

Special Report

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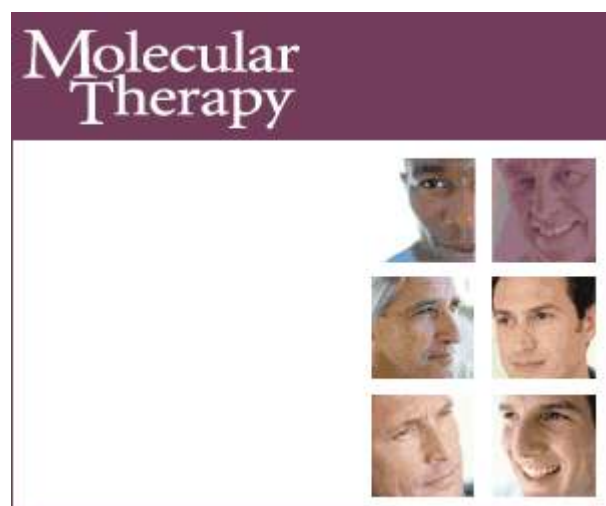
DNA masters

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1. Ricki Lewis is the author of *Human Genetics: Concepts and Applications*.

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As genetic testing becomes more common, the need rises for experts to interpret the results. Ricki Lewis reports.



As the fruits of the Human Genome Project continue to make headlines, a related field has been quietly gestating — genetic counselling. Its practitioners are hybrid professionals, combining expertise in human genetics with the ability to communicate their knowledge to patients and families. "This profession

is very much like a small, quaint community that is well known among some circles, but is on the verge of being discovered by the rest of the world," says Janey Youngblom, associate director for the master's degree program in genetic counselling at California State University, Stanislaus in Turlock.

With the recent expansion of genetic-disease screening programmes in newborns, the improved safety of amniocentesis for prenatal diagnosis and a slew of direct-to-consumer genetic-testing websites, genetic counsellors are more in demand than ever.

The term 'genetic counselling' was coined by geneticist Sheldon Reed in 1947, referring to the advice he provided to physicians whose patients had inherited diseases. The inaugural master's degree programme in genetic counselling — at Sarah Lawrence College in Bronxville, New York — saw its first class graduate in 1971, and this course has served as the model for the current 32 programmes in the United States.



P. SALOUTOS/CORBIS

The field is still small — the National Society of Genetic Counselors (NSGC) in Chicago, Illinois, has just 2,300 members, says Caroline Lieber, director of the graduate programme at Sarah Lawrence. In Europe, the first genetic-counselling programme started in 1992, from the University of Manchester, UK, and 14 other nations are playing catch-up, with the most new efforts in Japan.

At the inaugural meeting of the Transnational Alliance for Genetic Counselling in Manchester in May 2006, educators representing 45 programmes and 15 professional organizations, from 18 nations, met to discuss training. There, the term was consolidated. Jacquie Greenberg, an associate professor in human genetics at the University of Cape Town in South Africa, summarizes it: "Genetic counselling helps people understand how genetic disease affects their lives."

Evolving roles

When the field began, patients weren't genetics-savvy. Today, the Internet has changed that picture dramatically — but experts are needed to explain the science and to take psychosocial factors into account.

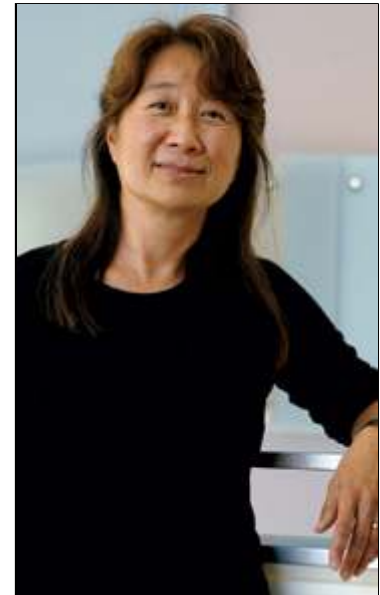
"People often form initial perceptions based on what they read on the web, and it can be difficult to make them see something from a different point of view," says Siobhan Dolan, associate professor of reproductive genetics at the Albert Einstein College of Medicine of Yeshiva University in New York. A nutrigenetics website, for example, might present population-level information on the association between a gene variant and a disease before studies have been done on the validity of extrapolating it to individuals.

Likewise, patients using direct-to-consumer genetic-testing services might confuse high risk with general population risk, notes Dolan. Generally, high risk for, say, the *BRCA* gene and breast cancer, is suspected for patients who have a young first-degree relative with breast cancer.

The counsellor's role transcends the provision of information and correction of misinformation. "We help patients make sense of the options, and provide reassurance and support," says Jennifer Fitzpatrick, director of the genetic-counselling programme at McGill University in Montreal, Canada. For example, if a prenatal screen shows a high risk of Down syndrome, the parents-to-be could very well think about termination. "Our role is to do damage control and put the information in perspective," she says. In this common scenario, the counsellor explains that a positive result from the screening indicates elevated risk, not a diagnosis, and explains further testing options.

The genetic counsellor is an advocate for the family, says Karen Marder, professor of neurology at Columbia University in New York. As a physician, Marder works closely with a genetic counsellor, who navigates families through the labyrinth of choices that accompany testing for Huntington's disease. "I can't imagine a multidisciplinary team without one," she says.

A genetic counsellor is especially helpful in rare diseases with which the physician is not familiar. Helga



C. EDMONDSON

Janey Youngblom: "This profession is well known among some circles, but is on the verge of being discovered by the rest of the world."

Toriello, director of genetics services at Spectrum Health in Grand Rapids, Michigan, gives the example of spinal muscular atrophy. There's no need to sequence the gene to diagnose it, she notes. "Don't test for all the mutations for a disease; do the common ones first."

Perhaps the counsellor's greatest contribution is time. In an hour-plus session or in multiple meetings, a counsellor discusses the effects of mutations and how they are transmitted, sketches a pedigree, talks to relatives about risks, tests and treatments and explores feelings such as fear, anxiety and guilt.



J. COATE/MARCH OF DIMES

Siobhan Dolan: "People often form initial perceptions based on what they read on the web."

And the field is growing fast. "The profession began with paediatric and prenatal care, but now includes cancer, cardiovascular disease, neurology and ophthalmology," says Beverly Yashar, director of the genetic-counselling graduate programme at the University of Michigan in Ann Arbor. A newer turf is public policy. "Policy-makers, like the public, have genetic terms in their lexicon but are not always sure what they mean," says Luba Djurdjinovic, executive director of the non-profit Ferre Institute in Binghamton, New York, which provides community-based genetics services. The institute's annual DNA Day lecture informs the state's legislature.

Training and traits

A genetic counsellor combines technical knowledge with characteristics such as empathy, curiosity, and comfort with new technology and with people who have disabilities, says Youngblom.



M. VLOET

Beverly Yashar: "The profession now includes cancer, cardiovascular disease, neurology and ophthalmology."

The career trajectory begins with an undergraduate degree in a biological or social science. Master's degree programmes in genetic counselling include coursework in all areas of genetics, as well as statistics, psychology, and 50–200 supervised cases. Certification in the United States requires passing an exam given by the American Board of Genetic Counseling in Olathe, Kansas. Licensing — a step beyond certification — is progressing on a state-by-state basis. Genetic counsellors are certified by the Health Professionals Council in South Africa and by the National Health Service in Britain.

Many genetic counsellors start their careers in science or medicine, but desire more time with patients, says Yashar, who earned her PhD in yeast genetics and then her master's in genetic counselling. The preponderance of females in the profession reflects the initial focus on prenatal care, the history of female domination of counselling fields and the flexibility of many jobs, which eases childcare concerns, says Youngblom.

Varied workplaces

Most genetic counsellors work in clinical settings. According to the NSGC's 2006 workplace survey, 38% of its members work in university medical centres, 31% in hospitals, 8% in diagnostic laboratories and 5% in private practices. The rest work at health-maintenance organizations, universities and in biotech, pharma or Internet companies.

Although the day-to-day tasks may be similar in the different settings, the degree of patient involvement varies. At Columbia University, says Marder, a genetic counsellor develops long-term relationships with families. In the corporate sphere, the job may be less personalized and broader. A genetic counsellor for Myriad Genetics in Salt Lake City, for example, interprets the results of hereditary cancer tests, keeps up with the literature, prepares educational material and presents training programmes.

Research is also possible. At Myriad, counsellors help improve breast-cancer tests. In universities, they can work with families enrolled in clinical trials, says Yashar. Opportunities are also opening up in the direct-to-consumer market. For example, people who seek testing for some mutations online can take a tissue sample at home and send it to a lab. The results are then sent to the patient and a genetic counsellor, with whom the patient can discuss the results — imperative for disorders in which not all people who inherit the mutation develop the disease.

The future

As genetic discoveries proliferate, more genetic counsellors will be needed to bridge the gap between what consumers think they know and what scientists have learned.

Prenatal care is at the forefront of the integration of genetic counselling into medical practice. Some obstetric practices, for example, routinely test for cystic fibrosis carriers, with nurses or physicians explaining common findings, says Yvette Conley, assistant professor of health promotion and development at the University of Pittsburgh in Pennsylvania. Patients with rare allele combinations are referred to genetic counsellors. "We will be educating all kinds of health-care professionals," says Lieber.

Djurđjinovic predicts that the marriage of new information and outreach may even create a new type of practitioner. "We can expect to see a new wave of genetic counsellors practising in small communities to support the application of new genetic tests for common conditions," she says. As microarray technologies become more common, Djurđjinovic sees genetics professionals becoming more important to the appropriate application of testing and interpretation of test results. "Maybe a new professional will



M. TABER-LIND

Helga Toriello: "Don't test for all the mutations for a disease; do the common ones first."

emerge: the genomic counsellor," she says.

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