MEDICAL CAREERS

Clinical Genetics: The Art and Science of Helping People Understand Their Genes

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When I tell people that I want to be a clinical geneticist when I am finished my medical training, usually I get a puzzled look – and not only from lay people. Fellow medical students, residents, and even some of my attending physicians are taken by surprise that such a specialty exists. One of my supervisors even told me that since I have such a good rapport with patients, it is a shame that I will be spending the rest of my life in a lab. Well, that couldn't be farther from the truth! This article will describe the roles played by clinical geneticists; it will also outline the training processes and special qualities required for the job. Finally, it will provide a brief description of a typical work environment for a physician working in this capacity.

ROLES OF THE CLINICAL GENETICIST

Diagnostician and Genetic Disease Management Consultant

Clinical geneticists are experts in inherited diseases. They diagnose genetic diseases, chromosomal anomalies, and dysmorphic syndromes. Their patients range from expectant couples to neonates, and from children with developmental delay to adults with cancer or neurodegenerative disease. Most of the conditions diagnosed by clinical geneticists cannot be cured, but accurate diagnosis often has major implications for prognosis, management, family planning and psychological acceptance. For example, a couple whose unborn child has been diagnosed with Trisomy 18, an invariably fatal chromosomal disorder, may choose to either: (a) end the pregnancy; or (b) emotionally prepare for the birth of severely handicapped child with a short life expectancy. A diagnosis of Marfan syndrome, a connective tissue disorder which puts the patient at increased risk for aortic dissection, will ensure regular monitoring by a cardiologist and appropriate lifestyle choices.[1] A young woman known to carry the BRCA1 gene, which predisposes to breast and ovarian cancer, may choose to have her children early and undergo elective oophorectomy, as well as having frequent breast examinations and yearly mammograms.[2]

Genetic Counselor

Contrary to popular belief, clinical genetics is a patient-centered specialty, in that clinical geneticists spend a great deal of time with each patient.[3] A typical new patient encounter will include a full medical history, detailed family history, physical examination, and lengthy discussion about the genetic basis of the disease or condition in question. An important aspect of a clinical geneticist's job is genetic counseling; this means explaining genes and chromosomes, how a particular disease occurs or is inherited, and the implications for diagnosis, treatment and prognosis, to both the patient and the patient's family.

Information Resource for Patients and Physicians

Clinical geneticists must keep up-to-date regarding the genetic tests that are available to their patients. With genetic testing becoming simultaneously more available and more complex, the results of these tests must be carefully explained to families at length. Patients can become inundated with media hype about genetic testing, and clinical geneticists help them separate fact from fiction. They can also be a valuable resource to other physicians, who may not be familiar with the less common genetic conditions; geneticists are consulted by family doctors, obstetricians, internists, pediatricians and neurologists, to name just a few.[3,4]

Dysmorphology "Detective"

Patients are referred to clinical geneticists for many reasons, a common one being to seek genetic or chromosomal causes for developmental delay. The clinical geneticist is trained to recognize dysmorphisms (differences in the physical appearance of a person that arose during fetal development); dysmorphisms can be as subtle as low-set ears or widely spaced eyes, or as obviously manifest as a cleft lip or extra digits. Particular combinations of dysmorphisms can sometimes be found in unrelated people with the same underlying condition. For example, people with Down syndrome, a common chromosomal condition in which there is an extra chromosome 21, typically have similar features including upslanting eyes, flat facial profile, short stature, single palmar crease, and a wide gap between the first and second toes.[5]

Risk Assessor

Clinical geneticists are like medical actuaries in that they estimate the disease risk posed to an individual or their offspring. Sometimes this is straightforward, as for conditions that are inherited in an autosomal recessive pattern. For example, cystic fibrosis is a serious lung disease that is only present when a child inherits a mutated gene from both parents; a couple with one affected child thus has a 25% recurrence risk (i.e., the chance that they will have another affected child with subsequent pregnancies). Increasingly, however, patients and clinicians are requesting risk estimates for such non-Mendelian, multifactorial diseases as Alzheimer's disease, epilepsy and schizophrenia, for which no one single gene is likely to be the cause.[4]

Researcher

Research is an important part of clinical genetics; geneticists will commonly publish case reports of interesting patients or families.[3] Because so many of the diseases that geneticists see are rare, reviewing scientific literature is an important part of the job.[3,6] They may also be involved in molecular, epidemiological, psychosocial or other types of research. Clinical geneticists will often adopt an area of special interest, and will focus much of their research in that area. For example, Dr. Margaret Nowaczyk, head of clinical genetics at McMaster, is the leading Canadian authority on Smith-Lemli-Opitz syndrome, a disorder of cholesterol biosynthesis.[7] Dr. Victoria Siu of the University of Western Ontario is studying genetic disease in the Amish Mennonites.[8] Dr. Michael Hayden of the University of British Columbia heads a laboratory investigating the role of genes in the development of Huntington disease and premature heart disease;[9] he is also the Editor-in-Chief of the scientific journal Clinical Genetics.[10]

BECOMING A CLINICAL GENETICIST

Currently there are two training routes for those interested in a career in clinical genetics. The Royal College of Physicians and Surgeons offers a five-year training program in medical genetics.[11] This is a residency program to which you apply directly from medical school. Medical genetics residency programs are currently offered at six institutions across Canada: University of British Columbia, University of Calgary, University of Manitoba, McGill University, University of Ottawa, and University of Toronto.[11] A second option is fellowship training under the Canadian College of Medical Genetics (CCMG).[12] Most people who have gone this route have completed a pediatric residency first, but other specialties (such as internal medicine or obstetrics and gynecology) are also applicable.[3] This option also takes a minimum of five years – for example, three years of pediatric training followed by two years of clinical genetics fellowship.[3]

What is residency in Clinical Genetics like?

Dr. Linlea Armstong is a recent graduate from the medical genetics residency program at Children's Hospital of Eastern Ontario in Ottawa; she found that residency in medical genetics was very diverse, including rotations through pediatrics, internal medicine and obstetrics during the first two years, with the last three years spent focusing on clinical genetics. "You have a chance to shape a good deal of your five-year curriculum yourself – for example, spending some time on a research project", says Dr. Armstrong. On the down side, since the existing programs have only one or a few residents at one time, there is comparatively little formal teaching relative to a larger program.[13] That being said, however, clinical genetics residency programs are growing. For example, the University of Toronto program currently has seven residents training under the Royal College. Dr. Sarah Nikkel, a clinical geneticist at CHEO, completed the medical genetics residency at the University of Manitoba; she feels that although the call requirements are not as onerous as in some other specialties, there is a significant knowledge base that needs to be mastered, as genetics touches on every single body system at every stage of the life cycle. Dr. Nikkel states, "learning the genetics of how these things occur... is fascinating."[14]

What qualities are required to succeed as a Clinical Geneticist?

A strong interest and background in genetics are obvious assets. Research experience is also an asset, but is certainly not required.[3] Communication skills are of utmost importance, as geneticists must convey complex scientific information in a simplified manner to patients, as well as being able to communicate with other physicians, scientists, and members of the genetics team. Compassion and empathy are required, as geneticists often must give distressing news about prognosis to their patients and deal with strong emotions such as grief, guilt, and anxiety. Also required are sensitivity and respect for the rights of individuals to make their own reproductive choices based on religious and cultural beliefs.

Where do Clinical Geneticists work?

Most geneticists in Canada work in hospitals and are affiliated with an academic centre. This obviously limits the geneticist as to where they can live and practice. Most centers have salaried physicians versus fee for service, as the numbers of patients seen by clinical geneticists are few compared to other specialties.[15] Starting salaries are usually around $150 000 per year, and range from $120 000 to $270 000 per year.[3,14,16] According to Dr. Nikkel, currently the job market for clinical geneticists in Canada is very good. "There are lots of jobs available, and many centres are expanding their genetics services [with the increasing demand]." Currently, there are 42 cities offering Clinical Genetics services across Canada.[17]

WHAT DO CLINICAL GENETICISTS ENJOY MOST ABOUT THEIR JOB?

Dr. Sandra Farrell is a McMaster medical graduate and head of clinical genetics at Credit Valley Hospital in Mississauga. "The opportunity to continuously be stimulated to learn is the one of the most positive aspects. Since we don't see an individual disorder very often and because the literature is so rapidly

evolving, one must review the topic for almost all referrals. For those who like to continue to learn, it is very stimulating. For those of us who like to teach, we spend a lot of time educating both patients and physicians."

Like all careers in medicine, the only way to know if clinical genetics is truly for you is to spend some time in a clinical genetics department. Dr. Nikkel gives this advice to students considering a career in clinical genetics: "Consider the type of patients you like seeing. If you like adrenaline rushes, this is not the career for you. However, if you like to learn new things everyday, taking the time to research and spending time with patients, this is a good choice."

Table 1. Job Description for a Clinical Geneticist

Roles
- Diagnostician and Genetic Disease Management Consultant
- Genetic counselor
- Information resource for patients and physicians
- Genetic risk assessor
- Dysmorphology "Detective"
- Researcher

Education
- M.D.
- Royal College residency program in Medical Genetics (5 years) or
- Pediatrics or other residency program (3 plus years), followed by a 2 year fellowship in Clinical Genetics

Skills and Qualities
- Strong interest in genetics
- Excellent communication skills
- Compassion, empathy
- Sensitivity to cultural and religious issues

Income
- Usually salaried
- $120 000 to $270 000 in Ontario; may vary by province and experience

Further Information
- Canadian College of Medical Geneticists website: www.ccmg.medical.org
- Canadian Association of Genetic Counsellors: www.cagc-accg.ca
- Canadian Residency Matching Service website: www.carms.ca

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