Databases in 2005 of Human Genes and Inherited Disorders

The vision community is fortunate to have several databases of human genes and inherited disorders. In most instances, the database user is prompted to insert a cue or cues (e.g., chromosome and symbol, sequence accession number, partial name and multiple species, gene name, Gene Ontology (GO) terms or identifiers, publication citation, chromosome and species, or Enzyme Commission (EC) numbers) to elicit a rich cache of data. Among the databases are Mendelian Inheritance in Man (OMIM), RetNet, POSSUM, and London Medical Databases (LMD).

OMIM is the most widely used and robust of the databases. It marries scientific literature, textual information, DNA and protein sequence databases, 3D protein structure and protein domain data, population study datasets, expression data, assemblies of complete genomes, and taxonomic information in an integrated system. OMIM was authored by Dr. Victor McKusick of Johns Hopkins and colleagues, and developed as an electronic product by the National Center for Biotechnology Information (NCBI) of the US National Library of Medicine. OMIM contains links to contents of NCBI's Entrez Gene cross-database search engine system and to numerous related resources elsewhere. Entrez Gene provides tracked, unique identifiers for genes and information associated with those identifiers.

RetNet is a service of the Laboratory for the Molecular Diagnosis of Inherited Eye Diseases, which is a joint program of the Hermann Eye Center and the Human Genetics Center of the University of Texas-Houston Health Science Center. RetNet contains tables of genes and loci causing inherited retinal diseases. It, too, has links to other resources including NCBI's Entrez Gene.

The database POSSUM contains x-rays, diagrams, histopathology slides, video clips, and detailed list of traits for aiding a clinician in diagnosing syndromes, including many where the eye is involved. It is managed by The Murdoch Childrens Research Institute at the Royal Children's Hospital in Melbourne, Australia. POSSUM is available for a fee. The website contains useful links to various foundations that raise awareness and funds to fight specific syndromes. Subscribers can follow a link within POSSUM to obtain additional information from OMIM.

Another database is the London Medical Database (LMD). It contains the Winter-Baraitser Dysmorphology Database (WBDD) with information on nearly 4000 multiple congenital anomaly and mental retardation syndromes and also the Baraitser-Winter Neurogenetics Database (BWND) with information on syndromes involving the central and peripheral nervous system. Both databases contain information on single gene disorders, sporadic conditions, and conditions caused by environmental factors. They also contain a photo library with a collection of many thousands of photographs and CT and MRI images and examples of characteristic electrophysiological recordings. Most recently, a database called GENEEYE was added to LMD. It contains similar data on over 2500 genetic eye disorders, for helping clinicians make accurate diagnoses. LMD is fee-based.