Cancer Genetics: Genetic Counseling, Ethical Issues and the Nurse

PART 1

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Advances through genetics and genomics research have begun to revolutionize health care

Genetic and Genomic Influences Across the Health Care Continuum

- Genetic and genomic science is redefining the continuum of human health and illness.
- There are over 1000 different genetic/genomic tests available.

Availability of Genetic Testing

- Prenatal diagnosis
- Carrier testing
- Confirmation of a genetic diagnosis
- Genetic susceptibility

Awareness of Genetic Risk

- Facilitate informed health care decisions
- Pre-implantation genetic diagnosis
- Promote risk reduction behaviors to reduce morbidity and mortality
- Risk reduction surgery
- Identify at risk individuals

Demand for Cancer Predisposition Testing

- Advances in molecular genetic technology have made testing for some hereditary cancer mutations available
- Press coverage of scientific advances has created consumer awareness of testing availability
Direct to Consumer Marketing

Patients will have questions.
Patients will need education.
Ethical Issues Arise.

Introductory Question

- What is your framework for ethical decision making?

1. There are some absolute rights and wrongs.
2. Everything must be taken in context.

Ethical Principles

- Autonomy
  - Right to Self Determination—Informed consent
  - Right to Know vs. Right not to Know
  - Confidentiality—When are Individual Rights Superseded?
  - Maintaining Sense of Self
  - Non-directiveness vs. Prescription for risk management

Ethical Principles

- Beneficence
  - Altruism vs. Paternalism
    - Do the right thing vs. the best thing
    - Who decides what is right or best
  - Best for whom?
    - Individual
    - Provider
    - Family
    - Common good
  - Risk of discrimination

Ethical Principles

- Nonmaleficence
  - Do no harm
    - Licensing vs. certification
    - Up-to-date, evidence based practice
    - Scope of practice and Practice standards
  - Do the least harm
    - Harm to whom?
      - Patient
      - Relatives
      - Society
      - Institution/business
      - Self
    - Protect from harm?
      - Ongoing care for lifelong risk
Ethical Principles

• Justice
  – Fairness vs. Equity
  – Getting What is Deserved vs. Inalienable Rights
  – Cost vs. Outcome
  – Who pays
  – Access

• Veracity
  – Right to be told the truth
  – Balanced presentation of information

• Fidelity
  – Right to confidentiality
  – Expectation that care will be provided
  – Advocacy—patient’s best interest is promoted
  – Respect for personhood
  – Dignity of the individual

Ethical Frameworks for Decision Making

• Consequential Utilitarianism Model
  – Casuistry
  – Situational Ethics
  – Case by case consideration with the implication that a paradigm case exists
  – Outcomes are the focus: Most good and least harm

• Nonconsequential Deontological Model
  – Normative Ethics
  – There are absolute rights and wrongs
  – Decisions are based on unchanging principles

CASE STUDIES

CASE STUDIES

Case Study 1

• 29 year old unaffected female
• Community wellness clinic
• Breast findings
• Compelling family history of breast and pancreatic cancer.

  – “She could use some conversation about her family history – would testing help her?”
Who should have testing?

Ethical Issues

- Does BF need testing?
- Will results change her care?
- Could other family members be helped by the information?
- How much does it cost?
- Who will pay?
- Does she want testing?
- What are the protections for confidentiality?

Personalizing Management

- Multigene Panel testing
- Prior to biopsy
- Biopsy positive
- No Pathogenic mutation
- Concerns about ovarian cancer potential
- 2 children
- Single mother

Risk Management Guidelines/Recommendations

- Family history evolving
- Consider bilateral mastectomies
- Consider bilateral oophorectomies
- Increased risk of breast, prostate, ovarian, melanoma, pancreatic cancers with known BRCA2 pathogenic mutation.
- BRCA 2 Variant of uncertain significance (VUS) = insufficient data for guidance in management modification (reclassification?).

Question 2

- Should all unaffected women be tested for hereditary breast and ovarian cancer?
  - Yes
  - No
Case Study 2

- 73 year old female with ovarian cancer
- Referred for hereditary cancer risk consultation by Gyn/Onc Surgeon
  - “Does she need BRCA 1 & 2 Testing”

Question 3
A 73 year old with ovarian cancer. Does she need genetic testing?

- A. No
- B. Yes
- C. Depends on family history

Question 4
If genetic testing is offered, what gene(s) should be tested?

- A. BRCA 1 & 2
- B. Lynch Syndrome genes
- C. Depends on family history
- D. Depends on insurance
- E. All of the above must be considered
**Ethical issues**

- Does she need testing?
- Will results change her care?
- Could other family members be helped by the information?
- How much does it cost?
- Does insurance cover?
- What is the cost to society as a whole?
- Does she understand the issues & give true informed consent?
- Does she want testing?
- What are the protections for confidentiality, any risk of loss of health care insurance coverage?

**Personalizing Management**

Further exploration of family history revealed a 30 year old maternal aunt had survived colon cancer, died in her 80s with no subsequent cancer.

Recommendations for screening in PMS2 mutations to begin colonoscopy at age 30 must be amended based on this family history.

Herding cats: getting all family members to come in for testing, come back for test results, follow-up with screening recommendations, maintaining confidentiality when some want to tell all and others don’t want to talk about it.

**Risk Management Guidelines/Recommendations**

- PMS2 mutations carriers start surveillance later than other Lynch Syndrome mutations (moderated by family history)
- Upper GI and Small bowel exams not needed unless family history indicates.

**Cancer Genetics: Genetic Counseling, Ethical Issues and the Nurse**

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**Question 5**

- Does your facility/practice have a process for referral to genetic counseling or testing?
  - Yes
  - No
  - OPINION
Question 6

- Identify the most significant barrier to genetic counseling and testing in your practice.

  A. Distance
  B. Insurance coverage
  C. Access to genetics professionals
  D. Knowledge about when to refer for counseling
  E. None of the above

- Opinion

Drivers of Change

- NCCN Guidelines
- American College of Surgeons, Commission on Cancer Standards
- USPSTF recommendations
- Supreme Court decision re: Myriad patents
- Angelina Effect

Most cancers are not inherited

- 10-15% familial
- 5-10% hereditary
- 75-85% sporadic

Red Flags of Hereditary Cancers

- Early age at onset
- Bilateral or multiple primaries
- Multiple FM affected
- Rare cancers
- Founder families
- Patterns
Explore the Family Tree

• Three generations – maternal and paternal
• Major health conditions
• Age developed disease
• Age and cause of death
• Ethnic background
• Pregnancy problems
• General lifestyle of family

Relatives……
Who to Include?

• First degree = 50% of genes
  Parents, siblings, children

• Second degree = 25% of genes
  Aunts, uncles, grandparents, nieces, nephews

• Third degree = 12.5% of genes
  Cousins, great-uncles, great-uncles, great-grandparents

Limitations

• Privacy issues
• Small family
• Accidental deaths
• Absence of targeted organ
• Male dominance in female hereditary syndrome
• Adoption/Separation

Hereditary Syndrome Patterns

• Hereditary = known genetic predisposition for cancer

• Assumed or putative hereditary = hallmarks and features of syndrome

• Familial = not clear pattern, may be nature/nurture

• Sporadic = acquired mutation or environmental exposure

Misconceptions About Family History

• “Cancer on the father’s side of the family doesn’t count.”

• “Ovarian cancer in the family history is not a factor in breast cancer risk.”

• “The most important thing in the family history is the number of women with breast cancer.”

Misconceptions About Family History

• Cancer on the father’s side of the family doesn’t count
  – Half of all women with hereditary risk inherited it from their father

• Ovarian cancer is not a factor in breast cancer risk
  – Ovarian cancer is an important indicator of hereditary risk, although it is not always present

• The most important thing in the family history is the number of women with breast cancer
  – Age of onset of breast cancer is more important than the number of women with the disease
Role of the Nurse - in general

Assess personal and family history
Educate
Initiate referrals
Personalize surveillance and management.
Evaluate the holistic impact of health condition, therapeutics and testing on client and family.
Expand professional knowledge and expertise through continuing education.

Role of the Nurse - in genetics

Assess personal and family history for increased genetic risk.
Educate about basic genetics to clients and families.
Initiate referrals to credentialed genetics professionals for counseling.
Personalize surveillance and management with genetic information.
Evaluate the holistic impact of genetic condition, therapeutics and testing on client and family.
Expand professional knowledge and expertise through continuing education in genetics and genomics.

Family History Collection: Tree Hugging 101

Make it a part of your routine
Ask the question: Do you have a family history of cancer?
Clarify - maternal and paternal family history
Ask the question again more specifically: Does anyone in your family have a history of breast, ovarian, colon cancer, colon polyps, or other cancers?
Ask the question again at follow up visits, as family histories change over time.

When Should Genetic Counseling or Testing Be Considered?

Significant family cancer history
Reasonable likelihood of carrying a mutation (affected first)
Results will influence medical management
Patient wants information (empowerment)

Many Testing Options

FOR BREAST CANCER
BRCA1 and BRCA2 only
Panel for “actionable genes” – with published screening guidelines: BRCA1, BRCA2, P53, CDH1, STK11, PTEN
Comprehensive panel (any gene that has been associated with a hereditary breast cancer family): BRCA1, BRCA2, P53, CDH1, STK11, PTEN, ATM, BARD1 BRIP1, MRE11A, MUTYH, NBN, PALB2, RAD50, RAD51C

FOR OTHER CANCERS
Single gene testing (i.e. APC only, MLH1 only, MSH2 only, etc.)
Panel for multiple genes associated with that particular cancer

Types of Testing

Sequencing – looks for the "single" nucleotide changes
Rearrangements – looks for large rearrangements of the gene ("BART")
Multisite – targeted mutation test typically done in an particular ethnic group
Single site – for a known mutation in a family
**Possible Results**

**Positive** (informative)

**Negative**
- True negative
  - Negative result when family mutation known
- Negative result in affected person
  - Different gene?
  - Can't find mutation?

**Uninformative**
- Negative in unaffected individual
- Variant of uncertain significance
- Additional information/testing needed

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**Question 7**

What does a “positive” genetic testing result mean?

A. A positive test result means that the person does not have a genetic mutation.
B. A positive test result means that the person has a genetic mutation that increases cancer risk.
C. A positive test result means that the person has cancer.
D. A positive test result means that the person will get cancer within a year.

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**American College of Surgeons, Commission on Cancer Standard 2.3**

“Qualified Genetics Professional”

- ABGC or ABMG Licensed Genetic Counselor
- An American College of Medical Genetics physician board certified in medical genetics
- A Genetics Clinical Nurse (GCN) credentialed through the Genetic Nursing Credentialing Commission (GNCC)
- Advanced Practice Nurse in Genetics credentialed through the Genetic Nursing Credentialing Commission (GNCC)
- An advanced practice oncology nurse with specialized education in cancer genetics and hereditary cancer predisposition syndromes
- A board-certified physician with experience in cancer genetics

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**Original Genetics Nursing Credential**

APNG, GCN (renewal only after 2014)

- Professional Portfolio review process
  - 300 hours of Genetics Practicum
- Completion of Log of 50 cases within five years.
- Four Case Studies
- Graduation from graduate program in nursing.
- 50 hours of genetic content in the past 5 years.

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**American Nurses Credentialing Commission**

Advanced Genetics Nurse – Board Certified

Professional Portfolio

- RN license or legally recognized equivalent in another country.
- Two years full time as a registered nurse
- Graduate degree (masters, post-graduate or doctorate) in nursing.
- 1500 practice hours in the specialty
- 30 hours of genetics nursing continuing education
- Fulfill two additional professional development categories

- READ and follow the Rubric!!
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<thead>
<tr>
<th>Preferred Order of Credentials</th>
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<tbody>
<tr>
<td>Highest earned degree <strong>permanent</strong></td>
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<tr>
<td>- Licensure <strong>required to practice</strong></td>
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<tr>
<td>- State designations or <strong>requirements</strong></td>
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<tr>
<td>- National certifications <strong>relevance</strong> to practice</td>
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<tr>
<td>- Awards and honors <strong>voluntary</strong></td>
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<tr>
<td>- Other recognitions <strong>professional</strong> first</td>
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<tr>
<th>Question 8</th>
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<tbody>
<tr>
<td>American College of Surgeons. Commission on Cancer Standard 2.3 does not include any nurses in their definition of qualified genetics professionals.</td>
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<tr>
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<tr>
<td>- <strong>TRUE</strong></td>
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<tr>
<td>- <strong>FALSE</strong></td>
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<tr>
<th>Take Home Message</th>
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<tr>
<td>The first step to screening, prevention and management of hereditary cancer syndromes is <strong>RECOGNITION</strong>!</td>
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<td>Increased surveillance and/or interventions may identify cancers early or reduce the risk of cancers.</td>
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<tr>
<td>Ethical issues are present in every situation.</td>
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<td>Genetic counseling/risk assessment does not always lead to genetic testing.</td>
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