

Pennsylvania Cancer Control Genetics/Genomics Toolkit

A Toolkit from the Pennsylvania Cancer Coalition
Genetics/Genomics Workgroup

April 2021



Dear Reader:

We hope you find the multi-component Genetics/Genomics Toolkit useful. It is designed to provide healthcare professionals with the tools and resources that will assist them in effectively integrating hereditary cancer risk assessment into practice, provide practical information on the role of genetics in care, increase recognition and the appropriate referral of individuals at risk for hereditary cancer syndromes, and resources to talk with individuals about risk factors.

The resources within the toolkit were assembled by members of the Genetics/Genomics Workgroup of the Pennsylvania Cancer Coalition (PCC), whose role is to integrate genetics into Pennsylvania's cancer control activities. Pennsylvania's 2019-2023 Cancer Control Plan lays out a path for genetics/genomics services by setting a clear goal and comprehensive, evidence-based strategies. Genetics/Genomics information is integral to early cancer diagnosis, treatment, and improved outcomes in reducing cancer incidence, morbidity, and mortality.

There is a lot to be accomplished to eliminate barriers and increase access to genetic/genomic services to achieve a healthier Pennsylvania and reduce the state's cancer burden. We urge you to join with us to be effective and successful in this endeavor. Please check out pacancercoalition.org for more information on the PCC Genetics/Genomics Workgroup and how you can join the effort.

A handwritten signature in black ink that reads "Andrea L. Durst".

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Pennsylvania Cancer Coalition Genetics/Genomics Toolkit

Introduction

Following a charge from the secretary of health, the Pennsylvania Cancer Control, Prevention, and Research Advisory Board (CAB) convened an ad-hoc Cancer Genetics/Genomics Committee to enhance Pennsylvania's approach to cancer genetics. The committee released its 2019 report, [Recommendations for a Public Health Approach to Cancer Genetics/Genomics](#), which addresses the importance of surveillance, education, insurance coverage, and the promotion of policy and systemic change to advance cancer genetics/genomics to save lives and improve health and quality of life. The goal of increasing availability and access to genetic counseling and testing was integrated into the 2019-2023 Pennsylvania Cancer Control Plan (Cancer Plan).

A permanent workgroup was formed within the [Pennsylvania Cancer Coalition](#) (PCC) to continue the ad-hoc committee's work and implement the report and Cancer Plan recommendations. The PCC is the statewide cancer coalition that engages stakeholders in implementing the Cancer Plan through its subcommittees and workgroups. Membership is open to all cancer stakeholders, including private and nonprofit organizations and individuals.

The PCC Genetics/Genomics Workgroup identified the need for ongoing and updated provider education as the workgroup's initial focus. Increasing awareness of genetic testing among healthcare providers requires access to appropriate education to identify candidates for genetic counseling and testing and knowing where to refer for counseling and testing, if applicable. The Genetics/Genomics Toolkit was created to expand awareness of resources for genetic counseling, testing, and services.

Pennsylvania Cancer Control Genetics and Genomics Toolkit

This toolkit is part of a statewide goal within the 2019-2023 Pennsylvania Cancer Control Plan's Cancer Genetics/Genomics approach to provide education to address genetic counseling, testing, and risk management services and promote policy and systemic change. This toolkit is a direct result of an objective to provide ongoing and updated education on hereditary cancer risk for the public and medical providers.

This toolkit is an interactive, curated, and annotated resource to find information related to hereditary cancer, including genetic counseling and testing for hereditary cancer, advocacy & support organizations, risk assessment tools, current assessment and care guidelines, policy information, and general education for the public and medical providers.



Elements of Informed Consent



- Informed Consent and Pretest Counseling Checklist

<https://www.jax.org/education-and-learning/clinical-and-continuing-education/cancer-resources/informed-consent-checklist>

This resource from The Jackson Laboratory provides an easy-reference checklist of the elements of informed consent that should be reviewed with patients before genetic testing.

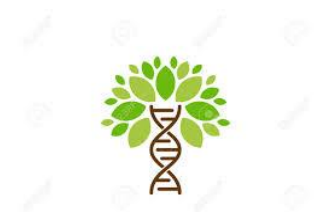
- HHS Elements of Informed Consent

(Attachment 1)

This handout from the US Department of Health and Human Services outlines the elements of informed consent required for research participants, including those individuals who are participating in genetics/genomics research and/or submitting biospecimens.

Family Health History Tools

Patients can input family history details, and the following tools will create a family history.



- My Family Health Portrait: A Tool from the Surgeon General

<https://phgkb.cdc.gov/FHH/html/index.html>

My Family Health Portrait is a family history drawing tool developed by the Surgeon General's Office and maintained by the CDC. Individuals can enter their family history, see how conditions run in their family, and use the risk assessment tools. In addition, the family history can be saved, printed, and shared with a healthcare provider.

- Susan G. Komen Family Health History
<https://apps.komen.org/FamilyHealthHistoryTool/>
The Susan G. Komen Family Health History tool allows individuals to enter in their family history information for each relative in their family. The tool contains a link to more information about risk factors for breast cancer, allows the user to save their family history, and allows the user to share the family history with another relative who can then edit or add to the information collected. Unlike some other tools, no pedigree is drawn.
- It Runs In My Family
<https://itrunsinmyfamily.com/>
This family history tool utilizes a chatbot that walks the user through a set of questions to gather their family history. An individual must get through the entire set of questions for a report to be emailed. The questions are straightforward and understandable.

Resources providers can recommend to patients on how to gather family health history information:

- Family History - National Society of Genetic Counselors
<https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/how-to-draw-your-family-tree>
This resource from the National Society of Genetic Counselors offers tips on how to draw out a family tree (pedigree). It includes a list of conditions and details that are particularly important to note when writing out a family tree and other relevant details health care providers may ask about family history. This webpage links to <https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/frequently-asked-questions-resources> for additional details.
- Medical history: Compiling your medical family tree – Mayo Clinic
<https://www.mayoclinic.org/healthy-lifestyle/adult-health/in-depth/medical-history/art-20044961>
Information on this web page from Mayo Clinic explains why it is important for healthcare providers to know a family medical history. This resource gives practical advice on how to ask family members for details on their medical history.
- A guide to family health history - Geisinger
<https://www.geisinger.org/-/media/OneGeisinger/pdfs/ghs/research/family-history/guide-to-family-health-history.pdf?la=en>
This pamphlet from Geisinger was compiled as a “manual” for collecting family health history information. Sample questions are listed for use as a starting point for conversations with family members.

Cancer Risk Assessment Tools

- NCI Gail

<https://bcrisktool.cancer.gov/index.html>

The Gail model takes into account a woman's personal medical history, familial history, and reproductive history. Specifically, it includes current age, age at menarche, age at first live birth, number of biopsies (0, 1, 2 or more), biopsy outcome (atypical hyperplasia: yes, no, unknown), and number of first-degree relatives (mothers/sisters/daughters) with breast cancer (none, one, more than one, unknown).



- The Gail Model should not be used in women with a history of breast cancer, DCIS or LCIS, women who are known to have a hereditary predisposition to breast cancer, or women with a significant family history suggestive of a hereditary predisposition.

There are important limitations of the Gail model regarding family history of breast cancer and other tumors. The model considers only first-degree relatives and does not include the paternal history of breast cancer or male breast cancer, history of ovarian cancer, and age at cancer diagnosis.

The Gail Model was initially published in 1989 and validated. The original model was developed to predict the risk of developing invasive cancer or carcinoma in situ using data derived from a Breast Cancer Detection Demonstration Project subset. It was then modified to predict the risk of invasive cancer precisely. The model was subsequently validated in 1999 using data from the Breast Cancer Prevention Trial and later updated to adjust the estimates for black, Asian, Pacific Islander, and Hispanic women. The tool may underestimate risk in black women with previous biopsies and Hispanic women born outside the United States. Because data on American Indian/Alaska Native women are limited, their risk estimates are partly based on data for white women and may be inaccurate. Mathematically, it is an unconditional logistic regression model that provides a ratio of risk in women with specified risk factors compared to women's risk without risk factors. The base race-specific population risk is derived from SEER estimates.

- CancerGene: Contains BRCAPRO, MMRpro, PANCpro, and MelaPRO

<http://www4.utsouthwestern.edu/breasthealth/cagene/>

In the late 1990s, Dr. David Euhus and UT Southwestern developed the initial version of CancerGene to aid in the cancer risk assessment of patients based on the algorithms available, including BRCAPro, MMRPro, MelaPro, and PancPro.

The software was updated in 2009 (CancerGene Connect) to a cloud-based system that incorporates even more integral health information about patients and automatically generates reports that include health information and risk assessment models.

BRCAPRO is a statistical model for assessing the probability that an individual carries a pathogenic variant in BRCA1 and BRCA2, based on his or her family's history of breast and ovarian cancer, including male breast cancer and bilateral synchronous and asynchronous diagnoses. BRCAPRO uses a Mendelian approach that assumes autosomal dominant inheritance. BRCAPRO also provides breast and ovarian cancer risk estimates based on the estimated carrier probability and utilizing published age-dependent cancer penetrance and population prevalence in the literature.

Factors that can be input into CancerGene: Invasive breast cancer in affected FDRs, SDRs, and TDR (males and females), unilateral and bilateral breast cancer, affected proband tumor hormone status, ovarian cancer in all affected FDRs, SDRs, and TDRs, age of onset for all cancers, age of unaffected relatives, Ashkenazi Jewish status, carrier status of a BRCA pathogenic variant, if known, and history of oophorectomy.

MMRpro assesses the probability that an individual carries a germline pathogenic variant in *MLH1*, *MSH2*, and *MSH6* MMR genes, based on family history of colorectal and endometrial cancer, based on his or her family's history of colorectal and endometrial cancer. The model also provides information on colon cancer risk.

MelaPRO assesses the probability that an individual carries a pathogenic variant in *CDKN2A* (p16) based on family history of single primary and multiple primary melanomas. It provides three separate prediction models based on penetrance previously estimated from 1) high-risk melanoma families in a high baseline incidence area -- Australia, the United States, and Sweden, 2) high-risk melanoma families in low a baseline incidence area -- Europe except for Sweden, and 3) all melanoma families from Australia, the United States, and Italy. The model gives information about an individual's cutaneous melanoma risk.

PancPRO estimates the probability an individual has a hereditary susceptibility gene (or gene effect) for pancreatic cancer, based on his or her family history of pancreatic cancer, and further estimates the probability an individual develops pancreatic cancer in the future.

- IBIS <http://www.ems-trials.org/riskevaluator/>

The International Breast Cancer Intervention Study (IBIS) tool is a risk-calculating model used to estimate a person's likelihood of carrying a pathogenic variant in BRCA1 or BRCA2. It also estimates the likelihood of developing breast cancer in the following ten years and over the individual's lifetime (up to age 80).

The model was developed with data from the International Breast Cancer Intervention Study (IBIS), including a cohort of daughters of patients diagnosed with breast cancer. The input for the model development was the estimated probability of carrying a BRCA1

or BRCA2 pathogenic variant and the estimated breast cancer risks based on family history, hormonal and reproductive factors, and personal characteristics.

The risks of developing breast cancer for the general population were taken from data on the first breast cancer diagnosis (ICD-10 code C50) in the Thames Cancer Registry area (UK) between 2005-2009. The risk from family history (caused by the adverse genes) is modeled based on the findings from a Swedish population-based study on cancer risk associated with a family history of breast and/or ovarian cancer in mothers. As the model incorporates the BRCA genes and a low penetrance gene, family history is used in conjunction with Bayes' theorem to iteratively produce the likelihood of carrying any genes predisposing to breast cancer, which in turn affects the chance of developing breast cancer. This risk was further refined based on the woman's personal history, including age, age at menarche, age at first-child, body mass index, benign breast disease (hyperplasia, atypical hyperplasia, LCIS), age at menopause, and use of hormone replacement therapy. The latest version of the model (v8) incorporates mammographic density (applicable only for those age 40 and older).

Note of caution: The IBIS model tends to overestimate breast cancer risk in the presence of atypia. The study by Boughey et al. found that the Tyrer-Cuzick model significantly overestimated the risk of breast cancer in women with atypical hyperplasia. The model was subsequently revised to correct for this. Also, the risk associated with mammographic density is included in the estimates for those older than 40; a study shows that mammographic density is not associated with subsequent breast cancer risk for those with atypical hyperplasia. Thus, the model might not be appropriate to use for women with atypia.

- CanRisk <https://canrisk.org/>

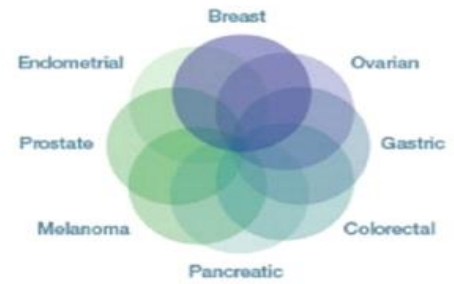
BOADICEA (CanRisk) is a comprehensive model that can be used to calculate the future risks of developing breast or ovarian cancer using the information on family history, lifestyle/hormonal risk factors, rare pathogenic variants in moderate and high-risk breast/ovarian cancer susceptibility genes, common breast/ovarian cancer genetic susceptibility variants (Polygenic Risk Scores), and mammographic density. It can also calculate the likelihood of carrying mutations in the moderate to high-risk genes BRCA1, BRCA2, PALB2, ATM, and CHEK2. Registration to create an account is required to access the tool.

Hereditary Cancer Syndromes – General

- NYMAC Cancer Genetics Referrals: When to Refer to Genetics
<https://www.wadsworth.org/cancer-referrals>
This resource provides a brief review of red flags that would indicate a patient may need a referral to genetic cancer services.

- Genetic and Rare Disease Information Center (GARD) Hereditary Cancer Syndromes
<https://rarediseases.info.nih.gov/diseases/diseases-by-category/28/hereditary-cancer-syndromes>

This website is a landing page that links to more in-depth descriptions of various hereditary cancer syndrome summaries included on the GARD website. Information on symptoms, inheritance, cause, diagnosis and treatment for each condition are reviewed.



Each cancer site is associated with multiple hereditary cancer syndromes

- GeneReviews
<http://www.genereviews.org>
GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition, which can be searched from the link to the homepage.

From the GeneReviews Website: *GeneReviews*, an international point-of-care resource for busy clinicians, provides clinically relevant and medically actionable information for inherited conditions in a standardized journal-style format, covering diagnosis, management, and genetic counseling for patients and their families. Each chapter in *GeneReviews* is written by one or more experts on the specific condition or disease and goes through rigorous editing and peer review process before being published online.

- National Society of Genetic Counselors (NSGC): About Genetic Counselors
<https://www.aboutgeneticcounselors.org/Resources-to-Help-You/Post/hereditary-cancer-hereditary-breast-and-ovarian-cancer-and-brca1-and-brca2>

This site contains some general information, videos, and other linked resources about hereditary cancer syndromes. The website is patient-friendly and may be helpful to providers as they talk to patients about genetic counseling and testing for hereditary cancer syndromes.

Lynch syndrome

- GeneReviews: Lynch syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1211/>
GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.



- Lynch Syndrome Screening Network (LSSN)
<https://www.lynchscreening.net/>
The Lynch Syndrome Screening Network promotes universal tumor screening for Lynch syndrome. The website contains helpful information about Lynch syndrome, screening for Lynch syndrome, and tools for implementing universal tumor screening.
- Collaborative Group of the Americas on Inherited Gastrointestinal Cancers (CGA-IGC)
<https://www.cgaigc.com/>
The CGA-IGC's mission is to advance the science and clinical care of inherited gastrointestinal cancers through research and education as the leading authority in the Americas. Their website contains a registry tool, a clinic tool, and a resource library.

Hereditary Breast and Ovarian Cancer syndrome

- GeneReviews: BRCA1 and BRCA2-Associated Hereditary Breast and Ovarian Cancer
<https://www.ncbi.nlm.nih.gov/books/NBK1247/>
GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- GeneReviews: PTEN Hamartoma Tumor Syndrome (Cowden syndrome)
<https://www.ncbi.nlm.nih.gov/books/NBK1488/>
GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- GeneReviews: LiFraumeni syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1311/>
GeneReviews provides detailed reviews of multiple cancer syndromes, each written by experts on the condition.
- BRCA Exchange
<https://brcaexchange.org/>
The BRCA Exchange site provides information about the pathogenicity of *BRCA1* and *BRCA2* variants that have been curated by ENIGMA, an international expert panel.



Guidelines

- NCCN Guidelines
https://www.nccn.org/professionals/physician_gls/default.aspx
The National Comprehensive Cancer Network provides clinical practice guidelines related to multiple hereditary cancer syndromes, including hereditary breast and ovarian cancer



syndromes and hereditary colorectal cancer syndromes. To access the guidelines, you must sign up for a free account.

- USPSTF Recommendations
BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing
<https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing>
These recommendations, recently updated in 2019, are a guide on when to utilize familial risk assessment tools for women who have a personal or family history of breast, ovarian, and/or primary peritoneal cancer. These guidelines do not consider risk assessment for men with a personal or family history of breast cancer.
- A practice guideline from the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: Referral Indications for Cancer Predisposition Assessment
https://www.acmg.net/docs/ACMG_Practice_Guideline_Referral_Indications_for_cancer_predisposition.pdf
These practice guidelines provide information on when to refer to genetic counseling for several different hereditary cancer syndromes. The guidelines are organized by cancer type and offer brief explanations for many hereditary cancer syndromes.
- Evaluation of Genomic Applications in Practice and Prevention (EGAPP)
<https://www.cdc.gov/genomics/gtesting/egapp/recommend/lynch.htm>
The EGAPP Workgroup developed evidence-based guidelines for genetic testing for Lynch syndrome. These guidelines provide guidance and information on tumor testing and genetic testing for Lynch syndrome.
- ACG Clinical Guideline: Genetic Testing and Management of Hereditary Gastrointestinal Cancer Syndromes
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4695986/>
These Guidelines from the American College of Gastroenterology include guidelines for the genetic testing and management of Lynch syndrome, FAP, attenuated FAP (AFAP), MUTYH-associated polyposis (MAP), Peutz-Jeghers syndrome, juvenile polyposis syndrome, Cowden syndrome, serrated (hyperplastic) polyposis syndrome, hereditary pancreatic cancer, and hereditary gastric cancer.

Find a Provider Links

- National Society of Genetic Counselors (NSGC) Find a Genetic Counselor
<https://www.nsgc.org/page/find-a-genetic-counselor>
This directory can be used to find a genetic counselor by city, state, or zip code and specialty area.



- American College of Medical Genetics and Genomics (ACMG): Find a Genetic Clinic
<https://clinics.acmg.net/>
This directory contains a listing of genetics clinics, including those that offer telegenetics. The directory is searchable by city, state, and specialty area. It is a comprehensive directory that is not linked to organization membership and is updated regularly.
- OrphaNet
<https://www.orpha.net/consor/cgi-bin/Clinics.php?lng=EN>
OrphaNet is a portal for rare diseases and orphan drugs. Their directory allows the user to search for an expert center by condition and country.
- American Board of Genetic Counseling (ABGC): Find a Genetic Counselor
<https://www.abgc.net/about-genetic-counseling/find-a-certified-counselor.aspx/>
A searchable directory of all board-certified genetic counselors
- Pennsylvania Association of Genetic Counselors (PAGC)
<https://www.pennsylvaniagc.org/patients-and-providers>
Listing of genetics clinics in PA. Email for questions: pennsylvaniagc@gmail.com

Genetic Discrimination, Cost, Insurance Coverage

- US Equal Employment Opportunity Commission
Information on the Genetic Information and Non-Discrimination Act of 2008 (GINA)
<https://www.eeoc.gov/laws/statutes/gina.cfm>
This website contains the Genetic Information and Non-Discrimination Act of 2008.



<https://www.eeoc.gov/laws/guidance/fact-sheet-genetic-information-nondiscrimination-act>

This fact sheet contains key information important for providers and patients to know about GINA.

Summary Information: The Genetic Information Non-discrimination Act of 2008 (“GINA”) was signed into law on May 21, 2008, and prohibited employers and health insurance providers from using information about an individual’s family history or genetic test results in a discriminatory manner. GINA is made up of Title I and Title II. Title I prohibits health insurers from discriminating against individuals based on genetic information, and Title II prohibits employers from using genetic information to determine employment. The law sets a minimum standard of protection that all states must meet to protect individuals from genetic discrimination in employment and health insurance

while not weakening any protection provided by individual state laws against genetic discrimination.

The definition of genetic information includes 1) predictive genetic tests, 2) family members' genetic tests, 3) family history information, 4) request for and receipt of genetic services (genetic services include genetic testing, genetic counseling, and genetic education), either by an individual or a family, 5) participation in clinical genetic research, and 6) genetic information obtained from a fetus or embryo legally related to an individual or family.

Summary of Title I

Title I applies to both individual and group health insurance coverage. Health insurers may not request or require individuals or their family members to undergo genetic testing or provide genetic information to health insurers to determine eligibility, coverage, underwriting, or premium-setting for health insurance enrollment or coverage. Also, health insurers cannot use genetic information obtained intentionally or unintentionally in decisions about enrollment or coverage. Genetic information as a preexisting condition is prohibited in the Medicare supplemental policy and individual health insurance markets.

Limitations to Title I: It does not address life, disability, or long-term care insurance issues. Nor does it apply to active-duty military personnel, veterans seeking care through the Veteran's Administration, or the Indian Health Services.

Two exemptions to Title I: 1) Health insurers may request genetic information when coverage of a particular claim would only be appropriate if there is a known genetic risk. 2) Health insurers may request (but not require) in writing that an individual undergo a genetic test for research purposes only. An individual may do so voluntarily. Refusal to participate will have no negative effect on the premium or enrollment status of the individual.

Summary of Title II

Title II of GINA prohibits employers from discriminating against employees or applicants based on genetic information. Specifically, Title II 1) prohibits the use of genetic information in making employment decisions; 2) restricts employers, employment agencies, labor organizations, and joint labor-management training and apprenticeship programs from requesting, requiring, or purchasing genetic information; and 3) limits the disclosure of genetic information.

The law forbids discrimination based on genetic information related to employment, including hiring, firing, pay, job assignments, promotions, layoffs, training, fringe benefits, or any other term or condition of employment.

It is also unlawful for employers to disclose genetic information about applicants or employees. However, limited exceptions to this non-disclosure rule are permitted, such as when disclosure of relevant genetic information to government officials investigating

compliance with Title II of GINA is required or when disclosure must be made in compliance with a court order.

Exceptions:

- 1) If the employer inadvertently acquires the genetic information.*
 - 2) Genetic information (including family medical history) may be obtained when an employee chooses to participate in voluntary health or genetic services (for example, wellness programs) offered by the employer.*
 - 3) Family medical history may be acquired as part of the certification process for Family Medical Leave requests.*
 - 4) Genetic information may be acquired through commercially and publicly available documents, such as newspapers.*
 - 5) Genetic information may be learned through a genetic monitoring program that monitors the biological effects of toxic substances in the workplace. The monitoring is required by law or voluntary under carefully defined conditions.*
 - 6) For employers who engage in DNA testing for law enforcement purposes (for example, forensic or human remains identification), the employee's genetic information may be used for quality control to detect sample contamination.*
- *Cost and Insurance Coverage – Most insurance companies consider telegenetics visits a covered benefit; however actual out-of-pocket expenses will vary with the specific plan. Similarly, insurance plans also vary concerning coverage for genetic testing.*
 - Genetic Alliance: Gina & You Fact Sheet
http://www.ginahelp.org/GINA_you.pdf
This patient-friendly fact sheet provides a summary of key definitions and protections contained in GINA.
 - National Coordinating Center for the Regional Genetics Network (NCC) Genetics Policy Resources
<https://nccrcg.org/lift/>
The NCC's mission is to improve access to quality genetic services for medically underserved populations. The website contains several policy resources, including a State Medicaid Genetics Policy database, a tracking resource for state and federal legislation and regulations, and policy resource documents.

Direct-to-Consumer Testing

- GeneReviews: Resources for Genetics Professionals – Direct-to-Consumer Genetic Testing
<https://www.ncbi.nlm.nih.gov/books/NBK542335/>
GeneReviews provides detailed reviews written by experts in the



field.

- National Human Genome Research Institute: Direct-to-Consumer Genetic Testing
<https://www.genome.gov/dna-day/15-ways/direct-to-consumer-genomic-testing>
This website provides general information about direct-to-consumer genetic testing and links to multiple other resources for patients and providers on the topic.
- Watershed DNA
<https://www.watersheddna.com/>
Founded by a genetic counselor, this website provides several resources for individuals who have undergone direct-to-consumer genetic testing, including tests for hereditary cancer. In addition, the website includes options for a consultation with a genetic counselor, a blog, an online support group, and additional resources.
- My Gene Counsel
<https://www.mygenecounsel.com/>
Founded by a genetic counselor, this company offers services to provide genetic counseling information and ongoing updates for patients and providers.

CME Opportunities in Cancer Genetics

- City of Hope Course
<https://www.cityofhope.org/education/health-professional-education/cancer-genomics-education-program/intensive-course-in-cancer-risk-assessment-overview>
- International Symposium on Hereditary Breast and Ovarian Cancer
<https://www.brcasymposium.ca/>
- Cancer genetic certification program from National Consortium of Breast Centers
<https://www.cgrcertification.org/certification/>
- Basser Center Scientific Symposium
<https://www.basser.org/investigators-clinicians/scientific-symposium>
- American Society of Clinical Oncology (ASCO) Genetics Toolkit
<https://www.asco.org/practice-policy/cancer-care-initiatives/genetics-toolkit>



- International Society of Nurses in Genetics (ISONG) Webinars
<https://www.isong.org/page-1325047>
- Collaborative Group of the Americas Inherited Gastrointestinal Cancer Annual Conference and Lectureships
<https://www.cgaigc.com/events>
- The Jackson Laboratory Cancer Genetic Clinical Education Modules
<https://www.cgaigc.com/events>

Somatic Tumor Testing Resources

The purpose of somatic mutation profiling is to identify driver mutations that could serve as treatment targets.

Somatic testing is not a substitution for germline testing. The sensitivity of most somatic testing is lower than that of most dedicated germline tests. In addition, the absence of a pathogenic or likely pathogenic variant for a given gene in somatic testing does not rule out the possibility of a germline mutation.

Germline variants may be incidentally found in somatic testing. In this case, patients should be referred for genetic counseling and confirmatory germline testing.

A patient who meets the criteria for germline testing should be referred for genetic counseling and confirmatory germline testing whether the patient has had somatic mutation profiling.

Below is a list of websites that can be used to provide helpful information about individual mutations.

Somatic only

- My Cancer Genome
<https://mycancergenome.org/>
 - i. My Cancer Genome contains information on the clinical impact of cancer-related genes, proteins, and other biomarker types on the use of anticancer therapies in cancer. There are associated genetic biomarkers, diseases, and pathways. Links to applicable clinical trials are available.
- Clinical Interpretation of Variants in Cancer (CIViC)
<https://civicdb.org/>
 - i. CIViC is an open access, open-source, community-driven web resource disseminating knowledge about clinically significant cancer genome alterations. There are clinical interpretations of variants. Links to ClinVar and COSMIC are available.
- Catalogue Of Somatic Mutations In Cancer (COSMIC)
<https://cancer.sanger.ac.uk/cosmic/>

COSMIC is the world's largest and most comprehensive resource for exploring the impact of somatic mutations in human cancer. COSMIC can be searched by gene, cancer type, or mutation. Links to OMIM and NCBI EntrezGene are available.

Somatic and germline

- ClinGen
<https://clinicalgenome.org/>
ClinGen is an authoritative central resource that defines the clinical relevance of genes and variants for use in precision medicine and research. Genomic and phenotypic data is shared between clinicians, researchers, and patients through centralized and federated databases for clinical and research use. ClinGen has variant curation with gene-disease validity and clinical actionability. Links to ClinVar and Gene Review are present.

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar/>
ClinVar aggregates information about genomic variation and its relationship to human health. ClinVar is a public archive of reports of the relationships among human variations and phenotypes, with supporting evidence. ClinVar facilitates access to and communication about the relationships asserted between human variation and observed health status and the history of that interpretation. ClinVar processes submissions reporting variants found in patient samples, assertions about their clinical significance, information about the submitter, and other supporting data.

ClinGen and ClinVar have recently formed a partnership that includes data sharing, data archiving, and collaborative curation to characterize and disseminate the clinical relevance of genomic variation.*

- Online Mendelian Inheritance in Man (OMIM)
<https://www.omim.org/>
OMIM is a comprehensive, authoritative catalog of human genes and genetic phenotypes that is freely available and updated daily by the Johns Hopkins University McKusick Nathans Department of Genetic Medicine. OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. The full-text, referenced overviews in OMIM contain information on all known mendelian disorders and traits, and over 15,000 genes, focusing on the gene-phenotype relationships.

- MedGen
<https://www.ncbi.nlm.nih.gov/medgen>
MedGen is NCBI's portal to information about human disorders and other phenotypes having a genetic component. MedGen is structured to serve health care professionals, the medical genetics community, and other interested parties by providing centralized access to diverse types of content.

Pharmacogenomic testing applicable for cancer treatment

- Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines
<https://cpicpgx.org/guidelines/>
<https://www.pharmgkb.org/guidelineAnnotations>
CPIC addresses barriers to clinical implementation of pharmacogenetic tests by creating, curating, and posting freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug clinical practice guidelines. CPIC guidelines follow standardized formats, include systematic grading of evidence and clinical recommendations, use standardized terminology, are peer-reviewed, and published in a leading journal (in partnership with Clinical Pharmacology and Therapeutics) with simultaneous posting to cpicpgx.org, where they are regularly updated.

Patient Advocacy Organizations

- Alive & Kickin'
 - i. <https://www.aliveandkickn.org/>
Alive & Kickin' serves to improve the lives of individuals and families affected by Lynch syndrome and associated cancers through research, education, and screening.
- Association for Multiple Endocrine Neoplasia Disorders
<https://www.amend.org.uk/>
The Association for Multiple Endocrine Neoplasia Disorders (AMEND) serves to improve the well-being of all persons affected by MEN and endocrine tumors by 1) providing support to patients and their families and friends, 2) providing information about MEN and other relevant issues relating to genetic disorders, 3) promoting a broader knowledge of MEN among the medical profession to assist in early and accurate diagnosis, and 4) assisting in future MEN research.
- American Multiple Endocrine Neoplasia Support
<http://www.amensupport.org>
The American Multiple Endocrine Neoplasia Support is a voluntary organization whose mission is to provide education and support to patients, families, and medical personnel regarding multiple endocrine neoplasia (MEN) type 1, MEN type 2a, MEN type 2b. MEN type 1 is a rare genetic disorder in which benign (noncancerous) tumors arise from the cells of various glands of the endocrine system. Multiple endocrine neoplasia (MEN) type 2 is a rare genetic disorder characterized by an increased risk of developing a specific form of thyroid cancer (medullary thyroid carcinoma) and benign tumors affecting additional glands of the endocrine system. MEN affect the functions of the endocrine glands, resulting in hormonal imbalances, which can lead to diabetes and other medical conditions.
- Bright Pink
<https://brightpink.org/>
Bright Pink helps save lives from breast and ovarian cancer by empowering women to

know their risks and proactively manage their health.

- Fanconi Anemia Research Fund, Inc.
<http://www.fanconi.org/>
The Fanconi Anemia Research Fund is a nonprofit organization that raises funds for medical research into Fanconi anemia, an inherited condition that leads to a deficiency of certain blood cells that are produced by the bone marrow. Established in 1989, the organization supports numerous investigators who are working on various approaches to gene identification and therapy. The Fanconi Anemia Research Fund sponsors an annual international Fanconi Anemia Research Symposium to stimulate scientific progress and collaborative research among scientists. Additionally, the fund develops and maintains a communication network that supplies information and support to affected families and their physicians. Educational materials include the "FA Handbook," "Standards for Clinical Care," and twice-yearly Family Newsletters and a Science Letter. The fund also moderates an electronic listserv for FA patients and their parents.

- FORCE
www.facingourrisk.org
FORCE improves the lives of individuals and families affected by hereditary breast, ovarian, and related cancers. FORCE accomplishes this mission by creating awareness, supplying information and support to our community, advocating for, supporting research, and working with the research and medical communities to help people dealing with hereditary breast, ovarian, and related cancers.

- Gorlin Syndrome Alliance
<https://gorlinsyndrome.org/>
The mission of the Gorlin Syndrome Alliance is to support thoughtfully, comprehensively educate and aggressively seek treatments and a cure for Gorlin syndrome, its manifestations, and sporadic BCCs.

- Hereditary Colon Cancer Foundation
<https://www.hcctakesguts.org/>
The Hereditary Colon Cancer Foundation is a registered 501(c)(3) nonprofit organization serving the hereditary colon cancer community by connecting patients, caregivers, and medical professionals to educational, social, and financial resources while promoting requisite research and health care initiatives.

- Li-Fraumeni Syndrome Association
<http://www.lfsassociation.org/>
LFS Association provides a wide range of information, advocacy, and support services for individuals and families with LiFraumeni Syndrome. LFSA supports researchers, medical providers, and caregiver's consortium to further research and promote optimal care for

the LFS community.

- Lynch Syndrome International
<https://lynchcancers.com/>
The primary mission of Lynch Syndrome International (LSI) is to focus on providing support for individuals afflicted with Lynch syndrome, creating public awareness of the syndrome, educating members of the general public and health care professionals, and providing support for Lynch syndrome research endeavors.

- Neurofibromatosis Network
<http://www.nfnetwork.org>
The Neurofibromatosis, Inc. is a national, voluntary, nonprofit organization dedicated to providing information, support, and advocacy to individuals and family members affected by neurofibromatosis type I (NF1) and type II (NF2). NF1 is a genetic disorder characterized by the development of multiple benign tumors on the covering of nerve fibers and the appearance of brown spots and freckles on the skin. NF2 is a rare genetic disorder characterized by the development of benign tumors on both auditory nerves and in other areas of the body. Established in 1988, Neurofibromatosis, Inc. services the needs of affected individuals through coordinated educational, support, and clinical and research programs. The organization provides information about NF1 and NF2 to affected individuals, family members, healthcare and other professionals, and the general public; provides referrals to local medical resources; and assists in identifying community support services. Neurofibromatosis, Inc. also encourages and supports research.

- Neurofibromatosis Northeast
<http://nfnortheast.org/>
The Mission of NF Northeast is to find a treatment and the cure for neurofibromatosis by promoting scientific research, creating awareness, and supporting those affected by NF.

- PA Breast Cancer Coalition
<https://www.pabreastcancer.org/>
The Pennsylvania Breast Cancer Coalition is a grassroots advocacy organization developed in 1993 with one mission in mind - “to help find a cure for breast cancer NOW... so our daughters don’t have to.” The PBCC has contributed over \$4.3 million to Pennsylvania breast cancer researchers. They host a traveling photo exhibit that emphasizes the importance of early detection and shows the faces of real women across the state with breast cancer, and they provide resources and information by sending thousands of “FREE Friends Like Me” care packages to newly diagnosed women. By helping to enact legislation, they are changing and saving women’s lives. For example, over 46,000 uninsured and underinsured women have received FREE mammograms, and thousands of women who were diagnosed under this program have received free

treatment for breast cancer.

- **No Stomach for Cancer**
<https://www.nostomachforcancer.org/>
No Stomach for Cancer is an organization that was developed to support research and unite the caring power of people worldwide affected by stomach cancer. Their key priorities include 1) advancing awareness and education about stomach cancer, including Hereditary Diffuse Gastric Cancer (HDGC), 2) establishing and maintaining a network of support for affected families, 3) supporting research efforts for screening, early detection, treatment, and prevention of stomach cancer, and 4) sustaining a strong and high performing organization.
- **Pheo Para Alliance**
<http://www.pheopara.org>
Pheo Para Alliance invests in research to accelerate treatments and cures for pheochromocytoma and paraganglioma while empowering patients, their families, and medical professionals through advocacy, education, and a global community of support.
- **Sharsheret**
<https://sharsheret.org/brcagenetics/counseling-testing>
This is a national nonprofit organization that aims to improve Jewish women and families living with or at increased genetic risk for breast or ovarian cancer through personalized support and saves lives through educational outreach. They have expertise in young women and Jewish families related to breast and ovarian cancer, but Sharsheret programs serve all women and men.
- **Tuberous Sclerosis Alliance**
<http://www.tsalliance.org>
The Tuberous Sclerosis Alliance (TSA) is a voluntary nonprofit organization dedicated to finding a cure for tuberous sclerosis complex (TSC) while improving the lives of those afflicted. TSC (tuberous sclerosis complex) is a genetic disorder that causes tumors to form in various organs, primarily the brain, heart, kidney, liver, skin, and lungs. People with TSC often develop epilepsy, autism, and learning and behavioral problems. The TS Alliance is committed to promoting and sponsoring medical research related to the diagnosis, cause, management, and cure of TS and ensuring that affected individuals and families can access appropriate medical services, support services, and resource information. It is involved in developing public and professional educational programs aimed at increasing awareness of TS and prompting early diagnosis and effective treatment.
- **VHL Alliance**
<http://www.vhl.org>
The VHL Alliance is a voluntary, nonprofit organization dedicated to improving diagnosis,

treatment, and quality of life for individuals and families affected by von Hippel-Lindau disease (VHL). VHL is an inherited multisystem disorder characterized by the abnormal growth of blood vessels (angiomas) in certain body areas, such as the retinas, the brain, the spinal cord, and/or the adrenal glands. Established in 1993, the alliance is committed to distributing current information about VHL to affected individuals, family members, and physicians; promoting research studies and providing research grants; maintaining a VHL tissue bank; and enabling affected individuals to exchange information, resources, and mutual support through its networking programs. The alliance also helps establish standards for clinics that specialize in diagnosing and treating VHL and provides referrals to designated clinical care centers in the United States and worldwide. In addition, it offers telephone support, engages in patient and family advocacy, and assists in the development of local chapters. The organization provides a variety of educational materials in several languages.

General information about genetic counseling/testing (video links, etc.)

- Genetic Counselors: Personalized Care for Your Genetic Health
<https://www.aboutgeneticcounselors.org/>
This website contains basic information, including what a genetic counselor is, what to expect from a genetic counseling appointment, and answers to other basic patient questions. The website also contains several short videos that can be shared with patients.
- CDC Office of Genomics and Precision Health: Genetic Counseling and Testing
https://www.cdc.gov/genomics/gtesting/genetic_counseling_testing.htm
This website provides basic information on genetic counseling and testing with some specific information about hereditary cancer syndromes. Language is patient-friendly. The CDC Office of Genomics and Precision Health offers many resources, including the Public Health Genetics Knowledge Base, a weekly newsletter, a blog, webinars, etc.

Video Resources

- Kintalk Cancer Genetics and Prevention Patient Education Videos
<https://kintalk.org/videos/>
This website offers several videos covering introductory topics on cancer genetics, including genetic counseling, genetic testing, and family communication. Of note: One of the videos includes Kintalk to communicate with family members and covers other methods.

Podcast Resources

- Bring Your Brave Campaign Podcast Series (CDC)
https://www.cdc.gov/cancer/breast/young_women/bringyourbrave/stories/podcasts.htm
This podcast series from the CDC features the stories of six individuals and their experiences with breast cancer, including some with *BRCA* mutations.
- Grey Genetics Patient Stories Podcast
<https://www.greygenetics.com/podcast/>
This podcast shares the stories of patients and their experiences with genetic conditions and genetic counselors, including some episodes with individuals who have a hereditary cancer syndrome.
- Alive and Kickn's podcast
<https://www.aliveandkickn.org/podcast>
This podcast dedicated to Lynch syndrome features patient advocates and providers.