The Genetics Home Reference: A New NLM Consumer Health Resource

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Abstract:
The Genetics Home Reference (GHR) is a new information resource developed to be part of the National Library of Medicine's (NLM's) consumer health initiatives. The GHR's guiding principle is to make the health implications of the Human Genome Project accessible to the public. The GHR accomplishes this by providing a bridge between the NLM's consumer health systems MEDLINEplus and ClinicalTrials.gov on the one hand and the multiple resources emanating from the Human Genome Project on the other. The initial focus is on single gene conditions that are main topics in MEDLINEplus.

Description:
As knowledge of genetics expands, the interrelationships between genes and diseases or genetic conditions will continue to unfold. The research results made possible by the Human Genome Project[1] are available in scientific databases on the Internet, but, because of the often highly technical nature of these databases, they are not readily accessible to the lay public. And yet the public is actively searching online for genetic health information [2]. We are in the process of developing an integrated Web-based information system designed for consumers and others to learn about specific genetic conditions and the genes that are related to them. Our goal is to provide a bridge between the clinical questions of the public and the richness of the data emanating from the Human Genome Project.

The Genetics Home Reference provides basic information on the nature of genes and how they give rise to various diseases or conditions. For each condition, the site provides answers to these specific questions:

- What is this condition?
- What genetic subtypes of this condition exist?
- How common is this condition?
- What are the symptoms of this condition?
- What are the genetic causes of this condition?
- How do people inherit this condition?
- How do doctors diagnose this condition?
- What treatments are available for this condition?
- What other names do people use for this condition?

For each gene, the GHR system provides the answers to these specific questions:

- How does the product of this gene normally function?
- Are there different forms of the protein?
- What other genes interact with this gene or its protein?
- What conditions do mutations in the gene cause?
- Is gene therapy available for these conditionss?
- Where is the gene located?
- What other names do people use for the gene or gene product?

Each description includes a glossary definition for the terms used in that description. In addition, each condition or gene description links directly to pertinent information available on a variety of other resources, including MEDLINEplus, PubMed, ClinicalTrials.gov, Gene Tests, Gene Reviews, LocusLink, and Online Mendelian Inheritance in Man. As a further guide to users of the site, we have developed a section called, "Help me understand genetics", which explains, together with diagrams and links to other web resources, some basic concepts in genetics. Extensive use of the UMLS allows the system to link the information to definitions, MeSH, and the other NLM systems. Use of the Gene Ontology[3] allows for browsing of gene function, location and involvement in biological processes.

The Genetics Home Reference initially concentrates on genetic conditions that are already topics on MEDLINEplus, the NLM's consumer health site.

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