Cancer Survivorship Symposium

Cancer and Heredity

January 16, 2017

Jeanne P. Homer, MS
Licensed Certified Genetic Counselor
Outline

• Cancer and Heredity
• Hereditary Cancer Risk Assessment & Genetic testing at Hoag
• A case study of a family with Hereditary Breast and Ovarian Cancer
Outline

• Cancer and Heredity

• Hereditary Cancer Risk Assessment & Genetic testing at Hoag

• A case study of a family with Hereditary Breast and Ovarian Cancer
Hereditary Cancer

How frequent is it?

Sporadic 90%
Hereditary 10%
Review of Genetics

• 60-second review
Genetics
Chromosomes and Genes
Chromosomes and Genes

BRCA1

BRCA2
RETIRED BLUES: HOW SAFE IS YOUR PENSION?

The Hunt for the Breast Cancer Gene

Jo Cunningham, 59, had breast cancer. Her daughter Julie, 29, had a preventive mastectomy. Will Alexandra, 5, be spared?
THE ANGELINA EFFECT

Angelina Jolie's double mastectomy puts genetic testing in the spotlight. What her choice reveals about calculating risk, cost and peace of mind

BY JEFFREY KLUGER & ALICE PARK
Hereditary Cancer Genes

- BRCA2
- BRCA1
- MYH
- FH
- MSH2
- MSH6
- MLH1
- VHL
- APC
- PMS2
- ATM
- MEN1
- SDHD
- RET
- PTEN
- CDH1
- p53
- BRCA1
- STK11
- CHEK2
DNA
Mutation

Nonfunctional or missing protein
Dominant Inheritance

mutation

Aa   Aa   aa

aa   Aa   aa   Aa
Hereditary Cancer

What are the implications?

Sporadic 90%
Hereditary 10%
Features of Hereditary Cancer

- Higher chance of cancer

2-8% vs. at least 50%
“I’ve already had cancer”

- High chance of a second cancer
Your family
Two things that may be worse than getting cancer

– Getting it again
– Passing it to your children
Why would you want to know?

• Screening

• Chemoprevention

• Surgery
Hereditary Cancer

How to tell if it’s hereditary?

- Sporadic: 90%
- Hereditary: 10%
Genetic Testing
Red Flags
Features of Hereditary Cancer

- Young age at diagnosis
Features of Hereditary Cancer

• Person diagnosed with cancer twice
Features of Hereditary Cancer

- Rare tumor or cancer

“once in a blue moon”
Features of Hereditary Cancer

- Multiple family members on the same side of the family with the same or related cancers
Ashkenazi Jewish Ancestry
(breast, ovarian, prostate, pancreatic and colon only)
Outline

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• A case study of a family with Hereditary Breast and Ovarian Cancer
Hereditary Cancer Program Team
Making an appointment
### FAMILY HISTORY FORM

**YOUR BROTHERS AND SISTERS**

Please include only sisters and brothers who are related by blood. For brothers and sisters who are half-siblings, write in who the parent you share (e.g. half sister through mother).

<table>
<thead>
<tr>
<th>NAME</th>
<th>M/F</th>
<th>Living</th>
<th>Current Age (if living) or Age at Death (if deceased)</th>
<th>Cancer Diagnosis</th>
<th>Location or Type</th>
<th>Age at Diagnosis</th>
<th>Other Cancer</th>
<th>Location or Type</th>
<th>Age at Diagnosis</th>
<th>Cause of Death</th>
<th>COMMENTS</th>
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<tr>
<td>Judith</td>
<td>F</td>
<td>Yes</td>
<td>45</td>
<td>Yes</td>
<td>Breast</td>
<td>42</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Christine</td>
<td>F</td>
<td>Yes</td>
<td>42</td>
<td>Yes</td>
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<td>40</td>
<td></td>
<td></td>
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<tr>
<td>Helen</td>
<td>F</td>
<td>Yes</td>
<td>40</td>
<td>No</td>
<td></td>
<td></td>
<td></td>
<td></td>
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<td></td>
</tr>
</tbody>
</table>
Please answer the questions below in order to begin constructing your family tree.
Please provide the number of blood relatives you have for each of the following relations.
Your parents and grandparents will be automatically added. You can add half siblings and other family members later. Be sure to include both living and deceased family members.

Just a reminder: If you need time to gather information on family members, click the "Save, Finish Later" button on the top-right corner of the screen.
When you log back into the questionnaire, you will return to the same place you left off.

- Brothers
- Sisters
- Sons
- Daughters
- Paternal Uncles (your father's brothers)
- Paternal Aunts (your father's sisters)
- Maternal Uncles (your mother's brothers)
- Maternal Aunts (your mother's sisters)
The Family Tree

Grandparents

Parents

Children
Review Medical Records
Analyze for all known hereditary cancer conditions
Computer analysis
Consultation
Genetic Counseling – what it is

- Explain genetics & answer questions
- Analyze family
- Risk assessment
- Arrange genetic testing
- Explain results
- Written summary
Genetic Counseling – what it isn’t Therapy
Genetic Testing
New Genes
Gene Panels

- BRCA1: 40
- BRCA2: 23
- Other genes: RAD51C, RAD50, PALB2, NBN, MSH6, MRE11, CHEK2, BRIP1, BARD1
Genetic Testing - Cost

2013
$4000+

2017
$475
Follow-up

• Plan for surveillance
• Keep in touch
DNA banking
What Genetic Counselors Do

- Collection and documentation of detailed family history of cancer
- Review of appropriate medical records
- Pedigree assessment and recognition of cancer susceptibility syndromes
- Calculation of hereditary cancer risk
- Explanation of inheritance pattern and implications for family members
- Assistance in exploring the medical and psychological implications of genetic testing
- Review of costs, benefits and limitations of genetic testing
- Provision of informed consent, including issues of privacy, confidentiality and legal protections against genetic discrimination
- Determination of best strategy for testing within the family
- Help in obtaining insurance coverage for the testing
- Selection of appropriate genetic test and testing laboratory
- Sample collection (blood or saliva)
- Interpretation of genetic test results, which can be complex
- Provision of ongoing emotional support and assistance in informing family members
- Assistance in developing a plan for cancer screening and risk reduction, when appropriate
- Detailed summary letters sent to patients and their physicians
- Referrals to research and national support organizations, when appropriate
- For those with BRCA-positive results, participation in the Hoag-sponsored Orange County quarterly education meetings
GINA

Genetic Information Nondiscrimination Act
– Signed into federal law in 2008
– Provides protection from discrimination based on genetic status in health insurance and workplace
Outline

- Cancer and Heredity
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Positive (Deleterious Mutation)

Nonfunctional or missing protein
Options for BRCA carriers

• Screening

• Chemoprevention

• Surgery
...Or not
Jeopardy!
JEOPARDY!
Most cancers are hereditary. True or False
False

- Sporadic: 90%
- Hereditary: 10%

(Chart showing the percentage distribution between Sporadic and Hereditary cases.)
Which test is more accurate: blood or saliva?
Both tests are accurate
Do people with cancer gene mutations always get cancer?
No
Breast cancer genes only come from the mother’s side: True or False
False
50% of the time, the gene mutation comes from the father
Genetic testing costs thousands of dollars and is never covered by insurance.
False
(but it used to be true)
Can I get genetic testing even if I don’t meet insurance criteria?
Yes,
(out-of-pocket cost of $100-$475)
Genetic Counseling appointment is scary: True or False
False
Would my insurance cover genetic counseling?
Yes
Genetic counseling is covered by most insurance plans (exception is Medicare)
Would I lose my insurance if I have the “gene”?
No.
LAW (GINA)
CONGRATULATIONS!
Thank you

Please feel free to contact us for more information

How Genetics Works
Questions?