WE’RE WAY PAST PEAS:
USES OF GENETIC INFORMATION TO UNDERSTAND
HUMAN HEALTH AND GUIDE HEALTH CARE DECISION
MAKING

Diana Nelson Louden, University of Washington Health Sciences Library
Carolyn Martin, NN/LM Pacific Northwest Region
Introductions

Tell us about you:

- Name
- Job title
- Place of work
- City/State
How many people have had their DNA tested using a direct-to-consumer genetic test?

12 million +
Which clinical specialists might use genetic information in patient care?

a) Obstetricians  
b) Pediatricians  
c) Oncologists  
d) Cardiologists  
e) Pharmacists

All of them!
What is an example of a disease that could have a genetic component?

- a) Diabetes
- b) Breast cancer
- c) Osteoporosis
- d) Epilepsy
- e) Sickle-cell anemia

All of them COULD have a genetic component. Sickle-cell anemia is ALWAYS genetic.
Genomics in the News

- Scientists report CRISPR restores effectiveness of lung cancer treatment
- Baltimore Ravens to hand out free DNA test kits
- Genetically Modified People Are Walking Among Us: Global study finds 44 genetic risk factors for major depression
- Crime scene investigators couldn’t tell identical twins’ DNA apart. Until now
- We will find you: DNA search used to nab Golden State Killer can home in on about 60% of white Americans
- A NEW GENETIC TEST COULD HELP DETERMINE CHILDREN’S SUCCESS
  - CRISPR gene editing can cause hundreds of unintended mutations
- World’s First Baby Born Using New Technique That Combines DNA Of 3 Parents
- New Genetic Engineering Method Called Promising — And Perilous
- Tackling the Opioid Crisis: Genetic testing to identify addiction risk
- Harnessing CRISPR for rapid detection of viral and bacterial infection
Genomic Literacy

- Genomic health literacy
- Genomic science literacy
- Role of media in genomic literacy
Consumer Genomic Health Literacy

Lack biology basics
Lack mathematical concepts
Low health literacy
Definitions

- Genomic Health Literacy
  - The capacity to obtain, process, understand, and use genomic information for health related decision making.

- Genomic Science Literacy
  - The knowledge of basic genetics and genomics concepts and processes needed to build conceptual understanding, and the necessary mathematical knowledge to support this comprehension.
Leading causes of death

1. Heart disease: 635,260
2. Cancer: 598,038
3. Accidents (unintentional injuries): 161,374
4. Chronic lower respiratory diseases: 154,596
5. Stroke (cerebrovascular diseases): 142,142
6. Alzheimer's disease: 116,103
7. Diabetes: 80,058
8. Influenza and pneumonia: 51,537
9. Nephritis, nephrotic syndrome, and nephrosis: 50,046
10. Intentional self-harm (suicide): 44,965
Genomic Science Literacy

- K-12 education unable to keep up with scientific advancements
- Low emphasis on genomics
- Some teachers have misconceptions about genetics/genomics and little understanding
- Teachers need updated skills and have little access to genetic/genomic quality science curriculum
- Encourage partnerships with scientists
- Empower students entering the age of personal genomic medicine
Challenges for Clinicians – translating research to practice

“Despite the growing use of genomic applications in clinical practice, health professional knowledge about genomic information and confidence in using it have not kept pace....

Many health care providers do not have either the knowledge or the tools they need in order to apply genetic information in their day-to-day practices.

This lack of support is contributing to a substantial delay in the translation of genetic research findings, when appropriate, into improvement in patient outcomes within the health care system.”

— Institute of Medicine 2015 report
Challenges for Clinicians: Translating Research to Practice

- Medical geneticists, genetic counselors, & advanced practice genetic nurses are the ones who are rigorously trained to deliver genetic health services.
- There’s a shortage of genetic specialists.
- Primary care providers are on the “front line.”
- There isn’t sufficient evidence to rely on many genetic tests.
- New genetic variants are identified all the time; the significance of many of them is not known.
- Genomic information is not typically well integrated in electronic health records.
"I’m a community physician…I’ve got a medical license, I can order any blood test I want to, and I may or may not be well-trained in the precise implications of that test. Heck, I may not even be able to interpret the report because what used to be about a half page report has now grown to about a two or three page report and there’s more BRCAs that you have to know the meaning of.” – Breast Oncologist

Topics for Today

- Genomic health literacy
- Basic principles of genetics
- Uses of genetic information in health care
- NCBI’s MedGen portal & other clinical genetics resources
- Practice answering questions using MedGen. Discussion.
  - Break 10:15-10:30
- Genetic consumer health resources
- Ethics and privacy
- Practice answering questions using Genetics Home Reference. Discussion.
  - Show what you know! Prizes! Evaluations!

Presentation slides are available at: https://nnlm.gov/pnr/guides/training-resources-you-can-use/presentations
BUT FIRST, PEAS
Mendel Discovered Patterns of Inheritance by Studying Physical Traits

Before genes were discovered, Mendel realized that he could make mathematical predictions about the inheritance of physical traits—like flower color.

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<thead>
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By Madprime (Own work) [CC0, GFDL (http://www.gnu.org/copyleft/fdl.html), CC-BY-SA-3.0 (http://creativecommons.org/licenses/by-sa/3.0/)], via Wikimedia Commons
Mary-Claire King: Way Past Peas!
Mary-Claire King: Way Past Peas!

Demonstrated in 1990 that mutations in a single gene, which she named BRCA1, were responsible for breast and ovarian cancer in many families.

Classic BRCA1 Pedigree
“Most of inherited breast and ovarian cancer can be prevented, if mutation carriers know who they are.”

But “about half of women who inherit mutations in BRCA1 or BRCA2 have no family history of breast or ovarian cancer and have no idea they are carrying cancer-causing mutations.” [They’ve inherited the mutation from an “unaffected father.”]

Wants women to be offered genetic testing for BRCA1 and BRCA2 mutations at about age 30 as part of routine medical care.


JUST ENOUGH GENETICS...
The Animated Genome

Unlocking Life’s Code video
https://unlockinglifescode.org/media/animations/659#660
Chromosomes are made of DNA

By OpenStax College [CC BY 3.0 (http://creativecommons.org/licenses/by/3.0)], via Wikimedia Commons
Genes are discrete segments of DNA found on chromosomes.
Humans Receive 23 Chromosomes from Each Parent

*Egg and sperm cells only contain one set of 23 chromosomes.

By Courtesy: National Human Genome Research Institute (Public Domain), via Wikimedia Commons
This is Your Genome

- The DNA that contains your 20,000+ genes.
- The DNA that regulates the expression of your genes.
- The DNA of unknown significance.
Genetic Alleles: Different Versions of the Same Gene

An allele is one of two or more versions/variants of a gene within a population.

“F” represents the freckle gene – MC1R. Freckles are a dominant trait. If you receive at least one copy of the F allele, you’re likely to have freckles.
Many Traits Are Polygenic – the Product of Multiple Genes

Eye color is determined by variation at several different genes and the interactions between them.

Brown Eyes
How does a gene affect a physical trait or process?

- Genes encode proteins.
- The DNA sequence of the gene dictates the amino acid sequence of the protein.
- A protein’s sequence defines its 3-dimensional structure & how it can interact with other molecules.
- Proteins do the work in your body.

Image credit from Harvey Mudd College web page:
http://fourier.eng.hmc.edu/bioinformatics/intro/node8.html
Altered genes can lead to altered proteins. Altered proteins might not function properly.

**Normal BRCA1 Protein**
- Job Description: Repair Damaged DNA
- Job Location: Breast and Ovaries

**Mutated BRCA1 Protein**
- Damaged DNA Not Repaired.
- Mutations Accumulate.
- Can Lead to Uncontrolled Cell Growth and Tumor Formation
CATEGORIES OF DISEASES ATTRIBUTED TO GENES

- Chromosomal Diseases
- Monogenic Diseases/Mendelian Diseases
- Multifactorial Diseases
An individual may have a missing chromosome, extra copies of a chromosome, or a portion of a chromosome may be deleted, duplicated, or translocated.

Alteration may be inherited or de novo. Most originate in the egg or sperm.

Example: **Down’s syndrome**
(extra copy of chromosome 21)
Monogenic Diseases/ Mendelian Diseases

- Single-gene diseases follow the patterns of inheritance that Mendel discovered in his studies of pea plants.
- These rare inherited diseases tend to be caused by mutations in a single gene.
- Examples: cystic fibrosis, sickle-cell anemia, muscular dystrophy, and Huntington’s disease.

Genetic Science Learning Center, University of Utah, http://learn.genetics.utah.edu
Multifactorial Diseases

- Complex diseases typically involve more than one gene.
- Also influenced by environmental factors, such as smoking, viral infections, and diet.
- Vast majority of human diseases fall into this category.
- Difficult to identify genes contributing to these diseases because a single physical condition can have many different causes.
- Examples: cardiovascular disease, cancer, diabetes, and a number of birth defects and psychiatric disorders.

Credit: Nature’s Scitable website - http://www.nature.com/scitable/topic/genes-and-disease-17
NHGRI “Complex Disorders Background”
https://www.genome.gov/10000865/
https://commons.wikimedia.org/wiki/File:Deep_fried_pineapple_on_a_stick_(2746967219).jpg
Genetic Risk Assessments
Are NOT Psychic Predictions
Three Ways Genetics Is Intersecting Health Care

Genomic Testing

Tumor Testing

Pharmacogenomics

Credit: Wikimedia Commons
1. Clinical Genomic Testing (Germline Testing)

- **Germline Testing**: Mutations/variations found in all of your cells – cells you’ve had since your beginning.

- **Goals**
  - **Diagnosis** – can we attribute your observed condition to a genetic cause?
  - **Risk assessment** – are you more likely than the average person to develop a particular disorder because of variation in particular genes?

- **Types of Genetic Tests**:
  - **Single gene tests** – e.g., CFTR. [Mutations in both copies of CFTR cause cystic fibrosis.]
  - **Gene panels** – multiple genes associated with a condition, e.g., colorectal cancer
  - **Whole-exome sequencing & whole-genome sequencing** – looking for “something”
Clinical Genomic Testing in the Context of Patient Care

- Genomic testing is recommended after considering a patient’s personal medical history and family medical history.
- Tests are selected based on determining what information is needed to guide patient care.
- Genetic tests are performed by academic and commercial labs.
- Clinician reviews test results and explains them to the patient.
- Next steps and management of patient care take genetic test results into consideration.
Vision for Genomic Medicine

“A vision for genomic medicine is that germline genome sequencing will be routinely conducted in health systems to provide healthcare and preventive services tailored to each individual [1].

For the most part, sequencing is not yet routinely used in clinical practice but is prioritized among people with certain diseases (e.g., ill newborns, and people with cancer or rare diseases) [2] or genetic predisposition to certain diseases (e.g., BRReast CAncer susceptibility gene 1 [BRCA1] and BRReast CAncer susceptibility gene 2 [BRCA2] testing for hereditary breast and ovarian cancer susceptibility) [3].”

### Classification Criteria

**Tier 1**
- FDA label requires use of test to inform choice or dose of a drug
- FDA cleared or approved companion diagnostic device
- CMS covers testing
- Clinical practice guidelines based on systematic review supports testing

**Tier 2**
- FDA label mentions biomarkers
- FDA premarket approval (PMA)
- FDA 510(k) substantially equivalent decision
- 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

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**CDC’s Office of Public Health Genomics (OPHG) ranks genomic tests, and family health history applications, by levels of evidence**

[phgkb.cdc.gov](http://phgkb.cdc.gov)
The Answer to a Genetic Test Is Often Something Other Than Yes or No.

Labs typically report an individual’s genetic test results using five categories:

1. Disease-causing mutation found.
2. Mutation found is *likely* disease-causing.
3. Mutation found is *probably* benign and not disease causing.
4. Mutation is *known* to be benign and does not cause disease.
5. Mutation is a “Variant of *Unknown* Significance” (VUS).
Variants of Unknown Significance (VUSs)

Yes! A variant (SNP) is present in a gene. (We don’t typically see that nucleotide in that location.)

No! We don’t know what the clinical significance is. It could be benign, or it could be pathogenic.

A VUS in a lab report really is UNKNOWN.

After more evidence is collected, VUSs can often be categorized as benign or pathogenic.
Genetic Test Results Require Interpretation

- Clinicians don’t order genetic tests unless the results are likely to improve patient management.
- Typically Medical Geneticists and Genetic Counselors are the clinicians who are most qualified to order and interpret genetic tests.

Genetic Test Registry
Polymorphism in Nature
Clinical Genomic Tests Often Look for Single Nucleotide Polymorphisms (SNPs)
>2,600 SNPs Have Been Identified in the BRCA1 Gene
>500 Variants are Considered “Causal”

In the BRCA1 gene – which is 193,689 nucleotides long – if this one particular nucleotide is an A instead of a G, a person is more likely to develop breast cancer.

https://www.snpedia.com/index.php/Rs80357292
BRCA1 SNPs –
“You have the breast cancer gene.”

What should a person with a **pathogenic BRCA1 variant** expect?

- Increased risk of developing breast and/or ovarian cancer at an earlier age
- Lifetime risk of breast cancer of 80-90% (compared to 12% of women in the general population)
- Lifetime risk of ovarian cancer of 40-50% (compared to 1.3% of women in the general population)
- Increased risk of bilateral breast cancer

Credit: OMIM
http://www.omim.org/clinicalSynopsis/604370

Credit: National Cancer Institute
Clinicians Are Concerned with Clinically Actionable or Clinically Useful Genetic Test Results

- If you have a pathogenic BRCA1 variant, a clinician may be able to provide:
  - Closer surveillance (MRI in addition to mammogram)
  - Surgery (if warranted)
  - Chemoprevention
  - Genetic Counseling

- Sometimes the significance of a gene variant isn’t known.
- Sometimes genetic testing is not likely to significantly improve patient outcomes and may lead to detrimental outcomes.
23andMe Genetic Test for Breast Cancer Risk

- In 2018 FDA approved the marketing of the 23andMe “Personal Genome Service Genetic Health Risk Report for BRCA1/2 (Selected Variants)”
- Consumers receive a qualitative report on increased risk based on only three of the BRCA1 and BRCA2 mutations known to correspond to increased cancer risk
University of Michigan BRCA1 & BRCA2 Mutation Panel

- Looks for ALL variants in these two genes.
- Attempts to interpret any novel variants.
- If variant interpretation changes over time, lab will contact the ordering physician.
2. Tumor Testing: Identifying Genetic Mutations for Targeted Therapy

- Study a sample of tumor cells from a patient with cancer.
- Did an identifiable mutation lead to out-of-control cell growth?
- Do we have a drug that targets that specific out-of-control pathway?
Two clinically significant genetic variants in EGFR (epidermal growth factor receptor) identified in patient’s lung cancer sample.
Tumor Cells With Particular EGFR Mutations Can Be Treated With an EGFR Inhibiting Drug

Mutations are detected with an FDA-approved genetic test. If the patient has one of these mutations, then recommended to treat with Tarceva.

TARCEVA (erlotinib) tablets, for oral use
Initial U.S. Approval: 2004

INDICATIONS AND USAGE

TARCEVA is a kinase inhibitor indicated for:
- The treatment of patients with metastatic non-small cell lung cancer (NSCLC) whose tumors have epidermal growth factor receptor (EGFR) exon 19 deletions or exon 21 (L858R) substitution mutations as detected by an FDA-approved test receiving first-line, maintenance, or second or greater line treatment after progression following at least one prior chemotherapy regimen. (1.1)
- First-line treatment of patients with locally advanced, unresectable or metastatic pancreatic cancer, in combination with gemcitabine. (1.2)

Limitations of Use:
- Safety and efficacy of TARCEVA have not been established in patients with NSCLC whose tumors have other EGFR mutations. (1.1)
- TARCEVA is not recommended for use in combination with platinum-based chemotherapy. (1.1)
3. Pharmacogenomics: Should We Prescribe You a Little, a Lot, or None At All?

Pharmacogenomics

**Purpose:**
Study how genes affect an individual’s responses to specific drugs.

**Goal:**
Predict who will benefit from a medication, who will not respond, and who will experience adverse drug reactions.
Use that information to treat an individual with the appropriate drug and dose.
PRECISION MEDICINE

Plavix (Clopidogrel) and Pharmacogenomics

Plavix: anti-platelet drug; inhibits blood clots which could lead to heart attack and stroke
- Some people have genetic variants of an enzyme, CYP2C19, that cause them to metabolize Plavix more slowly.
- Slow metabolism of Plavix = increased risk of clotting/adverse events.
- Physicians will want to prescribe different anti-platelet drugs for these “poor metabolizers.”

WARNING: DIMINISHED EFFECTIVENESS IN POOR METABOLIZERS

See full prescribing information for complete boxed warning.
- Effectiveness of Plavix depends on activation to an active metabolite by the cytochrome P450 (CYP) system, principally CYP2C19. (5.1)
- Poor metabolizers treated with Plavix at recommended doses exhibit higher cardiovascular event rates following acute coronary syndrome (ACS) or percutaneous coronary intervention (PCI) than patients with normal CYP2C19 function. (12.5)
- Tests are available to identify a patient’s CYP2C19 genotype and can be used as an aid in determining therapeutic strategy. (12.5)
- Consider alternative treatment or treatment strategies in patients identified as CYP2C19 poor metabolizers. (2.3, 5.1)
Information on FDA-Approved Drugs with Pharmacogenomic Information in Their Labels

As of April 2019, 261 FDA-approved drugs have pharmacogenomics information in their labels. Sometimes genetic testing is mandatory for prescribing a drug.
Jean’s Genetic Testing Timeline
(get it, “Jean”?)

Age 1 day: **Newborn testing** for a few serious childhood diseases.

Age 30: **Carrier testing** for cystic fibrosis (with her partner) before trying to get pregnant. (Some members of extended family had CF.)

Age 35: **Breast cancer risk prediction testing** when her sister developed breast cancer at a young age.

Age 55: **Tumor testing** of breast cancer cells to determine the appropriate drug for treatment.

Age 70: **Pharmacogenomic testing** when Plavix (anti-platelet drug) was not effective.
TAKE A STRETCH BREAK!
Genetics and Medicine: Recommended Resources

Genetics and Medicine Guide – http://guides.lib.uw.edu/hsl/geneticmedicine

Genetic Medicine Resources: Starting Points for Clinicians

- MedGen
- GeneReviews
- Genetic Testing Registry
- PharmGKB
- OMIM
- PubMed - Medical Genetics

Clinical Genetics eBooks

Thompson and Thompson
Genetics in Medicine, 8th ed. (2016) by Robert Nussbaum; Rodenck R. McInnes; Huntington F. Willard

Emery and Rimoin’s
Principles and Practice of Medical Genetics and Genomics, 7th ed. (2019) by Reed E. Pyeritz; Bruce R. Korf; Wayne W. Grody

Key Genetic Medicine Resources

- MedGen
  - Summaries of medical genetics information compiled from GeneReviews, OMIM, ClinVar, Genetic Testing Registry, and PubMed.
  - Search for a gene, genetic disorder, or clinical feature.
  - Links to practice guidelines.
- Gene Reviews
  - Point-of-care information for inherited conditions - diagnosis, management, and genetic counseling information.
  - Peer-reviewed chapters typically focus on a single gene or phenotype/disorder

Medical Genetics Literature in PubMed

Two ways to efficiently identify PubMed references relating to medical genetics:

1. Use the Medical Genetics clinical filter which can be found under “Clinical Queries”
2. Locate a MeSH term that describes the topic you’re interested in, and then add the “genetics” subheading. For example: "Cardiomyopathy, Hypertrophic genetics" (Mesh)
PubMed Medical Genetics Query

PubMed Clinical Queries
Results of searches on this page are limited to specific clinical research areas. For comprehensive searches, use PubMed directly.

ventricular hypertrophy

Clinical Study Categories
Category: Therapy
Scope: Broad

Systematic Reviews

Results: 5 of 10459
A randomized controlled trial of metformin on left ventricular hypertrophy in patients with coronary artery disease without diabetes: the MET-REMODEL trial.

Results: 5 of 87
Systematic review of the clinical outcomes of mineralocorticoid receptor antagonist treatment versus adrenalectomy in patients with primary aldosteronism.

Medical Genetics
Topic: All

Results: 5 of 6022
Right ventricular pressure overload alters cardiac lipid composition.

Role of Myocardial Fibrosis in Hypertrophic Cardiomyopathy: A Systematic Review and Updated Meta-Analysis of Risk Markers for Sudden Death.
Bittencourt MI, Cadena SA, Araújo DV, Salles ALF, Albuquerque LF, Spina PFM, Albuquerque DC, Moutinho-Rocha R.

Moving Beyond the Sarcomere to Explain Heterogeneity in Hypertrophic Cardiomyopathy: JACC Review Topic of the Week.
Hypertrophy, Left Ventricular

Enlargement of the LEFT VENTRICLE of the heart. This increase in ventricular mass is attributed to sustained abnormal pressure or volume loads and is a contributor to cardiovascular morbidity and mortality.

Year introduced: 1983

PubMed search builder options

Subheadings:

- analysis
- anatomy and histology
- blood
- chemically induced
- classification
- complications
- congenital
- cytology
- diagnosis
- diet therapy
- drug therapy
- economics
- embryology
- enzymology
- epidemiology
- etiology
- genetics
- history
- immunology
- metabolism
- microbiology
- mortality
- nursing
- organization and administration
- parasitology
- pathology
- physiology
- physiopathology
- prevention and control
- psychology
- radiography
- radionuclide imaging
- radiotherapy
- rehabilitation
- statistics and numerical data
- surgery
- therapy
- ultrasonography
- urine
- veterinary
- virology
# Important Genetic Medicine Databases for Clinicians

<table>
<thead>
<tr>
<th>Database</th>
<th>Description</th>
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<tbody>
<tr>
<td>Gene Reviews</td>
<td>Point-of-care information for inherited conditions - diagnosis, management, and genetic counseling information. Peer-reviewed chapters. Search by gene or disorder.</td>
</tr>
<tr>
<td>OMIM</td>
<td>Overviews of Mendelian disorders and genes associated with disease. Can search by symptom.</td>
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<tr>
<td>ClinVar</td>
<td>Variants found in patient samples (primary data) along with assertions regarding the variants' clinical significance. Includes level of evidence available.</td>
</tr>
<tr>
<td>PharmGKB</td>
<td>Information on the impact of human genetic variations on drug response. Includes drug dosing guidelines.</td>
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NCBI’s MedGen Portal


MedGen

Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.
MedGen: NCBI Portal to Medical Genetics Content

- Single point of access to resources with information about human disorders and features/symptoms that have a genetic component.
- Designed for health care professionals & the medical genetics community.
MedGen: “One Stop Shop for Phenotypes with a Genetic Component”

- Integrated content from multiple sources.
- Professional guidelines – manually curated by NCBI staff

Types of Records
- Mendelian disorders
- Pharmacogenetics of drug responses
- Complex diseases with a genetic component
- Clinical features
Follow Links from PubMed References to MedGen

Cardiac Phenotypes, Genetics, and Risks in Familial Noncompaction Cardiomyopathy.


Abstract

BACKGROUND: There is overlap in genetic causes and cardiac features in noncompaction cardiomyopathy (NCCM), hypertrophic cardiomyopathy (HCM), and dilated cardiomyopathy (DCM).

OBJECTIVES: The goal of this study was to predict phenotype and outcome in relatives according to the

Cardiomyopathy
(CMYO)
MedGen UID: 209332 • Concept ID: C0878544 • Disease or Syndrome

Synonyms:
CMYO

SNOMED CT:
Disorder of myocardium (57809008); Disorder of heart muscle (57809008); Cardiomyopathy (85989001); Myocardial disease (57809008)

Related genes:
RBM20, NEXN, TMEM43, JPH2, MYOZ2, PRKAG2, ANKR1D1, LDB3, ABCG1, BAG3, TACAP, CSRP3, VCL, TTN, TPM1, TNN1, TNNC1, TMPO, TGFBS, TAZ, SGCD, SDHA, SCN5A, RYR2, PSEN2, PSEN1, PLN, PKP2, MYL3, MYL2, MYH7, MYH6, MYBPC3, LMNA, LAMA4, JUP, FKTN, EYA4, DSP, DSG2, DSC2, DES, CRYAB, ACTN2, ACTC1

HPO:
HP:0001638

Orphanet:
ORPHA167848

Definition

A disease of the heart muscle or myocardium proper. Cardiomyopathies may be classified as either primary or secondary, on the basis of etiology, or on the pathophysiology of the lesion: hypertrophic, dilated, or restrictive. [from NCI]
Exploration of MedGen Together

As you type your query, names of genetic disorders used in the NIH Genetic Testing Registry (GTR) will be provided. If you do not make a selection from the menu that appears under the search box as you type, your query is processed by looking for a match on a word or phrase. * is used as the wild card, and that wild card can be used only at the end of a word.

<table>
<thead>
<tr>
<th>Name</th>
<th>Example searches</th>
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<td>achondroplasia</td>
<td>As you type your query, names of genetic disorders used in the NIH Genetic Testing Registry (GTR) will be provided. If you do not make a selection from the menu that appears under the search box as you type, your query is processed by looking for a match on a word or phrase. * is used as the wild card, and that wild card can be used only at the end of a word.</td>
</tr>
<tr>
<td>LMNB1</td>
<td>If you enter a gene symbol followed by [gene], the diseases caused by or with some association to that gene will be retrieved.</td>
</tr>
<tr>
<td>short stature</td>
<td>If you enter the name of the feature followed by [clinical feature] the diseases with that feature will be retrieved.</td>
</tr>
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MedGen Summary: Search was BRCA1[gene]

Breast-ovarian cancer, familial 1 (BROVCA1)
MedGen UID: 382914 • Concept ID: C2676676 • Finding

**Synonyms:**
- BRCA1 Hereditary Breast and Ovarian Cancer; Breast cancer, familial 1; BREAST CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1; BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1; BROVCA1; OVARIAN CANCER, SUSCEPTIBILITY TO
- BRCA2 Hereditary Breast and Ovarian Cancer
- BRCA1 (17q21.31)
- BRCA2 (13q12.2)

**Modes of inheritance:**
- Autosomal dominant inheritance (HPO, OMIM, Orphanet)
- Multifactorial inheritance (HPO, Orphanet)
- 604370

**Gene (location):**
- BRCA1 (17q21.31)
- OMIM®: 604370

**Excerpted from the GeneReview:**

BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

BRCA1- and BRCA2-associated hereditary breast and ovarian cancer syndrome (HBOC) is characterized by an increased risk for female and male breast cancer, ovarian cancer (includes fallopian tube and primary peritoneal cancers), and to a lesser extent other cancers such as prostate cancer, pancreatic cancer, and melanoma primarily in individuals with a BRCA2 pathogenic variant. The exact cancer risks differ slightly depending on whether HBOC is caused by a BRCA1 or BRCA2 pathogenic variant. [from GeneReviews]

**Full text of GeneReview (by section):**
- Summary
- Diagnosis
- Clinical Characteristics
- Genetically Related (Allelic) Disorders
- Differential Diagnosis
- Management
- Genetic Counseling
- Resources
- Molecular Genetics
- References
- Chapter Notes
A patient is suspected to have the condition known as familial hypertrophic cardiomyopathy.

1) Find an information-rich summary of familial hypertrophic cardiomyopathy (more general preferred). Make a note of the MedGen UID.

2) What genes can be associated with familial hypertrophic cardiomyopathy?

3) What year were the most recent professional guidelines written regarding the diagnosis and management of hypertrophic cardiomyopathy?

4) According to Gene Reviews, mutations/pathogenic variants in what two genes cause most cases of familial hypertrophic cardiomyopathy?

5) BONUS: According to the Genetic Testing Registry, is there a lab in the United States that offers a diagnostic panel (multiple genes) for hypertrophic cardiomyopathy?
A physician suspects that her patient doesn’t respond well to the drug clopidogrel (Plavix).

1) Find a MedGen record that addresses this phenomenon & note the UID.

2) What gene encodes the enzyme involved in metabolizing clopidogrel?

3) When were the most recent professional guidelines published that discuss how to use clopidogrel pharmacogenetic information in practice?

4) What percentage of Chinese people are thought to be poor metabolizers of clopidogrel?  [Hint: Medical Genetics summaries link]

5) Can you find some information that may be helpful for the patient?

6) BONUS: What database has detailed information on the effects of gene variants on drug response?  [Hint: It’s linked from MedGen record.]
TAKE A BREAK!
The importance of being: genomically literate

The importance of being: genomically informed
Patient/Consumer Resources
Section: Genetics/Birth Defects

Health Topic pages:
- Genes and Gene Therapy
- Genetic Brain Disorders
- Genetic Counseling
- Genetic Disorders
- Genetic Testing

text word search
Genetics/Birth Defects

Abnormalities see Birth Defects
Achondroplasia see Dwarfism
Adrenoleukodystrophy see Leukodystrophies
Alpha-1 Antitrypsin Deficiency
Ammiocentesis see Prenatal Testing
Anencephaly see Neural Tube Defects
Arnold-Chiari Malformation see Chiari Malformation
Ataxia see Friedreich's Ataxia
Ataxia Telangiectasia
Birth Defects
Blood Coagulation Disorders see Hemophilia
Brain Disorders, Inborn Genetic see Genetic Brain Disorders
Brain Malformations
Canavan Disease see Leukodystrophies
Cephalic Disorders see Brain Malformations
Cerebral Palsy
Charcot-Marie-Tooth Disease
Results 1 - 10 of 5,178 for genetics

Genetic Disorders

Genes are the building blocks of heredity. They can be passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in the body. They transport molecules from one place to another, build and repair cells, and do many other maintenance jobs.

Sometimes there is a mutation, a change in the gene's instructions. The protein does not work properly or is not made at all. This is called a genetic disorder.

Related Topics

Seizures
Epilepsy
Ovarian Cancer
Breast Cancer
Genetic Counseling

Refine by Type

All Results (7,721)
- Health Topics (363)
- External Health Links (4,007)
- Drugs and Supplements (82)
- Medical Encyclopedia (448)
- MedlinePlus Magazine (143)
- Multiple Languages (64)
- National Institutes of Health (2,514)

Genomics and Health Impact Update

1. Genomics and Health Impact Update
   - Centers for Disease Control and Prevention
   - National Human Genome Research Institute
   - National Human Genome Research Institute conducts genetic and genomics research, funds genetic and genomics research, and promotes research to advance genomics

2. Genomic Testing
   - Centers for Disease Control and Prevention
   - National Human Genome Research Institute
   - National Human Genome Research Institute conducts genetic and genomics research, funds genetic and genomics research, and promotes research to advance genomics

3. Genomics and Health Impact Update
   - Centers for Disease Control and Prevention
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4. Genomic Testing
   - Centers for Disease Control and Prevention
   - National Human Genome Research Institute
   - National Human Genome Research Institute conducts genetic and genomics research, funds genetic and genomics research, and promotes research to advance genomics

5. Frequently Asked Questions about Genetic and Genomic Science
   - National Human Genome Research Institute
   - Frequently Asked Questions about Genetic and Genomic Science

6. Precision Medicine
   - Centers for Disease Control and Prevention
   - Precision Medicine

7. Genomics and Health Impact Update
   - Centers for Disease Control and Prevention
   - Genomics and Health Impact Update

8. Genomic Testing
   - Centers for Disease Control and Prevention
   - Genomic Testing

9. Genomics and Health Impact Update
   - Centers for Disease Control and Prevention
   - Genomics and Health Impact Update

10. Genomic Testing
    - Centers for Disease Control and Prevention
    - Genomic Testing
Genetics Home Reference (GHR)

- Health conditions
- Genes
- Chromosomes and DNA
- Classroom
- Help Me Understand Genetics
Breast cancer

Description

Frequency

Causes

Inheritance Pattern

Diagnosis & Management Links

Additional Information & Resources

- Health Information from MedlinePlus (3 links)
- Genetic and Rare Diseases Information Center (2 links)
- Additional NIH Resources (4 links)
- Educational Resources (9 links)
- Patient Support and Advocacy Resources (10 links)
- Clinical Information from GeneReviews (5 links)
- Scientific Articles on PubMed (1 link)
- Catalog of Genes and Diseases from OMIM (3 links)
- Medical Genetics Database from MedGen (2 links)
Explore the normal functions of human genes and the health implications of genetic changes.

A: adenosine deaminase 2 (ADA)
B: adenosine deaminase 2 (ADA)
C: adenosine deaminase 2 (ADA)
D: adenosine deaminase 2 (ADA)
E: adenosine deaminase 2 (ADA)
F: adenosine deaminase 2 (ADA)
G: adenosine deaminase 2 (ADA)
H: adenosine deaminase 2 (ADA)
I: adenosine deaminase 2 (ADA)
J: adenosine deaminase 2 (ADA)
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M: adenosine deaminase 2 (ADA)
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P: adenosine deaminase 2 (ADA)
Q: adenosine deaminase 2 (ADA)
R: adenosine deaminase 2 (ADA)
S: adenosine deaminase 2 (ADA)
T: adenosine deaminase 2 (ADA)
U: adenosine deaminase 2 (ADA)
V: adenosine deaminase 2 (ADA)
W: adenosine deaminase 2 (ADA)
X: adenosine deaminase 2 (ADA)
Y: adenosine deaminase 2 (ADA)
Z: adenosine deaminase 2 (ADA)

Chromosomes & mtDNA

Read about each of the human chromosomes and mitochondrial DNA (mtDNA) and the health implications of genetic changes.

Information about specific chromosomes
If a genetic disorder runs in my family, what are the chances that my children will have the condition?

When a genetic disorder is diagnosed in a family, family members often want to know the likelihood that they or their children will develop the condition. This can be difficult to predict in some cases because many factors influence a person's chances of developing a genetic condition. One important factor is how the condition is inherited. For example:

- Autosomal dominant inheritance: A person affected by an autosomal dominant disorder has a 50 percent chance of passing the mutated gene to each child. The chance that a child will not inherit the mutated gene is also 50 percent. However, in some cases an autosomal dominant disorder results from a new (de novo) mutation that occurs during the formation of egg or sperm cells or early in embryonic development. In these cases, the child's parents are unaffected, but the child may pass the condition to his or her own children.

- Autosomal recessive inheritance: Two unaffected people who each carry one copy of the mutated gene for an autosomal recessive disorder (carriers) have a 25 percent chance with each pregnancy of having a child affected by the disorder. The chance with each pregnancy of having an unaffected child who is a carrier of the disorder is 50 percent, and the chance that a child will not have the disorder and will not be a carrier is 25 percent.

- X-linked dominant inheritance: The chance of passing on an X-linked dominant condition differs between men and women because men have one X chromosome and one Y chromosome, while women have two X chromosomes. A man passes on his Y chromosome to all of his sons and his X chromosome to all of his daughters. Therefore, the sons of a man with an X-linked dominant disorder will not be affected, but all of his daughters will inherit the condition. A woman passes on one or the other of her X chromosomes to each child. Therefore, a woman with an X-linked dominant disorder has a 50 percent chance of having an affected daughter or son with each pregnancy.

- X-linked recessive inheritance: Because the sex chromosomes differ between men and women, the probability of passing on an X-linked recessive disorder also differs between men and women. The sons of a woman with an X-linked recessive disorder are all affected; their daughters have a 50 percent chance of being carriers of the disorder.
Learning about genetics

Six Things Everyone Should Know About Genetics

1) Genes
2) Inheritance
3) Stability
4) Variation
5) Environment
6) Family
<table>
<thead>
<tr>
<th>Diseases</th>
<th>Guides</th>
<th>News</th>
<th>About GARD</th>
<th>En Español</th>
</tr>
</thead>
</table>

GARD Information Specialists can provide you with current, reliable, and easy to understand information about rare or genetic diseases in English or Spanish.

- **How to Find a Disease Specialist**
- **Tips for the Undiagnosed**
- **Support for Patients and Families**
- **Tips for Finding Financial Aid**
- **Help with Travel Costs**
- **How to Get Involved in Research**
- **FAQs About Chromosome Disorders**
- **Medical and Science Glossaries**
NIH Institutes, Centers, and Offices

- National Cancer Institute
- National Heart, Lung, and Blood Institute
- Eunice Kennedy Shriver National Institute of Child Health and Human Development
- National Institute of Neurological Disorders and Stroke
- etc...
National Organization for Rare Disorders (NORD)

- For Patients and Families
  - rare disease information
  - information to give to physicians
  - health insurance information
  - financial assistance
  - support and advocacy
Genetic Counseling Cultural Competence Toolkit

- For Genetic Counselors
- Offers CE
  - National Society of Genetic Counselors
  - American Board of Genetic Counseling
  - International Medical Interpreters Association
  - American Translators Association
Addressing Specific Diagnosis

Sickle Cell Disease
Sickle Cell Disease
Also called: Hemoglobin SS disease, Sickled cell disease

Start Here
- Facts about Sickle Cell Disease (Centers for Disease Control and Prevention)
  Also in Spanish
- Sickle Cell Disease (American Academy of Family Physicians)
  Also in Spanish
- Sickle Cell Disease (Centers for Disease Control and Prevention)
- Sickle Cell Disease (National Marrow Donor Program)
- What is Sickle Cell Disease? (National Heart, Lung, and Blood Institute)
  Also in Spanish
- When Blood Cells Bend: Understanding Sickle Cell Disease (NHLM)

Diagnosis and Tests
- Blood Smear (National Library of Medicine)
  Also in Spanish
- Complete Blood Count (CBC) (National Library of Medicine)
  Also in Spanish
- Hemoglobinopathy Evaluation (American Association for Clinical Chemistry)
- Sickle Cell Tests (American Association for Clinical Chemistry)

Treatments and Therapies
- Blood and Bone Marrow Transplantation (National Heart, Lung, and Blood Institute)
  Also in Spanish
- Bone Marrow Transplantation: MedlinePlus Health Topic (National Library of Medicine)
  Also in Spanish
- Complications and Treatments (Centers for Disease Control and Prevention)
  Also in Spanish
- Living Well with Sickle Cell Disease (Centers for Disease Control and Prevention)
  Also in Spanish
- Sickle Cell Crisis (For Teens) (National Heart, Lung, and Blood Institute)
  Also in Spanish

Living With
- Living Well with Sickle Cell Disease (Centers for Disease Control and Prevention)
  Also in Spanish
- Sickle Cell Disease Association of America

Related Issues
- Five Tips to Help Prevent Infections (Centers for Disease Control and Prevention)
  Also in Spanish
- For People of African, Mediterranean, or Southeast Asian Heritage

Genetics
- Genetics Home Reference: sickle cell disease (National Library of Medicine)
- Learning about Sickle Cell Disease (National Human Genome Research Institute)
- Prenatal Genetic Counseling (For Parents) (National Human Genome Research Institute)
- Sickle Cell Trait (Centers for Disease Control and Prevention)
  Also in Spanish

Health Check Tools
- Sickle Cell Disease Quiz (Centers for Disease Control and Prevention)
  Also in Spanish

Videos and Tutorials
- What Is Sickle Cell Disease? (Gibbs DNA Learning Center)

Statistics and Research
- Data and Statistics (Centers for Disease Control and Prevention)

Clinical Trials
- ClinicalTrials.gov: Anemia, Sickle Cell (National Institutes of Health)

Journal Articles
- References and abstracts from MEDLINE/PubMed (National Library of Medicine)
  - Article: Knowledge Deficit of Sickle Cell Trait Status: Can Nurses Help?
  - Article: Inhaled steroids associated with decreased macrophage markers in nonasthmatic individuals with...
  - Article: Association of Matched Sibling Donor Hematopoietic Stem Cell Transplantation With Transcranial...
  - Sickle Cell Disease – see more articles

Find an Expert
- Find a Genetic Counselor (National Society of Genetic Counselors)
- National Heart, Lung, and Blood Institute (National Library of Medicine)
- Sickle Cell Disease Association of America

Children
Sickle cell disease

Mutation in the **HBB** gene cause sickle cell disease. 

**Hemoglobin** consists of four protein subunits, typically, two subunits called alpha-globin and two subunits called beta-globin. The **HBB** gene provides instructions for making beta-globin. Various versions of beta-globin result from different mutations in the **HBB** gene. One particular **HBB** gene mutation produces an abnormal version of beta-globin known as hemoglobin S (HbS). Other mutations in the **HBB** gene lead to additional abnormal versions of beta-globin, such as hemoglobin C (HbC) and hemoglobin E (HbE). **HBB** gene mutations can also result in an unusually low level of beta-globin; this abnormality is called beta thalassemia.

In people with sickle cell disease, at least one of the beta-globin subunits in hemoglobin is replaced with hemoglobin S. In sickle cell anemia, which is a common form of sickle cell disease, hemoglobin S replaces both beta-globin subunits in hemoglobin. In other types of sickle cell disease, just one beta-globin subunit in hemoglobin is replaced with hemoglobin S. The other beta-globin subunit is replaced with a different abnormal variant, such as hemoglobin C. For example, people with sickle-hemoglobin C (HbSC) disease have hemoglobin molecules with hemoglobin S and hemoglobin C instead of beta-globin. If mutations that produce hemoglobin S and beta-thalassemia occur together, individuals have hemoglobin S-beta thalassemia (HbSBetaThal) disease.

Abnormal versions of beta-globin can distort red blood cells into a sickle shape. The sickle-shaped red blood cells die prematurely, which can lead to anemia. Sometimes the inflexible, sickle-shaped cells get stuck in small blood vessels and can cause serious medical complications.
Sickle Cell Disease

See also
Information for Health Professionals

Types
Causes
Risk Factors
Screening and Prevention
Signs, Symptoms, and Complications
Diagnosis
Treatment
Living With
Research for Your Health
Participate in NHLBI Clinical Trials
More Information

Related News
December 03, 2018  |  Media Availability
NIH researcher presents encouraging results for gene therapy for severe sickle cell disease

WHAT: A scientist from the National Institutes of Health will present promising, early results from a human...
Sickle cell anemia

Other Names: HbS disease, Hemoglobin S Disease, Sickling disorder due to hemoglobin S
Categories: Blood Diseases, Congenital and Genetic Diseases, Endocrine Disease

This disease is grouped under: Hemoglobinopathy

Summary

Sickle cell anemia is a disease in which the body produces abnormally shaped or sickle shaped red blood cells. These cells do not last as long as normal, round red blood cells. The sickle cells also get stuck in blood vessels, blocking blood flow. This can cause pain and other problems. Sickle cell anemia is inherited as an autosomal recessive pattern. Treatment typically includes pain medicines during crises; hydroxyurea to reduce the number of new sickle cells; and vaccines to prevent bacterial infections and blood transfusions. On July 2019, the FDA approved the use of Endari (prescription grade L-glutamine) to reduce the number of new sickle cells. This is the first FDA approved treatment that is also available for children with sickle cell anemia.

Treatment

The resources below provide information about treatment options for this condition. If you have questions about which treatment is right for you, talk to your healthcare professional.

Management Guidelines

- **Project OrphanAnesthesia** is a project whose aim is to create peer-reviewed, readily accessible guidelines for patients with rare diseases and for the anesthesiologists caring for them. The project is a collaborative effort of the German Society of Anesthesiology and Intensive Care, Orphanet, the European Society of Pediatric Anesthesia, anesthetists and rare disease experts with the aim to contribute to patient safety.

FDA-Approved Treatments

The medication(s) listed below have been approved by the Food and Drug Administration (FDA) as orphan products for treatment of this condition. Learn more orphan products.

- **Hydroxyurea (Brand name: Droxia)** - Manufactured by Bristol-Myers Squibb Co.
  FDA-approved indication: To reduce the frequency of painful crises and to reduce the need for blood transfusions in adult patients with sickle cell anemia with recurrent moderate to severe painful crises (generally at least 3 during the preceding 12 months).
  National Library of Medicine Drug Information Portal
  Medline Plus Health Information

- **L-glutamine oral powder (prescription grade) (Brand name: Endari)** - Manufactured by Emmaus Medical, Inc.
  FDA-approved indication: To reduce the acute complications of sickle cell disease in adult and pediatric patients 5 years of age and older.
  National Library of Medicine Drug Information Portal

- **Hydroxyurea (Brand name: Siklos)** - Manufactured by Addmedica Laboratories
  FDA-approved indication: To reduce the frequency of painful crises and to reduce the need for blood transfusions in pediatric patients, 2 years of age and older, with sickle cell anemia with recurrent moderate to severe painful crisis.
  National Library of Medicine Drug Information Portal
  Medline Plus Health Information

Find a Specialist

If you need medical advice, you can look for doctors or other healthcare professionals who have experience with this disease. You may find these specialists through advocacy organizations, clinical trials, or articles published in medical journals. You may also want to contact a university or tertiary medical center in your area, because these centers tend to see more complex cases and have the latest technology and treatments.
Sickle Cell Disease

NORD gratefully acknowledges MA Bender, MD, PhD, Department of Pediatrics, University of Washington, Fred Hutchinson Cancer Research Center, Seattle, Washington, for assistance in the preparation of this report.

Synonyms of Sickle Cell Disease
- SCD

Subdivisions of Sickle Cell Disease
- sickle cell anemia
- sickle cell hemoglobin C disease
- sickle cell thalassemia disease

General Discussion
Summary Sickle cell disease (SCD) is a rare blood disorder that is inherited in an autosomal recessive manner. It is characterized by the presence of sickle, or crescent-shaped, red blood cells (erythrocytes) in the bloodstream. These crescent-shaped cells are stiff and sticky and interfered with other cells and the

Search Rare Diseases
Enter a disease name or synonym to search NORD's
Addressing Specific Diagnosis

Genetic testing: prenatal, newborn, child
Healthy Lifestyle

Pregnancy week by week

Prenatal testing, including screening and diagnostic tests, can provide valuable information about your baby’s health. Understand the risks and benefits.

By Mayo Clinic Staff

Pregnancy is a time of great anticipation — and, sometimes, anxiety. You might worry that your baby will have health problems. While most babies are born healthy, it’s important to understand your options for obtaining details about your baby’s health.

The two main types of prenatal testing are:

• Screening tests. Prenatal screening tests can identify whether your baby is more or less likely to have certain birth defects, many of which are genetic disorders. These tests include blood tests, a specific type of ultrasound and prenatal cell-free DNA screening. Prenatal screening tests are usually offered during the first or second trimester. Screening tests can’t make a definitive diagnosis. If results indicate an increased risk for a genetic disorder, your health care provider will discuss your options for a diagnostic test to confirm the diagnosis.

• Diagnostic tests. If a screening test indicates a possible problem — or your age, family history or medical history puts you at increased risk of having a baby with a genetic problem — you might consider an invasive prenatal diagnostic test. A diagnostic test is the only way to be sure of a diagnosis. Some diagnostic tests, such as chorionic villus sampling and amniocentesis, carry a slight risk of miscarriage.

Prenatal screening tests include:

• First trimester screening tests. During your first trimester, your health care provider will offer a blood test and an ultrasound to measure the size of the clear space in the tissue at the back of a baby’s neck (nuchal transluency). In Down syndrome and in certain other conditions, the nuchal transluency measurement is abnormally large.

• Second trimester screening tests. During your second trimester, your health care provider will offer another blood test called the quad screen. This test measures levels of four substances in your blood. Results indicate your risk of carrying a baby who has certain chromosomal conditions, such as Down syndrome. The test can also help detect neural tube defects — serious abnormalities of the brain or spinal cord.

• Prenatal cell-free DNA screening. This blood test examines fetal DNA in the maternal bloodstream to screen for the increased chance for specific chromosome problems, such as Down syndrome. This screening can also provide information about a baby’s sex and the Rh blood type.

Prenatal screening tests for fetal abnormalities are optional. It’s important to make an informed decision about prenatal testing, especially if you’re screening for fetal conditions that can’t be treated. Before going forward, consider these questions:

• What will you do with the test results? Normal results can ease your anxiety. However, if prenatal testing indicates that your baby might have a birth defect, you could be faced with wrenching decisions — such as whether to continue the pregnancy. On the other hand, you might welcome the opportunity to plan for your baby’s care in advance.

• Will the information shape your prenatal care? Some prenatal tests detect problems that can be treated during pregnancy. In other cases, prenatal testing alerts your health care provider to a condition that requires immediate treatment after birth.

• How accurate are the results? Prenatal screening isn’t perfect. The rate of inaccurate results, known as false-negative or false-positive results, varies from test to test.

• What are the risks? Weigh the risks of specific prenatal tests — such as anxiety, pain or possible miscarriage — against the value of knowing the results.
Prenatal Genetic Screening Tests

- What is prenatal genetic testing?
- What are genetic disorders?
- What are the two main types of prenatal genetic tests?
- What are the different types of prenatal genetic screening tests?
- What is first-trimester screening?
- What is second-trimester screening?
- What is combined first- and second-trimester screening?
- What is cell-free DNA testing?
- What do the different results of prenatal screening tests mean?
- How accurate are prenatal genetic screening tests?
- What should I consider when deciding whether to have prenatal genetic testing?
- Glossary

Prenatal Genetic Diagnostic Tests

- What is prenatal genetic testing?
- What are genetic disorders?
- What are the two main types of prenatal genetic tests?
- What is amniocentesis?
- What is chorionic villus sampling?
- What is preimplantation genetic diagnosis?
- How are the cells analyzed in prenatal diagnostic testing?
- What do the different results of prenatal diagnostic tests mean?
- What should I consider when deciding whether to have prenatal genetic testing?
- How do I choose between prenatal screening and diagnostic testing?
- Glossary
Newborn Screening Welcome

The National Newborn Screening and Global Resource Center (NNSGRC) is an independent, nongovernmental, national resource center for newborn screening information. It serves as the official clearinghouse for newborn screening information and its purpose is to improve the availability and quality of newborn screening across the United States. The NNSGRC was established by the U.S. Department of Health and Human Services and the National Institutes of Health in January 2013.

Originally created as a federal focal point for newborn screening in the US, the NNSGRC is dedicated to providing newborn screening information both nationally and globally. Consultative services, program reviews, and international subcommittee affiliations are among the many benefits of newborn screening.

For Families

Frequently Asked Questions about Newborn Bloodspot Screening
Find out the answers to the following questions and more. Why does my baby need newborn screening tests? How will my baby be tested? How will I get the results of the test?

Conditions Required or Otherwise Included in State Newborn Screening Programs
A comprehensive listing (in chart format) that shows the implementation status of the various conditions on the nationally recommended Uniform Screening Panel (USP). The current USP includes 31 conditions. Other conditions may be required by some states and these are also included. Readers should be sure to understand which conditions are required by state law or rule and those that may not be required but may be included as a result of the technology being used. Notes at the top of the table explain the symbols used in the tables.

State Newborn Screening Program Contacts
A PDF file containing contact information for the laboratory and follow-up coordinators in each state's and territory's newborn screening program.

Hearing Screening
Hearing screening resources, state screening programs, legislation, organizations, and parent education links.

Disorder Fact Sheets
Disorder Fact Sheets for families provided by the Screening, Technology and Research in Genetics (STAR-G) Project.

Additional Testing
List of screening laboratories that may charge a fee for additional screening not offered in some screening programs.

Genetics Information
Links to genetics counseling, family forums, and fact sheets on genetics.
Baby’s First Test

What is Newborn Screening?

Many parents are unaware of the conditions included in screening, or that it varies from state to state. Baby’s First Test brings together resources to help guide parents and health professionals alike.

What To Expect

Newborn screening is just one of many things that happen in the first few days after a baby is born. The following information can help prepare expecting parents for the newborn screening process and answer common questions, such as:

- What should I do before birth?
- What are the screening procedures?
- What happens to the blood sample?
- How should I respond to results?

ASK AN EXPERT

Have a question that’s not answered on Baby’s First Test? Send it to our experts.

Newborn Screening 101

- Genetics & Family History
- Newborn Screening Community
- Recommended Uniform Screening Panel (RUSP)
- Glossary
- Ask an Expert

Family Experiences

Learn from other families, in their own words, what their journey has been like after a diagnosis. Their stories not only give us valuable insight into how they manage conditions, but also reaffirm that none of us are alone. You can also visit the Baby’s First Test YouTube channel for family stories and guides to support your family through the newborn screening process. Do you have a newborn screening story you would like to share?

9 Stories

- It’s His Heart

Billy Werner tells the story of her grandson, Lucas, who was born with critical
About Genetic Counselors

What Happens at a Prenatal Genetic Counseling Appointment?

Pregnancy is an exciting time for couples but it can also be a stressful one. Being referred to a genetic counselor can temporarily raise stress levels, but genetic counseling can give moms and couples helpful information, guidance and very often peace of mind. There are many reasons a couple may be referred for prenatal genetic counseling:

- There may be a personal or family history of a genetic condition or birth defect
- They may have a child with a genetic condition or birth defect
- They may have lost a baby or lost two or more pregnancies
- The results of an ultrasound or screening test might suggest the need for genetic testing

However, many couples with no specific indication can also benefit from genetic counseling. While prenatal genetic screening is a routine part of pregnancy, it has become increasingly complicated with more and more options for patients to consider.
Addressing Specific Diagnosis

Ovarian Cancer
Ovarian cancer

- Description
- Frequency
- Causes
- Inheritance Pattern
- Diagnosis & Management Links
- Other Names for This Condition
- Additional Information & Resources
- Sources for This Page
Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening (PDQ®) - Patient Version

ON THIS PAGE

- What is screening?
- General Information About Ovarian, Fallopian Tube, and Primary Peritoneal Cancer
- Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening
- Risks of Ovarian, Fallopian Tube, and Primary Peritoneal Cancer Screening
- About This PDQ Summary

What is screening?

Screening is looking for cancer before a person has any symptoms. This can help find cancer at an early stage. When abnormal tissue or cancer is found early, it may be easier to treat. By the time symptoms appear, cancer may have begun to spread.

Scientists are trying to better understand which people are more likely to get certain types of cancer. They also study the things we do and the things around us to see if they cause cancer. This information helps doctors recommend who should be screened for cancer, which screening tests should be used, and how often the tests should be done.

It is important to remember that your doctor does not necessarily think you have cancer if he or she suggests a screening test. Screening tests are given when you have no cancer symptoms.

If a screening test result is abnormal, you may need to have more tests done to find out if you have cancer. These are called diagnostic tests.
Ovarian cancer

Other Names: Ovarian carcinoma
Categories: Rare Cancers

Summary

Ovarian cancer is a form of cancer that occurs due to abnormal and uncontrolled cell growth in the ovaries. Many people with early ovarian cancer have no signs or symptoms of the condition. When present, symptoms are often nonspecific and blamed on other, more common conditions. Most cases of ovarian cancer occur sporadically in people with little to no family history of the condition; however, approximately 10-25% of ovarian cancers are thought to be "hereditary." Although the underlying genetic cause of some hereditary cases is unknown, many are part of a hereditary cancer syndrome (such as BRCA1 or BRCA2 hereditary breast and ovarian cancer syndrome, Lynch syndrome, and Peutz-Jeghers syndrome) and are inherited in an autosomal dominant manner. The best treatment options for ovarian cancer depend on many factors including the subtype and stage of the condition, but may include surgery, chemotherapy, radiation therapy, and/or targeted therapy (such as monoclonal antibody therapy).

Last updated: 3/30/2015

Symptoms

Many people with early ovarian cancer have no signs or symptoms of the condition. When present, symptoms
OVARIAN CANCER

There are five main types of cancer that affect a woman’s reproductive organs: cervical, ovarian, uterine, vaginal, and vulvar. As a group, they are referred to as gynecologic cancers (GYN nehl kahrz LEHN jik). A sixth type of gynecologic cancer is the very rare fallopian tube cancer.

This fact sheet about ovarian cancer is part of the Centers for Disease Control and Prevention’s (CDC) Inside Knowledge About Gynecologic Cancer campaign. The campaign helps women get the facts about gynecologic cancer, providing important “inside knowledge” about their bodies and health.

What is ovarian cancer?
Cancer is a disease in which cells in the body grow out of control. Cancer is always named for the part of the body where it starts, even if it spreads to other body parts later.

When cancer starts in the ovaries, it is called ovarian cancer. Women have two ovaries that are located in the pelvis, one on each side of the uterus. The ovaries make female hormones and produce eggs.

When ovarian cancer is found in its early stages, treatment is most effective.

What raises a woman’s risk for getting ovarian cancer?
There is no way to know if you will get ovarian cancer. However, a woman’s risk for ovarian cancer is higher if she has:

- Being middle-aged
- Having close family
- Using birth control pills

Inside Knowledge About Gynecologic Cancer

Knowledge Is Power: Ovarian Cancer

Inside Knowledge About Gynecologic Cancer provides resources to women, healthcare providers, and organizations to share with patients and communities.

- NEW Shareable buttons and badges
- NEW Short animated videos
- PSAs
- Posters
- Information about each of the most common gynecologic cancers: cervical, ovarian, uterine, vaginal, and vulvar
- NEW Family History and Cancer Fact Sheet
- Printable version
- Provider continuing education (CE) on gynecologic cancers

Inside Knowledge was developed by CDC and others to improve awareness of gynecologic cancer.
**Ovarian cancer**

Ovarian cancer is cancer that begins in the ovaries. The ovaries make female hormones and produce a woman's eggs. Ovarian cancer is a serious cancer that is more common in older women. Treatment is most effective when the cancer is found early. Screening for ovarian cancer is not recommended for most women.

Learn more about ovarian cancer at the National Cancer Institute.

**What is ovarian cancer?**

**Who gets ovarian cancer?**

**Are some women more at risk for ovarian cancer?**

Women with a high risk of ovarian cancer are those with a harmful mutation on the BRCA1 or BRCA2 genes. These mutations can be found with a blood test. Women with a family or personal history of breast or ovarian cancer also have a higher risk of ovarian cancer.

If you have family members in multiple generations with breast cancer or ovarian cancer, see your doctor to learn more about your risk of ovarian cancer. Research shows that certain steps, such as surgery to remove the ovaries and the fallopian tubes, may help prevent ovarian cancer in women who are at high risk. The sooner ovarian cancer is found and treated, the better your chance for recovery. But ovarian cancer is hard to detect early because its symptoms are also the symptoms of many other illnesses.

**Q: What are the symptoms of ovarian cancer?**

A: The following may be symptoms of ovarian cancer if they continue or get worse over time:
- Pain in the pelvis or abdomen (belly)
- Bloating in the abdomen
- Urinary urgency (need to pee right away)
- Urinary frequency (having to pee often)
- Constipation or diarrhea
- Feeling full quickly while eating
- Having difficulty eating
- Vaginal bleeding or other discharge that is different than normal
- Back pain

If you have any of these symptoms, talk to your doctor. He or she can determine if the cause is cancer or...
The Genetics of Cancer

ON THIS PAGE

- Genetic Changes and Cancer
- Hereditary Cancer Syndromes
- Genetic Tests for Hereditary Cancer Syndromes
- Identifying Genetic Changes in Cancer

Genetic Changes and Cancer

Cancer is a genetic disease—that is, cancer is caused by certain changes to genes that control the way our cells function, especially how they grow and divide.

Genes carry the instructions to make proteins, which do much of the work in our cells. Certain gene changes can cause cells to evade normal growth controls and become cancer. For example, some cancer-causing gene changes increase production of a protein that makes cells grow. Others result in the production of a missshapen, and therefore nonfunctional, form of a protein that normally repairs cellular damage.

Genetic changes that promote cancer can be inherited from our parents if the changes are present in germ cells, which
Whether you work in a hospital, public, or health sciences library you may have a patron or patient seeking information about a particular health condition such as Alzheimer's disease either for themselves or someone they know. As genetics becomes more commonly referred to in relation to Alzheimer's it is good to be able to have a variety of resources available to use or to refer others to as patients may request information about their own risk or the risk for their children. Using the resources mentioned in this class see if you can locate information for the following questions.

It is recommended you start with Genetics Home Reference and from there explore the other resources mentioned in this class.

Please state which resource you used to answer each question:

- What genes are associated with early onset Alzheimer's?
- What genes are associated with late onset Alzheimer's?
- What resource would you use that provides the patron with information about how Alzheimer’s runs in the family?
- What resource would you offer a patron to learn more about the basics of genetics?
Direct to Consumer Genetic Testing
Direct to Consumer Genetic Testing Companies
**DTC BRCA test**

**FDA announcement**

**23andMe announcement**
Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement

Nick Webborn,1 Alun Williams,2 Mike McNamee,3 Claude Bouchard,4 Yannis Pitsiladis,5 Ildus Ahmetov,6 Euan Ashley,7 Nuala Byrne8 Silvia Camporesi,9 Malcolm Collins,10 Paul Dijkstra,11 Nir Eymon,11 Noriyuki Fuku,12 Fleur C Garton,13 Nils Hoppe,15 Soren Holm,16 Jane Kaye,17 Vassilis Kliassouras,18 Alejandro Lucia,19 Kamel Maase,20 Colin Moran,21 Kathryn N North,14 Fabio Pigozzi,22 Guan Wang5

ABSTRACT

The general consensus among sport and exercise genetics researchers is that genetic tests have no role to play in talent identification or the individualised prescription of training to maximise performance. Despite the lack of evidence, recent years have witnessed the rise of an emerging market of direct-to-consumer marketing (DTC) tests that claim to be able to identify children’s athletic talents. Targeted consumers include mainly coaches and parents. There is concern among the scientific community that the current level of knowledge is being misrepresented for commercial purposes. There remains a lack of universally accepted guidelines and legislation for DTC testing in relation to all forms of genetic testing and not just for talent identification. There is concern over the lack of clarity of information over which specific genes or variants are being tested and the almost universal lack of appropriate genetic counselling for the interpretation of the genetic data to consumers. Furthermore, recent independent studies have identified issues relating to quality control by DTC laboratories with different results being reported from the same test. There is no evidence of the value in relation to genetic testing and the limitations of current knowledge. This article reviews the issues around the currently available evidence behind the genetic testing, comments on the ethical considerations and makes recommendations about such tests.

STATEMENT ON BACKGROUND TO THE CONSENSUS PROCESS

A group of world experts in the field of genomics, exercise, sport performance, disease, injury and antidoping gathered with the International Federation of Sports Medicine (IFMS) Scientific Commission for a symposium to discuss the current state of knowledge and to share ideas. One key concern was the misuse of research evidence and the misinformation about genetic testing, particularly when marketed directly to the public, coaches or parents. This is known as DTC testing for the purpose of talent identification and to assess potential for future sports performance. There have been
How Should Primary Care Physicians Respond to Direct-to-Consumer Genetic Test Results?

Kyle B. Brothers, MD, PhD and Esther E. Knapp, MD, MBE
Concerns

- Privacy
- Legality
- Who has access?
- How useful now?
- What all is being done now and in the future with the information?
- Unexpected surprises?
- Test results can vary among companies
- Validity of tests
- No counseling provided
- Who can get the testing?
Benefits

- Learn more about own health
- More effective medical treatments
- Learn more about ethnicity and family history
- Bring awareness to family health issues for future generations
- Motivation to work on health habits
- Encourages patient engagement
- Contributing to advancement of healthcare and science
- Moral obligation
### Direct-to-Consumer Genetic Testing

- What is direct-to-consumer genetic testing?
- What kinds of direct-to-consumer genetic tests are available?
- What is genetic ancestry testing?
- What are the benefits and risks of direct-to-consumer genetic testing?
- How do I choose a direct-to-consumer genetic testing company?
- How is direct-to-consumer genetic testing done?
- How much does direct-to-consumer genetic testing cost, and is it covered by health insurance?
- What do the results of direct-to-consumer genetic testing mean?
- What can raw data from a direct-to-consumer genetic test tell me?
- Can a direct-to-consumer genetic test tell me whether I will develop cancer?
- Can a direct-to-consumer genetic test tell me whether I will develop Alzheimer disease?
- What does it mean to have hunter/donovan DNA?
- How do direct-to-consumer genetic testing companies protect their customers’ privacy?
- Can the results of direct-to-consumer genetic testing affect my ability to get insurance?
- Where can I read more about the diseases and traits covered in my direct-to-consumer genetic testing report?

- What kinds are available
- Ancestry testing
- What do results mean
- Risk of developing a disease
- Privacy protection
Information for Patients/Consumers about DTC


Sheldon Krimsky and David Gay Johnston
Council for Responsible Genetics
A project funded by the Rose Foundation
March 2017

What You Need to Know about Direct-to-Consumer Genetic Testing

What is direct-to-consumer (DTC) genetic testing?

DTC refers to a genetic test you can complete at home without a health care provider. You collect a DNA sample and send it to the company. They analyze it and give you a report on your genetics.

What information can I get from a DTC genetic test?

There are many different types of DTC tests available. Some tell you about ancestry, kinship, lifestyle factors, and disease risk. Companies can analyze your DNA and give information about those things.

What do DTC companies do with my data?

That depends on the fine print! You should read up on each company’s policies. They vary on how they decide to store your sample and your data, and with
What is At-Home Genetic Testing?

Understanding what an at-home test will - and will not - tell you

For a price (typically several hundred to a thousand dollars) you can order a genetic testing kit online or by phone. You'll swab your cheek or spit into a test tube. Then you will mail it to a lab where it may be tested for a wide variety of things — from whether you inherited your intolerance to the lactose in dairy products to your risk of certain types of cancer to if you carry a gene for a serious illness such as Cystic Fibrosis and could pass it on to your children.

There are at-home tests for:

- Traits (e.g., male hair loss to dimples)
- Wellness (e.g., risk of certain types of cancer to restless leg syndrome)
- Ancestry reports (i.e., ethnicity and lineage)
- Carrier status (e.g., Tay-Sachs Disease to Sickle Cell Anemia)
- Paternity testing (i.e., determining a child's biological father)

There are three general ways to get genetic testing:

- Through your physician or a genetic counselor — These are the most detailed, comprehensive tests. They include cancer testing and testing for genetic disorders. They include large "panels."

Related Links

- FIND A GENETIC COUNSELOR
- PREGNANT CONDITIONS
- FAMILY HEALTH HISTORY
- PROTECTING YOUR PRIVACY
- ABOUT AT-HOME TESTING

Aboutgeneticcounselors.com What is At-Home Genetic Testing?
Genetic Counselors

- Evaluate family history and medical records
- Assist in making decisions regarding genetic testing
- Identify and interpret risks of inherited disorders, increase the family’s understanding of a genetic condition
- Discuss options regarding disease management and the risks and benefits of further testing and other options
- Help the individual and family identify the psychosocial tools required to cope with potential outcomes
- Reduce the family’s anxiety

National Society of Genetic Counselors
Deciding about genetic testing

Questions to ask:

- Am I in the group at risk and should I get tested?
- If I decide to get tested, what do the results mean?
- What are my treatment options based on results?
- How do I decide on treatment?
Knowing is Not Enough—Act on Your Family Health History

Have you or a family member had a heart attack, stroke, or diabetes? If yes, you're more likely to get the same disease as your parents or siblings. If you answered "yes," you're more likely to get heart disease, stroke, or diabetes. If you answered "no," you're less likely to get the same disease as your parents or siblings. If you answered "yes," you're more likely to get the same disease as your parents or siblings. If you answered "no," you're less likely to get the same disease as your parents or siblings.

How to Collect Your Family Health History

- Talk to your family. Write down the names of your close relatives on both sides of the family, including parents, siblings, grandparents, aunts, uncles, nieces, and nephews. Talk to these family members about what conditions they have or had, and at what age the conditions were first diagnosed.
- Ask questions. To find out about your risk for chronic diseases, ask your relatives about which of these diseases they have had and when they were diagnosed. Questions can include:
  - Do you have any chronic diseases, such as heart disease or diabetes, or health conditions, such as high blood pressure or high cholesterol?
  - Have you had any other serious diseases, such as cancer or stroke?
  - What type of cancer did you have?
  - How old were you when each of these diseases or health conditions was diagnosed?
  - What was the cause of death for relatives who have died?

CDC – Family Health History
Family Health History and Chronic Disease

If you have a family health history of a chronic disease like cancer, heart disease, diabetes, or osteoporosis, you are more likely to get that disease yourself. Share your family health history with your doctor, who can help you take steps to prevent disease and catch it early if it develops.

Learn more about what having a family health history of each of these conditions means for you:

- **Breast & Ovarian Cancer**
- **Colorectal Cancer and Lynch Syndrome**
- **Heart Disease, Genetics and Family History**
- **Diabetes**
- **Hereditary Hemochromatosis**
- **Osteoporosis**

**Family Health History**

The Basics

Expecting a baby? You might wonder about baby’s temper or daddy’s temper. But you might want to know about your parents’ family health history. If your parents have a family health history of conditions like cancer, heart disease, or diabetes, you may be at higher risk for certain conditions. If you have a family history of cancer, heart disease, or diabetes, you may need to be screened more often for those conditions.

Based on your family history, you may need to be screened more often for cancer, heart disease, and diabetes.

Other reasons for genetics tests:

- Infertility or trouble getting pregnant
- 2 or more miscarriages
- A previous pregnancy with a genetic condition
- A baby who died at birth from a genetic condition

After genetic counseling, you may decide to have a carrier test. A carrier test can tell you if you could have a child with the disease but does not tell you if you have the disease.

If the results show that you are a carrier for a disease, the other potential parent would also need to have carrier screening to know if you could have a baby with the disease.
Disclaimer: The Surgeon General’s My Family Health Portrait tool does NOT keep a government record of the information you fill in nor make your health information available to anyone else but you. It only provides the software for organizing your information. By accessing the tool on the web, you make use of that software. But the information you fill in is not transmitted back to our servers, and never available to anyone else, unless you choose to share or disclose it. After you fill in your information, it is available only to you for downloading. After that, it’s up to you whether you want to share the information with other family members or provide it to your health care practitioner. The Surgeon General’s tool helps gather information that will be useful for you and your health care practitioner, but it does not provide medical advice. You should consult with a health professional about advice based on your family health history information.

Your Personal Information

We start the family health history with you. Enter the required personal information and your health history information. At the bottom of the page (you may need to scroll), press the 'Next' button. You will then be asked to tell the system which family members you would like to add to the health history.

* Indicates required information.

- **Name:**
- **Sex assigned at birth:**
  - Male
  - Female
- **Date of Birth** (mm/dd/yyyy)
- **Were you born a twin?**
  - No
  - Yes - Identical (Same)
  - Yes - Not Identical (Fraternal)
- **Were you adopted?**
  - Yes
  - No
- **Height**
  - Feet
  - Inches
  - OR
  - Centimeters
- **Weight**
  - lbs

Your Health Information

In the list below, select a Disease or Condition (if any) from the dropdown box. Then select the Age at Diagnosis and press the Add button. You may repeat this process as necessary.

<table>
<thead>
<tr>
<th>Disease or Condition</th>
<th>Age at Diagnosis</th>
<th>Action</th>
</tr>
</thead>
<tbody>
<tr>
<td>Please Select a Disease</td>
<td>Select Age at Diagnosis</td>
<td>Add</td>
</tr>
</tbody>
</table>

Your Family Background Information

Check here if your parents are related to each other in any way other than marriage. Multiple races and ethnicities may be selected.

- **Race:**
  - American Indian or Alaska Native
  - Asian
  - Black or African-American
  - Native Hawaiian or Other Pacific Islander
  - White

- **Ethnicity:**
  - Hispanic or Latino
  - Ashkenazi Jewish
  - Not Hispanic or Latino

Why are we asking about Ashkenazi Jewish heritage?
Genetic Alliance guides

A Guide to Family Health History

A Guide to Genetics and Health

Genetic Alliance resources
Ethics and Privacy
Societal Concerns

- Who should have access to personal genetic information, and how will it be used?
- Who owns and controls genetic information?
- How does personal genetic information affect an individual and society's perceptions of that individual?
- What are the larger societal issues raised by new reproductive technologies?
- How will genetic tests be evaluated and regulated for accuracy, reliability and utility?
- How do we prepare healthcare professionals and the public?
- What is considered acceptable diversity?
- Where is the line between medical treatment and enhancement?
- Should testing be performed when no treatment is available?
In Hunt For Golden State Killer, Investigators Uploaded His DNA To Genealogy Site

by LAUREL WAMSLEY

April 27, 2018 • After failing to find a match within criminal databases, law enforcement uploaded the killer’s DNA profile to a no-frills website used to trace ancestry. The tactic has spurred privacy concerns.
Information protected by GINA

- Family health history
- Results of genetic tests
- Use of genetic counseling and other genetic services
- Participation in genetic research
- But....
GINA does not apply to...

- Members of the U.S. military who receive their care through the Tricare military health system
- Veterans who receive their care through the Veteran’s Administration
- The Indian Health Service
- Federal employees who get their care through the Federal Employees Health Benefits Plans
  - These have their own policies similar to GINA
H.R. 1313 Employee Wellness Program

The NEW ENGLAND JOURNAL of MEDICINE

Undermining Genetic Privacy? Employee Wellness Programs and the Law
Kathy L. Hudson, Ph.D., and Karen Pollitz, M.P.P.

Genetic information is becoming ubiquitous in research and medicine. The cost of genetic analysis continues to fall, and its medical and personal value continues to grow.

PMID: 28537794
Genomic Health Literacy

People in a scientifically and genetically literate society are:

- better able to understand the world they live in
- better able to understand the diversity of life
- better able to understand the basics of health and disease
- more empowered to make informed decisions about their healthcare
- more empowered to make informed decisions about privacy, security, ethical, and societal issues related to genetics
Policy Issues in Genomics

NHGRI is committed to driving the responsible use of genomics in society in order to advance knowledge and ensure that genomics benefits the health of all humans. To do this, we consider the ethical, legal, and social aspect of genomics research in our work, including these key issues.
Literacy Resources
Precision Medicine

“...a bold new research effort to revolutionize how we improve health and treat disease.”
Mission statement:

To enable a new era of medicine through research, technology, and policies that empower patients, researchers, and providers to work together toward development of individualized care.
Precision Medicine is...

- Precision medicine is a revolutionary approach for disease prevention and treatment that takes into account individual differences in lifestyle, environment, and biology.
- Instead of what treatment is right for this disease it is what treatment is right for this patient.
Genomic Research

Precision Medicine

What is precision medicine?

What is the difference between precision medicine and personalized medicine? What about pharmacogenomics?

What is the Precision Medicine Initiative?

What are some potential benefits of precision medicine and the Precision Medicine Initiative?

What are some of the challenges facing precision medicine and the Precision Medicine Initiative?
All of Us & NNLM Community Engagement Team

**MidContinental Region**
George Strawley
(CO, KS, MO, UT, & WY)

**Greater Midwest Region**
Darlene Kaskie
(IL, IA, KY, MI, MN, ND, OH, SD, & WI)

**New England Region**
Catherine Martin
(CT, ME, MA, NH, RI, & VT)

**Middle Atlantic Region**
Veronica Leigh Milliner
(DE, NJ, NY, & PA)

**Southeastern/Atlantic Region**
April Wright
(AL, DC, FL, GA, MD, MI, NC, PR, SC, TN, VI, VA, & WV)

**Pacific Northwest Region**
Michele Spatz
(AK, ID, MT, OR, & WA)

**Pacific Southwest Region**
Kelli Ham
(AZ, CA, HI, NV & US Territories in the Pacific Basin)

**South Central Region**
Rachel Tims
(AK, LA, NM, OK, & TX)
Participating in Genomics Research

- What is the purpose of the study?
- How much effort will I be expected to commit?
- What healthcare will I receive as part of the study?
- How will participating affect me and my family?
- Is it physically safe to participate in genomics research?
- How will my privacy be protected?
- Might I be discriminated against as a results of information about my genome being revealed?
Participating in Genomics Research, continued
“Preparing the public to make educated personal and family health decisions in a time of rapidly evolving genetic and genomic knowledge will require new partnerships between the education system, health care systems, the government, community advocacy organizations, consumers and the media.”

PMID: 23448722

#thinkbeforeyouspit
#geneticsandhealth
#familyhealthhistory
1) What is the revolutionary healthcare approach that takes into account individual differences in lifestyle, environment, and biology?

2) Clinicians are not concerned about all genetic variants – only those that are ____________.

3) Do you need to have a genetic test in order to be prescribed some FDA-approved drugs?

4) If you have a known pathogenic variant of BRCA1, will you get breast cancer before you’re 80 years old?

5) True or False? GINA (Genetic Information Nondiscrimination Act) protects you from life insurance discrimination.

6) What resource would you recommend to patients who wanted to learn more about a genetic condition?

7) What is a good starting place for finding genetic information for clinicians?
Questions?

Diana Louden, MLib
Biomedical & Translational Sciences Librarian
UW Health Sciences Library
DKNL@uw.edu

Carolyn Martin, MLS, AHIP
Consumer Health Coordinator
NNLM PNR
martinc4@uw.edu