VHL & Family Planning:
Ethical Considerations & the Importance of Counseling

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This presentation is derived from my dissertation research submitted to King’s College London in 2015.
Children of VHL-positive couples

50% chance of inheritance
Infertility

Tumors of the epididymis may block sperm and consequently cause infertility.

VHL Alliance 2014

Progesterone (P) → Tumor Growth

Some research suggests that high exposure to P (ie: during pregnancy) or sustained exposure to P (ie: from P-based contraceptives) may be associated with increased VHL tumor manifestation / growth.

Adekola et al. 2013; Franzten et al 2012
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Can I have children?

SHOULD I have children?

Are there other questions I should be asking?

Tumor Growth

Are there other questions I should be asking?
REPRODUCTIVE CHOICE

What are the options? Who gets to choose?
The Alternative Options:

- **PnGT**: Prenatal diagnosis (PnGT) with termination of VHL-positive fetus(es)
- **IVF (+PGD)**: In vitro fertilization (IVF) with pre-implantation genetic diagnosis (PGD), plus selective implantation of VHL-negative embryos
- **Adoption**: Acquiring existing children instead of reproducing (creating new children)
“I always hoped to have children one day, but I risk passing on VHL and that [having children] may be both selfish and irresponsible....

VHL has forced me to take a more pragmatic approach to the whole procreation thing and to plan the best possible future for a child....

I know my father feels incredibly guilty about passing his illness on, even though it was not in any way his fault. I feel lucky that science and technology have given me the opportunity to at least try to have children without passing on VHL.”

SenGupta et al., 2012, p103
EUGENIC CERTIFICATE

THIS GUARANTEES that I have examined the sender of this card and find a perfect Physical and Mental Balance and unusually strong Eugenic Love possibilities, well fitted to promote the happiness and future welfare of the race.

3/0/10.

Sara S. Salkin, M.D.

Courtesy of the Robert Bogdan Collection
Is this negative eugenics?

Some say NO.
These are no longer governmentally mandated but rather individual decisions (Habermas 2003).

Some say YES, but with a revised goal.
The “aim is, first and foremost to relieve... all human beings of the burdens of genetic disease” (Buchanan 1996).
PnGT: Prenatal Genetic Testing

The original option?
Prenatal Genetic Testing (PnGT)

Two methods:

• Amniocentesis (mid 2\textsuperscript{nd} trimester)
• CVS (late 1\textsuperscript{st} trimester)

"To alleviate the "humiliation" that women faced in obtaining post-amniocentesis "therapeutic abortions".

Cowan 1993, p14
Why are women humiliated?

“Earlier diagnosis seemed particularly necessary because of the social and religious attitudes of some ethnic groups at risk. For instance, British Muslims originating from Pakistan are as distressed as anyone else by having children suffering from [a rare genetic disease], but most find mid-trimester diagnosis and abortion unacceptable. However, they expressed a lively interest in the possibility of first trimester diagnosis.”

Cowan 1993, p15 – quoting Modell 1986
Why are women humiliated?

“Earlier diagnosis seemed particularly necessary because of the social and religious attitudes of some ethnic groups at risk. For instance, British Muslims originating from Pakistan are as distressed as anyone else by having children suffering from [a rare genetic disease], but most find mid-trimester diagnosis and abortion unacceptable. However, they expressed a lively interest in the possibility of first trimester diagnosis.”

Is PnGT “appropriate” for VHL?

“Requests for [PnGT] for conditions which (like VHL) do not affect intellect and have some treatment available are not common. Differences in perspective may exist... regarding the use of [PnGT], particularly if the testing is being considered for the purpose of termination rather than early diagnosis.

Although most centers would consider decisions about prenatal testing to be the choice of the parents, discussion of these issues is appropriate.”

Frantzen et al. 2012, VHL Chapter of Gene Reviews
Most women who are given a positive test result terminate their pregnancy (Press 2008).

Women who actively choose to continue a pregnancy with a positive test result may be socially shunned (Lippman 1991, p28).

Within pediatric medicine, it is widely recommend to wait to perform genetic testing until the patient can consent as a competent adolescent / adult (Borry et al. 2006).
“Expressionist Objection”
Buchanan, 1996

What do these technologies say about VHL-positive individuals?

1. The lives of individuals with disabilities are not worth living.
2. Only perfect individuals should be brought into the world (and imperfect individuals have no right to exist.)

Buchanan 1996, p28
PnGT: an undesirable contingency plan

Decision-making within the context of this contingency plan is
“extremely burdensome for both the couple and the professionals involved.”
IVF WITH PGD

A more ethical option? (Cameron & Williamson 2002)
IVF Counseling Agenda

- The “mechanics”
- The welfare of the child
- “Spare” embryos
- Questions?
[Legally] we must consider the welfare of any existing or future child(ren) born as a result of treatment. As part of this initial assessment, we will ask you to complete a questionnaire. If any information given raises concerns, these will be discussed with you and you will be asked to meet with a counsellor. Very occasionally, it may be necessary to delay or cancel your treatment.

National Health Service, 2008, p30
Welfare of the Child

VHL-positive or VHL-negative, the child will still be VHL-affected because YOU have VHL.
‘Spare’ embryos

• ‘Spare’ == ‘suboptimal’ or in excess
• ‘Utility’ of the fresh embryos

“Couples should not be approached for the donation of fresh embryos to research... subject to some exceptions [such as PGD-positive embryos, because]... a couple who has chosen PGD for a serious inheritable condition is unlikely to wish to have an ‘affected’ embryo transferred or frozen and is more likely to consent to such embryos [for research]. “

Scott et al. 2012, p43-45 – emphasis in original
ADOPTION

The forgotten option?
External limitations?

Assisted Reproductive Technology (ART) is marketed as enhancing reproductive choice, [but] many feminists counter that ART actually narrows choice by discouraging alternatives like child-free living and adoption.

“People who emphasize the importance of genetic background... [and those] who perceive potential genetic problems with adoptees will be less willing to adopt a child.”

(Bausch 2006, p50, p60)
Is there anything ELSE we should consider?
Concluding Thoughts

• NO reproductive option guarantees the genetic outcome of the child.

• An “informed decision” requires understanding of complex ethical considerations.

• Social factors impact the ethical frameworks from which a patient (or counselor) approaches family planning.

• Patients may benefit from comprehensive counseling.
**Acronyms**

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<tr>
<th>Acronym</th>
<th>Description</th>
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<tr>
<td>ART</td>
<td>Artificial Reproductive Technology</td>
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<tr>
<td>IVF</td>
<td>In-vitro Fertilization</td>
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<td>P</td>
<td>Progesterone</td>
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<td>PGD</td>
<td>Pre-implantation Genetic Diagnosis</td>
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<td>PnGT</td>
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