

DNA Analysis: Part II: Genetic Testing

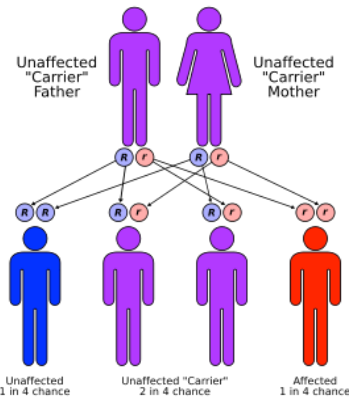
US275 Scientific Ethics
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Genetic testing identifies the alleles of an individual's genes.

- Usually examines genes for inherited diseases.
- determine if a person carries an affected allele
 - potential to develop disorder
 - carrier that could pass allele to offspring.



2 *Methods of inheritance of autosomal recessive genes.*
Wikimedia Commons, 2007

The laboratory doing the genetic testing examines the DNA in a person's cells

- Normally part of a genetic consultation ordered by a clinician
 - buccal smear collecting cheek cells from inside of mouth
 - blood cells from newborn infant



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Genetic counselors discuss the results of the tests with patients

- Implications of results may be complicated
 - emotional
 - health decisions
 - prevention, monitoring, treatments
 - impacts life decisions

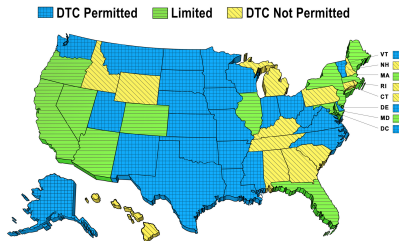


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Now genetic testing is available through direct-to-consumer at-home tests kits

- over the counter at pharmacy (or internet)
- tests may be unproven, invalid or incomplete so results may be misleading
- often results provided without adequate explanation of implications

State DTC Testing Statutes and Regulations



Source: Genetics and Public Policy Center, <http://www.dnapolicy.org/resources/DTCStateLawChart.pdf>

In 2008, FDA regulates diagnostic but not other genetic testing.
http://www.dnapolicy.org/policy/issue.php?action=detail&issuebrief_id=32

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The availability of genetic testing is associated with a risk of genetic discrimination.

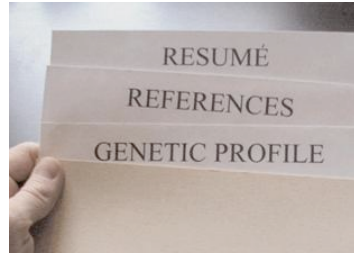
- Genetic predisposition to an inherited disorder brings higher risks
- Insurance companies try to minimize risks
 - pre-existing conditions
 - likelihood of disability or death



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There are laws against genetic discrimination, but this is a rapidly developing area of concern.

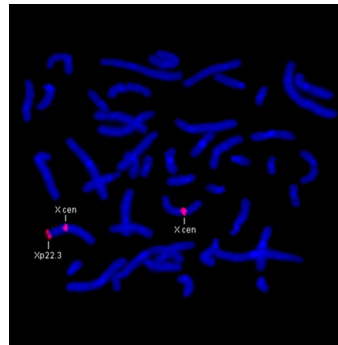
- 2008 Genetic Information Nondiscrimination Act (GINA)
 - prohibits the use of genetic information for discrimination by health insurers and employers.
 - does not cover life insurance, disability insurance, or long-term care insurance.



<http://www.councilforresponsiblegenetics.org/genewatch/GeneWatchPage.aspx?pagelid=189>

Molecular cytogenetics is a modern version of karyotyping.

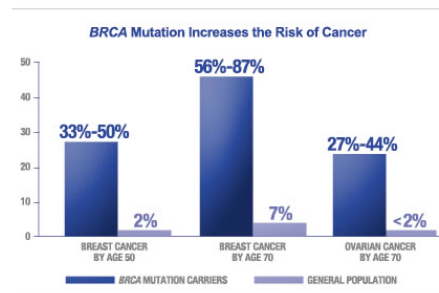
- Fluorescent In Situ Hybridization (FISH)
- locate mutations on chromosomes
 - find specific alleles



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BRCA1 is a gene for a tumor suppressor.

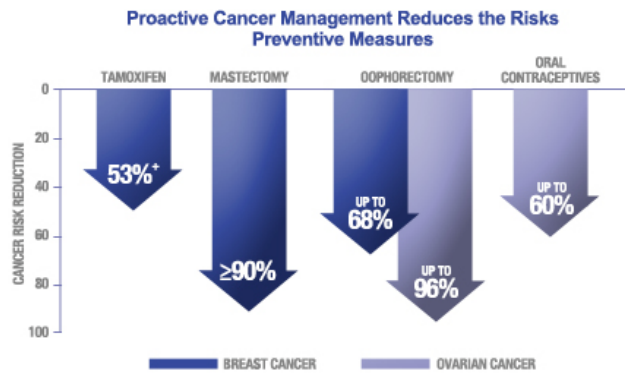
- rare mutations in the gene are associated with a significantly higher risk of
 - breast cancer
 - ovarian cancer



<http://www.bracnow.com/considering-testing/hboc-basics.php>

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A patient aware of the presence of BRCA mutation can take steps to minimize their risk of developing cancer.



<http://www.bracnow.com/considering-testing/hboc-basics.php>

The ACLU and others have filed suit to end the patenting of genes.

- about 20% of Human genes are patented
- Gene patent
 - patent on specific isolated gene sequence
 - lasts 20 years from filing date
 - promotes investment of resources for development of diagnostic and therapeutic tests
 - rewards discoverer



In March 2012, the U.S. Supreme Court emphasized that in order to be patentable a gene or product must not occur in nature.

- Myriad genetics
 - molecular diagnostic company
 - holds an exclusive patent on the BRCA gene
- issue licenses for
 - research
 - controls results
 - testing
 - about \$3,500





Assignment

Gene Patenting?

Review the article Genetics and Patenting (2010) Humane Genome Project Information at

http://www.ornl.gov/sci/techresources/Human_Genome/elsi/patents.shtml

In your discussion group:

Consider is it ethical to patent genes in the human genome?

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Readings

Genetic Testing: Benefits, cost, risks, and limitations of genetic testing (2012). U.S. National Library of Medicine, National Institutes of Health. <http://ghr.nlm.nih.gov/handbook/testing>

Wadman (2010). Breast cancer gene patents judged invalid. Nature. <http://www.nature.com/news/2010/100330/full/news.2010.160.html>

Devi (2012) BRCA patent dispute may head to US Supreme Court. Lancet 379(9813):300. [http://www.thelancet.com/journals/lancet/article/PIIS0140-6736\(12\)60127-5/fulltext](http://www.thelancet.com/journals/lancet/article/PIIS0140-6736(12)60127-5/fulltext)



Reading on the Rocks Grand Manan
John George Brown, 1877

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Any Questions?

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<http://www.vipitbullkennels.com/images/animated-question-mark.gif>

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