A State Population Approach to Promoting Genomics through Surveillance, Education and Policy

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What is Public Health Genomics? (Bellagio Statement, 2006)

- A multidisciplinary field concerned with the effective and responsible translation of genome-based knowledge and technologies to improve population health.
Healthy People 2020 (HP 2020)

- Started in 1979
- 10-year national objectives for promoting health and preventing disease
- HP 2020 marks first time for genomics objectives
- Encourage collaborations across sectors, guide individuals toward making informed health decisions, and measure the impact of prevention activities
- Works to achieve increased quality and years of healthy life and the elimination of health disparities.
Three Core Public Health Functions and Ten Essential Services

- **Assessment**: The regular systematic collection, assembly, analysis, and dissemination of information, including genetic epidemiologic information, on the health of the community.
  - Monitoring how genomic factors are contributing to health outcomes
Three Core Public Health Functions and Ten Essential Services

- **Policy Development**: The formulation of standards and guidelines, in collaboration with stakeholders, which promote the appropriate use of genomic information and the effectiveness, accessibility, and quality of genetic tests and services.
Three Core Public Health Functions and Ten Essential Services

- **Assurance**: That genomic information is used appropriately and that genetic tests and services meet agreed upon goals for effectiveness, accessibility, and quality.
CDC Funding Announcement

Genomics Applications in Practice and Prevention (GAPP): Translation Programs in Education, Surveillance, and Policy

- 3 year cooperative agreement (2008-2011) awarded to four projects
  - Large, well-defined populations in US (greater than 100,000)
- **Goal**: move human genome applications into health practice to maximize health benefits and minimize harm through non-research activities

- Expected measurable outcomes:
  - **Surveillance**: measure use of counseling and testing for BRCA1/2; knowledge of providers or public on use of BRCA1/2 or EGAPP-identified genetic test(s); use of EGAPP genetic test(s); use of family history tools
  - **Education**: increase knowledge of validity, utility, harms and benefits of EGAPP-identified genetic test(s); increase use of family history, counseling and BRCA1/2 tests as recommended by USPSTF
  - **Policy**: increase use of family history, counseling, and BRCA1/2 tests as recommended by USPSTF
Availability of Genetic Tests

• “...efforts are needed not only to implement what is known in genomics to improve health but also to reduce potential harm and create the infrastructure needed to derive health benefits in the future.”

Genomic Advances to Improved Population Health

Women whose family history is associated with an increased risk for deleterious mutations in BRCA1 or BRCA2 genes should be referred for genetic counseling and evaluation for BRCA 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)
Evaluation of Genomic Applications in Practice and Prevention (EGAPP)

www.egappreviews.org
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

*Gen Med* 2009;11:35-41 & 42-65
Three-Tier Classification of Recommendations on Genomic Applications

- **Tier 1: Ready for implementation** (per evidence-based recommendation on clinical utility)
  - Encourage use; can save lives
  - Examples: BRCA, Lynch syndrome, familial hypercholesterolemia, newborn screening

- **Tier 2: Informed decision making** (adequate information on analytic and clinical validity, promising but not definitive information on clinical utility)
  - Provide information for shared decision making
  - Examples: Gene expression profiles in breast cancer, family history assessment in primary care

- **Tier 3: Discourage use** (no or little information on analytic, clinical validity or clinical utility; or evidence of harm)
  - Discourage use; reduce potential harms and save unnecessary healthcare costs
  - Examples: Population screening for hereditary hemochromatosis, personal genomic tests sold directly to consumers

Importance of Informed Ordering Providers

“Errors in Delivery of Cancer Genetics Services: Implications for Practice”
Conn Med 2010; 74(7): 413-423
http://yalecancercenter.org/education/grand-rounds.html#videotop

- Case reports series demonstrates need for better provider education and importance of framework for referral to board certified genetics professional
- Inappropriate cancer genetic testing performed without counseling by a qualified provider:
  - Wrong genetic test ordered
  - Genetic test results misinterpreted
  - Inadequate genetic counseling
- Adverse outcomes:
  - Unnecessary prophylactic surgeries
  - Unnecessary testing
  - Psychosocial distress
  - False reassurance resulting in inappropriate medical management
Michigan Informed Consent Law for Genetic Testing

- Michigan law states that a provider shall not order “a presymptomatic or predictive genetic test without first obtaining the written, informed consent”
- Nature and purpose of the test
- Effectiveness and limitations
- Implications of taking the test, including, but not limited to, the medical risks and benefits.
- The future uses of the sample taken and the information gained from the test.
- The meaning of the test results and how results will be disclosed
- Who will have access to the patient’s sample and result and the right to confidentiality
Promoting Cancer Genomics
Best Practices through Surveillance, Education and Policy Change in the State of Michigan

A reduction in early cancer deaths (before age 50) through statewide surveillance and implementation of systems of care for inherited breast, ovarian, colorectal and other Lynch syndrome (HNPCC) related cancers that use best practice recommendations for family history assessment, cancer genetic counseling and testing
Our Program’s Goals
2008-2011

- Develop and implement a model for surveillance of inherited cancers and use of relevant genetic tests; and share with other cancer registries and national programs
- Identify model provider education programs to increase use of appropriate screening, counseling and evidence-based genetic tests; and share with public health and/or clinical practice organizations
- Identify a model health insurance policy for BRCA1 & 2 cancer genetic testing; and share with health plans in Michigan and other states

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Target Population

State of Michigan

♦ Public
  ~ 10 million residents
  ~ 6.9 million under age 50

♦ Health systems and providers
  – ~200 facilities reporting to the Michigan Cancer Surveillance Program (excludes labs, dermatology and dental offices)
  – ~64,000 new reportable cancer cases per year

♦ Health insurance plans
  – 24 health plans
Reduction in early cancer deaths in Michigan residents from appropriate use of cancer genetic services and tests

Health Plan Champion; Michigan Association of Health Plans (MAHP)

Surveillance

Michigan Cancer Surveillance Program (MDCH)

12 Clinical Cancer Genetics Sites

MDCH Genomics Program

CDC Office of Public Health Genomics

CDC Division of Cancer Prevention and Control

Policy

Michigan Cancer Genetics Alliance

Education

Michigan Cancer Consortium
Provider Education Goals

• Change provider knowledge, attitudes and intentions to ultimately reduce the number of early cancer deaths
• Use statewide surveillance data in conjunction with EGAPP and USPSTF guidelines
• Increase provider awareness about EGAPP recommendations for Lynch syndrome/HNPCC and gene expression profiling tests for early stage breast cancer
• Increase use of USPSTF clinical practice guidelines for USPSTF

- 853 charts reviewed from cancer patients reported to Michigan Cancer Surveillance Program (MCSP)
- 82% documented presence or absence of family history of cancer
  - 30% had positive family history of cancer
  - Over 80% documented relationship to patient and gender of affected family member
  - Over 94% missing age of onset/diagnosis of affected family member’s cancer
- Resulted in mandatory family history reporting for MCSP starting in 2007
Family History Provider Collection
Chart Reviews (2005-2007)

- 668 Primary Care Provider charts reviewed by Michigan Health Plan
  - 60% from Family Practice
  - 25% from Internal Medicine
  - 15% from Pediatrics
- Providers are collecting family history information.
- 92% of charts documented family history
  - 42% documented family history of cancer
  - 93% documented relationship of affected
  - Over 98% of charts never documented age of onset of affected
Key Informants and Focus Groups: Family Health History Collection (2003-2008)

Common Themes Identified for Michigan Providers:

- Do not believe they see patients with high-risk cancer family history
- Do not feel confident in ability to identify high-risk family history
- Uncertain where to refer
- Would use a pocket tool in practice
Development of Cancer Family History Guide©

How can we help providers to:

• Identify who is at risk?
• Determine level of risk?
• Ensure proper screening?
• Ensure proper preventive services?
• Improve health outcomes?
• Possibly even save health care resources?
Development of Cancer Family History Guide

- Modeled after standard obstetrical pregnancy wheel
  - Same size
  - Same concept as hand held pocket tool
- Developed by 3 board-certified genetic counselors employed at MDCH with input from CDC Office of Public Health Genomics and Michigan Cancer Genomics Best Practices Steering Committee
- Overwhelming positive feedback in focus group testing with 2 primary care provider groups and 2 oncology groups
- Recommendations based on national publications
  - 2005 USPSTF BRCA Recommendation
  - Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Guidelines for Lynch syndrome
  - National Comprehensive Cancer Network (NCCN) Guidelines
  - Society of Gynecologic Oncologists (SGO) Education Committee Statement
Cancer Family History Guide©

For More Information

www.migeneticsconnection.org

Or call 1-866-852-1247
Cancer Family History Guide©

Evaluation & Dissemination

• 5,740 guides disseminated since July 2010
  – Available at no cost until supplies last (6,760 available)
• Michigan Cancer Consortium (MCC) Breast Cancer Advisory Committee
  – 688 surveys on family history of breast and ovarian cancer completed by women waiting for clinical visits
  – Compared 5 risk assessment tools to determine appropriate referral to cancer genetics
  – Same 71 women (13%) identified appropriate for referral by Cancer Family History Guide and B-RST
    • Lowest referral rate compared to other tools rates (18-54%)
  – Cancer Family History Guide and B-RST identified same 28% of women at moderate risk

• Oakwood Hospital & Medical Center based in Dearborn, Michigan
  – New provider referrals to cancer genetics measured since May 2010 (6 months prior to dissemination)
  – Oakwood Communications sent tools to 700 Oakwood providers in October 2010
  – New provider referrals to cancer genetics to be measured until April 2011 (6 months after dissemination)
• Washington Health Department measuring impact following distribution to 500 internal medicine and BCCCP providers
• Cook County measuring impact after distribution to oncologists serving low SES black population in Chicago

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**Surveillance Objectives**

- To examine the epidemiology of multiple primaries, early onset breast, male breast, ovarian and Lynch syndrome cancers
- To evaluate the use of genetic counseling and tests:
  - Who is accessing genetic counseling? and testing?
  - What providers are referring for genetic counseling?
  - Is referral for counseling appropriate using USPSTF family history guidelines?
  - For patients having BRCA testing, what are their test results?
  - Do health plan policies that are consistent with USPSTF guidelines influence visits?
- To assess barriers/facilitators to cancer survivors knowledge and attitudes about family health history, genetic counseling and testing
- To provide data that will reinforce educational messages to health care providers
Network of Michigan Clinical Cancer Genetics Clinics

Collecting all BRCA counseling cases from October 2007-March 2011 seen by a board certified genetics professional in Michigan
Cancer Genetics Clinic BRCA Counseling Data, 2007 - 2010

Demographics
- Total of 4,426 patients
  - 3,159 (71.4%) patients had BRCA testing
- Mean age = 50 years old at initial visit
- 56.3% have a personal history of breast and/or ovarian cancer
- 4.6% of those counseled are male
- 84.3% white, 9.2% black, 2.0% Arabic, 1.9% Asian

Risk Factors
- 10.6% Ashkenazi Jewish ancestry
- 11.5% have a known mutation in their family
- 41.2% have a family history that meets USPSTF suggested family history guidelines for referral

Other Findings
- Only 38.5% of female relatives with a reported cancer history were from the paternal side of the family
- Most common referring provider type:
  - Oncologist for those with personal history of cancer (36%)
  - OB/GYN for those with family history of cancer (27%)
Comprehensive BRACAnalysis®

$3,340.00

Sequencing

**BRCA1**

**BRCA2**

5-Site Rearrangement

**BRCA1**

1. Dutch Ancestry: deletions in exon 13 and exon 22
2. European (primarily British) ancestry: duplication of exon 13
3. European ancestry: deletion of exons 8 and 9
4. Deletion of exons 14-20
Multisite 3 BRACAnalysis®
For Individuals of Ashkenazi Jewish Ancestry
$575.00

BRCA1
185delAG

BRCA2
6174delT

5382insC
Single Site BRACAnalysis®

For Individuals with a Known Familial Mutation in *BRCA1* or *BRCA2*

$475.00

★ ‘True Negative’
- Negative result in a patient with a known deleterious BRCA mutation in the family
- Same risk and screening recommendations as the general population
Types of BRCA tests:
- **Multisite 3**: 338 tests (10.3%)
  - 18.1% positive
- **Comprehensive**: 2572 tests (81.4%)
  - 6.5% positive; 4.6% variant
- **Single Site**: 383 tests (11.6%)
  - 44.9% positive; 2.1% variant
  - 53% were ‘true negative’ (203 patients)
    - Huge cost savings when find true negative patient
    - No increased risk for cancer; general population screening

13% of all patients tested were positive; 4% were variants
- **Personal history of cancer**: 10% positive
- **No personal history of cancer, met USPSTF guidelines**: 22% positive
- **No personal history of cancer, did not meet USPSTF guidelines**: 13% positive

Potential cost savings of up to $1.097 million with Single Site testing (compared to if Comprehensive ordered)

True negatives save millions of $ in cancer screening, prophylactic surgeries, and chemoprevention
Reasons For Not Testing

• 1,283 female patients seen for BRCA counseling and did not pursue testing
  ▪ Not the best test candidate - 314 (24.5%)
  ▪ Not clinically indicated – 239 (18.6%)
  ▪ Inadequate insurance – 188 (14.7%)
  ▪ Many are not clinically indicated

• Among patients with breast and/or ovarian cancer (462 patients)
  ▪ Inadequate insurance – 118 (25.5%)
  ▪ Not clinically indicated – 78 (16.9%)

• Among patients with family history (819 patients)
  ▪ Not the best test candidate- 314 (38%)
  ▪ Not clinical indicated- 166 (20%)
  ▪ Inadequate insurance- 74 (9%)

• Other reasons given for not testing include ‘not a good time,’ ‘don’t want to know,’ and ‘need to discuss options with relatives’

553 patients found to be inappropriate for testing at this time (potential cost savings of up to $1.85 million)
Policy Objectives

- Understand current status of Michigan health insurance policies for BRCA1/2 counseling and testing with respect to USPSTF guidelines and related clinical services for BRCA1/2 mutation carriers
- Understand current status of Michigan health insurance policies for Lynch syndrome genetic testing with respect to EGAPP recommendations
  - No known health plans with written policies for Lynch syndrome testing in alignment with EGAPP recommendations
- Increase the number of health plans that have policies consistent with USPSTF guidelines
Surveillance of Health Plan BRCA 1/2 Policies

Methods:

- Contracted with “health plan champion” and Michigan Association of Health Plans (MAHP)
- Identified total of 24 Michigan health plans
- Using multiple search types (i.e. websites, key administration contacts, list servs, newsletters, conferences) request and/or identify policies

Conduct ongoing surveillance to determine:

1. Does the health plan have a written BRCA counseling and testing policy?
2. If written policy, does it include coverage for female members with a significant family history of breast and/or ovarian cancer without a personal history (aligned with USPSTF)?
3. Does the policy ‘require’ or ‘strongly recommend’ counseling by a qualified health care professional or genetic counselor prior to BRCA testing?

Consider aligned with USPSTF if all three criteria are fulfilled
### Michigan Health Plan Policies Consistent with USPSTF and Cancer Genetic Counseling Visits

<table>
<thead>
<tr>
<th>Health Plan</th>
<th>BRCA1/2 written policy (Y/N)</th>
<th>Consistent with USPSTF</th>
<th>Distribution (%) of Patients at 8 Clinical Sites Receiving Cancer Genetic Counseling*</th>
<th>Number of Michigan Members*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aetna</td>
<td>Y (2008)</td>
<td>Y</td>
<td>110 (2.8)</td>
<td>280,000</td>
</tr>
<tr>
<td>BCBSM; BCN</td>
<td>Y (2009)</td>
<td>Y</td>
<td>2189 (56.3)</td>
<td>4.6 million; 625,000</td>
</tr>
<tr>
<td>Cigna</td>
<td>Y (2009)</td>
<td>Y</td>
<td>55 (1.4)</td>
<td>pending</td>
</tr>
<tr>
<td>Beaumont Employee</td>
<td>Y (2008)</td>
<td>Y</td>
<td>96 (2.5)</td>
<td>34,818</td>
</tr>
<tr>
<td>Molina</td>
<td>Y (2008)</td>
<td>Y</td>
<td>10 (0.3)</td>
<td>230,000</td>
</tr>
<tr>
<td>United Health</td>
<td>Y (2009)</td>
<td>Y</td>
<td>94 (2.4)</td>
<td>805,000</td>
</tr>
<tr>
<td>Health Alliance Plan</td>
<td>Y (2010)</td>
<td>Y</td>
<td>509 (13.1)</td>
<td>470,000</td>
</tr>
<tr>
<td>Priority Health</td>
<td>Y (2008)</td>
<td>Y</td>
<td>117 (3.0)</td>
<td>600,000</td>
</tr>
<tr>
<td>12 Health Plans</td>
<td>N</td>
<td>N</td>
<td>110 (2.8)</td>
<td>~1 million</td>
</tr>
<tr>
<td>1 Health Plan</td>
<td>Y</td>
<td>N</td>
<td>2 (0.1)</td>
<td>187,000</td>
</tr>
<tr>
<td>Medicaid</td>
<td>Y</td>
<td>N</td>
<td>106 (2.7)</td>
<td>500,000+</td>
</tr>
<tr>
<td>Medicare</td>
<td>Y</td>
<td>N</td>
<td>493 (12.7)</td>
<td>~1.5 million</td>
</tr>
</tbody>
</table>

*Patients may be counted more than once because of multiple insurers*
Coverage of Management Services for BRCA Positive Patients (2011)

- Services reviewed for those with a personal history of cancer and those without:
  - Clinical breast exam (semiannually beginning at age 25\textsuperscript{1})
  - Mammography (annually beginning at age 25\textsuperscript{1})
  - Breast MRI (annually beginning at age 25\textsuperscript{1,2})
  - CA-125 testing (semiannually beginning at age 35\textsuperscript{1})
  - Transvaginal ultrasound (semiannually beginning at age 35\textsuperscript{1})
  - Anti-estrogen therapies such as Tamoxifen
  - Prophylactic mastectomy
  - Prophylactic oophorectomy
  - Breast reconstruction

- 7 health plans written policies that cover all highlighted services
  - Aetna
  - BCBS/BCN
  - Cigna
  - Priority Health
  - HAP
  - Molina

- Other covered services vary among plans
- Other 17 health plans do not have readily available policies and will be contacted by phone in 2011

Honoring Health Plans Aligned with USPSTF Grade B Recommendation

- Michigan Association of Health Plans (MAHP) 25th Anniversary Conference held on July 17-20, 2010
- Announcement regarding regulations requiring new health insurance plans to cover preventive care for USPSTF Grade A & B Recommendations on July 14, 2010
- Pinnacle Awards to honor health plans aligned with USPSTF Grade B BRCA Recommendation on September 21, 2010
- CME Best Practices event to educate health plan directors on December 8, 2010
- MAHP Summer 2011 Conference and 2011 Pinnacle Awards
Promoting USPSTF Grade B BRCA Recommendation to Health Plans

• Educate health plans about USPSTF Grade B BRCA Recommendation and Best Practices
  ▪ Health plan conferences
  ▪ CME events
  ▪ Displays
  ▪ Provider tools
  ▪ Articles in newsletters
Notification to Health Plans Not Aligned with USPSTF

- MDCH staff provided individualized packets to Michigan health plans at key events
  - Discuss in person with key health plan administrators
  - Emphasize USPSTF Grade B Recommendation
  - Provide summary of project and partnership with CDC and MAHP
  - Highlight three criteria required to receive honors
  - Report individualized information for each health plan regarding their assessment
  - Encourage to contact MDCH or MAHP for technical assistance
Educational Materials for Health Plans

Packet of educational materials includes:

- 2005 USPSTF BRCA Recommendation
- Michigan Informed Consent Law for Pre-symptomatic and Predictive Genetic testing
- Cancer Family History Guide©
- Directory of Michigan Cancer Genetic Counseling Services
- Model BRCA Policies with permission from:
  - Aetna
  - Priority Health
  - UnitedHealthcare
Summary

• Understand current status of Michigan health insurance policies for BRCA1/2 testing with respect to USPSTF guidelines
  – 11 out of 24 health plans with written policies for BRCA coverage
  – 9 in alignment with USPSTF recommendations
• Increase the number of health plans that have policies consistent with USPSTF guidelines
  – Increased the number of health plans that have policies consistent with USPSTF recommendations from four to nine out of 24 Michigan plans
• CDC Division of Cancer Prevention & Control using process as a model to investigate BRCA health plan policies in 20 states in 2011
“…no important health problem will be solved by clinical care alone, or research alone, or by public health alone- But rather by all public and private sectors working together…..”

JS Marks. Managed Care 2005;14:p11
Supplement on “The Future of Public Health”
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