Examples of Research Questions

- What is the natural history of these syndromes?
- Genotype/phenotype alignment?
- Why is there a difference in presentation between siblings?
- Are *de novo* mutations more aggressive?
- What is the role of epigenetics?
- What is the role of environmental factors?
- What role does lifestyle play?
  - Anxiety/Stress/Depression, Nutrition/Diet, Exercise
- Do medications impact tumor growth?
- Is thyroid disease associated with VHL?
- Does tumor aggression correlate with …?
- Does oral health correlate with …?
- Does treatment with targeted therapies or SRS modify the course of the disease?
- Does pregnancy modify the course?
  - What about other sources of hormones?
- Does early pre-symptomatic screening really help?

To participate, log on to

www.vhl.org/databank

VHL Alliance
THE CURE FOR CANCER IS IN OUR GENES

VHL Alliance
2001 Beacon Street, Suite 208
Boston, MA 02135-7787

Phone: +1-800-767-4845 x4
Fax: +1-858-712-8712
info@vhl.org
www.vhl.org

A member of

BEST IN AMERICA
INDEPENDENT CHARITIES OF AMERICA

BBB ACCREDITED CHARITY
bbb.org/charity

NORD
National Organization for Rare Disorders
2011 National Organization Member

CANCER IN OUR GENES

International Patient Databank

VHL Alliance
THE CURE FOR CANCER IS IN OUR GENES

A Partnership between Patients and Clinical Researchers

Patient participation helping to find a cure

VHL
BHD
HLRCC
SDHB

and other related cancers

www.vhl.org/databank

Goals of the CGIP Databank

- Create a unified resource for both researchers and participants
- Include a significant number of patients to allow meaningful statistical analysis.
- Give a complete "picture" of each VHL, HLRCC, BHD, SHDB patient
- Provide a natural history of genetic disorders leading to kidney and/or adrenal tumors
- Reveal correlations between known effects and other conditions and between diseases, learning from commonalities and differences
- Expedite matching patients to clinical trials

Why Multiple Conditions?

- VHL, HLRCC, BHD, and SDHB all increase the odds of getting kidney cancer.

  - Each is caused by a different genetic alteration, in a different gene
  - If any one of these four genes can cause kidney cancer, what do they have in common? What is it that they all help to control?
  - What can be learned from their similarities and differences?

Advantages of a Patient-Driven Databank

- Only the patient can provide the complete "picture" of their experience and quality of life
- Provides insights into lifestyle and quality of life (i.e., nutrition, exercise, mental outlook)
- Includes usage of both prescription and non-prescription medications
- Shows family history (blood relatives)

Data Privacy and Confidentiality

- The VHL Alliance specifically owns and controls the data
- Authorized third parties, including researchers, will receive only "**de-identified**" data
  - No names
  - No addresses or phone numbers
  - No other identifying information

Benefits to the Participants

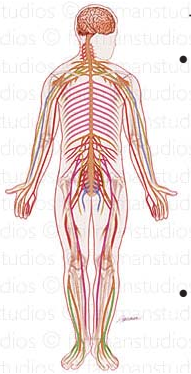
- Expedited matching to clinical trials
  - Participants will be notified of researcher interest by the VHLA and must initiate contact if they want to be considered for a clinical trial or other research project
- Documents on medical history and medications can be downloaded to share with physicians during office visits
- Optional Screening reminders sent to participants

Contribute Your Experiences to Advance Research

- Today researchers are limited to the data compiled by a single hospital or research team
- This clinician-sourced data provides only a limited picture of the disease, often focused on a single organ
- With the global CGIP databank, researchers have access to the annually updated compiled information of thousands of patients
- ***We need as many people as possible to contribute experiences and information!***

CGIP: Support of Clinical Trials

- Expedite Clinical Trials
  - Identify candidate patients
    - Includes questions recommended by the FDA for clinical drug trials
  - Linked to tissue bank
  - Provide natural history data for comparison
  - Help determine promising approaches for drug development
  - Define "normal" to know if the drug make a difference
  - Learn from all experimentation
  - Learn from off-label use experiences and interactions with other prescribed or over-the-counter drugs and supplements
- Better and more harmonized collection of outcomes



Global Rare Disease Initiative

- VHLA is working with NORD and others around the world to create a global repository for rare disease information
- Many sources of data will feed into this common data set
- Incorporates FDA feedback on questionnaire design