

CAREERS

TRANSITIONS From building houses to building molecules **p.153**

FUTURE PLANS Three steps to prepare for the next five years go.nature.com/bpd1rc

NATUREJOBS For the latest career listings and advice www.naturejobs.com

DRA_SCHWARTZ/GETTY



GENETICS

Fluent in DNA

As genomics migrates to the clinic, job options are emerging for genetic counsellors to explain the meaning in mutations.

BY MICHAEL EISENSTEIN

Jehannine Austin spent years at the laboratory bench developing techniques for detecting genetic mutations, and was well into her PhD programme before dissatisfaction began to set in. “I was working on the genetics of schizophrenia and bipolar disorder, but I didn’t have the communication skills or knowledge to explain how what I was doing was relevant at a personal level,” she says. She wanted to help patients and their families to understand the potential medical implications of genetic changes. And so, after earning her

degree, she enrolled in a master’s programme in genetic counselling at the University of British Columbia in Vancouver, Canada.

Clinicians routinely recommend genetic tests as part of diagnosing disease and predicting disease risk, but many people find it hard to understand exactly what it means when a result indicates greater risk or the potential to pass on diseases to their children. Genetic counsellors help individuals to see how their genomic variation could affect their own health and that of their family members. A counsellor’s role is to explain what testing may or may not reveal, and how to prepare for the

medical and psychological implications of those results. That includes helping people to respond proactively to findings — for example, genetic counsellors often encourage early cancer screening for women who have a *BRCA1* mutation that strongly elevates their risk of breast cancer.

The ranks of genetic counsellors have swelled in recent years. Throughout the United States and Canada, there are more than 3,500 licensed practitioners, according to the American Board of Genetic Counseling (ABGC), the certifying organization for these two countries. And at least another 300 work in the United Kingdom. But there are more jobs than eligible candidates: a 2012 assessment from the US Bureau of Labor Statistics determined that prospects in the field are growing much more rapidly than the average for other occupations.

FROM LAB TO CLINIC

A genetic counsellor’s job straddles the fields of biology and psychology, and the position draws many scientists who are looking beyond the bench. “When people apply to genetic-counselling programmes, their letters typically say, ‘I love genetics, but the lab isn’t for me — I want to be at the people end of things,’” says Austin, now a genetic-counselling faculty member at her alma mater and the incoming president of the US National Society of Genetic Counselors.

Those who wish to pursue the career need a master’s degree in genetic counselling and a certification or licence to practise. Professional societies administer such certification in Australia, North America, Japan, South Africa and the United Kingdom; in mainland Europe, individual programmes handle licensing. Worldwide, graduate programmes emphasize a similar foundation of genetic-literacy and counselling skills paired with hands-on experience in the clinic. Training focuses on the situations that most commonly require genetic counselling, including hereditary cancer risk, prenatal diagnosis of birth defects and early-onset or developmental disorders in children.

The first genetic-counselling programme was established in 1969 at Sarah Lawrence College in Bronxville, New York. For decades, the job centred mainly on understanding patterns of inheritance, and attempting to diagnose genetic maladies on the basis of physiological or biochemical indicators. But the field of clinical genetics shifted dramatically in the 1990s as technological advances began to ►

► enable physicians to interpret the medical consequences of specific sequence variations, such as the link between mutations in two *BRCA* genes and the risk of breast and ovarian cancer. The field advanced further in the 2000s with an explosion in genomics — next-generation sequencing platforms for fast, low-cost genetic analysis. This greatly accelerated the discovery of gene–disease associations, and gave clinical geneticists a much deeper well of knowledge from which to draw.

AT EASE WITH UNCERTAINTY

Today, most genetic counselling is clinical, focusing on helping patients to understand the consequences of mutations either in individual genes or in panels of specific genes that have ties to conditions such as cancer or neurological disease. But a growing number of academic centres, hospitals and private companies are exploring genome-scale sequencing and the extent to which information about thousands of genes can guide clinical diagnosis of inherited diseases and cancer. Some countries are pursuing national-scale medical-genomics programmes, such as the ‘100,000 Genomes’ initiative in the United Kingdom, which formally began recruitment early this year (see go.nature.com/ri9rn5).

That translates into an unprecedented demand for genetic counsellors who can tackle the surge in the number of people who are choosing to have their genomes analysed. Professionals must grapple with the challenge of results from thousands of genes simultaneously, rather than a carefully selected — and well-understood — handful.

The shift to large-scale sequencing does not necessarily rewrite the counsellor’s job description, but it does add complexity. One major issue is uncertainty — genome-scale analysis still routinely fails to definitively identify a causative mutation, and may instead return ‘variants of unknown significance’. These reside in genes that are thought to be clinically important, but they have not been clearly

demonstrated to interfere with gene function.

Such mutations are relatively common, says Kelly Hagman, managing director of clinical genomics at Ambry Genetics, a genetic-testing company in Aliso Viejo, California. She estimates that 10% of Ambry’s ‘exome tests’ (which sequence all the protein-coding genes in a given genome) return these undetermined variants. Although the emotional effects of this uncertainty has been a major source of concern, genetic counsellors generally find their patients to be resilient, mainly because genomic analysis is often just the latest stop on a years-long diagnostic odyssey.

More challenging is the potential for ‘incidental findings’ that might be uncovered in a person’s genome while searching for a specific clinical result, such as assessing cancer risk. To try to pre-empt undesired, disturbing disclosures, patients are now asked before undergoing sequencing whether they would want to receive such results.

Still, the array of results and decisions can make the post-sequencing meeting harder for a counsellor to navigate. “Previously, you knew the condition you were testing for when the family arrived, and you could counsel them on it,” says Sarah Scollon, a genetic counsellor involved with a large-scale clinical exome-sequencing programme at Baylor College of Medicine in Houston, Texas. “Now you’re throwing a wider net and can’t do that kind of specific counselling up front; that now comes more at the back end.”

TO THE LAB AND BEYOND

On the plus side, the exploding interest in clinical genomics has created a wealth of opportunities for genetic counsellors. The boom in exome- and genome-research programmes, and the rapid growth of commercial sequencing providers such as Ambry, have shifted the field to applied research: now, genetic counselling beckons even those who most enjoy the bench. “When I first started around 2004, less than 10% of counsellors worked in labs, and now it’s well over 20%,” says Hagman.

At Geisinger Health System, a large health-care provider in Danville, Pennsylvania, the counsellor workforce has grown by 75% since 2006, according to Janet Williams, the company’s director of research genetic counsellors. And Hagman reports that Ambry employs 70 or 80 genetic counsellors, and is “always hiring”.

Many of these counsellors have little direct contact with patients; instead, they write educational materials for physicians and patients, explain the technology to clients, consult on test selection and publish research. These

counsellors are also working on cutting-edge capabilities, adapting to rapidly evolving genetic knowledge. Across Ambry’s exome tests, roughly one-quarter of results flagged in diagnostic sequencing are mutations that were discovered to be clinically relevant only in the past two years, says Hagman.

Public-education and policy specialists who have genomics expertise are also in demand, says Barbara Biesecker, who directs a joint genetic-counselling programme between Johns Hopkins University (JHU) in Baltimore, Maryland, and the US National Human Genome Research Institute (NHGRI). She notes that the programme helps students to explore careers in policy and industry.

There has also been discussion about creating advanced-degree programmes for genetic counsellors who are interested in further specialization. A task force formed by the ABGC in 2011 found that counsellors want more opportunities for academic growth. “There is a lot of interest in the idea of genetic counsellors gaining higher-level, research-based training,” says Austin. In 2013, the ABGC followed up on this to form the Committee of Advanced Training for Certified Genetic Counselors, aiming to assess the skills and education that counsellors may require for career tracks outside the clinic.

Some worry that the rate of training is failing to keep pace with demand. The 34 accredited genetic-counselling master’s degree programmes in the United States and Canada attract many more applicants than they can accommodate. The prestigious 2.5-year JHU–NHGRI programme, established in 1996, receives 80 to 100 applicants a year, yet accepts only 4 or 5 students. Several more programmes are under development, but funding is tight — multiple programmes have folded in past years because of budgetary constraints.

The United Kingdom offers only two training programmes, and in most other countries, the field is just now emerging as a career option. “They’ve either had no counsellors or they’ve had medics or nurses doing genetic counselling,” says Anna Middleton, a genetic counsellor at the Wellcome Trust Sanger Institute in Cambridge, UK.

Nonetheless, the profession is expected to become more deeply embedded in a wide range of medical and scientific settings. A July paper by a team from the American Society of Human Genetics (J. R. Botkin *et al. Am. J. Hum. Genet.* **97**, 6–21; 2015) cited the low number of counsellors as one of the barriers hindering the incorporation of genetic medicine into standard practice. “I think that in medicine, for a long time, we’ve taken the approach that the information itself is all you need, but that’s blatantly wrong,” says Austin. “It is exquisitely important how you deliver that information.” ■

Michael Eisenstein is a freelance writer in Philadelphia.



Sarah Scollon advises people about gene testing.