Genetic counseling on VHL

Ignacio Blanco, MD PhD
Genetic Counseling and Clinical Genetics Program
Germans Trias Hospital
Badalona, Spain
Counseling of patients and closely related family members has to take a central place in management of hereditary diseases.
"Counseling" is a word that is often misunderstood.

In many countries, like Spain, it is a foreign concept.

Thus it is of importance to establish some common understanding of what counseling is and what it involves.

There is a difference between having a discussion, giving information, and counseling.
Counseling

- **Discussion** assumes that there is a dialogue, but does not imply that a method is used to ensure that the information being given by the health care provider is understood and accepted.

- **Providing information** is an integral part of all health care, but does not necessarily allow for a dialogue or an assessment of the patient’s understanding and beliefs.

- **Counseling**, on the other hand, is a way of addressing the implications of the information and of reaching a better understanding between the healthcare worker and patient about a range of issues.
Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.
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Genetic Counseling

This process integrates the following:

- **Interpretation of family and medical histories** to assess the chance of disease occurrence or recurrence.

- **Education** about inheritance, testing, management, prevention, resources and research.

- **Counseling** to promote informed choices and adaptation to the risk or condition.
Genetic Counseling

This process integrates the following:

- **Risk Assessment.**

- **Health education and health promotion.**

- **Counseling.**
Genetic Counseling is much more than to order a Genetic Test
Genetic Counseling. **Aims**

- Define and share the aims of the genetic counseling process.

- Being clear about the aims of genetic counseling in each circumstance is of first importance both for the person conducting the session and for the person seeking help or being informed.

- Clarity about the aims of counseling helps to achieve the tasks effectively in a limited amount of time.
The basic principles guiding practice include:

- Do not make assumptions about people’s wishes, beliefs, knowledge, or concerns, but ask questions;
- Approach the subjects with sensitivity in accordance with beliefs, religion, and family values.
- Set small goals for the session that are achievable;
- Remember that everything said has an impact so use words carefully and avoid jargon;
- Share responsibility with patients and do not try to reassure them about issues that cannot be resolved easily or where there is uncertainty.
Genetic Counseling on VHL

This process integrates the following:

- **VHL Risk Assessment**.
- **VHL Health education and health promotion**.
- **VHL Counseling**.

http://www.zazzle.es/mordeduras_de_von_hippel_lindau_camiseta-235733023758933821
Risk Evaluation

Family dynamics

Emotional aspects

Patient experiences about VHL

Patient experiences about screening procedures

http://olioshealth.com/services/electronic-medical-record-implementation/
http://shamaneileen.deviantart.com/art/Damian-s-Family-Tree-303067603
Increasing the knowledge about VHL improves patient’s autonomy and modifies attitudes about the disease.

Knowledge about carrier status enables early diagnosis and intervention.

In addition, knowledge decreases uncertainty and anxiety and assists in making decisions about lifetime plans.
Education about VHL

- VHL, its prognosis, and the therapeutic options;
- how VHL is transmitted;
- tests for diagnosis of VHL and carrier status and their reliability;
- procedure for giving information about test results;
- implications for future and existing children and other family members;
- Family planning;
- ...

To enable and encourage patients to be informed and to be part of the decision-making process about their own health.
Education about VHL

Information has to be tailored
Education about VHL

The VHL Handbook
What You Need to Know About VHL
A reference handbook for people with von Hippel-Lindau, their families, and support personnel
Edition 4.1 Revised 2014

VHL Handbook
Kids’ Edition
A Handbook for Parents and Kids Living with von Hippel-Lindau

http://www.vhl.org/wordpress/patients-caregivers/resources/vhl-handbooks/
Education about VHL

DNA = A Library

Genes = Books in a Library

Gene Mutation = A Spelling Mistake In A Book

Parent 1: With VHL
A = VHL Gene Mutation
B = VHL Working Gene

Parent 2: Without VHL
C = VHL Working Gene
D = VHL Working Gene

Each child gets one copy of the VHL gene from each parent.
In this way, there are four possible arrangements of these four genes:

AC: Child With VHL
AD: Child With VHL
BC: Child Without VHL
BD: Child Without VHL

Your genes make you special!

http://www.vhl.org/wordpress/patients-caregivers/resources/vhl-handbooks/
Education about VHL

<table>
<thead>
<tr>
<th>CNS</th>
<th>Ages at Diagnosis</th>
<th>Most Common Ages at DX</th>
<th>Frequency in Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Retinal hemangioblastomas</td>
<td>0-68 yrs</td>
<td>12-25 yrs</td>
<td>20-60%</td>
</tr>
<tr>
<td>Endolymphatic sac</td>
<td>1-50 yrs</td>
<td>16-28 yrs</td>
<td>11-16%</td>
</tr>
<tr>
<td>Cerebellar hemangioblastomas</td>
<td>9-78 yrs</td>
<td>18-25 yrs</td>
<td>44-72%</td>
</tr>
<tr>
<td>Brainstem hemangioblastomas</td>
<td>12-46 yrs</td>
<td>24-35 yrs</td>
<td>10-25%</td>
</tr>
<tr>
<td>Spinal cord hemangioblastomas</td>
<td>12-66 yrs</td>
<td>24-35 yrs</td>
<td>13-50%</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Viscera</th>
<th>Ages at Diagnosis</th>
<th>Most Common Ages at DX</th>
<th>Frequency in Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Renal cell carcinoma or cysts</td>
<td>16-67 yrs</td>
<td>25-50 yrs</td>
<td>25-60%</td>
</tr>
<tr>
<td>Pheochromocytomas*</td>
<td>4-58 yrs</td>
<td>12-25 yrs</td>
<td>10-20%**</td>
</tr>
<tr>
<td>Pancreatic tumors or cysts</td>
<td>5-70 yrs</td>
<td>24-35 yrs</td>
<td>35-70%</td>
</tr>
<tr>
<td>Epididymal cystadenomas</td>
<td>17-43 yrs</td>
<td>14-40 yrs</td>
<td>25-60% males</td>
</tr>
<tr>
<td>APMO or broad ligament cystadenomas</td>
<td>16-46 yrs</td>
<td>16-46 yrs</td>
<td>estimated 10% of females</td>
</tr>
</tbody>
</table>

*Includes the 20% of these tumors that occur outside the adrenal gland, also called paragangliomas.
**Frequency of pheochromocytoma varies widely depending on genotype.

Visual Art: © 2013 The University of Texas MD Anderson Cancer Center

Risk Communication

S. Richard et al. / Seminars in Cancer Biology 23 (2013) 26–37

Truncated protein ++
or deep missense mutations

Surface missense mutations ++

HIF level

http://norma-ohsas18001.blogspot.com.es/2013/06/analisis-preliminar-de-riesgos.html
Risk Communication

- It is important that patients understand what risk is and how it can be altered.
Risk Communication

- Difficulties in communicating **diagnostic information** are inherent in doctor-patient interactions, where **specialized knowledge** has to be **interpreted** and **understood** in a context quite different from that in which the knowledge was created.

- The difficulties in such task are further exacerbated when the **diagnosis** is a **risk** for a severe disease with a variety of **clinical manifestations**.

- In scientific contexts, risks are calculated on the basis of a variety of systematically established factors; while the risk judgments of individuals are assumed to be influenced to a greater extend by **personal or familial experiences, moral values** and **social norms**.
Risk Communication. Be Prepare

Where?

What are my chances of developing cancer, doc?

When?
Risk Communication. *Be Prepare*

Share responsibility with patients and do not try to reassure them about issues that cannot be resolved easily or where there is uncertainty.
Family Planning on VHL

- Sensitive Genetic Counseling
- Discuss all the options:
  - Adoption
  - Sperm or egg donation
  - Having children, testing after
  - Having children, testing prenatally
Family Planning on VHL

Transcervical Chorionic Villus Sampling
- Catheter taking tissue samples
- Ultrasound Transducer
- Uterus
- Placenta
- Fetus

Amniocentesis
- Ultrasound Transducer
- Amniotic Fluid
- Fetus
- Placenta
- Uterus

Non-invasive prenatal diagnosis of single-gene disorders from maternal blood

http://www.hopkinsmedicine.org/healthlibrary/test_procedures/gynecology
Family Planning on VHL

- Sensitive Genetic Counseling

- Discuss all the options:
  - Adoption
  - Sperm or egg donation
  - Having children, testing after
  - Having children, testing prenatally
  - Pre-implantation genetic diagnosis
Family Planning on VHL

Step 1: Ovulation induction
Step 2: Oocyte aspiration
Step 3: Fertilization and Embryo Culture
Step 4: Polar body removal and/or blastomere biopsy
Step 5: Preimplantation Genetic Diagnosis in blastomeres
Step 6: Embryo transfer and implantation

Biopsied cell

Affected

Transfer only unaffected embryos to the patient


http://www.nature.com/scitable/content/principles-of-preimplantation-genetic-diagnosis-7871
Family Planning on VHL

Polar body-based preimplantation genetic diagnosis for Mendelian disorders

Anver Kuliev* and Svetlana Rechitsky

Reproductive Genetics Institute, 2825 North Halsted St., Chicago, IL 60657, USA
Family Planning on VHL

- Individuals have options depending on personal preferences and circumstances.
- We have to encourage patients to explore options with their partners, doctors and genetic counselors.
Emotional aspects on VHL

- Screening for tumors associated with VHL is complex and involves multidisciplinary care and the monitoring of multiple organ systems over a lifetime.

- Screening may commence in children as young as 2 years of age.

- Within the same family individual family members may show one or several features of the disease.

- Consequently, there is a great deal of **unpredictability** regarding how and **when** the disease will manifest across family members.
Emotional aspects on VHL

- Lifelong commitment to various medical screening regimens may generate diverse emotional responses, including denial, anger, fear, sadness and anxiety, and may influence one’s sense of self, particularly for children, adolescents, and young people.

- The ways in which patients think and feel about VHL, and the meaning of lifelong surveillance, may inevitably influence their ability to accept and practice behaviors that promote health and wellbeing.

- It is very important to explore and address the emotional aspects during the genetic counseling process
Emotional aspects on VHL

- While there is currently a good deal of information available on the psychosocial impact of the more common forms of hereditary cancer, there are few data available on the psychosocial impact of hereditary cancer syndromes in which individuals are at high risk of developing multiple tumors at various sites and ages with limited preventive options, as is the case with VHL.
Forty-eight families with a VHL mutation: 123 family members (carriers, 50% at-risk, non-carriers).

40% of the VHL family members reported clinically relevant levels of distress,

50% among the carriers

36% among the non-carriers

Having lost a first degree relative due to VHL during adolescence was related significantly to heightened levels of distress.

Only one-third of those who reported heightened levels of distress had received professional psychosocial support.
Emotional aspects on VHL

Approximately **one-quarter of the partners** exhibit clinically relevant levels of distress.

Levels of distress and worries of the partners and their high-risk **spouse** were **significantly correlated**.

The majority of partners (76%) believed that professional psychosocial support should be routinely offered to them.

The distress levels of the ‘patient’ could potentially be used to identify partners at risk of developing clinically relevant levels of distress.
Despite well-established protocols for the medical management of VHL, families reported a range of emotional, social, and practical challenges, including:

- sustained **uncertainty** about future tumor development,
- **frustration** regarding the need for **lifelong screening**, 
- **strained family relationships**, 
- **perceived isolation** from peers and colleagues, 
- **limited career opportunities**, 
- **concerns** about **financial security**, 
- **complex decisions** in relation to **childbearing**, and 
- **difficulties** gaining access to appropriate and timely support.
New forms for connecting VHL patients and their families with holistic, empathic, and person-centered medical and psychosocial care are urgently needed.
Ethical Considerations
Ethical Considerations

- Ethical considerations include human rights, issues surrounding consent, and those pertaining to confidentiality.

  *These apply to genetic counseling in both national and international contexts and vary according to the legal and social customs in particular settings.*

- Concerns about disclosure of personal information (such as the results of genetic testing) to others (family members, insurance companies, etc.) and maintenance of confidentiality can raise difficulties for some individuals.
Ethical Considerations

- Although individuals have a right to confidentiality regarding personal issues, this raises concerns about the rights of those who might be affected through inheriting genetic disease to such pertinent information.

  Uninformed family members might suffer in different ways. i.e., someone who does not know he has VHL might lack appropriate medical care and treatment.

- Disclosure of information by genetic counselors to family members should uphold accepted standards of confidentiality by encouraging individuals to disclose VHL inheritance themselves to those who might benefit from the information.
Ethical Considerations

- Key issues in VHL are diagnosis of symptomatic children and adults, and presymptomatic testing of those who may benefit from preventative management, including children.
Conclusions

- Genetic counseling, including risk assessment, health education and counseling, is an essential aspect of VHL care.

- The laboratories offer technical knowledge and expertise whilst genetic counseling can translate this knowledge into practice to help those affected make more informed decisions. Having one without the other is an inefficient way forward.

- It is important to achieve a balance between giving information and discussing the implications so that people do not leave confused and unclear about the main issues for them.
Conclusions

- Childbirth and disability are sensitive issues which are influenced by family, practical, social, religious, and personal beliefs. Genetic counseling is complex because it covers both factual and non-factual issues.

- In developing countries, genetic counseling is one relatively inexpensive way to help people deal with VHL whilst conditions for diagnosis and treatment improve.

- This might seem hard to do, but offering people time to explore the issues is a first step towards helping them cope in their particular situation.
Thanks

iblanco.germanstrias@gencat.cat