

# Genetic Testing

## Introduction

Genetic testing has been an advancing area of research over the past few years that has allowed the medical field greater understanding about particular diseases. It has also provided patients with an ability to find out if they are a carrier of a genetic disease, screen embryos for disease, perform prenatal and newborn testing, confirm diagnoses of certain diseases, take preventive action against later developing genetic disease, and determine pharmacological treatments that are individual specific.

I chose to do my report on genetic testing because it is a subject I am familiar with, but do not know a lot about. Currently, I work with a number of young adults with special needs and various genetic abnormalities, so I was curious about some of those disorders. I was also interested because my friend is about to have a baby, and she had gone through some of the standard genetic testing earlier in the year. In conjunction with genetic abnormalities at birth, I am interested in the way genetics influence disease later in life. I took this as an opportunity to educate myself more on the subject.

## Section 1: Background and Problem Statement

**Web site #1:** Medline Plus: A service of the U.S. National Library of Medicine and the National Institutes of Health. (Score: 28)

**Web address:** <http://www.nlm.nih.gov/medlineplus/genetictesting.html>

- **Information:**

This Web site provides an overview of what genetic testing is used for, and provides a plethora of reliable and informative links regarding the specifics of genetic testing. The site was easy to follow, and is a great starting point to find general and specific information on genetic testing. The site includes many links to resources, references, and current research in the field. I have used this site instead of Healthy People 2010 because there was no information on that site regarding genetic testing, and this is a good supplemental government Web site.

**Web site #2:** National Human Genome Research Institute (Score: 28)

**Web address:** <http://www.genome.gov/19516567>

- **Information:**

This Web site gives basic background information to the reader, while providing a deeper look at what genetic testing is used for and what types of tests are currently available. This site also gives information on 'consumer genetics.' Consumer genetics are tests individuals can purchase online to find out about familial genetic diseases, nutrigenetics (what type of diet they should be on), ancestry, and/or general genome scanning. This Web site also provides a number of other helpful links.

**Web site #3: Kids Health: Genetic Testing (Score: 27)**

**Web address:** <http://kidshealth.org/parent/system/medical/genetics.html>

- **Information:**

This site is an informational page for parents and parents to be. It covers the basics about genetic testing during pregnancy and the types of tests one can expect when having prenatal genetic testing done. The site also provides information about why a doctor may suggest genetic testing for an expecting mother and/or her child. The Web site issues a word of caution about genetic testing which is useful for parents to understand before going into it. The only problem with the site is that it has not been updated in two years, and there may be new, pertinent information available.

**Web site #4: Genetic Home Reference (30)**

**Web address:** <http://ghr.nlm.nih.gov/>

- **Information:**

This Web site is sponsored by the National Institutes of Health and US National Library of Medicine. It provides information regarding genetic conditions, testing, and resources. It includes a section called 'What's New?', which provides very up to date information on information and research pertaining to certain genetic diseases. The site also includes information about each chromosome and genes related to genetic disorders. This Web site is easy to navigate and provided a glossary and handbook, which would be useful for someone who is new to learning about genetic testing, diseases, and current research.

## **Section 2: Research**

Because genetic testing is a broad topic, I have chosen to review research examining genetic tests used to diagnose certain conditions

and/or identify predispositions for certain conditions. Some conditions that genetic tests are used for are Down's Syndrome, Cystic Fibrosis, breast cancer, and Fragile X syndrome.

**Web site #1:** Prenatal detection of Down's syndrome by rapid aneuploidy testing for chromosomes 13, 18, and 21 by FISH or PCR without a full karyotype: a cytogenetic risk assessment (Score:25)

**Web address:** <http://www.ncbi.nlm.nih.gov/pubmed/16005334> (Abstract only, full text available through Buley)

- **Summary of the research:**

This article examined the effectiveness of current genetic testing methods used to diagnose Down's Syndrome in utero. The authors compare the standard method of karyotyping with newer methods called FISH and PCR. The newer genetic tests are much quicker than karyotyping. This study sought to examine the efficacy of their use in replacement of the lengthier process of karyotyping. In addition to being faster, the FISH and PCR recommended by the UK National Screening Committee tests would only test for trisomies on chromosomes 13, 18, and 21. The authors conclude that for best results karyotyping and PCR or karyotyping and FISH should be used.

**Web site #2:** 'Laboratory standards and guidelines for population-based Cystic Fibrosis carrier screening' from: Journal of the American College of Medical Genetics. (Score:25)

**Web address:**

[http://www.geneticsinmedicine.org/pt/re/gim/pdfhandler.00125817-200103000-](http://www.geneticsinmedicine.org/pt/re/gim/pdfhandler.00125817-200103000-00010.pdf)

[00010.pdf](http://www.geneticsinmedicine.org/pt/re/gim/pdfhandler.00125817-200103000-00010.pdf);jsessionid=JtMLV8yhhsJTF61Z4hJcTLQFTGgbyd2QfR1VCHxFJmQmps1T1MLY!1553038018!181195628!8091!-1

- **Summary of the research:**

This research provided information regarding best practices for Cystic Fibrosis (CF) screening as presented by the American College of Medical Genetics. The authors identify several issues to take into consideration when testing individuals for CF carrier genes. First, certain ethnic groups are more likely to carry this gene than others. The AMCG suggests that those of non-Jewish Caucasians of European descent and Ashkenazi Jews should be tested because of their increased likelihood of carrying the CF gene. However, the authors note that the increased genetic diversity in the US should be considered as well. The AMCG suggest that all couples should be educated about the

disease and given the option for testing. Next, the panel suggests that couples should be screened before conception, and explain the types of mutation panels that should be used to identify CF carrier genes. This article provides a good analysis of the current testing methods used for CF and is generally easy to follow. However, it is an article from 2001, so there may be more up to date info on the subject available today.

**Web site #3:** 'BRCA1/BRCA2, and Prevalence of Other Breast Cancer Susceptibility Genes' from: Journal of Clinical Oncology (Score: 27)

**Web address:** <http://jco.ascopubs.org/cgi/content/abstract/20/11/2701>

- **Summary of the research:**

This article presented current research on the sensitivity of a genetic test called BRCA1/BRCA2 used to detect breast cancer susceptibility genes. Specifically, the test looks for genetic markers called BRCA1 and BRCA2. The authors found that the BRCA1/BRCA2 was a reliable test and an 'accurate counseling tool' for determining genetic susceptibility to breast cancer. The abstract summary of this journal article was easy to read and understand. The Web site itself is easy to use and provides a list of links to further research, which this article has been cited in.

**Web site #4:** 'An improved Diagnostic PCR Assay for identification of Cryptic Heterozygosity for CGG Triplet Repeat Alleles in the Fragile X Gene (FMR1)' from: PubMed. (Score:26)

**Web address:**

[http://www.ncbi.nlm.nih.gov/pubmed/18471319?ordinalpos=21&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed\\_ResultsPanel.Pubmed\\_DefaultReportPanel.Pubmed\\_RVDocSum](http://www.ncbi.nlm.nih.gov/pubmed/18471319?ordinalpos=21&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum)

- **Summary of the research:**

This article examined the genetic tests used to screen for Fragile X syndrome. Because Fragile X is a X-linked chromosomal disorder, a certain test (PCR) was not as good at diagnosing the disorder in females. This problem with the PCR test often leads to a follow up test called the Southern blot. The Southern blot is more expensive and time consuming than the PCR test. The authors describe a new and effective type of the PCR test which is more effective at identifying females with Fragile X, and leads to less of a need for the expensive follow up of Southern blot tests.

## Section 3: Statistics

**Web site #1: Bio Medicine: Genetic mutation Linked to more aggressive breast cancer found more often in African American women. (Score: 23)**

**Web address: <http://news.bio-medicine.org/biology-news-2/Genetic-mutation-linked-to-more-aggressive-breast-cancer-found-more-often-in-African-Americans-593-1/>**

- **Summary of the statistics:**

This article summarized the findings from a research project completed at the Yale School of Medicine. It was found that African American women were four times more likely than white women to show differences in a tumor suppressor gene that make them less able to inhibit breast cancer growth. This statistic was easy to understand, as it had already been summarized by the author from the Bio Medicine Web site. The Web site itself is very poorly designed.

**Web site #2: Wellness Consumer Health Information (Score: 24)**

**Web address: <http://www.netwellness.org/healthtopics/idbd/2.cfm>**

- **Summary of the statistics:**

This Web site provided statistics about the prevalence of genetic disorders in the world. The Web site information is based on a review of a reference book about genetics in medicine and was last reviewed by a professor of genetics in 2008. The site reports that 3-4% of the 4 million babies born each year will have a genetic disease or birth defect. About 1% of all babies are born with a chromosomal abnormality, and more than 20% of infant deaths are caused by birth defects or genetic abnormalities. In conjunction with the rates of infant related problems with genetic disease and abnormality, about 10% of adults and 30% of children are in hospital because of a genetically related disorder. This Web site was short, to the point, and easy to read. Unfortunately, there was not a terrible amount of information on the site besides the above stats and a brief description of genetics disorders.

**Web site #3: Genetic Home Reference (Score: 30)**

**Web address: <http://ghr.nlm.nih.gov/handbook/inheritance/riskassessment>**

- **Summary of the statistics:**

I used this site in my background information section, and found a great page about statistics and genetics in the 'Handbook' section. In short, this page explains the terminology used to explained statistical inheritance rates. For example, someone with an autosomal dominant inheritance gene has a 50% chance of passing it along the mutated gene to a child. A person with an autosomal recessive gene has a 25% chance of passing that gene along to a child. The site goes on to explain how genes are transferred when they are X-linked, co-dominant, or mitochondrial. This site is an excellent resource for those who are carriers of a genetic disorder and need to understand the statistics of how their genes may be passed down. It is easy to read and make a lot of the more complicated genetic statistical information easier to understand.

**Web site #4: University of Kansas Hospital: Prevalence of Genetic Conditions / Birth Defects (Score: 25)**

**Web address: <http://www.kumc.edu/gec/prof/prevalnc.html>**

- **Summary of the statistics:**

This site used a variety of references to list widespread data about the prevalence of certain genetic disorders and their affects on infant mortality rates, hospital admissions, and considerations for managed care. It has statistics similar to the Wellness Consumer Health page, but also included a statistic about mental retardation that I found interesting: 50% of mental retardation is due to genetics. I would have thought that number would have been higher. The site also reports that 15% of cancers have an inherited susceptibility, and 10% of all chronic disease has a significant genetic component. The Web site stats are easy to understand, but seem outdated. The information is mostly based on statistics from the early 90s. Perhaps not much has changed since then, but certainly one can assume that the advances in genetic technology would have made a difference in the availability of new genetic information.

## **Section 4: Consumer Information**

**Web site #1: Access Excellence: Understanding Gene Testing (Score: 30)**

**Web address: <http://www.accessexcellence.org/AE/AEPC/NIH/>**

- **Summary of the information:**

From the homepage, this Web site provides a number of links explaining the basics about genetic testing. The consumer who knows little about genetic testing can easily find out the basics about genetic testing, genetic disorders, and what one's options are when considering genetic testing for one's health needs.

**Web site #2: Human Genome Project (Score: 30)**

**Web address:**

[http://www.ornl.gov/sci/techresources/Human\\_Genome/medicine/genetest.shtml](http://www.ornl.gov/sci/techresources/Human_Genome/medicine/genetest.shtml)

- **Summary of the information:**

This Web site is easy to understand for a person who knows little or nothing about genetic testing. It outlines why genetic testing is done, the pros and cons of genetic testing, and provides a list of some of the disorders for which genetic tests are currently available. The site also provided information about regulation and insurance coverage. The Human Genome Project site provides a number of links to resources covering a wide variety of topics dealing with genetic testing.

**Web site #3: Genetics Home Reference (Score: 30)**

**Web address:** <http://ghr.nlm.nih.gov/>

- **Summary of the information:**

I used this Web site in the background information sections as well. It is a very thorough and easy to navigate Web site. I think anyone who needs a good introduction to this topic would find this Web site very useful. It also provides links to many up to date resources.

**Web site #4: National Cancer Institute (Score: 28)**

**Web address:**

<http://www.cancer.gov/cancertopics/understandingcancer/genetesting>

- **Summary of the information:**

This is a good site for people who are interested in learning about genetic testing and cancer. It gives very simple, easy to understand slide presentations on each of the bulleted topics. It provides a lot of information regarding cancer and genetic testing, but has not been updated since 2006.

## Conclusions

### Section 1: Background and Statement

#### The best Web site: Genetic Home Reference

- This Web site was thorough, easy to understand, and had a plethora of references and links to further resources. It was also up to date and recognized as a site for reliable health information by the government. It provides a handbook and glossary for basic information, but also explains each chromosome and gene as it relates to genetic disorders. All around, this Web site was the best site for providing all the info one needs in one place.

#### The worst Web site: Kids Health

- This Web site only provided a limited amount of information about genetic disorders and genetic testing. Obviously, it is geared towards parents, and it is a good resource for that population, it just does not provide information about genetic testing in a broad sense. However, in of itself, it is a good Web site.

### Section 2: Research

- New information I learned include: I did not realize how many varieties of genetic tests were actually used, and I had no idea that certain tests were specific to detecting certain disorders (e.g. Fragile X syndrome). I was surprised to learn that disorders could actually be missed in a genetic test. I had assumed that our modern technology had totally figured genetic testing out already.

### Section 3: Statistics

- New statistics I learned include: One very interesting statistic that I learned was that African American women are genetically less able to suppress breast cancer tumor growth. Additionally, it was interesting to learn the percentages of children (30%) and adults (10%) that are in hospital are due to genetic diseases.

### Section 4: Consumer Information

- The best Web site: Genetic Home Reference  
This Web site is really one of the best, most organized, and complete resources I found. It is easy to understand, but

provides links to more in depth information. It is also up to date and supported by the National Institutes of Health.

- The worst Web site: National Cancer Institute  
I wouldn't say that this was a bad Web site in general, but it was specific to cancer. For a consumer looking for information about genetic testing and cancer, it is a great resource. However, the Web site simply doesn't cover the general information and overview of genetic testing like some of the other sites in the 'consumer' section.

<b>Back to Betty C. Jung's Web site</b>	<a href="http://www.bettyjung.net/">http://www.bettyjung.net/</a>
<b>Back to Web site Critique Reports Directory</b>	<a href="http://www.bettyjung.net/Pch201wsreports.htm">http://www.bettyjung.net/Pch201wsreports.htm</a>

# Presentation Outline Template

## ○ Background/Problem Statement

- Why is your topic a Public Health Issue?

Genetic disease affects so many people's health, and new technology in genetic testing offers an opportunity to identify, treat, and prevent genetic disease in our population.

- Definition: Genetic Testing = A test that analyzes DNA to look for a genetic alterations that may indicate an increased risk for developing a specific disease or disorder

- Types of Genetic Testing

- Carrier screening: Cystic Fibrosis
- Pre-implantation diagnosis: Embryo screening
- Prenatal diagnosis: Down's Syndrome
- Newborn screening: PKU
- Pre-symptomatic prediction of adult onset disorders: Huntington's Disease
- Pre-symptomatic prediction for risk of developing adult onset diseases: Alzheimer's, certain cancers
- Confirm diagnosis
- Forensics/Identity testing
- Treatment plans: Pharmacological treatments that are individual specific

## ○ Research

- Most interesting finding: In general, it was interesting to learn that there are several genetic tests that can be used in diagnosing disorders, and that often several tests should be used together for the most accurate diagnosis.

- Why is it interesting?

I had just assumed that at this point in technology that all genetic tests would be fail proof, but in fact, there is a good deal of research still assessing the efficacy of testing methods with disorders such as Down's Syndrome.

## ○ Statistics

- Most interesting finding: More than 20% of infant deaths are caused by genetic abnormalities, and 30% of all children are in the hospital because of a genetic abnormality. Also interesting, 10% of adults are in hospital because of a genetic disease.

- Why is it interesting?

These stats illustrate how much of the population will be affected by genetic disease at some point in either their own lives, or the lives of their children.

- **Consumer Information**
  - **Most Interesting information: The Genetic Home Reference Web site provides information about specific chromosomes and genes. For instance, abnormalities on chromosome 9 may lead to bladder cancer.**
  - **Why is it interesting?**

**This information is extremely interesting because one can go through each chromosome and see the health issues that have been associated with that chromosome thus far. The site also gives current information about gene specific abnormalities. One could really learn a lot about a particular disorder, or any number of disorders, from this Web site.**