

## The VHL Patient: Putting It All Together

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## How do I find a genetic counselor?

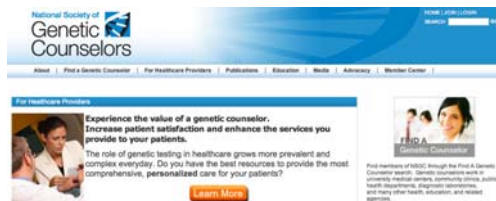
Ask your physician

## “Comprehensive”

Fact	Medical	Psychological
Multi-organ involvement	Multiple specialists	How do I manage my own care? Who is there to help me?
Chronic	-Frequent monitoring -Illness and wellness	-How do I adapt and cope differently through life's transitions? -Illness and wellness
Not predictable	-Cannot say if, when and where a tumor will form -Growth pattern unpredictable	What will my next scan show?
Hereditary	-Genetic condition -Impacts at-risk family members -Reproductive choices	Why? How do I tell my siblings/parents/children? How does this impact our decisions to start or grow our family?

## How do I find a genetic counselor?

[www.nsgc.org](http://www.nsgc.org)



## How can a genetic counselor help?

- ▶ Translate complex information
- ▶ Connect you with necessary medical specialists
- ▶ Help your family understand and adapt to
  - Medical information about the disease
  - Psychological implications
  - Familial implications
  - VHL disease
  - Recommendation for at-risk relatives to consider genetic testing
- ▶ Help you understand and adapt to a new diagnosis

## Initial Diagnosis of VHL

- ▶ VHL is diagnosed from clinical features, family history, and genetic test results
- ▶ Common reactions: fear, anxiety, surprise, anger, disappointment, surprise, sadness
- ▶ Having a firm diagnosis may bring relief
- ▶ May feel urgency regarding medical decisions
- ▶ May feel overwhelmed by medical information

## What May Impact News of VHL

- ▶ The person's age
- ▶ The person's current health status
- ▶ Whether or not the news was anticipated
- ▶ The person's familiarity with VHL
- ▶ The experiences of relatives with VHL
- ▶ The underlying reason for the diagnosis or genetic test

## What Relatives Need to Know



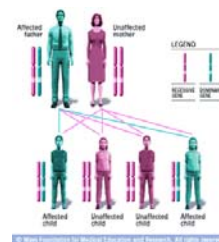
- ▶ Basic info about VHL
- ▶ The risk of having VHL
- ▶ Option of genetic test
- ▶ Where to get more info



## Option of Genetic Testing

- ▶ The VHL gene (3p25-26)
- ▶ Possible results:
  - Positive: The person has VHL
  - Negative: The person does not have VHL
  - Variant: The result is inconclusive
- ▶ Issues to Consider:
  - Logistics, including cost
  - Timing
  - Emotional readiness

## VHL is Dominantly Inherited

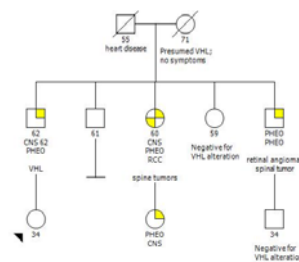


Each child has a 50% chance of having VHL and a 50% chance of not having VHL

## Risks & Benefits of VHL Testing

- ▶ Benefits of Testing:
  - May lead to better options for medical care
  - May clarify risk of VHL for children & other relatives
  - Allows relatives to have a targeted genetic test
  - May test negative (person does not have VHL)
- ▶ Risks of Testing:
  - May cause increased distress and worry
  - Discrimination concerns (although this is low risk)
  - May introduce tension in the family

## Genetic Pedigree



## Telling the News to Relatives

- ▶ Having to describe VHL and answer relatives' questions can be difficult
- ▶ Being "the messenger" can also be hard; relatives may have unpredictable reactions
- ▶ There may be issues with wanting to share the news, but also maintain your privacy
- ▶ It may be especially difficult (or impossible) to share the news with certain relatives

## Dana-Farber Cancer Institute

- ▶ Cancer Genetics & Prevention Center
  - Katherine Schneider, MPH, Genetic Counselor
  - Shelley McCormick, MS, Genetic Counselor
  - Dr. Huma Rana, MD, Cancer Geneticist
  - Toni Choueiri, MD, Renal Oncologist
  - Andrea Patenaude, PhD, Psychologist
  - Dr. Judy Garber, MD, MPH, Director
- ▶ To make an appt, please call:
  - Fatiana Cardoso at 617-632-2178



## Massachusetts General Hospital Cancer Center

### *VHL Clinical Care Center*

- Othon Iliopoulos, MD, Medical Director
  - Gayun Chan-Smutko, MS, Genetic Counselor  
[gchansmutko@partners.org](mailto:gchansmutko@partners.org)
  - ▶ To make an appointment, please call:  
Karen McDonough at 617-724-1971
- [www.massgeneral.org/cancer/about/ccra\\_rcc.aspx](http://www.massgeneral.org/cancer/about/ccra_rcc.aspx)

## How Genetic Counselors Can Help

- ▶ To discuss pros and cons of gene testing
- ▶ To determine insurance coverage for testing
- ▶ To arrange testing and provide support during and after the testing process
- ▶ To help you strategize about how to share the news about VHL with your relatives
- ▶ To help refer you to the appropriate medical specialists