Objectives

By the end of this talk, you will be:

- Able to identify the clinical characteristics of patients who should be referred for cancer genetic counseling.
- Familiar with the components of a genetic counseling consultation.
- Familiar with the administrative considerations for starting a genetic counseling program.
Purpose

The purpose of cancer genetic counseling is to:

- Educate clients about their chance of developing cancer.
- Help them derive personal meaning from cancer genetic information.
- Empower them to make educated, informed decisions about genetic testing, cancer screening, and cancer prevention.
Why is genetic counseling important? For patients…

- Cancer risks in hereditary cancer families are much higher than the general population.
- Hereditary cancers tend to develop at a younger age than expected, often prior to the time at which general population screening would be initiated.
- Clinical management may be altered.
- Identify individuals in the family who are at risk. Psychosocial impact on patient and family.
Why is genetic counseling important? For physicians…

- Complex hereditary syndromes
- Changing information on diagnosis and testing
- Psychosocial impact on patient and family
- Long-term follow-up
Why is genetic counseling important? For the practice...

- Ann Chadwick, RN at the University of Washington
- Died at age 41 of ovarian cancer.
  - Breast cancer at 28.
  - Contralateral breast cancer at 37.
- Lawsuit asserting that doctors should have warned a woman to have her ovaries removed because she had a high risk of ovarian cancer was settled in 2001 for $1.6 million.
Who should be referred?

- A referral for cancer genetic risk assessment and counseling should be considered for clients with personal or family history features suggestive of familial or hereditary cancer.
- Should not be limited to just those individuals who are potential candidates for genetic testing.
Features

- Individual Patient
  - Multiple primary tumors.
  - Bilateral primary tumors in paired organs.
  - Younger-than-usual age at tumor diagnosis
  - In the sex not usually affected.

- Patient’s Family
  - One first degree relative with the same or a related tumor.
  - ≥ two 1st degree relatives with tumors at the same site or related tumors.
  - ≥ two relatives in two generations with tumors of the same site or related tumors.
  - Known genetic mutation in the family.
Making a Referral

- Referral Form
  - Patient demographics
  - Indication
  - Patient Signature

- Who introduced the idea?
- Is the patient still in the clinic?
- Schedule the consultation.
The Initial Consultation

- Psychosocial Assessment
- Personal medical history
- Family medical history
- Risk Assessment
  - Classify
  - Calculate
  - Counsel
- Molecular Testing
- Pretest Counseling
- Informed Consent
- Sample Collection
Psychosocial Assessment

- Motivations for seeking a cancer risk consultation.
- Beliefs about cancer etiology and perception of risk.
- Ethnocultural information.
- Socioeconomic and demographic information.
- Psychosocial factors.
- Current cancer screening.
- Health behaviors.
- Coping strategies.
Personal Medical History

- Age
- Personal history
- Major illnesses
- Hospitalizations
- Surgeries
- Biopsy history
- Reproductive history

- Organ in which tumor developed
- Age at time of diagnosis
- Number of tumors
- Pathology, stage, and grade of malignant tumor
- Pathology of benign tumors
- Treatment regimen
Family History

- Cornerstone of cancer genetic risk assessment.
- Minimum three to four-generation pedigree
- Standardized pedigree nomenclature
- Paternal and maternal history
- Ancestry/Ethnicity
- Consanguinity
Taking a Family History

- Organ in which tumor developed
- Age at time of diagnosis
- Number of tumors
- Pathology, stage, and grade of malignant tumors
- Pathology of benign tumors
- Treatment regimen
- Environmental exposures
- Surgeries
- Current screening
- If deceased, age and cause

Dominant Pedigree

Red = Affected
Cancer Family History

Key
- Breast CA
- Ovarian CA

58
37
Ruth 35
28
Dx 49, 53
60 yrs
Dx 42
82 yrs
Dx 55
d. 56

- Breast CA
- Ovarian CA
Risk Assessment - Classify

- **Hereditary Cancer (5-10%)**
  - Families with known cancer syndromes or histories that exhibit the classic features of hereditary cancer.

- **Familial Cancer (10-15%)**
  - More cases of a specific type of cancer than expected, but not necessarily exhibiting the classic features of hereditary cancers.

- **Sporadic Cancer (75-80%)**
  - Cancer in the family is mainly due to nonhereditary causes.
Risk Assessment - Calculate

- Develop differential diagnosis
- Utilize model that estimates risk of carrying a mutation in a cancer predisposition gene. (e.g. BRCAPro)
- Utilize model that estimates absolute risk of developing specific cancers in the absence of a mutation in a known gene. (e.g. Gail or Claus model)
Risk Assessment - Counsel

- Risk of having mutation in susceptibility gene vs. risk of developing cancer
- Patient’s perception of risk
- Risk for patient’s children / other family members
- Assess patient understanding.
Molecular Testing

- Client has significant personal and/or family history of cancer.
- Test can be adequately interpreted.
- Results will affect medical management.
- Client can provide informed consent.
Molecular Testing (cont.)

- Clinical testing
  - Available on a clinical, fee-for-service basis.
  - CLIA-approved laboratory.
  - Note: Not all labs offer same clinical testing.

- Research testing
  - Available in the context of a research study.
  - IRB-approved.

- Both available?
  - Discuss pros and cons with the patient.
Informed Consent

- Purpose of the test and who to test.
- General information about the gene(s).
- Possible test results.
  - Positive result
  - Negative result
  - Variant
- Likelihood of a positive result.
Informed Consent (cont.)

- Technical aspects and accuracy of the test.
  - False-positive or false-negative result
- Economic considerations
- Risk of genetic discrimination
- Psychosocial aspects
- Confidentiality issues
Informed Consent (cont.)

- Utilization of test results: Medical surveillance and preventive measures.
- Alternatives to genetic testing.
- Storage and potential reuse of genetic material.
Sample Collection

- Coordinate sample collection and shipment.
- Preauthorize with insurance company?
- Provide client with estimated turnaround time.
- Establish a plan for disclosing results.
The Second Consultation - Result Disclosure

- Combine with an MD visit
- Results disclosure
- Significance of test results (e.g. cancer risk)
- Assess emotional impact of test results
- Medical management
- Informing other relatives
- Future contact
- Resources
Program Infrastructure

- Space (sites, staffing, equipment needs)
- Human Resources
- Other Resources
  - Budgets
  - Billing structure (96040 – Genetic Counseling)
  - Information Management
  - Patient Confidentiality
Program Development

- Structure and Function
  - What clinic model will be used?
  - Team meetings
  - Scheduling
  - Patient flow and patient communication policies
  - Development of forms
Additional Considerations

- Genetic Testing Fees ($400 - $4000)
  - Clinic pays for testing and seek reimbursement from patient.
  - Patient seeks pre-approval from insurance company.
  - Seek reimbursement from insurance company.
  - Patient pays out of pocket.
Resources

http://www.nsgc.org/

Development of consensus document around prenatal screening and diagnosis as concurrence: Understanding Prenatal Screening and Diagnosis of Down Syndrome and Advocacy Community Perspectives. A newsletter is now available. Click here to view the newsletter.

A new award is now available! Click here to announce.

Prenatal counseling. Created in collaboration with Genetic Alliance, this guide to genetic counseling is, what to expect from a genetic counseling visit, and the different types av and other emails? Download instructions on how to set your email to

Frequently Asked Questions about Genetic Counseling Services
Resources (cont.)

Welcome to the NSGC Cancer Genetics Healthcare Provider Link

- Fact Sheets about Hereditary Cancers *(Under Construction)*
- **What can a Genetic Counselor do for you?**
- Pocket Referral Guidelines
- Find a Genetic Counselor
- **What should I tell my patients about genetic discrimination?**
- Who needs genetic counseling and genetic testing? *(Under Construction)*
- Clinical Genetic Testing vs. Research Genetic Testing Fact Sheet
- DNA Banking
- NSGC Practice Guidelines
  - Hereditary Cancer Risks
  - Hereditary Breast Ovarian Cancer Syndrome
- NSGC Position Statements
- Need a SPEAKER for an educational event? Go to the [NSGC Speaker’s Bureau](http://www.nsgc.org/provders/cancer/cancer_genetics_provider.cfm)
- Additional Cancer Genetics Resources
- Resources for your Patients *(Under Construction)*

Back to Cancer Genetics Healthcare Providers Main Page