

## LAUNCHING PAD &gt; HUMAN GENETIC CONDITIONS

In beginning a research project on human genetic conditions, it is a good idea to proceed in a series of steps. First, you may wish to begin with reliable noncommercial websites designed for the public. This should help you to learn some of the vocabulary and background that you can use later, when you conduct more focused searches.

Second, you can use general online search engines, such as Google. This strategy often returns resources that are very helpful and that would otherwise have been difficult to find. However, please keep in mind that some of the resources you retrieve may be outdated or inaccurate. As you become familiar with your subject, you will develop a keen eye for evaluating the information that you find.

Third, graduate to the vast world of the genetics literature. Original journal articles are at the cutting edge of science, so this is where you can get an up-to-the-minute picture of research as it advances. Don't worry if original research papers are difficult to read at first; you can start out with review articles, which are usually written for a less specialized audience. The language in the research papers will gradually become more understandable, and soon you will be comfortable in this new environment.

**Step One: Using Reliable Public Websites****The U.S. Surgeon General's Family History Initiative**

This site presents well-designed print and computer-based family history tools to assist individuals and families with constructing their family medical history.

<http://www.hhs.gov/familyhistory/>

**The National Alliance of Genetic Support Groups**

This nonprofit advocacy organization offers a nationwide network of referrals to support groups for patients and families with genetic conditions. This comprehensive web site is a valuable resource for those who are struggling to come to terms with a diagnosis or for those who need information and support from others with their condition. In addition, they work to protect patients and families from discrimination or loss of insurability.

<http://www.geneticalliance.org>

**GeneTests**

This site was originally developed as a voluntary and comprehensive listing of diseases and testing laboratories (both clinical and research tests are included) so that health professional could contact the laboratories that offer genetic tests. Now the site includes many well-written summaries of medical information about genetic conditions (GeneClinics and GeneReviews).

<http://www.genetests.org>

**Facing Our Risk of Cancer Empowered (FORCE)**

The nonprofit organization is dedicated to providing information and networking for those at risk for hereditary breast and ovarian cancer. In addition to educational resources, they offer online communication tools to enhance formation of communities while maintaining individual privacy.

<http://www.facingourrisk.org>

#### U. S. Department of Energy Human Genome Program

This extensive web site presents easily understood educational information on the Human Genome Program, genetics, technology, ethical, legal, and social implications, and medical issues. Several continuing medical education options are listed. The site has resources and links that are appropriate for students, providers, and the public.

<http://www.ornl.gov/hgmis>

#### The University of Kansas Medical Center Genetics Education Center

This web site offers comprehensive links to a wide variety of educational resources about genetics and human genetic conditions.

<http://www.kumc.edu/gec>

#### National Human Genome Research Institute (The National Institutes of Health)

This site is funded by the National Institutes of Health, and presents wide-ranging materials on the Human Genome Project that are suitable for students, healthcare providers, and the public.

<http://www.genome.gov/>

#### The National Cancer Institute (National Institutes of Health)

These tutorials present the basic concepts of how cancer occurs, how cancer risk can be inherited, and issues relevant to genetic testing. The intended audience is the general public.

<http://newscenter.cancer.gov/sciencebehind/>

#### National Newborn Screening & Genetics Resource Center

Though newborn screening is emphasized, there are links to listings posted by regional genetics organizations, and a number of educational resources are listed on this site.

<http://GENES-R-Us.uthscsa.edu/resources.htm>

#### National Coalition for Health Professional Education in Genetics (NCHPEG)

This site provides access to a wide variety of genetics resources for professionals. Because members of professional groups are often just beginning to learn about genetics, it may be useful to check out the various educational programs.

<http://www.nchpeg.org>

### **Step 2: Using Search Engines**

While you were investigating public websites, you expanded your genetics vocabulary. Now is your chance to use that vocabulary to retrieve information using search engines. Try out many different terms and observe the links that are returned after each search. Think of the links as data that have been generated by an experiment, and if one approach seems not to be fruitful, try another. This is exploratory research, and you may be surprised at what you find, from online courses to slide presentations!

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### Step 3: Using Research Databases

#### PubMed

The National Library of Medicine and the National Institutes of Health offer a comprehensive resource called PubMed that indexes the academic literature of virtually all health-related and biological journals. This resource is so vast that it includes over 17 million citations.

PubMed offers a useful tutorial on the left-hand menu because it can be challenging to design a selective search to retrieve the information that you are looking for without also recovering thousands of other papers. Two tips to keep in mind are to limit the search to languages that you understand and to begin with articles that are classified as Reviews. This strategy should give you sufficient background to refine your search terms as you begin to include original scientific research papers in your readings. PubMed searches can retrieve abstracts, but full text articles are sometimes only available online if you are affiliated with an educational institution that subscribes to the journal. If you lack full text access to a particular journal article, check with a reference librarian at your school or public library and see if you can obtain a copy through interlibrary loan.

<http://www.ncbi.nlm.nih.gov/sites/entrez/>

#### Online Mendelian Inheritance in Man (OMIM)

This is a comprehensive resource designed for medical professionals and researchers. OMIM lists all known human genetic conditions, including history, research summaries, clinical synopsis, and references. Entries are updated regularly. This is a great resource, but very detailed and technical.

<http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>

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