The Internet and genetic disease

The Human Genome Project and the Internet came of age together, so it’s no surprise that information on genetics and genetic disease abounds on the Net. The definitive resource is the Online Mendelian Inheritance in Man (OMIM) database at the National Center for Biotechnology Information (www.ncbi.nlm.nih.gov/omim/), which contains more than 10,000 entries. It may be searched using the database’s own search engine or via a newly added portal, Entrez (www.ncbi.nlm.nih.gov/entrez/). A search for “cystic fibrosis” results in 98 entries, topped by those for the CFTR gene and cystic fibrosis itself.

Entries for individual diseases feature an up-to-date summary of the knowledge that is accumulating under headings such as description, clinical features, molecular genetics, pathogenesis, diagnosis and clinical management. Given the magnitude of the task and the rapid progress within the field, the summaries are a work-in-progress rather than an elegantly written review, but they are comprehensively referenced to original articles and reviews.

Hyperlinks from each citation connect to details in Entrez. For larger entries — and some are very large indeed — a distillation (MINI-MIM) and clinical synopsis are offered. Links from the NCBI main page (www.ncbi.nlm.nih.gov/) lead to general educational material on genetic disease and the Human Genome Project.

Many genetic diseases are rare, and the Office of Rare Diseases at the National Institutes of Health (rarediseases.info.nih.gov/ord) is a clearinghouse for information on them. (In most cases, a rare disease is defined as one that affects fewer than 200,000 people in the United States.) The site is designed to inform patients and physicians of ongoing research, contacts and resources.

Other useful sites are www.geneclinics.com, which provides clinical information relating to testing for and managing inherited disorders, and www.modimes.org, which discusses common disorders. Visitors to www.geneticalliance.org will find a coalition of genetic support groups, while www.genetests.org offers a directory of laboratory tests.

In Canada, the Canadian Association for Rare Disorders (www.cord.ca) was launched by Maureen Gaetz-Faubert of Lethbridge, Alta., after she was diagnosed with a rare connective-tissue disorder. This site sports links to numerous organizations and support groups and the organization itself provides information and electronic networking for families, physicians and researchers.

There are also well-appointed Canadian sites dealing with specific genetic diseases. The Canadian Cystic Fibrosis Foundation (www.ccff.ca) provides information for patients and their caregivers, while the Canadian Hemophilia Society (“We’re all related by blood”) has a comprehensive site (www.hemophilia.ca) dealing with different types of inherited bleeding disorders. — Alison Sinclair, CMAJ

Return-to-work guide now available online

Disability, whether work related or not, is seldom discussed during the training of Canada’s physicians. For that matter, neither are the compensation issues surrounding disability, even though most doctors will have to deal with them at some time. Now a new guide that helps physicians manage patients with workplace injuries and deal with disability and return-to-work issues is available to aid them in overcoming that past neglect.

Injury/Illness and Return to Work/Function: a Practical Guide for Physicians emphasizes the importance of the physician’s role in enabling a patient’s safe return to work. The guide also includes information on income support for people unable to work because of illness or injury. The guide is a result of the Physician Education Project in Workplace Health, an 1994 initiative of the Ontario Medical Association and the Institute for Work and Health.

The design allows it to be modified for use elsewhere and allows for future expansion; among the possibilities under consideration is a return-to-work guide for people with psychiatric illness and a physicians’ guide on insurance issues. MDs can download the guide at www.wsib.on.ca. — Alison Sinclair, CMAJ

New boss at Health Canada

A new player entered Canada’s health care debate in January, when the federal government named career civil servant Ian Green to replace David Dodge as deputy minister at Health Canada. (Dodge left Health Canada to assume responsibility for the health of the Canadian dollar — he was named governor of the Bank of Canada.) Green, 52, began his career by working for Progressive Conservative leader Joe Clark from 1974 to 1980. He then joined the Privy Council Office and rose quickly through the ranks to become an assistant deputy minister in the health department in 1987.

Since then he has held senior posts in several large departments. Green’s CV is not studded with the kind of academic credentials that his predecessor’s boasted, but he is known as an unflappable manager. Green will have to carry through on the work started by Dodge, who conducted a major shakeup at Health Canada that included a total structural reorganization and the naming of a chief scientist. — Charlotte Gray, Ottawa