Title: Linking Diseases and Genes through Informatics Knowledge Bases and Ontologies

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Abstract: The National Library of Medicine is working on methods to connect clinical questions to the information from the Human Genome Project. The core data related to diseases, gene products, gene function, genetic testing and gene therapy would be used to connect clinical questions with specific data of the Human Genome Project and the NLM databases. Major considerations of the project are to

(1) Identify the difficulties that arise when traversing multiple information resources.
(2) Examine the coverage of the Unified Medical Language System (UMLS) related to the Gene Ontology.
(3) Consider methods by which the ontologies and information resources could be used more effectively to access the information resources.
(4) Examine the relationships and potential for integration of the ontologies and knowledge bases.
(5) Present the phenotype-genotype information in understandable representations.

The information resources included in the project include those from the National Library of Medicine [MeSH, LocusLink, OMIM, UMLS, MEDLINEplus, Genes and Disease, ClinicalTrials.gov] and other genomic resources and sites [Gene Ontology, KEGG, GeneCards, GDB, HGNC, Proteome, Swiss-Prot, HGMD, GeneClinics/GeneTests].

The initial evaluation of concept representation and difficulties between the GO and the UMLS and an in-depth analysis of selected gene products will be presented.