REVIEWS OF WEB SITES

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Online Mendelian Inheritance in Man
URL: http://www3.ncbi.nlm.nih.gov/Omim/
Sponsor: Johns Hopkins University, Baltimore, Maryland
Author: Dr. Victor A. McKusick and colleagues
Web Version: National Center for Biotechnology Information

Readers of this column may be interested in *Online Mendelian Inheritance in Man* (*OMIM*) for two reasons: Firstly, some patients have genetic conditions that must be considered when planning anesthesia. Secondly, it is an excellent demonstration of the use of the Internet to make a vast amount of medical information widely available in a format that is more convenient to use and more up-to-date than would be possible in any other way.

OMIM is a database with 9,145 entries, each reviewing a specific genetic disease, which is continually being updated at the rate of 60 new entries and 500 revisions per month by Dr. Victor A. McKusick and his colleagues at Johns Hopkins University and elsewhere. It has been made freely available on the Web by the National Center for Biotechnology Information. The site begins with a warning: "*OMIM* is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. Non-medical users are strongly urged to seek the assistance of an expert in the interpretation of *OMIM* text and images."

Presentation

The site consists of plain text with small link buttons. Animations, Java, frames, plug-ins, and multimedia are not used. Unfortunately, a link to the Cedars-Sinai Medical Center Genetics Image Archive was not available at the time of this review.

Search Capabilities

For anesthesiologists, the most important feature is the search engine. Type in almost any variation of any name for an inherited disease, and, if spelling is correct, a link will be provided to the appropriate entry for that disease. Because many genetic syndromes have various names, this is more convenient than most textbook indices. A facility that offers alternative spelling when a search produces no results would be a useful addition.

Format

Each disease entry is presented as a Web page. A table of contents provides shortcuts to sections that typically include "Clinical Features," "Diagnosis," "Clinical Management," and "References," and several sections that would only interest a geneticist, such as "Population Genetics." A clinical synopsis is sometimes available as a separate Web page.

Links

Each entry is referenced heavily, with links from the text to the relevant PubMed abstract. The full text can be ordered online through "Loansome Doc." There are extensive links to MEDLINE and to other genetics resources. Each page has a link to a form that can be used to add comments or new information.

Comparison with Established Textbooks

I compared *OMIM* to two standard anesthetic textbooks, *Anesthesia and Co-Existing Disease*, 3rd edition, 1993, by Stoelting and Dierdorf, and *Anesthesia and Uncommon Diseases*, 4th edition, 1998, by Benumof. I searched each resource for information about five genetic conditions of interest to anesthetists, which I saw recently in the preadmission clinic.

Malignant Hyperthermia

The anesthetic textbooks both had extensive and detailed accounts of this condition, indexed under both words. *OMIM* has 11 pages about "hyperthermia of anesthesia." This contains a good history of the disease from the first case report in 1962 to recent developments, but no practical information for anesthesiologists. There was no link to clinical resources such as the Malignant Hyperthermia Association of the United States Emergency Hotline.

Pseudocholinesterase Deficiency

The anesthetic texts mentioned pseudocholinesterase only in the context of liver disease. *OMIM* had no relevant information. Perhaps an inherited enzyme deficiency is not really a disease, but it was surprising that none of the resources mentioned this condition.

Acute Intermittent Porphyria

Both anesthetic textbooks had more than a page of information, although *Anesthesia and Uncommon Diseases indexed* acute intermittent porphyria under "H" for "hepatic porphyrias." Both textbooks listed safe and unsafe anesthetic drugs. *OMIM* has 23 pages of text with 82 references. Although the section about clinical features mentions that attacks can be precipitated by barbiturates or sulfonamides, there was not enough information to plan a safe anesthetic.

Freeman-Sheldon Syndrome (Whistling Face-Windmill-Vane Hand Syndrome)

Both anesthetic texts had brief entries that mentioned difficult tracheal intubation and the risk of malignant hyperthermia. *OMIM* produced five pages of information, including 26 references (compared with five between the two textbooks), but none of the references were to anesthesia journals, and there was no mention of difficult tracheal intubation or malignant hyperthermia.

Hereditary Hemorrhagic Telangiectasia (Osler-Rendu-Weber disease).

Anesthesia and Co-Existing Disease had 134 words, half of which was about anesthetic implications, indexed under both names, whereas *Anesthesia and Uncommon Diseases* provided 118 words, indexed only under "Osler-Weber-Rendu." Each had one reference. *OMIM* had 16 pages and 82 references. An extensive history section included fascinating nuggets, including the correct pronunciation ("OHz-ler, ren-DYU, and VAY-ber"), with a note that Weber "pronounced his name in the Germanic manner event though he was born in England."

Summary

OMIM is an invaluable resource for physicians dealing with patients with genetic diseases. It uses the Web to make medical information accessible in a way that should inspire other specialties to develop similar resources. Although it does not contain enough anesthesia-related information to act as a sole source of information about genetic conditions for anesthesiologists, it would be of great benefit to any anesthesiologist writing a consult note, preparing rounds, or publishing a case report about a patient with an inheritable condition.

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