Eye: ELST

- Marie Luise Bisgaard re audiology to get early warning of ELST
- Dr. Jose de Campos, Spain, re cochlear implant after ELST damage
- Early diagnosis and prompt action are critical to preserving hearing
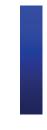
Renal Manifestations: Management of renal tumors

- Peter Pinto, NIH
- Before ANY surgery, check adrenal and CNS status
- Nephron-sparing enucleation, 3 cm rule
- Evolving techniques:
  - Radical nephrectomy
  - Partial nephrectomy
  - Laparoscopic
  - Robotic
- Balance risk of cancer with risk of dialysis
- RFA and cryo – caution: experience counts!

Renal manifestations: Hope from the drug world

- Anti-angiogenic drugs
- Angiogenic escape (Wood)
- New drug targets (Koh & Jonasch)
- Predicting outcomes through gene expression profiling (Monzon)

Prevention

- How can we prevent tumors from developing?
  - Early detection, appropriate treatment
  - Managing Stress (exercise, meditation, etc)
  - Nutrition, nutrition, nutrition
  - Sleep for self-healing
  - Environmental toxins
  - Attitude
- Recommend:
  - VHL Handbook v4
  - The China Study

The Take-Away Message

- YOU have a role in preventing tumors from developing
- It's not all fate
- Enroll children early in taking responsibility for their health
  - Make good choices (e.g. no smoking)
  - Learn to be an educated consumer
  - Reach out to get and give support
- AND help us fund research!

Teamwork for Health

Families and Health Care Professionals
working together
to improve diagnosis, treatment, and
quality of life
for individuals and families
affected with von Hippel-Lindau

VHL Family Alliance