NLM E-Minute:
Genetics for Fun and Research – Patient and Professional Databases
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In this rapidly changing world, where discoveries are being made on an almost daily basis, it can be hard to keep up with what is new and what is suddenly old. Nowhere is this truer than in the evolving field of genetics. Here are a few helpful resources designed to point you, your patrons, researchers, and family members in the right direction.

Genetics Home Reference (GHR)

Launched in 2003, this database offers consumer-friendly information about the genetic variations on human health and is an excellent place for students and family members to start their research. Focused for the non-geneticist (and written at the 8-12 grade level), the GHR includes the genetics effects of over 900 health conditions, diseases, and syndromes and more than 1,100 genes, health effects of genetic differences, and gene families.

Access to other databases for further information is provided, including links to: MedlinePlus, Genetic and Rare Diseases Information Center, the research studies in ClinicalTrials.gov, PubMed, the OMIM Genetic disorder catalog, Gene Reviews, and the Genetic Testing Registry. Educational resources, patient support options, tools for researchers, and a handbook entitled Help Me Understand Genetics, are also available.

Additional Consumer Resources

- Genetic and Rare Diseases Information Center (GARD)
  http://rarediseases.info.nih.gov/GARD/

The goal of the Genetic and Rare Diseases Information Center (GARD) is to help people find useful information about genetic conditions and rare diseases. Search for a specific disease or browse by the first letter or number in the name or by a disease category. Started in 2002, GARD is the collaborative effort of the National Institutes of Health (NIH), The Office of Rare Diseases Research (ORDR) and the National Human Genome Research Institute (NHGRI).

- MedlinePlus (NLM)
  http://medlineplus.gov/

Launched in 1997, this consumer database provides links to authoritative genetic information. Many diseases will have a specific health topic page. Others will link directly to the Medical Encyclopedia, to the GHR, to NIH Institutes with resources focused on that disease, and/or to patient handouts on that topic.
Professional Resources


OMIM is a comprehensive, authoritative compendium of human genes and genetic disorders, with links to literature references, sequence records, maps, a clinical synopsis, and related databases such as: PubMed, ClinicalTrials.gov, and Gene Reviews. It is a continuation of the book, *Mendelian Inheritance in Man*, and is currently bio-curated at the McKusick-Nathans Institute of Genetic Medicine, The Johns Hopkins University School of Medicine. OMIM can also be assessed from the NCBI drop-down menu found in PubMed.


This new database (still under development) is the National Center for Biotechnology Information (NCBI)'s curated portal to Medical Genetics information. Sources such as GeneReviews, OMIM, and the Genetic Testing Registry (GTR) are integrated into concepts and assigned a unique identifier. Information includes clinical features of a disorder and related genes, medical and research literature consumer resources, and ontologies. MedGen is linked from the NCBI drop-down menu.


On the NCBI Bookshelf and linked from PubMed, GeneReviews are expert-authored, peer-reviewed disease descriptions presented in a standardized format. Updated regularly, these reviews are focused on clinically relevant and medically actionable information on the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.

Educational Opportunities

The NN/LM offers courses which include free online materials and presentations, such as:

- A Librarian's Guide to NCBI
- ABCs of DNA: Unraveling the Mystery of Genetics Information for Consumers
- Chemicals, Drugs and Genetics Oh My!: Searching PubMed and Beyond


Another new addition to the NLM pantheon is, this portal is geared for high school science teachers. Topics include cell biology, heredity and genetic conditions, DNA and chromosomes, epigenetics and the environment, and evolution. Labs, experiments, latest news and issues, teacher resources and career options are included.

Enjoy! And please feel free to contact me if you would like more information on these or on any other NLM databases and resources.

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