touching base

The New Genethics

The growing need for intelligent debate on the difficult ethical issues associated with genetic research and the Human Genome Project will be helped by a new site on the Internet. The Gene Letter (http://www.geneletter.org.) has been created by Philip Reilly, Dorothy Wertz and Robin Blatt, of the Shriver Center for Mental Retardation in Waltham, Massachusetts, with support coming in the form of a two-year grant from the US Department of Energy/Ethical, Legal and Social Implications Program. The Gene Letter appears every other month, with regular columns on health, law, society and international developments. The November issue contains interesting articles on DNA evidence in the courtroom, genetic discrimination and insurance, and consumer information on teratogens. In addition, the site contains an unedited discussion forum named GeneTalk, and a useful feature on 'Two months of genetics in the news', highlighting coverage of genetics stories in major US newspapers.

Testing, Testing

On October 30, Myriad Genetics began offering a complete sequence test for the two known hereditary breast and ovarian cancer genes, *BRCA1* and *BRCA2*. For $2,400, the company will completely sequence both genes — a process that takes up to four weeks. If any specific mutations are found, they can be screened in additional family members for $395 per person. The Myriad strategy differs from that already available from OncorMed, which offers three levels of screening, initially focusing on specific mutations and culminating in a complete sequence test for *BRCA1*.

At a press conference announcing the availability of the BRACAnalysis™ screen, Myriad staff seemed at odds on who the test is aimed at. Janet Haskell, president of Myriad Genetic Laboratories, said that the test will be confined largely to women who have a strong family history of the disease. But company co-founder Mark Skolnick believes that the test may prove popular in a broader spectrum of women, not necessarily just those with early-onset disease. Citing new unpublished data based on genetic analysis of hundreds of women, Skolnick said that his team has uncovered a surprisingly high frequency of *BRCA1* mutations in older women, including 30–40% previously undescribed flaws, attesting to the virtues of the complete sequence test. Myriad is setting up a registry of mutations at the Dana Farber Cancer Institute in Boston, and pledges to maintain patient confidentiality.

The virtues of taking any kind of genetic test for breast cancer, let alone one offered commercially, have divided women's advocacy groups, but in some cases it can be a life-saver. In the United Kingdom, advertizing executive Annmarie Blomfield recently learned that she (and her two young daughters) had not inherited a mutation in *BRCA2* that had triggered breast cancer in 15 of her relatives. A year ago, Mrs Blomfield was advised to postpone her planned surgery to have a prophylactic mastectomy, following the identification of *BRCA2* by Michael Stratton and colleagues at the Institute of Cancer Research.

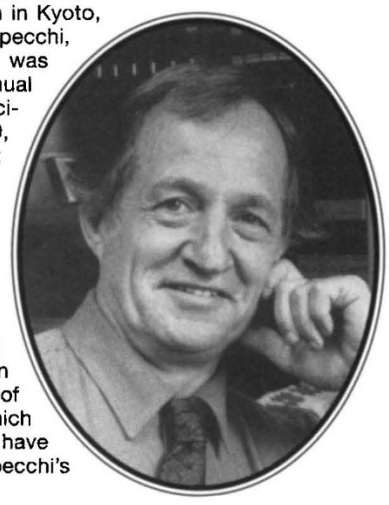
Go Dutch

Among the extensive personnel changes expected to be announced by President Clinton following his re-election last month is the replacement of Hazel O'Leary, the Department of Energy Secretary. O'Leary was a controversial member of Clinton's cabinet for her extensive (and some suggested unwarranted) jet-setting trips around the world at the taxpayers' expense. However, O'Leary was able to celebrate some good news just before the presidential election, as Dutch scientists from Leiden University, in collaboration with researchers from the Lawrence Livermore National Laboratory in California (part of the Department of Energy's Joint Genome Institute), announced that they had found the gene for episodic ataxia type 2 (EA-2) and familial hemiplegic migraine (Ophoff, R.A. *et al. Cell* **87**, 543-552; 1996). The *CACNL1A4* gene on chromosome 19 encodes a brain-specific, voltage-gated calcium channel subunit, and harbours missense mutations in some migraine patients and putative loss-of-function abnormalities in two patients with EA-2. O'Leary congratulated the Dutch team, headed by Rune R. Frants, and (clearly warming to her task) commented: "The Department of Energy's work in mapping genetic blueprints holds enormous promise for the future — improving health, the environment and the economy for Americans and people around the globe."

Must See TV

Y.W. Kan could be forgiven for looking a little jaded as he was interviewed recently by Katie Couric, the popular host of NBC's breakfast television programme, *The Today Show*. Kan was summoned at 3.30 a.m. local time for the satellite link with New York to discuss his group's recent success in performing two non-invasive prenatal diagnoses by enriching fetal cells from maternal blood (Cheung, M.-C. *et al. Nature Genet.* **14**, 264–268; 1996). The previous afternoon during the American Society of Human Genetics conference in San Francisco, Mei-Chi Cheung described the results of a third diagnosis, this time for cystic fibrosis. The mother and father were carrying an N1303K and ΔF508 mutation, respectively. Analysis of fetal erythroblasts isolated from the maternal blood showed that the fetus was heterozygous for the father's mutation, having also inherited the mother's normal *CFTR* allele. As in the previous two cases, the diagnosis was confirmed by chorionic villus sampling. Kan expects other researchers to visit his laboratory to learn the technique first hand.

Knock, Knock



At a ceremony held last month in Kyoto, molecular geneticist Mario Capecchi, from the University of Utah, was awarded one of the 12th annual Kyoto Prizes, in the 'Basic Science' category. Dr Capecchi, 59, receives not only Japan's richest award of ¥50 million ($460,000), but also its most prestigious — the Kyoto Prize is often called the Nobel Prize of Japan. In the citation accompanying the award, Dr Capecchi was praised for his work in creating 'knockout' mice. The testimonial concluded: 'It would not be an overstatement to say that many of the fruitful areas of research which comprise today's life sciences have been made possible by Dr Capecchi's work.'