Learning Objectives

- Describe the current status of access to appropriate cancer genomics services in the State of Michigan
- Identify specific strategies employed in Michigan to increase access in underserved populations and limitations of these strategies
Presenters and Topics

- Deb Duquette, MS, CGC, MDCH
  - Overview and Background
- Kara Milliron, MS, CGC, University of Michigan
  - Planned Parenthood
  - Informed Medical Decisions, Inc
- Dana Zakalik, MD, Beaumont Health System
  - Federally Qualified Health Center
- Julie Zenger-Hain, PhD, FACMG, Oakwood Hospital
  - Hispanic Community

Genomics Goal:
Increase availability of cancer-related genetic information to the Michigan public and decrease barriers to risk-appropriate services

http://michigancancer.org/
Healthy People 2020 Genomics Objectives

Genomics

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G-1 Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling

G-2 (Developmental) Increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify Lynch syndrome (or familial colorectal cancer syndromes)

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Commission on Cancer (CoC)  
Genetic Counseling Standard

STANDARD 2.3  
Risk Assessment and Genetic Counseling

Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.

DEFINITION AND REQUIREMENTS

Cancer risk assessment and genetic counseling are the processes to identify and counsel people at risk for familial or hereditary cancer syndromes. The purpose of genetic counseling is to educate patients about their chance of developing cancers, help them understand the risk information from genetic counseling, and empower them to make educated, informed decisions about genetic testing, cancer screening, and cancer prevention. Identifying patients at increased risk of developing cancer because of a family history of cancer or a known hereditary cancer syndrome can have dramatic effects on early detection and cancer outcome. For this reason, cancer risk assessment and genetic counseling are rapidly becoming standards of care for patients with personal and/or family history of cancer who are at high risk of having a hereditary syndrome.

The program provides cancer risk assessment and genetic counseling on-site or by referral to another facility or community-based organization.

Cancer risk assessment and genetic counseling are performed by a cancer genetics professional who has extensive experience and educational background in genetics, cancer genetics, counseling, and hereditary cancer syndromes to provide accurate risk assessment and comprehensive genetic counseling to patients with cancer and their families.

Cancer risk assessment and the potential for referral may be discussed as part of the multidisciplinary cancer conference.

Genetics professionals include people with the following:

- A Board of Genetic Counseling (ABGC) or American Board of Medical Genetics (ABMG) board-certified/board-eligible or (in some states) a licensed genetic counselor.
- An American College of Medical Genetics physician board certified in medical genetics.
- A Genetic Counseling Board (GCN) or an Advanced Practice Nurse in Genetics (APNCG).
- A Genetics Clinical Nurse (GCN) or an Advanced Practice Nurse in Genetics (APNG).
- A board-certified physician with experience in cancer genetics (defined as providing cancer risk assessment on a regular basis).
- Please note, specialized training in cancer genetics should be ongoing, educational seminars offered by commercial laboratories about how to perform genetic testing are not considered adequate training for cancer risk assessment and genetic counseling.

The Cancer Committee defines the appropriate individuals who will provide risk assessment and counseling for major cancer disease sites (such as breast and colon/cervix). In addition, the programs not providing immediate access to formal genetic counseling services should identify resources for referral.

Cancer risk assessment and genetic counseling involve pretest and posttest counseling. At a minimum, this counseling includes the following:

- Pretest counseling
  - Collecting relevant information needed to assess a patient's personal and family medical history
  - A 3- to 7-generation pedigree, including detailed medical information about the patient's father, mother, and siblings
  - Evaluating the patient's risk
    - One aspect of risk assessment is discussing the absolute risk that the patient will develop a specific type of cancer or cancer based on the family history. The second aspect is the risk that the patient carries a hereditary or germline mutation in a cancer susceptibility gene.
  - Performing a psychosocial assessment
    - Educating the patient about the suspected hereditary cancer syndrome, if appropriate
    - The provider reviews and discusses with the patient the cancer risks associated with gene mutations, including basic concepts such as genes and inheritance patterns and more advanced concepts of penetrance and variable expressivity and the possibility of genetic heterogeneity.
    - Obtaining informed consent for genetic testing
      (If genetic testing is recommended).

- Posttest counseling

SPECIFICATIONS BY CATEGORY

All programs fulfill the standard as written.

DOCUMENTATION

The program completes the SAR.

During the on-site visit, the surveyor will discuss the process for providing cancer risk assessment and genetic counseling services on-site or by referral.

MEASURING COMPLIANCE

Rating

(1) Compliance: The program fulfills the following criterion:

Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.

(5) Noncompliance: The program does not fulfill the following criterion.

Cancer risk assessment, genetic counseling, and testing services are provided to patients either on-site or by referral, by a qualified genetics professional.

Hereditary Breast and Ovarian Cancer (HBOC)

- Accounts for 5-10% of all breast cancers
- At least 11% of Michigan adult women meet USPSTF and NCCN criteria for referral for BRCA counseling (2011 and 2012 MiBRFS)
- Approximately 1/200-1/500 are carriers in the general population; 1/40 in Ashkenazi Jewish population
- Caused by mutations in BRCA1/BRCA2
- Autosomal dominant inheritance – 50% risk to each child/sibling/parent
- For those women with a deleterious BRCA mutation, the risk of developing breast cancer by age 70 is ~35-84% and the risk of developing ovarian cancer by age 70 is ~10-63%
- For men with a deleterious BRCA mutation breast cancer risk increased to 6%
- Management by risk-reducing surgery, enhanced screening regimen and chemoprevention


http://abcnews.go.com/blogs/health/2013/05/14/angelina-jolies-choice-should-you-get-bra-gene-testing/
Primary care providers should screen women with a family history of breast, ovarian, fallopian tube or peritoneal cancer to identify those potentially at increased risk for a BRCA mutation. Women with a significant family history receive genetic counseling, and, if indicated, they be offered genetic testing

(Grade B Recommendation)

USPSTF also recommends against routine referral or routine BRCA testing for women whose family history is not associated with increased risk

(Grade D Recommendation)
CRITERIA FOR FURTHER GENETIC RISK EVALUATION

An affected individual with one or more of the following:

- A known mutation in a breast cancer susceptibility gene within the family
- Early-onset breast cancer
- Triple negative (ER-, PR-, HER2-) breast cancer
- Two breast cancer primaries in a single individual
- Breast cancer at any age, and
  - ≥1 close blood relative with breast cancer ≤50 y, or
  - ≥1 close blood relative with epithelial ovarian cancer at any age, or
  - ≥2 close blood relatives with breast cancer and/or pancreatic cancer at any age
- From a population at increased risk
- ≥1 family member on same side of family with a combination of breast and ovarian cancer and ≥1 of the following (especially if early onset): pancreatic cancer, prostate cancer (Gleason score ≥7), sarcoma, adrenocortical carcinoma, brain tumors, endometrial cancer, leukemia/lymphoma; thyroid cancer, dermatologic manifestations, and macrocephaly, hamartomatous polyps of GI tract; and diffuse gastric cancer

Ovarian cancer
Male breast cancer

An unaffected individual with a family history of one or more of the following:

- A known mutation in a breast cancer susceptibility gene within the family
- ≥2 breast primaries in single individual
- ≥2 individuals with breast primaries on the same side of family
- ≥1 ovarian cancer primary from the same side of family
- First- or second-degree relative with breast cancer ≤45 y
- ≥1 family member on same side of family with a combination of breast and ovarian cancer and ≥1 of the following (especially if early onset): pancreatic cancer, prostate cancer (Gleason score ≥7), sarcoma, adrenocortical carcinoma, brain tumors, endometrial cancer, leukemia/lymphoma; thyroid cancer, dermatologic manifestations, and macrocephaly, hamartomatous polyps of GI tract; and diffuse gastric cancer

Male breast cancer

For populations at increased risk, requirements for inclusion may be modified (eg, women of Ashkenazi Jewish descent with breast or ovarian or pancreatic cancer at any age).

For dermatologic manifestations, see COAD.

For hamartomatous colon polyps in conjunction with breast cancer and hyperpigmented macules of the lips and oral mucosa, STK11 testing should be considered. See NCCN Guidelines for Genetic/Familial High-Risk Assessment: Colorectal, Peutz-Jeghers syndrome. Melanoma has been reported in some HBOC families.

For lobular breast cancer with a family history of diffuse gastric cancer, CDH1 gene testing should be considered.

Genetic counseling is highly recommended when genetic testing is offered and after results are disclosed. A genetic counselor, medical geneticist, oncologist, surgeon, oncology nurse, or other health professional with expertise and experience in cancer genetics should be involved early in counseling patients who potentially meet criteria for an inherited syndrome.

Note: All recommendations are category 2A unless otherwise indicated.

Clinical Trials: NCCN believes that the best management of any cancer patient is in a clinical trial. Participation in clinical trials is especially encouraged.
Continual growth of appropriate cancer genetic counseling and BRCA testing of individuals with a personal and/or family history of breast and/or ovarian cancer.

Extraordinary increase in number of cancer genetic clinics with board-certified genetic professionals in Michigan including new clinics in previously underserved areas.

- 17 clinics in 2014 compared to 8 clinics in 2010

Reduced barriers for appropriate BRCA testing with continued decrease in percentage of individuals who had genetic counseling but were not able to pursue BRCA testing due to inadequate insurance.

- 8.3% of those not testing in 2014 compared to 21.7% in 2008
Populations in Need of Greater Cancer Genetic Services in Michigan

- All women with a significant family history of breast and ovarian cancer
  - ~11% of adult Michigan women met USPSTF family history criteria for BRCA counseling and testing (MiBRFS 2011 and 2012)
  - Of these, 8.8% have had cancer genetic counseling
  - Of these 22.4% had at least one family member who had cancer genetic counseling

- Individuals of a relative with a known deleterious mutation
  - 50% risk to inherit known deleterious mutation for first degree relatives
  - Single site testing is extremely informative and much less expensive
  - Rate of single site testing has remained relatively low and steady in Michigan (BRCA Clinical Network database, Myriad data)
Populations in Need of Greater Cancer Genetic Services in Michigan (continued)

- **Ovarian cancer patients**
  - Of 137 ovarian cancer charts reviewed by Michigan Cancer Surveillance Program, only 3.6% of ovarian cancer cases in 2006-2010 had received cancer genetic counseling (MCSP data)
  - 29.4% of patients with a history of both breast and ovarian cancer; and 14.0% of patients with ovarian cancer found to have deleterious BRCA mutation (BRCA Clinical Database)
  - Of women who did not have testing, 50.0% (5) with both breast and ovarian cancer and 30.6% (15) with ovarian cancer alone had to decline testing due to inadequate insurance coverage or high co-pay (BRCA Clinical Database)

- **African American adults with a significant personal or family history** (BRCA Clinical Database)
  - 9.4% of all patients receiving BRCA genetic counseling were African American
  - 56.1% of African American patients had BRCA testing (lowest of all race/ethnicity groups) vs. 67.6% of white patients
  - Only 3.3% of African Americans referred with a known family mutation vs. 13.4% of whites
Use of Cancer Genetic Services among Michigan Young Breast Cancer Survivors (YBCS)

Use of cancer genetic services | Total (n=828) | Black (n=317) | Other (n=511)
--- | --- | --- | ---
Had genetic counseling* | 32.9% | 26.6% | 37.1%
Had genetic testing* | 28.5% | 19.9% | 33.7%
Had genetic counseling and testing* | 27.5% | 18.3% | 32.9%

* Significant at the 0.001 level for Black vs. Other

Black YBCS were less likely than White/Other YBCS to use cancer genetic services

Study conducted by University of Michigan School of Nursing, MDCH Cancer Genomics, MCSP and Prevention Research Center of Michigan with funding from CDC in 2011-2014
# Reasons for not seeking genetic services among Michigan YBCS

<table>
<thead>
<tr>
<th>Most common reasons for not seeking genetic services*</th>
<th>Total (n=547)</th>
<th>Black (n=228)</th>
<th>Other (n=319)</th>
</tr>
</thead>
<tbody>
<tr>
<td>No one ever suggested</td>
<td>67.8%</td>
<td>74.6%</td>
<td>63.0%</td>
</tr>
<tr>
<td>Out-pocket expense/Not covered</td>
<td>13.0%</td>
<td>6.6%</td>
<td>17.6%</td>
</tr>
<tr>
<td>Unknown benefit</td>
<td>2.9%</td>
<td>1.3%</td>
<td>4.1%</td>
</tr>
</tbody>
</table>

* Significant at the 0.001 level for Black vs. Other

The most common self-reported reason among all groups for not seeking genetic services was that no one ever suggested.

Study conducted by University of Michigan School of Nursing, MDCH Cancer Genomics, MCSP and Prevention Research Center of Michigan with funding from CDC in 2011-2014
Patient-Powered Network for Hereditary Breast and Ovarian Cancer

- Established in 2014 under the Affordable Care Act
- Goal is patient-centered, representative, large-scale, rapid comparative effectiveness research studies by collecting, sharing and integrating health data
  - 11 health system networks – each includes >7 million patients
  - 18 condition-focused patient-powered networks – each targeting enrollment of 0.5% of U.S. population with the condition
  - Integrate EHR, health claims and/or patient-reported outcomes data on 70 million Americans by September 2015
One of PCORnet’s 18 patient-powered, condition-focused networks
- Hereditary breast, ovarian and related cancer risks
- Goal: to improve informed decision-making and health outcomes by answering important questions high-risk patients and their providers face every day
- Led by patients, public health professionals and researchers
  - Patients driving governance and research – identifying the research questions, priorities, design, recruitment, analysis and dissemination
- Representativeness is key – across geographic, socioeconomic, clinical severity, racial, ethnic, age groups
EGAPP Recommendation on Genetic Testing for Lynch Syndrome

- Sufficient evidence to offer counseling & genetic testing for Lynch syndrome to patients newly diagnosed with colorectal cancer to reduce morbidity & mortality in relatives

- Relatives of patients who test positive for Lynch could be offered counseling, testing & if positive, increased colonoscopy

- Evidence of benefit to the patient’s relatives

What is Lynch Syndrome (LS)?

- Autosomal dominant hereditary cancer syndrome
  - Most common hereditary colorectal (CRC) and uterine cancer syndrome
  - 20-80% lifetime risk for CRC cancer ~3% of CRCs with LS
  - Mean age of onset of CRC is ~45 years old
  - Increased risk of endometrial, ovarian, urinary tract, gastric tract, small bowel, pancreas, sebaceous cancers
**LS Screening & Management**

- **Screening is complex**
  - Multiple approaches including IHC and/or MSI testing on tumor with DNA testing
  - Different genes involved in LS
    - MSH2, MSH6, MLH1, PMS2

- **Cancer surveillance & prophylactic survey options**
  - Colonoscopy every 1-2 years beginning at ~20-25 years old or 10 years earlier than youngest case in family
  - Annual endometrial sampling and transvaginal ultrasound beginning at 30 years old
  - History and exam annually begin at 21 years
  - Annual urinalysis
  - Prophylactic surgery including subtotal colectomy, total abdominal hysterectomy and bilateral salpingo-oophorectomy
Michigan & National Data on Lynch Syndrome Screening

- No source of national data
  - HP2020 objective is developmental
  - MSI included in cancer registry reporting since 2010

- Michigan surveillance efforts
  - In 2006-2009 Michigan Colorectal Cancer Screening Program provided screening for low income, uninsured in three counties with high mortality rates
    - Of 1500 adults screened, 177 referred to genetic counselor
  - 2010 MiBRFS indicates nearly 80% of individual at risk for familial CRC syndrome report no knowledge of genetic test
    - Only 3% at risk for familial CRC syndrome had genetic test
  - Of 610 CRC charts reviewed from 2006-2010 diagnoses, less than 2% had Lynch syndrome screening
    - 119 cases aligned with NCCN guidelines
    - 6 had MSI testing; 11 had IHC; 0 had BRAF; 5 had MMR; 6 had genetic counseling
CDC Funding Announcement


- **5 year** cooperative agreement awarded to four projects
  - Authorized from Affordable Care Act
  - State health departments and Tribal governments eligible

**Purpose:** Enhance state health department’s capacities to promote and apply evidence-based breast and ovarian cancer genomics guidelines in public health practice

- Develop, enhance and evaluate education, surveillance and policy/systems change
- Emphasis on partnerships
- Focus on HBOC but may also include Lynch syndrome
- May identify target populations disproportionately affected by HBOC and lack genetic services

Figure 5: Cancer Genomics Best Practices
For More Information

www.migrc.org

www.michigan.gov/genomics
www.michigan.gov/cge

Or call 1-866-852-1247

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