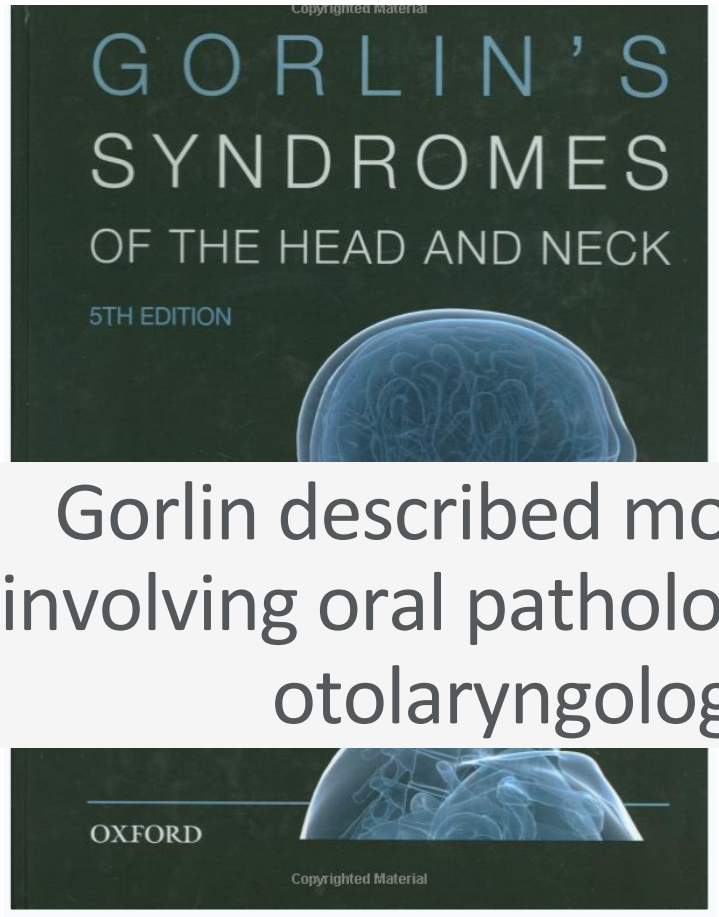
Northwestern Medicine®
Feinberg School of Medicine

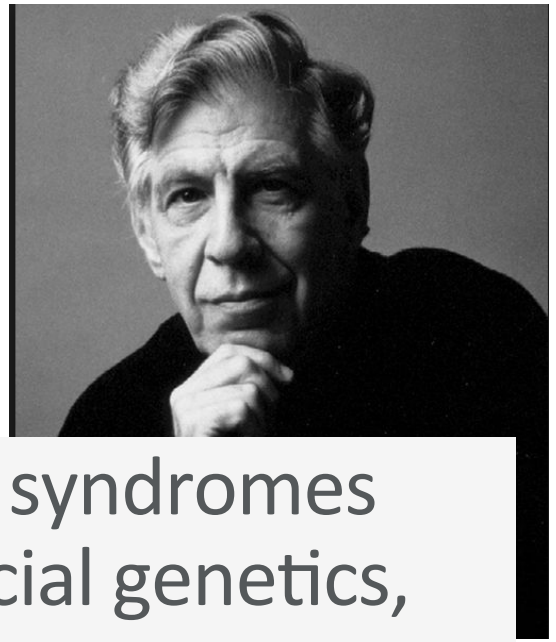
Implementation of Public Health Genomics & Possible Future Dental Applications

Debra Duquette, MS, CGC
Graduate Program in Genetic Counseling
Feinberg School of Medicine
Center for Genetic Medicine
Northwestern University

January 26, 2017

Robert (Bob) Gorlin, DDS, MS, 1923-2006





Gorlin described more than 100 syndromes involving oral pathology, craniofacial genetics, otolaryngology and obstetrics

"In human genetics, we think Bob belongs to us, but the dentists, the pathologists, the dermatologists, the oncologists, the reconstructive surgeons, and the craniofacial specialists all think he belongs to them too." - Judy Hall

Multitude of Genetic Conditions with Dental Implications

- Of the approximately 5,500 known human genetic orders,
  - More than 700 are craniofacial disorders
  - More than 200 genes involved in the embryogenic development, morphogenesis and differentiation of the teeth
  - Chromosome abnormalities, genetic syndromes, and non-syndromic isolated and multifactorial genetic factors
  - Consider if dental phenotype isolated finding or possible associated with syndrome with broader clinical implications
    - Evaluate family and medical history
    - Consider associations with other conditions (i.e. oligodontia and colon cancer, enamel defects and kidney disease, microdontia and deafness)
    - Consider routinely collecting genetic conditions with potential oral health implictions
    - Consider referral to genetic counselor or other specialists
    - When possible dentist should be part of personalized medicine team
  - However, only one dental school requires one semester of molecular biology or genetics for admission (Hart & Hart, 2016)

Hart PS & Hart TC, Molecular Genetics and Genomic Medicine, 2016, 123-125

What is Genetic Counseling?

- Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:
  - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
  - Education about inheritance, testing, management, prevention, resources and research.
  - Counseling to promote informed choices and adaptation to the risk or condition.

Resta et al, J Genet Counseling, 2006 Apr;15(2):77-83

Genetic Counseling Specialties

Historically	Adult	Treatment
• Prenatal • Pediatrics	• Cancer • Neurogenetic • Cardiology • Psychiatry • Endocrine • General	• Somatic sequencing/with or without germline to dictate treatment • Germline sequencing to dictate treatment • Pharmacogenomics

- Industry
- Laboratory utilization
- Research
- Public Health

National Society of Genetic Counselors

ABOUT US | EVENTS | PUBLICATIONS | NEWSROOM | JOB CONNECTION | EDUCATION | BECOME A MEMBER

Find a Genetic Counselor

This directory has been developed to assist physicians, patients and genetic counselors in accessing genetic counseling services.

- DISCLAIMER
- SEARCH TIPS
- MEET BY PHONE
- MEET IN PERSON
- ABOUT GENETIC COUNSELORS
- PATIENT RESOURCE SITE
- MEMBER DIRECTORY

FIND A GENETIC COUNSELOR

The Find a Genetic Counselor directory offers access to over 3,300 genetic counselors (US and Canada).

Check with your insurance company to verify coverage of genetic counseling, testing and authorized providers. For more information, visit AboutGeneticCounselors.com.

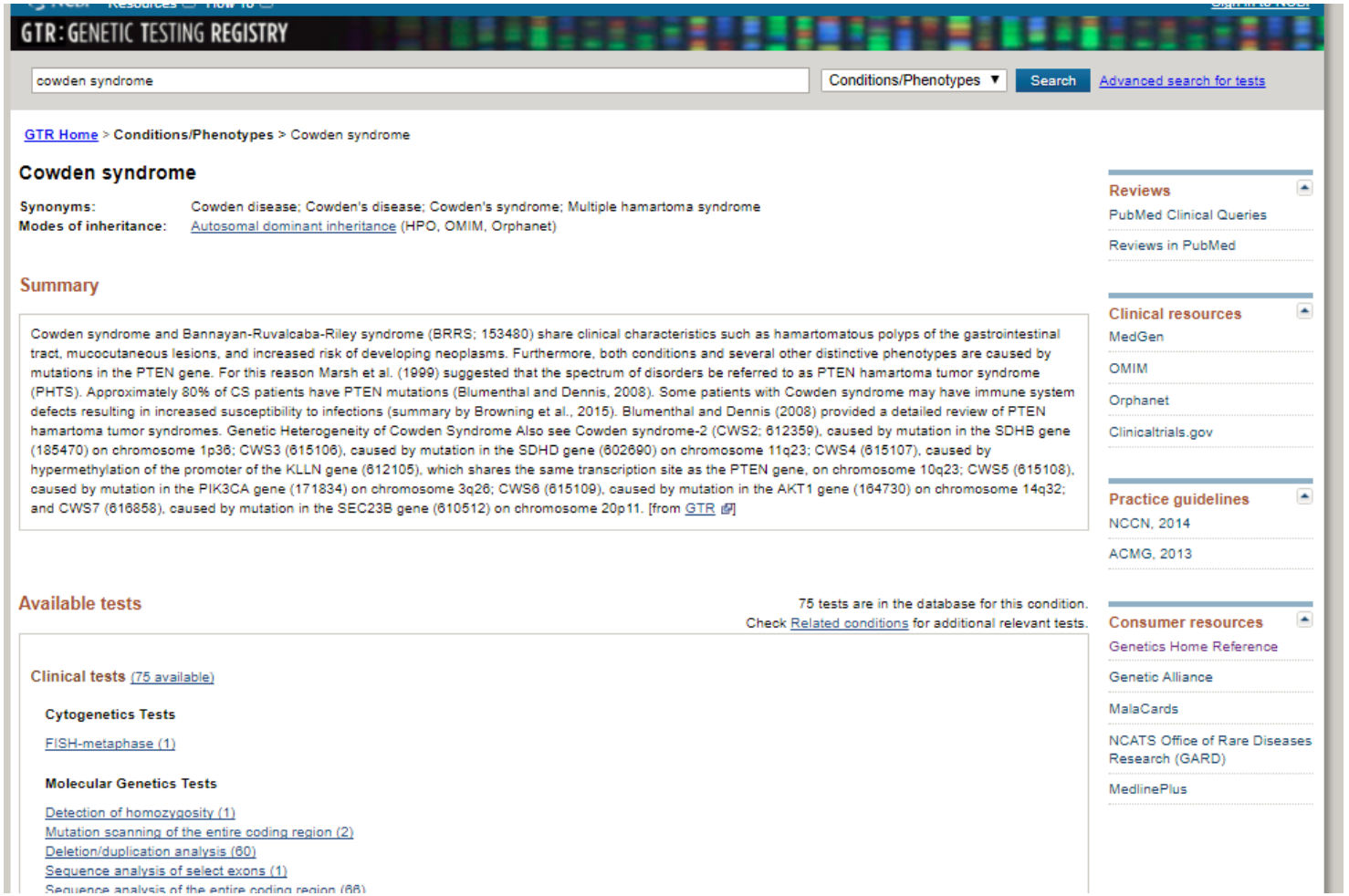
To start your search, first tell us **how you would prefer to meet with a genetic counselor:**

Additional searches:

- If you are a student, healthcare provider or other individual interested in speaking with a genetic counselor, click here.
- NSGC members are offered an expanded directory that contains additional information for use in searching for colleagues. Access the NSGC Member Directory.

https://www.nsgc.org/page/find-a-genetic-counselor

GTR: Genetic Testing Registry



https://www.ncbi.nlm.nih.gov/gtr/conditions/C0018553/

NATIONAL SOCIETY OF GENETIC COUNSELING (NSGC) VISION

"Integrating genetics and genomics to improve health for all"

-NSGC 2016-2018 Strategic Plan

http://www.nsgc.org/page/about-nsgc

…FOR ALL

- ~324 million people of the United States
- Ensure access to all regardless of race, gender, income, geography, and ability to pay
- ~5,600 hospitals in the United States
  - ~4,900 community hospitals
    - ~1,800 rural community hospitals
    - ~3,185 part of health system
    - ~1,700 part of health network
- 61 dentists practicing per 100,000 US population (2017)

http://www.aha.org/research/rc/stat-studies/fast-facts.shtm
https://www.ada.org/en/science-research/health-policy-institute/dental-statistics/workforce

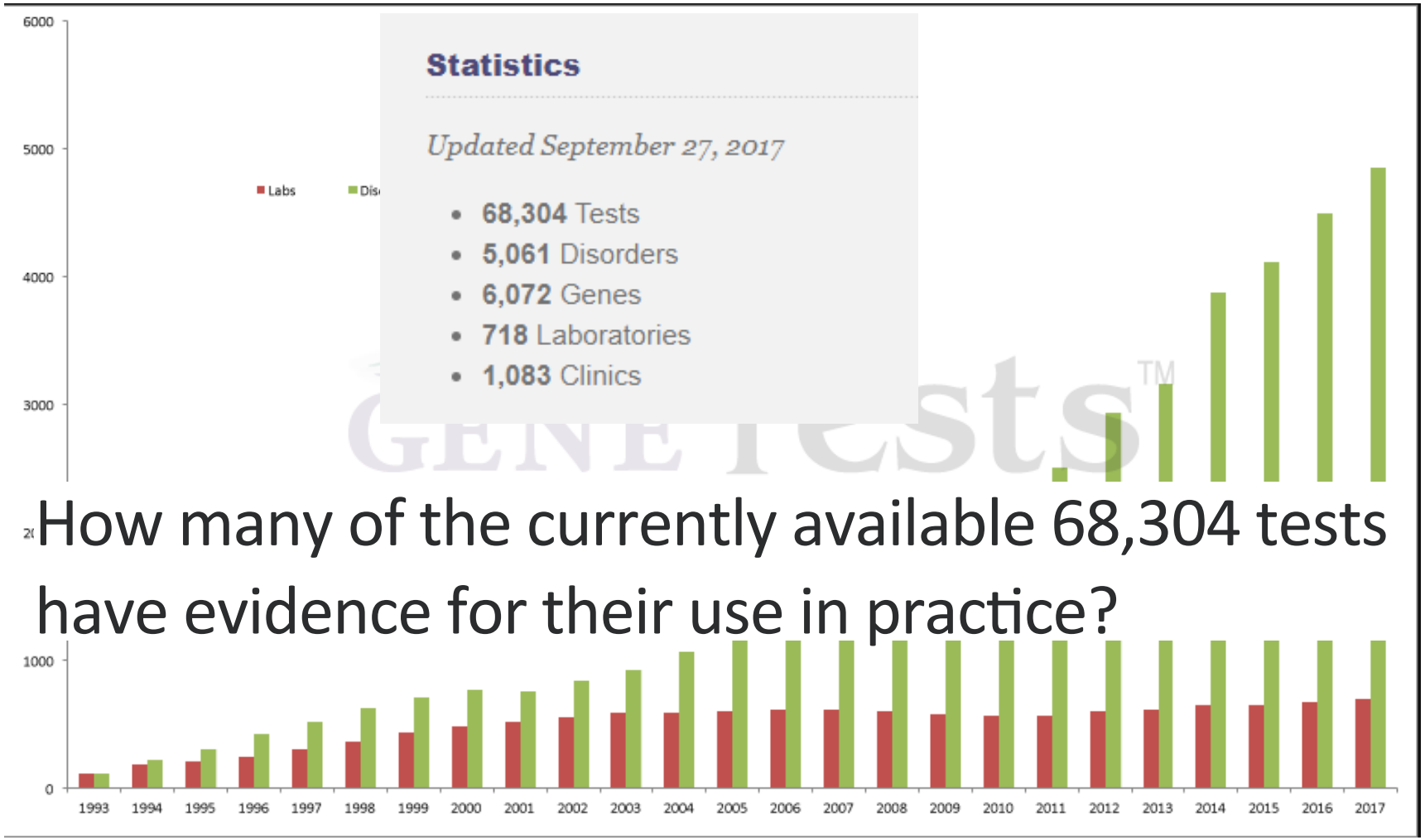
VISION FOR PUBLIC HEALTH GENOMICS

"Genomics will be to the 21st century what infectious disease was to the 20th century…

Genomics should be considered in every facet of public health: infectious disease, chronic disease, occupational health, environmental health, in addition to maternal and child health"

Gerard et al. Journal Law, Medicine , Ethics 2002; vol 30(suppl):173-176

Increase in Number of Genetic Tests & Disorders



How many of the currently available 68,304 tests have evidence for their use in practice?

https://www.genetests.org/

Key Questions to Consider about Genetic Tests

- ✓ How valid and reliable are the genomic tests/services?(**analytic validity**)
- ✓ How well does the test/service predict outcomes? (**clinical validity**)
- ✓ What are the benefits and harms when the test/service is used to influence patient management? (**clinical utility**)
- ✓ How should the medical community, public health, policy makers respond? (**ethical, legal, social issues**)

https://www.cdc.gov/genomics/gtesting/acce/

Public Health Genomics Knowledge Base (v1.0)

PHGKB

About

Genomics & Health Impact Weekly Scan (Current Edition)

Advanced Molecular Detection Weekly Clips (Current Edition)

Genomics (A-Z)

Evidence Classification Database

https://phgkb.cdc.gov/PHGKB/topicStartPage.action

Last data update: Dec 16, 2015. (Total: 159 Documents)

mody Search

Note: Simple Boolean operators are all

Recommend Tweet Share

Classification Criteria

Tier 1

- FDA label requires use of test to inform choice or dose of a drug
- CMS covers testing
- Clinical practice guidelines based on systematic review supports testing

Tier 2

- FDA label mentions biomarkers
- CMS coverage with evidence development
- Clinical practice guideline, not based on systematic review, supports use of test
- Clinical practice guideline finds insufficient evidence but does not discourage use of test
- Systematic review, without clinical practice guideline, supports use of test
- Systematic review finds insufficient evidence but does not discourage use of test
- Clinical practice guideline recommends dosage adjustment, but does not address testing

Tier 3

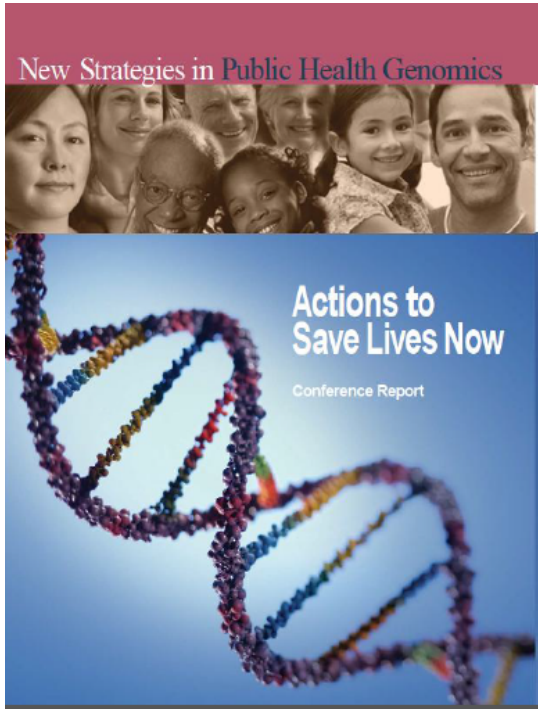
- FDA label cautions against use
- CMS decision against coverage
- Clinical practice guideline recommends against use of test
- Clinical practice guideline finds insufficient evidence and discourages use of test
- Systematic review recommends against use
- Systematic review finds insufficient evidence and discourages use
- Evidence available only from published studies without systematic reviews, clinical practice guidelines, FDA label or CMS labels coverage decision

Small Number of Tests Known to Save Lives

- Tier 1 Tests per CDC Office of Public Health Genomics
  - Proven analytic validity, clinical validity and clinical utility
  - Can save lives!
  - Often underused in clinical practice

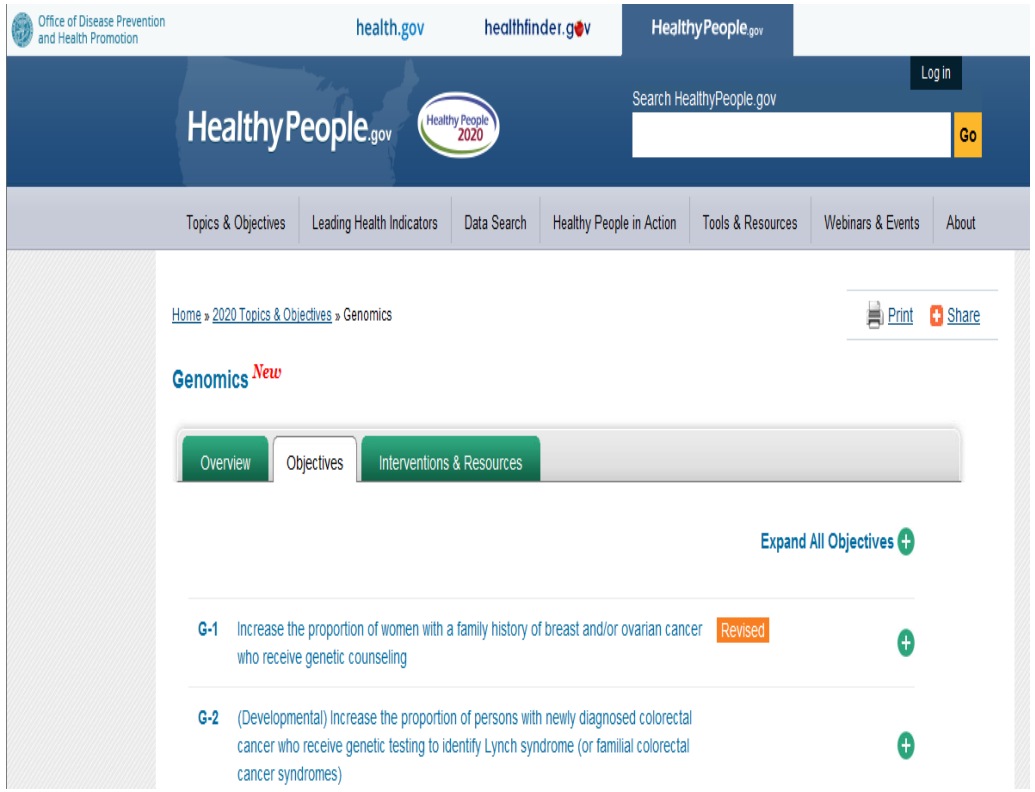
- >48 genomic tests supported by evidence for use in practice
  - >35 cancer-related tests
  - Examples include cascade testing of relatives of people with FH; universal Lynch syndrome screening on newly diagnosed colorectal cancer; BRCA counseling with consideration of testing for women with significant family history

- Many intended uses include
  - Diagnosis
  - Prognosis
  - Risk prediction to inform prevention
  - Treatment, including choice of medication and dosage
  - Screening



Centers for Disease Control and Prevention. genomicsforum.org/editoruploads/ActionstoSaveLivesNowReport.pdf

Healthy People 2020
(HP 2020) Cancer Genomics Objectives



- HP 2020 marks first time for genomics objectives
  - Drafted by multiple federal agencies and one state health department (MDHHS) in 2009 and approved by HP2020 in 2010

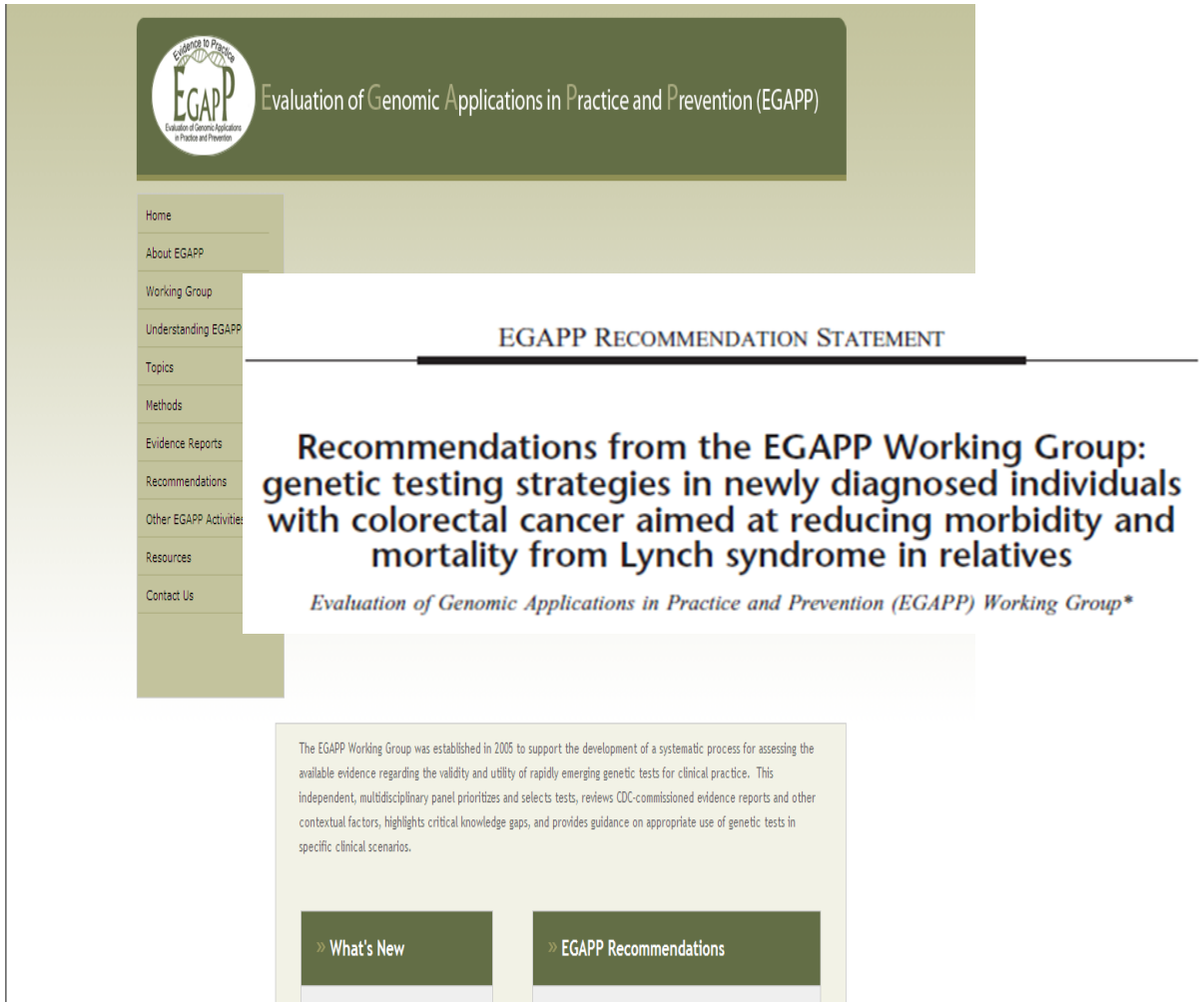
- Increase the proportion of women with a family history of breast and/or ovarian cancer who receive **genetic counseling**

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

http://www.healthypeople.gov/2020/topics-objectives/topic/genomics/objectives

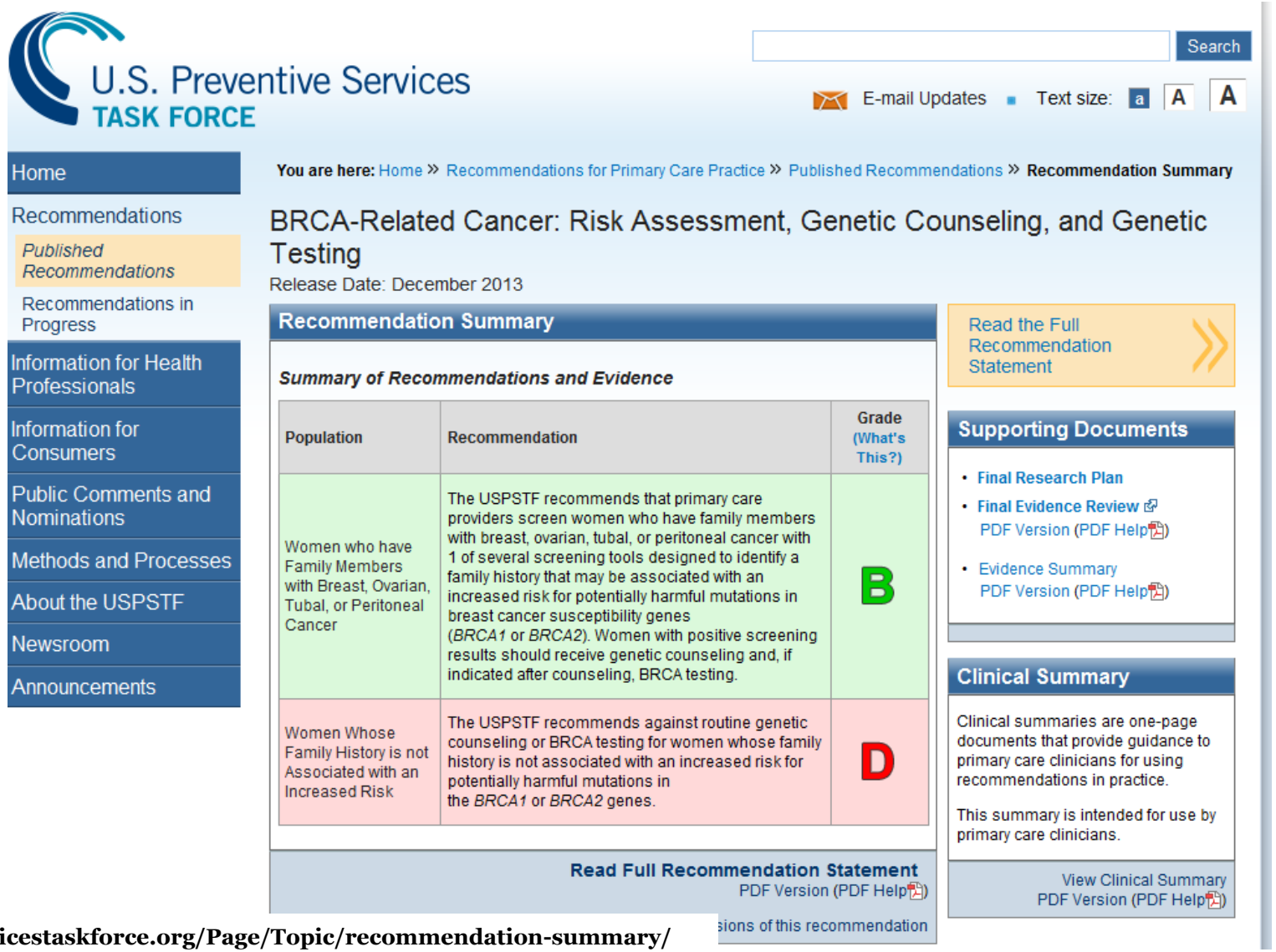
2009 EGAPP Recommendation on Genetic Testing for Lynch Syndrome

- 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

2013 USPSTF *BRCA* EVIDENCE-BASED RECOMMENDATIONS (UPDATED FROM 2005)



U.S. Preventive Services TASK FORCE

Search

E-mail Updates | Text size: a A A

Home
Recommendations
Published Recommendations
Recommendations in Progress
Information for Health Professionals
Information for Consumers
Public Comments and Nominations
Methods and Processes
About the USPSTF
Newsroom
Announcements

You are here: Home » Recommendations for Primary Care Practice » Published Recommendations » Recommendation Summary

BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing

Release Date: December 2013

Recommendation Summary

Summary of Recommendations and Evidence

Population	Recommendation	Grade (What's This?)
Women who have Family Members with Breast, Ovarian, Tubal, or Peritoneal Cancer	The USPSTF recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with 1 of several screening tools designed to identify a family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (*BRCA1* or *BRCA2*). Women with positive screening results should receive genetic counseling and, if indicated after counseling, BRCA testing.	B
Women Whose Family History is not Associated with an Increased Risk	The USPSTF recommends against routine genetic counseling or BRCA testing for women whose family history is not associated with an increased risk for potentially harmful mutations in the *BRCA1* or *BRCA2* genes.	D

Read Full Recommendation Statement
PDF Version (PDF Help)

...sions of this recommendation

Read the Full Recommendation Statement

Supporting Documents

- Final Research Plan
- Final Evidence Review
  PDF Version (PDF Help)
- Evidence Summary
  PDF Version (PDF Help)

Clinical Summary

Clinical summaries are one-page documents that provide guidance to primary care clinicians for using recommendations in practice.

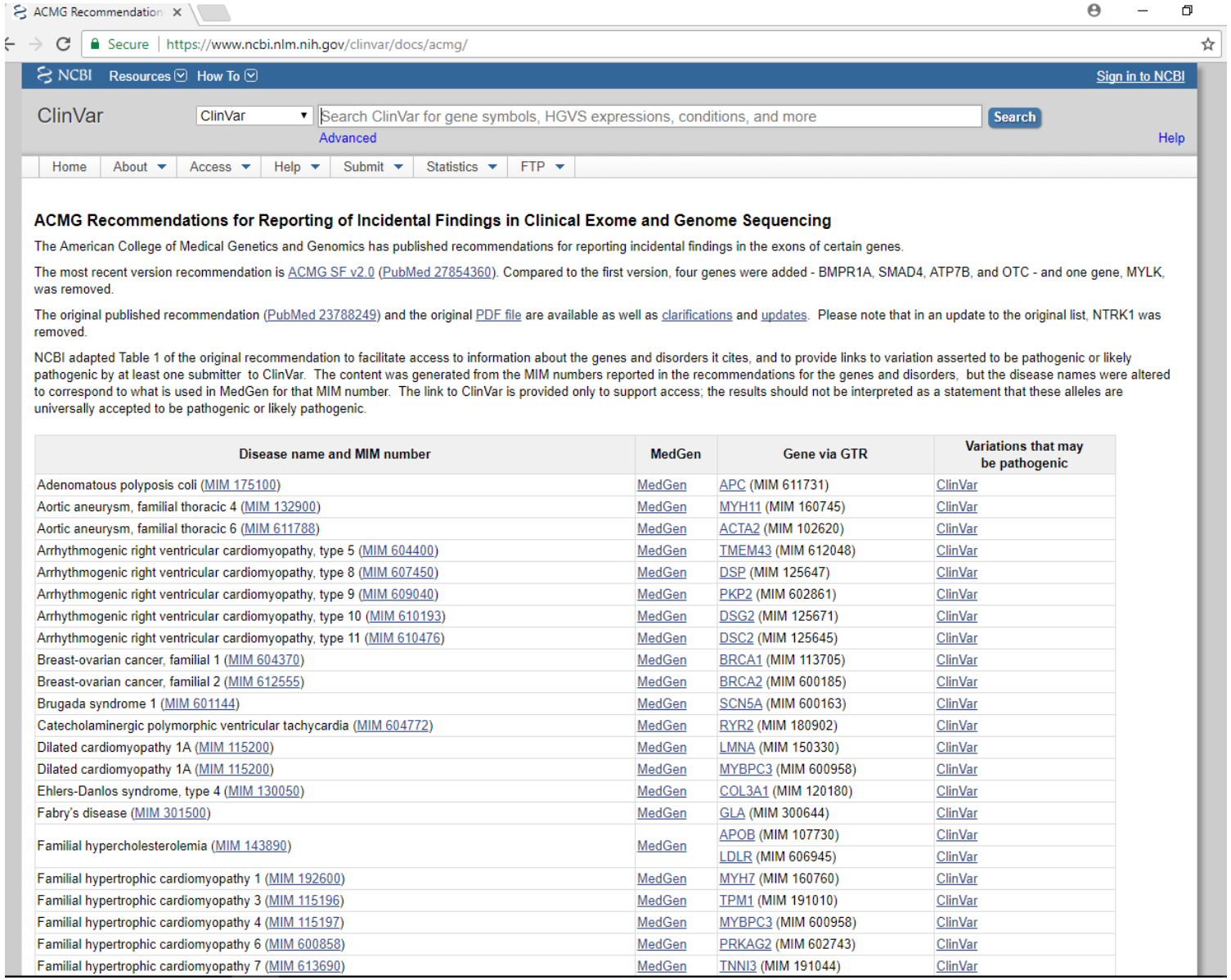
This summary is intended for use by primary care clinicians.

View Clinical Summary
PDF Version (PDF Help)

http://www.uspreventiveservicestaskforce.org/Page/Topic/recommendation-summary/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing

American College of Medical Genetics and Genomics (ACMG) Secondary Findings Recommendations

- Recommends labs report secondary findings when perform clinical exome and genome sequencing tests on list of genes known to cause severe disease and clinically relevant actions available
- List currently includes 59 genes (updated in 2016)
- ACMG working group curate and updates list periodically



ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing

The American College of Medical Genetics and Genomics has published recommendations for reporting incidental findings in the exons of certain genes.

The most recent version recommendation is ACMG SF v2.0 (PubMed 27854360). Compared to the first version, four genes were added - BMPR1A, SMAD4, ATP7B, and OTC - and one gene, MYLK, was removed.

The original published recommendation (PubMed 23788249) and the original PDF file are available as well as clarifications and updates. Please note that in an update to the original list, NTRK1 was removed.

NCBI adapted Table 1 of the original recommendation to facilitate access to information about the genes and disorders it cites, and to provide links to variation asserted to be pathogenic or likely pathogenic by at least one submitter to ClinVar. The content was generated from the MIM numbers reported in the recommendations for the genes and disorders, but the disease names were altered to correspond to what is used in MedGen for that MIM number. The link to ClinVar is provided only to support access; the results should not be interpreted as a statement that these alleles are universally accepted to be pathogenic or likely pathogenic.

Disease name and MIM number	MedGen	Gene via GTR	Variations that may be pathogenic
Adenomatous polyposis coli (MIM 175100)	MedGen	APC (MIM 611731)	ClinVar
Aortic aneurysm, familial thoracic 4 (MIM 132900)	MedGen	MYH11 (MIM 160745)	ClinVar
Aortic aneurysm, familial thoracic 6 (MIM 611788)	MedGen	ACTA2 (MIM 102620)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 5 (MIM 604400)	MedGen	TMEM43 (MIM 612048)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 8 (MIM 607450)	MedGen	DSP (MIM 125647)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 9 (MIM 609040)	MedGen	PKP2 (MIM 602861)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 10 (MIM 610193)	MedGen	DSG2 (MIM 125671)	ClinVar
Arrhythmogenic right ventricular cardiomyopathy, type 11 (MIM 610476)	MedGen	DSC2 (MIM 125645)	ClinVar
Breast-ovarian cancer, familial 1 (MIM 604370)	MedGen	BRCA1 (MIM 113705)	ClinVar
Breast-ovarian cancer, familial 2 (MIM 612555)	MedGen	BRCA2 (MIM 600185)	ClinVar
Brugada syndrome 1 (MIM 601144)	MedGen	SCN5A (MIM 600163)	ClinVar
Catecholaminergic polymorphic ventricular tachycardia (MIM 604772)	MedGen	RYR2 (MIM 180902)	ClinVar
Dilated cardiomyopathy 1A (MIM 115200)	MedGen	LMNA (MIM 150330)	ClinVar
Dilated cardiomyopathy 1A (MIM 115200)	MedGen	MYBPC3 (MIM 600958)	ClinVar
Ehlers-Danlos syndrome, type 4 (MIM 130050)	MedGen	COL3A1 (MIM 120180)	ClinVar
Fabry's disease (MIM 301500)	MedGen	GLA (MIM 300644)	ClinVar
Familial hypercholesterolemia (MIM 143890)	MedGen	APOB (MIM 107730)	ClinVar
		LDLR (MIM 606945)	ClinVar
Familial hypertrophic cardiomyopathy 1 (MIM 192600)	MedGen	MYH7 (MIM 160760)	ClinVar
Familial hypertrophic cardiomyopathy 3 (MIM 115196)	MedGen	TPM1 (MIM 191010)	ClinVar
Familial hypertrophic cardiomyopathy 4 (MIM 115197)	MedGen	MYBPC3 (MIM 600958)	ClinVar
Familial hypertrophic cardiomyopathy 6 (MIM 600858)	MedGen	PRKAG2 (MIM 602743)	ClinVar
Familial hypertrophic cardiomyopathy 7 (MIM 613690)	MedGen	TNNI3 (MIM 191044)	ClinVar

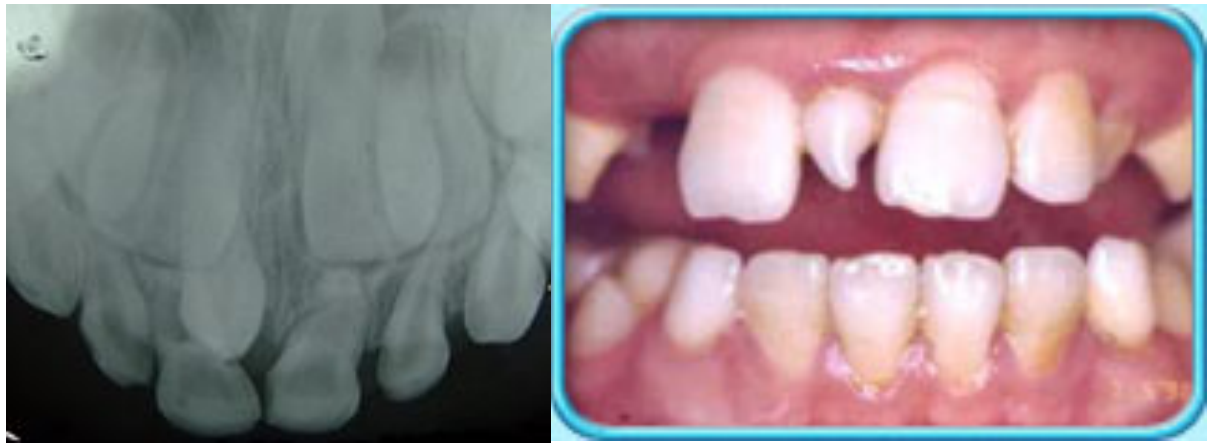
https://www.ncbi.nlm.nih.gov/clinvar/docs/acmg/

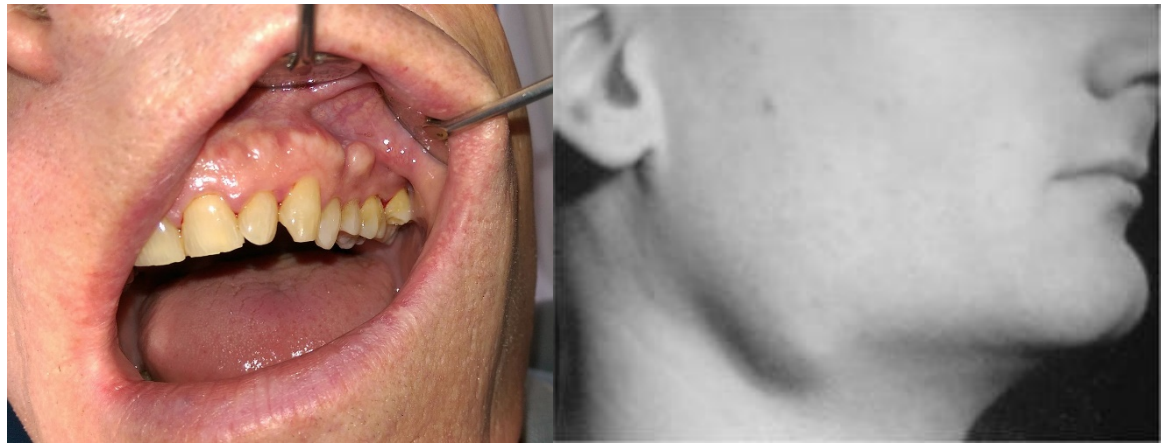
Examples of 59 ACMG Genes of Potential Importance to Precision Public Oral Health

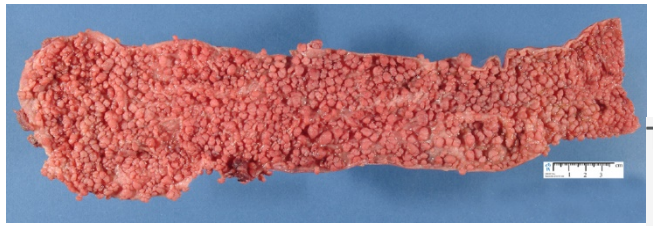
Condition	Gene	Relevance to Oral Health
Adenomatous polyposis coli (FAP)	APC	Teeth and tongue findings could be key to referral for genetic screening/diagnosis
Peutz-Jegher syndrome	STK11	Mouth features could be key to referral for genetic screening/diagnosis
PTEN hamartoma tumor syndrome (Cowden syndrome)	PTEN	Teeth and tongue findings could be key to referral for genetic screening/diagnosis
Long QT syndrome	KCNQ1, KCNH2, SCN5A	Medications and surgical environment for dental procedures
Malignant hyperthermia	CACNA1S	Medications and surgical environment (general anesthetics and stress) for dental procedures consider
Loeys-Dietz syndrome	TGFBR1, TGFBR2	Craniofacial features and bifid uvula/cleft palate consider for genetics referral
Brugada syndrome 1	SCN5A	Surgery complicated and consider avoiding local anesthesia during dental procedures

Adenomatous polyposis coli (FAP)

- Accounts for 1% of all colorectal cancers
- Autosomal dominant
  - 100% penetrance
  - Untreated polyposis leads to ~100% risk of cancer
- Risk of extracolonic tumors
  - Upper GI
  - Desmoids
  - **Osteomas (50-90% of patients)**
  - **Thyroid**
  - Brain
  - Hepatoblastoma
- **Dental anomalies**
  - **Supernumerary teeth (11-27% of patients with FAP)**
  - **Unerupted teeth**
  - **Congenital absence of one or more teeth**





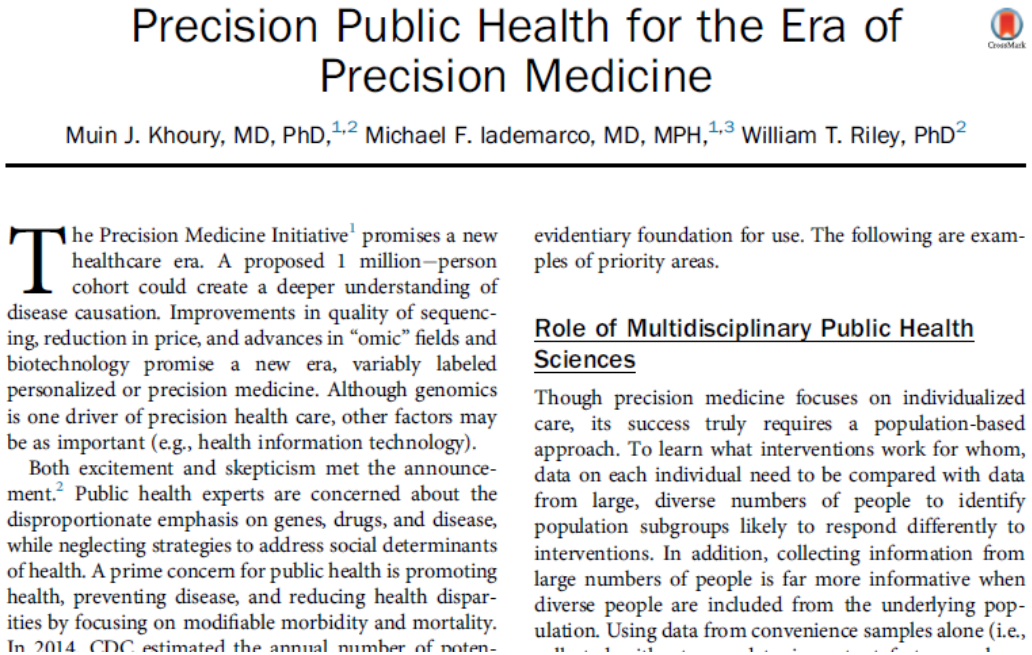


Thank you to Jess Stoll, MS, CGC
from University of Chicago for sharing slides

Precision Public Health
Can We Conduct Public Health Functions With More "Precision"?

The 3 Core Public Health Functions

- **Assessment**
  - More "precision" in measuring population health problems
- **Policy Development**
  - Developing the right intervention for the right population
- **Assurance**
  - More "precision" in delivering interventions & addressing health disparities



Precision Public Health for the Era of Precision Medicine

Muin J. Khoury, MD, PhD,[1,2] Michael F. Iademarco, MD, MPH,[1,3] William T. Riley, PhD[2]

The Precision Medicine Initiative[1] promises a new healthcare era. A proposed 1 million–person cohort could create a deeper understanding of disease causation. Improvements in quality of sequencing, reduction in price, and advances in "omic" fields and biotechnology promise a new era, variably labeled personalized or precision medicine. Although genomics is one driver of precision health care, other factors may be as important (e.g., health information technology).

Both excitement and skepticism met the announcement.[2] Public health experts are concerned about the disproportionate emphasis on genes, drugs, and disease, while neglecting strategies to address social determinants of health. A prime concern for public health is promoting health, preventing disease, and reducing health disparities by focusing on modifiable morbidity and mortality. In 2014, CDC estimated the annual number of potentiary foundation for use. The following are examples of priority areas.

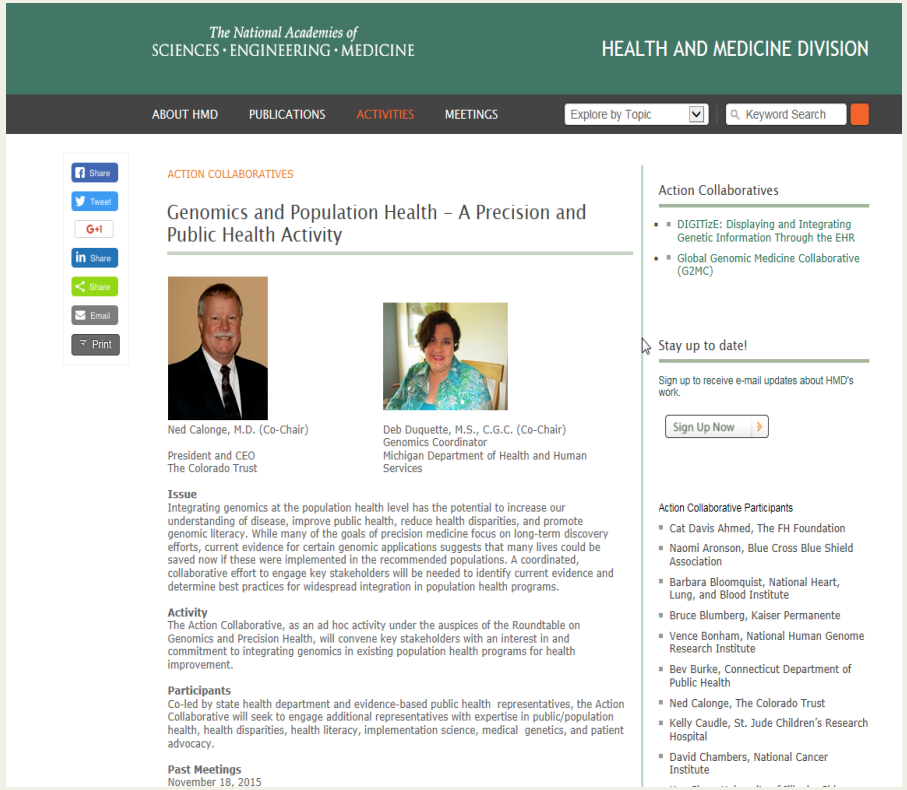
Role of Multidisciplinary Public Health Sciences

Though precision medicine focuses on individualized care, its success truly requires a population-based approach. To learn what interventions work for whom, data on each individual need to be compared with data from large, diverse numbers of people to identify population subgroups likely to respond differently to interventions. In addition, collecting information from large numbers of people is far more informative when diverse people are included from the underlying population. Using data from convenience samples alone (i.e.,

AJPM, 2016

THE NATIONAL ACADEMY OF MEDICINE ACTION COLLABORATIVE ON GENOMICS & POPULATION HEALTH

- *Goal:* To explore opportunities for genetics and genomics-based research to improve public health, reduce health disparities, and promote genomic literacy
  - Formed in 2016 following diverse stakeholders meeting
  - Significant increases in membership and activities in first year
  - Focus of first year on state public health; evidence-based approaches to improve early detection and clinical care of individuals with pathogenic variants for *BRCA* and Lynch syndrome
  - Second year begin to focus on health systems with three work streams on implementation, cascade screening and population genetic screening
  - *Please contact Deb if would like to join*



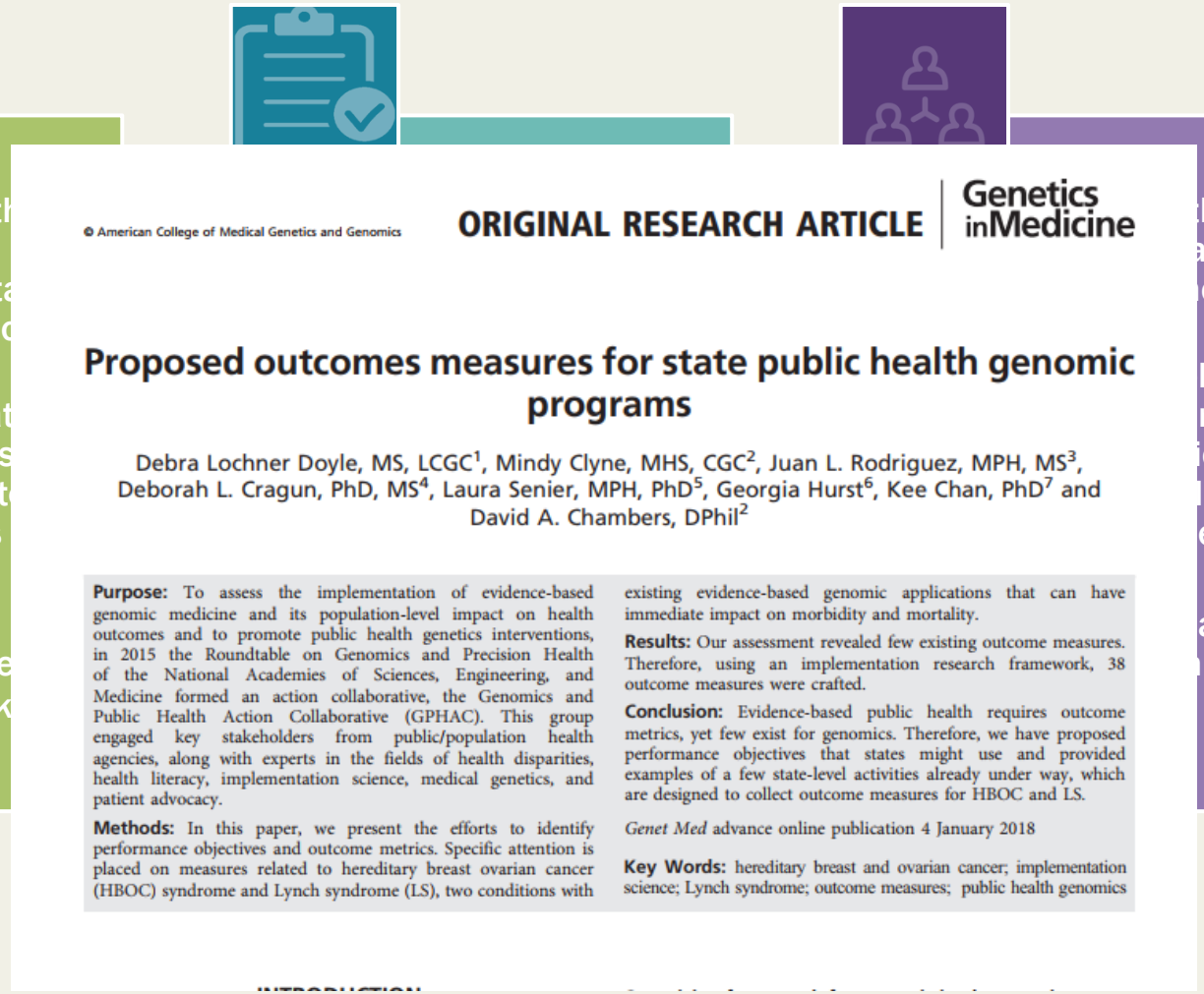
http://www.nationalacademies.org/hmd/Activities/Research/GenomicBasedResearch/Innovation-Collaboratives/Genomics-and-Population-Health.aspx

IMPLEMENTATION WORKGROUP, YEAR 1-2 PROJECTS

PROJECT 1

- Assess factors th
  determine the
  'readiness' of sta
  to carry out genc
  programs
- Perform qualitat
  interviews with s
  health officials t
  identify barriers
  facilitators

- Led by: Laura Se
  and Ridgely Fisk
  Green



ORIGINAL RESEARCH ARTICLE | Genetics inMedicine

Proposed outcomes measures for state public health genomic programs

Debra Lochner Doyle, MS, LCGC[1], Mindy Clyne, MHS, CGC[2], Juan L. Rodriguez, MPH, MS[3], Deborah L. Cragun, PhD, MS[4], Laura Senier, MPH, PhD[5], Georgia Hurst[6], Kee Chan, PhD[7] and David A. Chambers, DPhil[2]

Purpose: To assess the implementation of evidence-based genomic medicine and its population-level impact on health outcomes and to promote public health genetics interventions, in 2015 the Roundtable on Genomics and Precision Health of the National Academies of Sciences, Engineering, and Medicine formed an action collaborative, the Genomics and Public Health Action Collaborative (GPHAC). This group engaged key stakeholders from public/population health agencies, along with experts in the fields of health disparities, health literacy, implementation science, medical genetics, and patient advocacy.

Methods: In this paper, we present the efforts to identify performance objectives and outcome metrics. Specific attention is placed on measures related to hereditary breast ovarian cancer (HBOC) syndrome and Lynch syndrome (LS), two conditions with existing evidence-based genomic applications that can have immediate impact on morbidity and mortality.

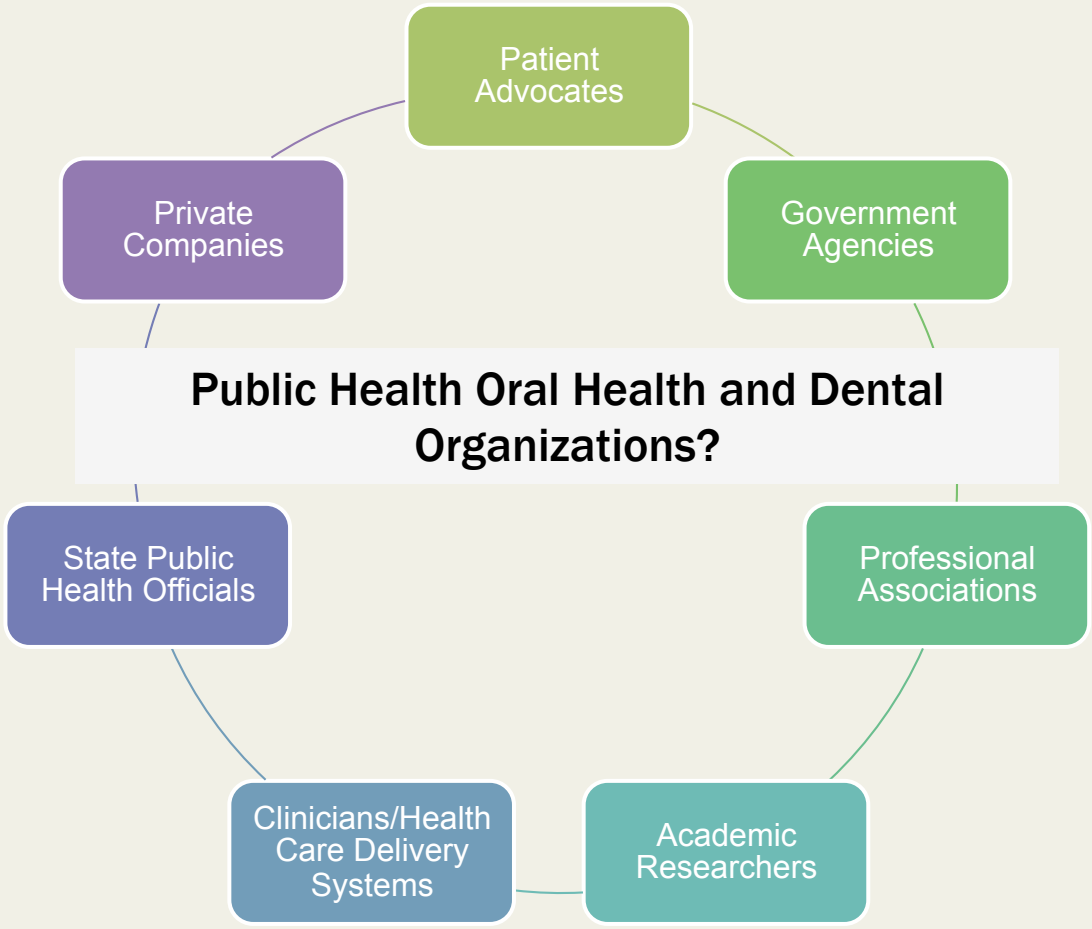
Results: Our assessment revealed few existing outcome measures. Therefore, using an implementation research framework, 38 outcome measures were crafted.

Conclusion: Evidence-based public health requires outcome metrics, yet few exist for genomics. Therefore, we have proposed performance objectives that states might use and provided examples of a few state-level activities already under way, which are designed to collect outcome measures for HBOC and LS.

Genet Med advance online publication 4 January 2018

Key Words: hereditary breast and ovarian cancer; implementation science; Lynch syndrome; outcome measures; public health genomics

Collaboration Among Many Stakeholders



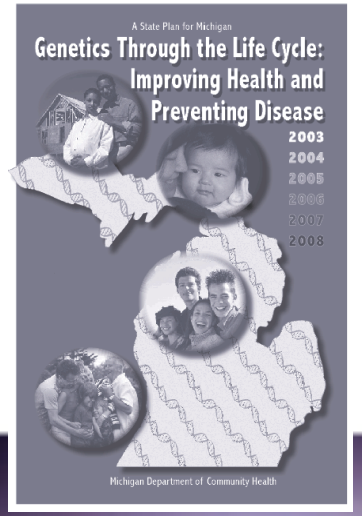
"...no important health problem will be solved by clinical care alone, or research alone, or by public health alone- But rather by all public and private sectors working together"

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

CDC State Cooperative Agreements in Cancer Genomics

- **Enhancing Cancer Genomic Best Practices through <u>Education, Surveillance, and Policy</u>**
- **Goal: Provide leadership and build capacity for cancer genomics activities in state public health departments**
  - 2003-2008: Michigan, Minnesota, Oregon, and Utah
  - 2008-2011: Michigan and Oregon
  - 2011-2014: Georgia, Michigan, and Oregon
  - 2011: Connecticut (Healthy People 2020 Action Award)
  - 2014-2019: Colorado, Connecticut, Michigan, Oregon, and Utah

http://www.cdc.gov/cancer/breast/what_cdc_is_doing/genomics_foa.htm



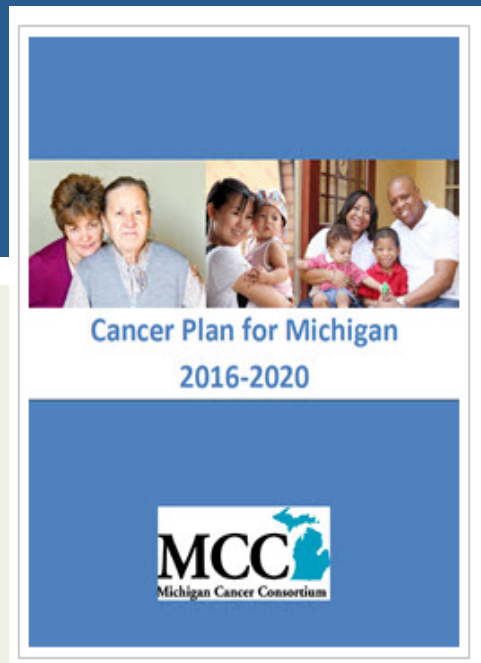
PUBLIC HEALTH GENOMICS IMPLEMENTATION TO SAVE LIVES: FROM NATIONAL VISION TO STATE SUCCESS

https://www.youtube.com/watch?v=OfjkY1lLxbE&feature=youtu.be

- Video created by CDC and Genetic Alliance
- Highlights Michigan as state public health genomics model
- Successful strategies highlighted:

1. Set Goals by Assessing Data and Available Resources
2. Build Partnerships
3. Conduct Surveillance
4. Provide Info to Policy Makers
5. Make Education Available to the Public
6. Implement Bi-directional Reporting
7. Conduct Surveillance and Assess Results

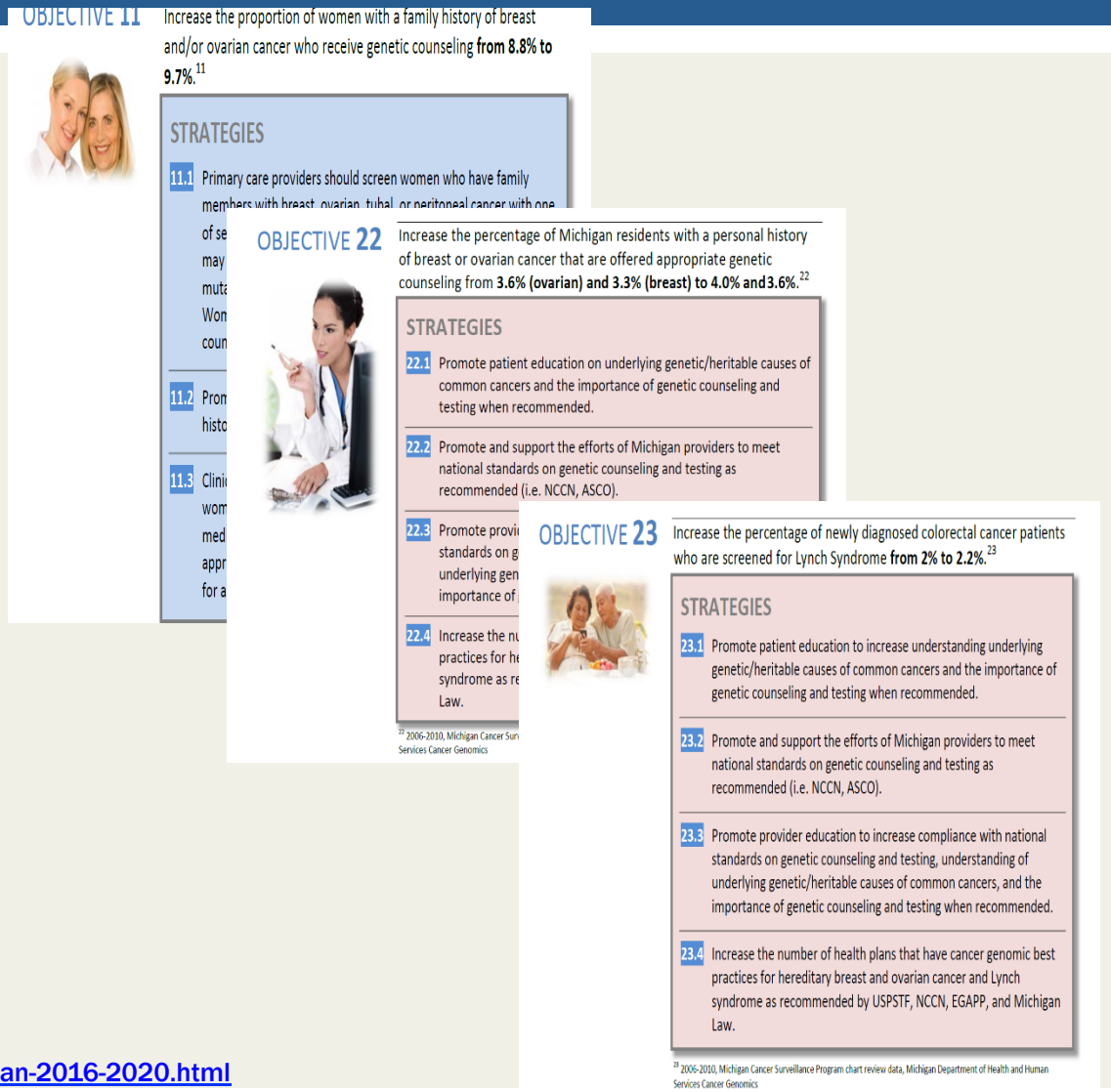
CANCER PLAN FOR MICHIGAN, 2016-2020



Goals

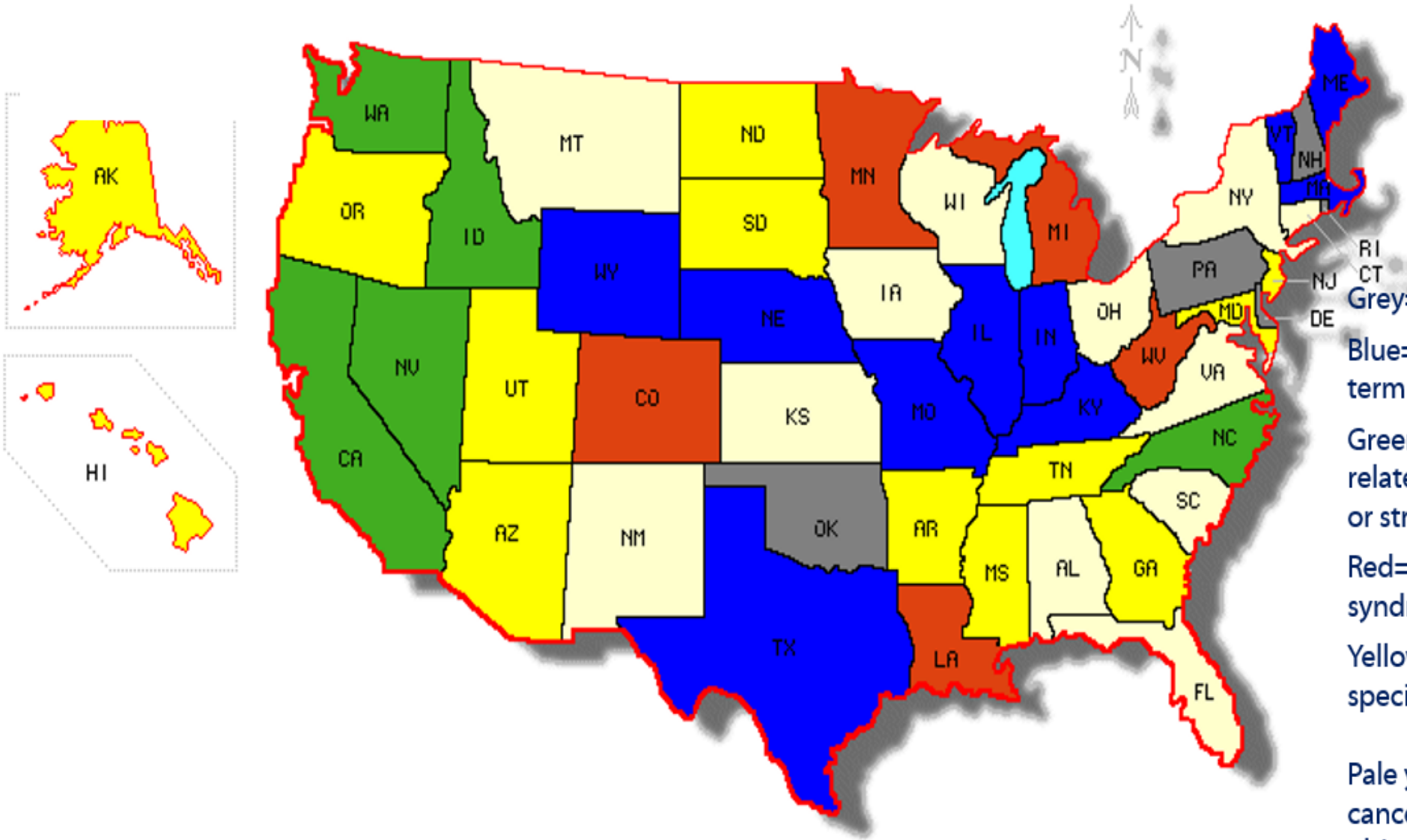
Broad general statements about the underlying purpose of the cancer plan. Modeled after the cancer care continuum, there are four goals for the cancer plan:

- **Prevent** cancer from occurring.
- Promote **early detection** of cancer using tests that have been shown to reduce mortality.
- **Diagnose and treat** all patients using the most effective and appropriate methods.
- Optimize **quality of life** for every person affected by cancer.



http://www.michigancancer.org/CancerPlan/ComprehensiveCancerControlPlan-2016-2020.html

Genomics in State Cancer Control Plans



Grey= None

Blue= genetics-related term

Green= + genomics-related goal, objective, or strategy

Red= + universal Lynch syndrome screening

Yellow=Lynch syndrome specific objective

Pale yellow=Hereditary cancer or FHH of cancer objective

Questions to Consider

- Do any state oral health plans have genomics-related or precision medicine goals? objectives? strategies?

- What are possible state oral health plan genomics-related or precision public health goals and/or objectives to promote?

Possible Idea?

ADA American Dental Association®
America's leading advocate for oral health

About the ADA | Contact Us | Join/Renew | Login

MEMBER CENTER EDUCATION/CAREERS SCIENCE/RESEARCH ADVOCACY PUBLICATIONS PUBLIC PROGRAMS

Oral Health Topics

Return to Oral Health Topics

Home > Member Center > Oral Health Topics > Genetics and Oral Health

Share

Oral Health Topics

Genetics and Oral Health

Key Points

- Many common diseases are not inherited as a single gene defect but instead result from gene-environment interactions.
- A predictive test for dental caries or for periodontal disease does not currently exist; both of these are complex diseases with multiple gene and environmental risk factors.
- No gene to date has been identified that has as large an impact on periodontal disease as do environmental influences, such as smoking or diabetes.
- While genetic testing holds potential for clinical application in the future, clinical measurements remain the best approach to assessment of caries and periodontal disease at this time.

https://www.ada.org/en/member-center/oral-health-topics/genetics-and-oral-health

Another Possible Idea?

ADA Adopts Policy on Genetic Testing

October 27, 2017

Contact Information:
mediarelations@ada.org

CHICAGO, October 27, 2017 — The American Dental Association at its annual meeting in Atlanta adopted a policy on genetic testing calling for insurers to:

- demonstrate that genetic tests used to determine eligibility for benefit coverage of specific oral health services are scientifically valid
- disclose financial relationships between manufacturer and payer
- be transparent about conflicts of interest between the test manufacturer, payer and study investigators
- provide independent third party agency confirmation of test validity and reliability for the intended purpose
- and an analysis of how utilization of the test will affect health outcomes and plan costs.

The policy states, "Health professions will experience a growth of such products and tests in the coming years and [dentists] will need a mechanism to assess the claims and counter claims so that we may best serve our patients and advocate for the needs of the public."

The Journal of the American Dental Association

Volume 146, Issue 3, March 2015, Pages 164-173.e4

Original Contributions

Genetic Screening

Interleukin 1 genetic tests provide no support for reduction of preventive dental care

Scott R. Diehl PhD, Fengshen Kuo MS, PhD, Thomas C. Hart DDS, PhD

Available online 25 February 2015.

https://www.ada.org/en/press-room/news-releases/2017-archives/october/ada-adopts-policy-on-genetic-testing

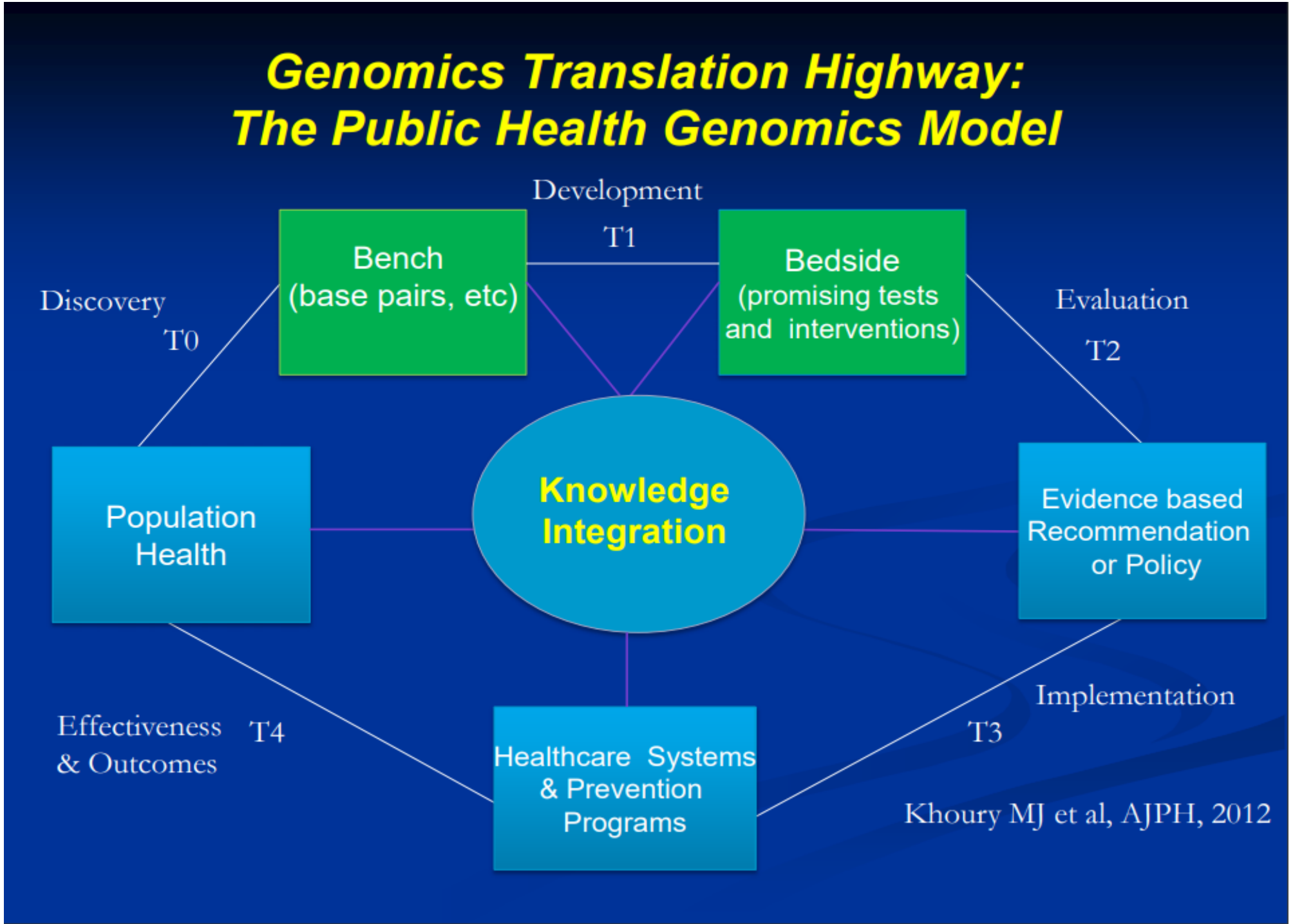

Genomics Translation Highway:
The Public Health Genomics Model
Development
T1
Bench
(base pairs, etc)
Bedside
(promising tests
and interventions)
Discovery
T0
Evaluation
T2
Knowledge
Integration
Population
Health
Evidence based
Recommendation
or Policy
Implementation
T3
Effectiveness
& Outcomes
T4
Healthcare Systems
& Prevention
Programs
Khoury MJ et al, AJPH, 2012

Table 1: The continuum of translational research in cancer genetics: types of research and examples from the portfolio analysis

From

How can we stimulate translational research in cancer genomics beyond bench to bedside?

Sheri D. Schully Ph

Genetics in Medicine

What part of translational highway has most genomics publications?

◂ back to article

Table 1: The continuum of translational research in cancer genetics: types of research and examples from the portfolio analysis

Translation research phase	Notation	Examples of types of research	Example from portfolio analysis
T0	Gene and other discoveries	GWASs, candidate gene studies	A GWAS of prostate cancer in African
TI	Discovery to candid		
T2	Health application to evidence-based practice guidelines	Phase III clinical trials; observational studies; evidence synthesis and guidelines development	Clinical utility of genetic markers associated with early-onset prostate cancer Programs in clinical effectiveness of cancer pharmacogenomics
T3	Practice guidelines to health practice	Dissemination research; implementation research; diffusion research; phase IV clinical trials	Sociocultural factors and BRCA genetic counseling for diverse Latinas Building a genome-enabled electronic medical record
T4	Practice to population health impact	Outcomes research; population monitoring of morbidity, mortality, benefits, and risks	Lung cancer model: risk, progression, and intervention

GWAS, genome-wide association study.

98% of published genomics research in T0 to T1;

Only 2% in T2 to T4

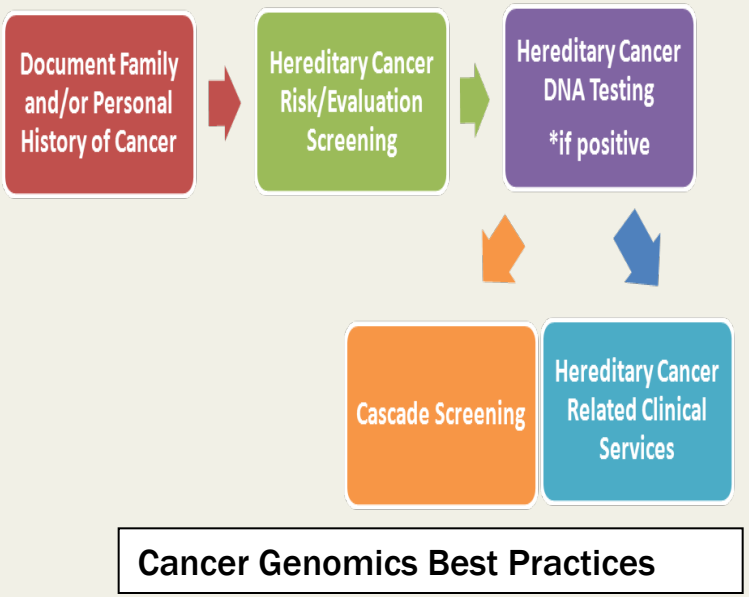
Tables index

PROMOTING SYSTEM CHANGE THROUGH EDUCATION, SURVEILLANCE & POLICY TO ADVANCE CANCER GENOMICS BEST PRACTICES IN MICHIGAN, 2014-2019

- **Purpose:**

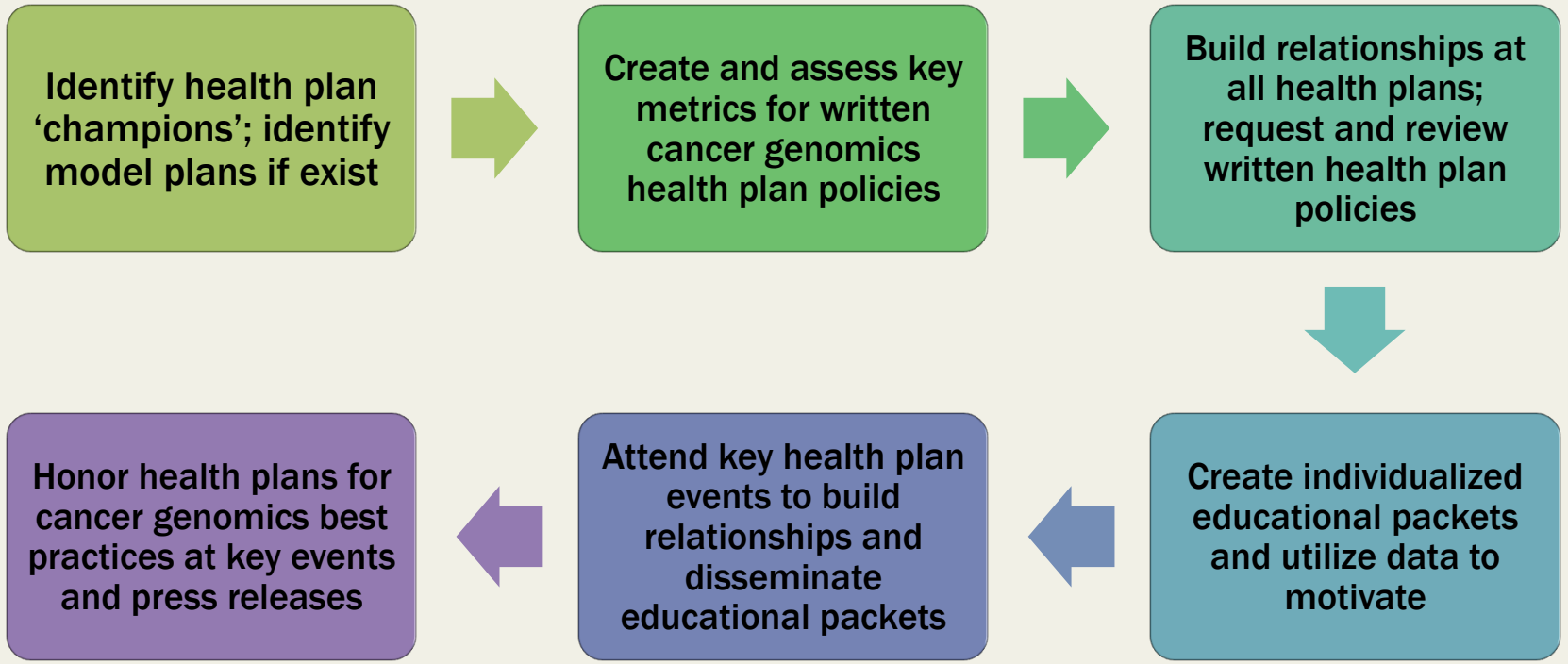
  Reduce breast, ovarian and colorectal cancer incidence and mortality rates by overcoming barriers and advancing health system changes to promote cancer genomics best practices

- **Short- and intermediate term outcomes:**
  - Increase knowledge among key clinical and policy stakeholders about cancer genetic best practices; improved access to and coverage of cancer genomics best practices **[Policy/system change]**
  - Improve ability to assess the burden of hereditary cancers and use of cancer genomics best practices; increased production and dissemination of periodic cancer surveillance reports. **[Surveillance]**
  - Increase knowledge of hereditary cancers and appropriate use of cancer genomics best practices among the public and health care providers. **[Education]**
  - Improve partnerships and coordination among key stakeholder groups regarding cancer genomics services and care. **[Partnerships]**

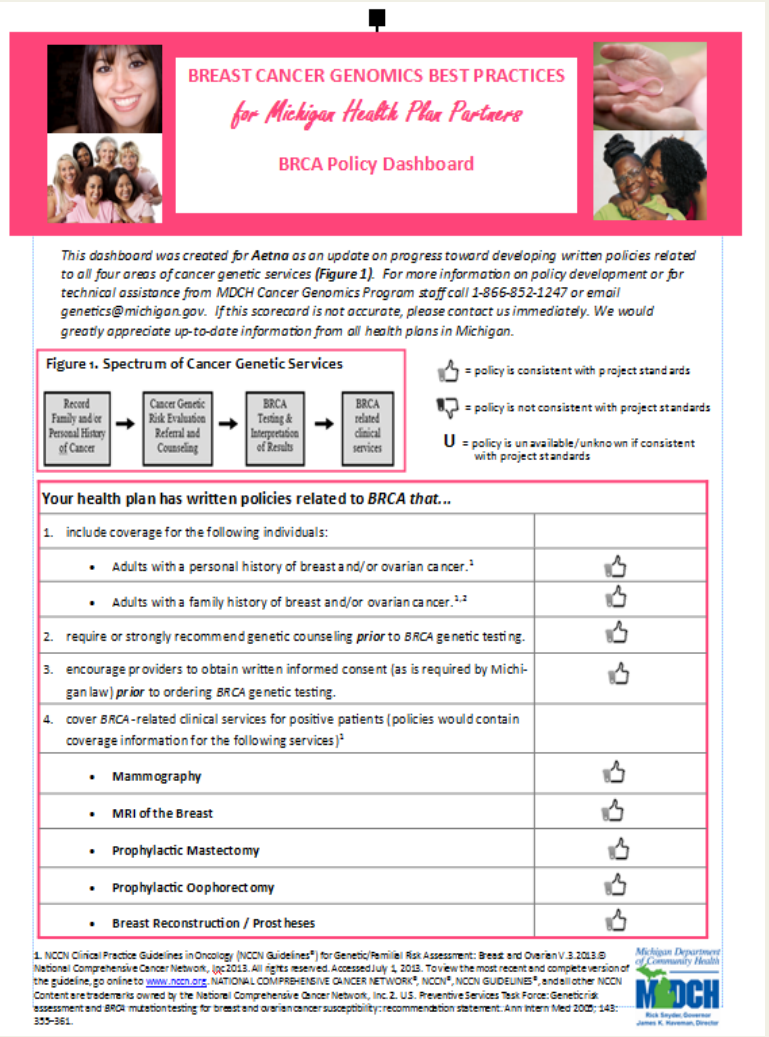


WRITTEN HEALTH PLAN POLICIES FOR HBOC BEST PRACTICES

Activity: Michigan Department of Health and Human Services (MDHHS) Cancer Genomics Program will continue to partner with Michigan Association of Health Plans and Michigan Cancer Genetics Alliance to recognize health plans that are aligned with Cancer Genomics Best Practices for Hereditary Breast and Ovarian Cancer and Lynch syndrome as recommended by USPSTF, NCCN, EGAPP and Michigan law



EXAMPLE OF *BRCA* HEALTH PLAN POLICY DASHBOARD & UTILIZING MICHIGAN CANCER GENETIC CLINICAL DATA, 2015



BREAST CANCER GENOMICS BEST PRACTICES

for Michigan Health Plan Partners

BRCA Policy Dashboard

*This dashboard was created for **Aetna** as an update on progress toward developing written policies related to all four areas of cancer genetic services **(Figure 1)**. For more information on policy development or for technical assistance from MDCH Cancer Genomics Program staff call 1-866-852-1247 or email genetics@michigan.gov. If this scorecard is not accurate, please contact us immediately. We would greatly appreciate up-to-date information from all health plans in Michigan.*

Figure 1. Spectrum of Cancer Genetic Services

Record Family and or Personal History of Cancer → Cancer Genetic Risk Evaluation Referral and Counseling → BRCA Testing & Interpretation of Results → BRCA related clinical services

= policy is consistent with project standards

= policy is not consistent with project standards

U = policy is unavailable/unknown if consistent with project standards

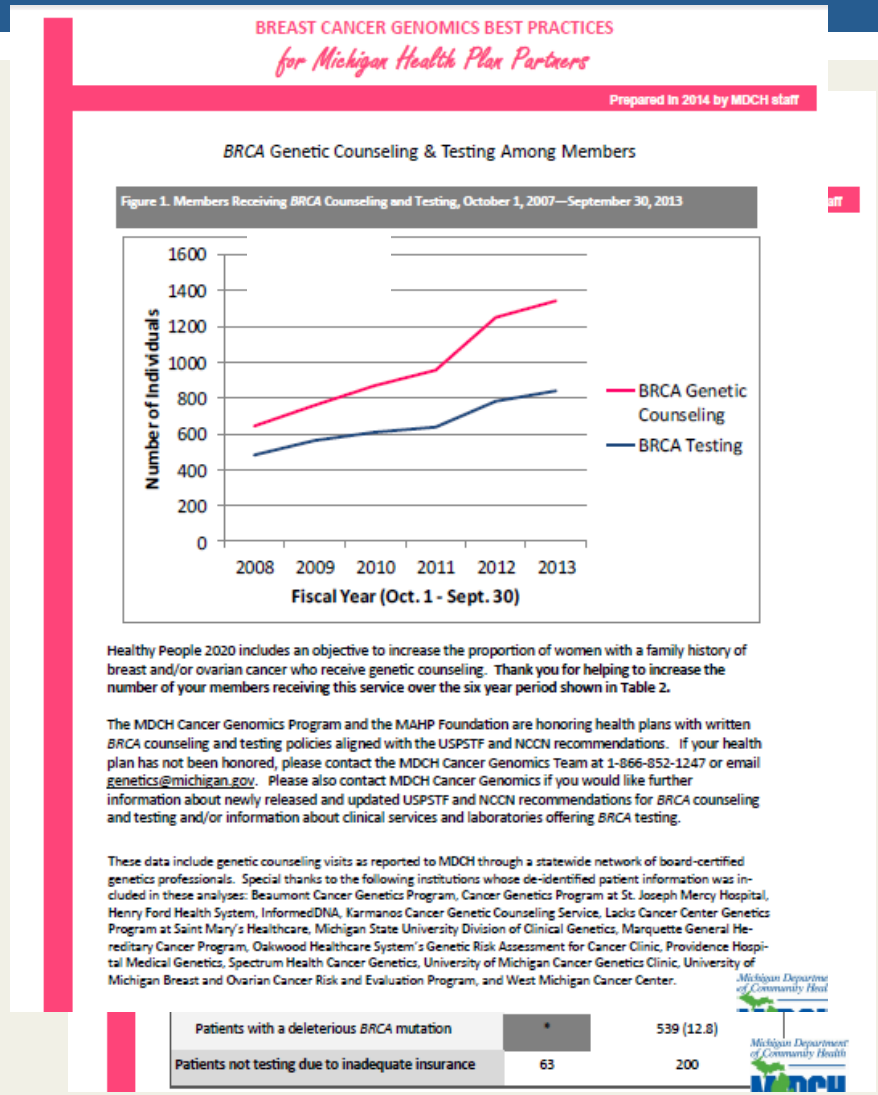
Your health plan has written policies related to *BRCA that...*	
1. include coverage for the following individuals:	
• Adults with a personal history of breast and/or ovarian cancer.[1]	👍
• Adults with a family history of breast and/or ovarian cancer.[1,2]	👍
2. require or strongly recommend genetic counseling *prior* to *BRCA* genetic testing.	👍
3. encourage providers to obtain written informed consent (as is required by Michigan law) *prior* to ordering *BRCA* genetic testing.	👍
4. cover *BRCA*-related clinical services for positive patients (policies would contain coverage information for the following services)[1]	
• Mammography	👍
• MRI of the Breast	👍
• Prophylactic Mastectomy	👍
• Prophylactic Oophorectomy	👍
• Breast Reconstruction / Prostheses	👍

1. NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial Risk Assessment: Breast and Ovarian V.3.2013.© National Comprehensive Cancer Network, Inc 2013. All rights reserved. Accessed July 1, 2013. To view the most recent and complete version of the guideline, go online to www.nccn.org. NATIONAL COMPREHENSIVE CANCER NETWORK®, NCCN®, NCCN GUIDELINES®, and all other NCCN Content are trademarks owned by the National Comprehensive Cancer Network, Inc. 2. U.S. Preventive Services Task Force: Genetic risk assessment and *BRCA* mutation testing for breast and ovarian cancer susceptibility: recommendation statement. Ann Intern Med 2005; 143: 355-361.

Michigan Department of Community Health

MDCH

Rick Snyder, Governor
James K. Haveman, Director



BREAST CANCER GENOMICS BEST PRACTICES

for Michigan Health Plan Partners

Prepared in 2014 by MDCH staff

BRCA Genetic Counseling & Testing Among Members

Figure 1. Members Receiving *BRCA* Counseling and Testing, October 1, 2007—September 30, 2013

Healthy People 2020 includes an objective to increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling. **Thank you for helping to increase the number of your members receiving this service over the six year period shown in Table 2.**

The MDCH Cancer Genomics Program and the MAHP Foundation are honoring health plans with written *BRCA* counseling and testing policies aligned with the USPSTF and NCCN recommendations. If your health plan has not been honored, please contact the MDCH Cancer Genomics Team at 1-866-852-1247 or email genetics@michigan.gov. Please also contact MDCH Cancer Genomics if you would like further information about newly released and updated USPSTF and NCCN recommendations for *BRCA* counseling and testing and/or information about clinical services and laboratories offering *BRCA* testing.

These data include genetic counseling visits as reported to MDCH through a statewide network of board-certified genetics professionals. Special thanks to the following institutions whose de-identified patient information was included in these analyses: Beaumont Cancer Genetics Program, Cancer Genetics Program at St. Joseph Mercy Hospital, Henry Ford Health System, InformedDNA, Karmanos Cancer Genetic Counseling Service, Lacks Cancer Center Genetics Program at Saint Mary's Healthcare, Michigan State University Division of Clinical Genetics, Marquette General Hereditary Cancer Program, Oakwood Healthcare System's Genetic Risk Assessment for Cancer Clinic, Providence Hospital Medical Genetics, Spectrum Health Cancer Genetics, University of Michigan Cancer Genetics Clinic, University of Michigan Breast and Ovarian Cancer Risk and Evaluation Program, and West Michigan Cancer Center.

Michigan Department of Community Health

Patients with a deleterious *BRCA* mutation	*	539 (12.8)
Patients not testing due to inadequate insurance	63	200

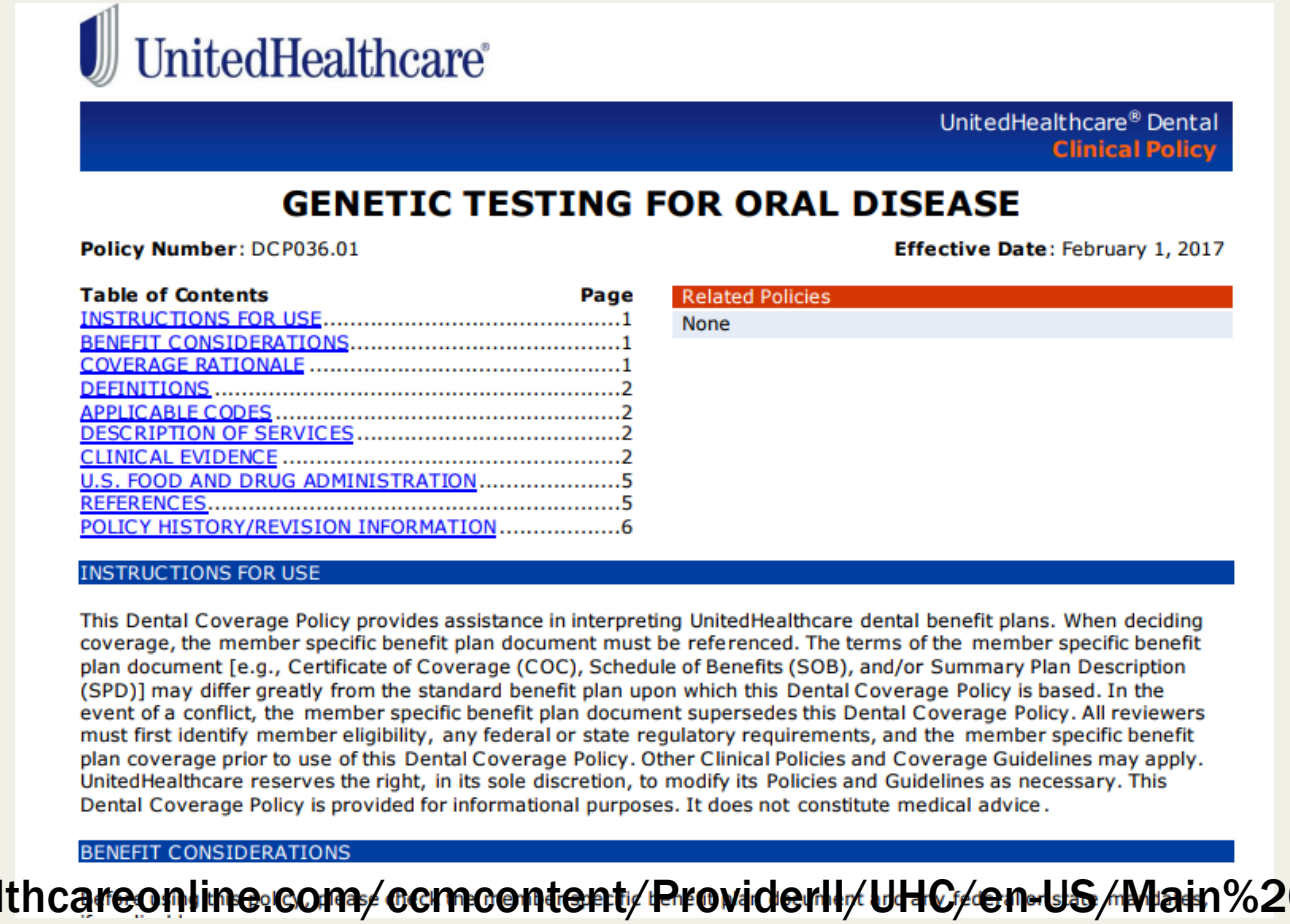
Michigan Department of Community Health

MDCH

EXAMPLES OF HEALTH PLAN CANCER GENOMICS BEST PRACTICES POLICY OUTCOMES

- Increased written health plan policies for appropriate BRCA counseling and testing to 18 of 25 health plans (increase from 4 health plans in 2009)
  - Over 8 million residents in Michigan residents of these 18 health plans
- Awarded 8 of 25 health plans in Michigan with written policies for *BRCA*-related clinical services for women with a known deleterious *BRCA* mutation aligned with NCCN guidelines
  - Most important written health plan policies needed to save lives from counseling and testing!
- 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
  - Reduced to 8.3% of those not testing in 2015 compared to 21.7% in 2008
- Received MCC Spirit of Collaboration, 2015 Award

ANOTHER IDEA?



UnitedHealthcare®

UnitedHealthcare® Dental
Clinical Policy

GENETIC TESTING FOR ORAL DISEASE

Policy Number: DCP036.01 **Effective Date**: February 1, 2017

Table of Contents **Page**

Related Policies
None

INSTRUCTIONS FOR USE

This Dental Coverage Policy provides assistance in interpreting UnitedHealthcare dental benefit plans. When deciding coverage, the member specific benefit plan document must be referenced. The terms of the member specific benefit plan document [e.g., Certificate of Coverage (COC), Schedule of Benefits (SOB), and/or Summary Plan Description (SPD)] may differ greatly from the standard benefit plan upon which this Dental Coverage Policy is based. In the event of a conflict, the member specific benefit plan document supersedes this Dental Coverage Policy. All reviewers must first identify member eligibility, any federal or state regulatory requirements, and the member specific benefit plan coverage prior to use of this Dental Coverage Policy. Other Clinical Policies and Coverage Guidelines may apply. UnitedHealthcare reserves the right, in its sole discretion, to modify its Policies and Guidelines as necessary. This Dental Coverage Policy is provided for informational purposes. It does not constitute medical advice.

BENEFIT CONSIDERATIONS

https://www.unitedhealthcareonline.com/ccmcontent/ProviderII/UHC/en-US/Main%20Menu/Tools%20&%20Resources/Policies%20and%20Protocols/Dental%20Clinical%20Policies%20&%20Coverage%20Guidelines/StaticFiles_PDFs/Genetic_Testing_for_Oral_Disease.pdf

BUILDING RELATIONSHIPS WITH PRIMARY CARE HEALTH NETWORKS IN MICHIGAN UNDERSERVED GEOGRAPHIC AREAS

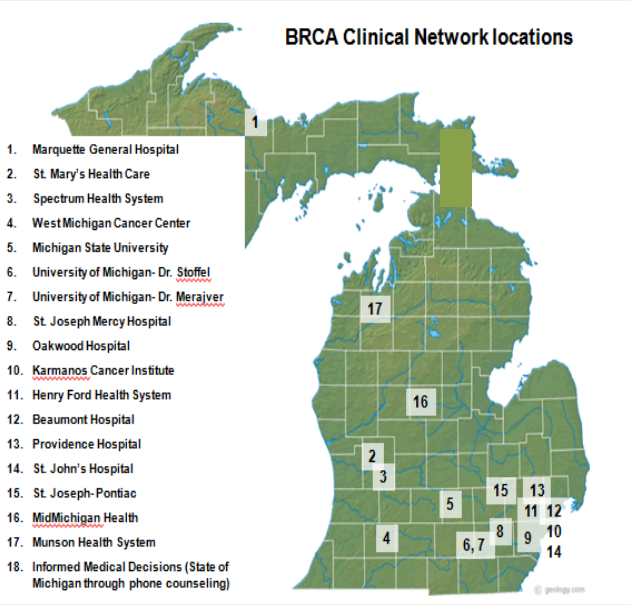
Locations of Michigan cancer genetic clinics with board-certified genetic professionals (*BRCA* Clinical Network)

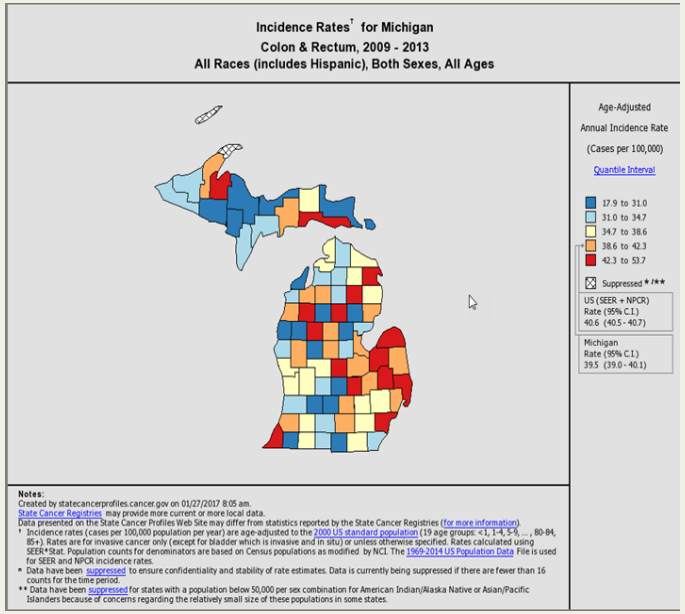
+

Counties with higher age-adjusted incidence and mortality of cancers of interest (State Cancer Registry)

→

Primary care provider engagement and education in underserved counties with higher incidence/mortality





https://statecancerprofiles.cancer.gov/map/map.withimage.php?26&001&020&00&0&01&0&1&5&0#results

Consider engagement and education of dental professionals in underserved counties with higher incidence/mortality of cancers of interest?

http://www.jaxge.org/workshops/

CANCER MOONSHOT BLUE RIBBON PANEL RECOMMENDATION:

PREVENTION AND EARLY DETECTION TOPICS

"Advances in implementation procedures would prevent additional cancer cases and unnecessary deaths in…populations with familial cancer risk attributable to known gene mutations, including those underlying Lynch Sydrome or Hereditary Breast and Ovarian Cancer"

- Identification of individuals with genetic predisposition to cancer
  - Prevention!
  - Early Detection!
  - Implementation!
  - Evidence-Base!
- Other topics considered:
  - CRC cancer screening
  - HPV vaccination
  - Tobacco control
- Estimated that half of cancer deaths could be prevented by these four topic areas

https://www.cancer.gov/research/key-initiatives/moonshot-cancer-initiative/blue-ribbon-panel/blue-ribbon-panel-report-2016.pdf

Cancer Moonshot Blue Ribbon Recommendation:

Recommendation G:

Prevention and Early Detection: Implementation of Evidence-Based Approaches

ADVANCING CANCER PREVENTION: LYNCH SYNDROME DEMONSTRATION PROJECT

Some cancers run in families due to an inherited predisposition to cancer development. Included amoong this group are people with a condition known as Lynch syndrome. This condition is marked by the presence of inherited mutations in a group of specific genes that increase their risk of developing a number of cancers at an early age, including colorectal and endometrial cancer and, to a lesser extent, stomach, ovarian, pancreatic and several other cancers. It is estimated that 1,000,000 people in the United States have Lynch syndrome; less than 5% are aware of it. About 135,000 new cases of colorectal cancer are diagnosed each year, up to 7,000 of which are caused by Lynch syndrome.

Because of the widespread availability of genetic testing, we now have the opportunity to successfully identify families in which Lynch syndrome is often found and individual members of these families with these cancer-predisposing genetic mutations, which impair a type of DNA repair known as mismatch repair. Because early detection and prevention can also decrease the risk of dying from cancer in people with an inherited predisposition to cancer, these individuals are an important target population for cancer prevention and early detection strategies.

In fact, professional medical groups recommend that all people diagnosed with colorectal cancer and women diagnosed with endometrial cancer be tested to see if they have Lynch syndrome.

Not only can this inform their own care but it means that other members of their family may have Lynch syndrome and should be tested for it. Unfortunately, studies have shown that only a small portion of people diagnosed with colorectal and endometrial cancer are actually screened for Lynch syndrome.

This project would establish a new national network of individuals and families with Lynch syndrome. It would facilitate enrollment of patients with Lynch syndrome cancers into existing and new clinical trials and help to expand genetic counseling capabilities and access to genetic counseling services to areas where they have traditionally been lacking.

The Blue Ribbon Panel recommendations call for a nationwide demonstration project to systematically screen all people diagnosed with colorectal and endometrial cancer for Lynch syndrome

https://www.cancer.gov/research/key-initiatives/moonshot-cancer-initiative/blue-ribbon-panel/blue-ribbon-panel-report-2016.pdf

NCI Funding Opportunity (closed January 2018)

NIH NATIONAL CANCER INSTITUTE

1-800-4-CANCER Live Chat Publications Dictionary

ABOUT CANCER | CANCER TYPES | RESEARCH | GRANTS & TRAINING | NEWS & EVENTS | ABOUT NCI | search

Home > Research > Key Initiatives > Cancer Moonshot℠

CANCER MOONSHOT℠

- Blue Ribbon Panel +
- Implementation
- **Funding Opportunities**
  - Public Access Policy
  - Upcoming FOAs

Cancer Moonshot℠ – Funding Opportunities

The funding opportunity announcements (FOAs) listed below highlight research initiatives that align with the efforts of the Cancer Moonshot. They may be supported with existing funds or with the 21st Century Cures funding.

Planning for implementation of longer-term scientific initiatives is also underway. We will continue to update this page as new funding opportunities become available so check back often or sign up to receive automatic email updates on Cancer Moonshot-related activities.

My Family Health Portrait

A tool from the Surgeon General

Language English

Using My Family Health Portrait you can:

- Enter your family health history.
- Learn about your risk for conditions that can run in families.
- Print your family health history to share with family or your health care provider.
- Save your family health history so you can update it over time.

Talking with your health care provider about your family health history can help you stay healthy!

Learn more about My Family Health Portrait

Create a Family Health History

Use a Saved History

https://familyhistory.hhs.gov/FHH/html/index.html

THANK YOU TO MICHIGAN CANCER GENOMICS STAFF & KEY PARTNERS!

- MDHHS Cancer Genomics Team
  - Janice Bach, MS, CGC
  - Robert Wahl, DVM, MS
  - Jillian Schrager, MPH
  - Maricar Macalincag
  - Nancie Petrucelli, MS, CGC
  - Angela Trepanier, MS, CGC
  - Carol Davis
  - Erin Van Haaren
- CDC
  - Juan Rodriguez, MPH
  - Sun Rim, MPH
  - Marie Kumerow, MPH
  - Dave Dotson, PhD
- LSSN Board of Directors
  - Heather Hampel, MS, CGC
  - Cecelia Bellcross, PhD, MS, CGC
  - Debi Cragun, PhD, MS, CGC
  - Alanna Rahm, PhD, MS, CGC
- Advocacy Partners
  - Sherry Berry
  - Eileen Kastura
  - Mollie Smith
  - Alice Christensen
- Other National Partners
  - Tuya Pal, MD
  - Kate Reed, MPH, SCM, CGC
  - Michael Dougherty, PhD
  - Megan Myers, MS, CGC
- ABOUT Network
  - Sue Friedman
  - Rebecca Sutphen, MD
  - Lisa Rezende, PhD
  - Lisa Schlager
- MCSP
  - Glenn Copeland
  - Georgetta Alverson
- MDHHS Cancer Section
  - Ann Garvin, MS, CNM, RN
  - Sarah Mott, MPH, MS, RDN
- Michigan Medicaid
  - Catherine Reid, MD
- MCGA
  - Julie Zenger Hain, PhD
  - Kara Milliron, MS, CGC
- Michigan Association of Health Plans
  - Rick Murdock
  - Lisa Farnum
- BCBSM
  - Sarah Mange, MPH
  - Besty Wasilevich, PhD
- Clinical Partners
  - Beaumont Cancer Genetics Program
  - Beaumont Hospital-Dearborn Genetic Risk Assessment for Cancer
  - Henry Ford Health System Cancer Genetics Program
  - Karmanos Cancer Institute Cancer Genetic Counseling Service
  - InformedDNA
  - Michigan State University Hereditary Cancer Program
  - Munson Cancer Genetics Clinic
  - Providence Hospital Medical Genetics
  - Sparrow Hospital Cancer Genetics
  - Spectrum Health Cancer Genetics Program
  - St. Joseph Mercy Hospital Cancer Genetics Program
  - St. John Van Elslander Cancer Genetics Program
  - St. Mary Health Care Lacks Cancer Center Genetics
  - St. Mary Mercy Our Lady of Hope Cancer Center
  - University of Michigan Breast and Ovarian Cancer Risk and Evaluation Program
  - University of Michigan Cancer Genetics Clinic
  - UP Health System-Marquette
  - West Michigan Cancer Center

Some of this presented work was supported by the Cooperative Agreement Number 5U58DP003798 and NU58DP005357 from The Centers for Disease Control and Prevention (CDC). Its contents are solely the responsibility of the presenter and do not necessarily represent the official views of the CDC.

Northwestern Medicine®
Feinberg School of Medicine

Thank you!