

A Career in Clinical Genetics

Clinical genetics is a diagnostic and counselling service for individuals and families concerned about the suspected or confirmed diagnosis of a genetic disorder. There are a number of reasons why people ask to be referred to the genetics service, or why their doctor thinks that they should be referred. These include:

The Role of the Clinical Geneticist

One of the best-kept secrets in medicine is the wide variety of issues covered by clinical genetics. It is one of the last truly generalised disciplines, encompassing both adult and paediatric medicine. It is also unique in its approach to caring for families as well as individuals within a family.

The Clinical Genetics Society issued a document¹ summarising the role of the geneticist. This includes:

- Diagnosis of genetic disorders affecting all ages and all body systems, birth defects and developmental disorders.
- Investigation and genetic risk assessment.
- Genetic counselling.
- Where appropriate, follow-up support and co-ordination of health surveillance for specific genetic conditions.
- Where appropriate, the offer of genetic services to extended families.
- Liaison with genetic laboratories (biochemical, cytogenetic and molecular).
- Education and training of genetic professionals and other health care professionals.
- Research: clinical, biomedical, psychosocial and service-related.

On a day-to-day basis, genetics is an outpatient specialty with a strong emphasis on time spent with families. An average genetic consultation lasts between 45 minutes and 1 hour, allowing enough time to fully explore the issues with the family. This consultation may vary greatly in content: counselling a pregnant couple regarding a fetal anomaly; assessing a dysmorphic child and discussing potential recurrence risks; assessing risks of malignancy in a family with a history of cancer; explaining chromosomal rearrangements and their implications; guiding patients through predictive genetic testing protocols. Such a variety of problems requires both a detailed knowledge of the subject and an ability to communicate complex issues to families. The wide range of potential problems demands a broad understanding of the whole of medicine. It also requires an understanding of the background science and the ability to discuss potential problems and solutions with scientists in biochemical, cytogenetic and molecular laboratories.

The Team Approach

Clinical geneticists are part of a multidisciplinary team including biochemical, cytogenetic and molecular scientists and genetic co-workers. The co-workers, who may be either genetic nurses or genetic associates, are often present in the counselling session. They are usually involved in pre-clinic contact and work-up of cases before they are seen by a doctor. Independent counselling by genetic co-workers is common, so supervision of other professionals is now an important part of a clinical geneticist's work. Liaison with laboratory staff is invaluable in the interpretation of

complex or unusual test results.

Multidisciplinary clinics involving other specialties such as ophthalmology, neurology, maxillary-facial services and endocrinology are often part of the workload. As such, the ability to liaise with a variety of other health care professionals as well as other members of the genetic service is a vital skill.

Expanding Specialty

Traditionally, genetics has been seen as a paediatric specialty. However, adult genetics is a large part of the workload, and this is increasing with the rapid expansion of knowledge of the genetics of common disorders. Cancer genetics now accounts for 40–50% of the workload in the majority of centres, so physicians with an adult background are eminently suitable for a career in genetics. It is common for special interests to be carried over into genetics: oncologists specialize in cancer genetics; an interest in paediatric neurology may develop into an interest in neuromuscular genetics; psychiatry translates into an interest in the genetics of dementias; general paediatricians frequently become interested in dysmorphism.

The referral rates to regional genetics departments are increasing steadily, partly due to the increasing awareness of the genetic component of common disorders and the advances in technology and testing that have resulted from the human genome project. It is expected that an expansion of consultant posts in clinical genetics will follow this expansion of referrals.

Regional Genetic Centres

There are currently twenty-five regional genetics centres covering the UK, with one centre in the Republic of Ireland. To date, there are about 130 consultants in clinical genetics with around a third of these in clinical academic posts. This ensures that national meetings involve friendly, recognisable faces and are usually good sources of gossip. A small specialty also means that most of the trainees are known around the country as well as in their own department.

A potential disadvantage to a career in genetics is that there is usually only one department in each major city (with the exception of London). This means that if there aren't any training numbers within your region for a few months or years then relocation may become necessary. It also means that a training programme is likely to be based entirely within one department, although there are currently discussions to allow trainees from small centres to move into larger centres for a period of time. In London, the trainees do get the opportunity to move to other centres during their training.

The organisation of a consultant geneticist's work varies from centre to centre. Some consultants sub-specialize (for example, in cancer genetics) and will only see cases within their subspecialty. In other centres, one consultant will cover all referrals within a geographical region and will be involved with all facets of clinical genetics. Even in these cases, most consultants have an area of special interest. If covering a geographical area, a driving licence is usually necessary.

Many centres offer joint multidisciplinary clinics, for example with ophthalmologists, neurologists, oncologists or fetal management teams. Consultants are expected to be available to give out-of-hours advice, especially for regional neonatal units, although it is unusual for an SpR to be expected to cover out-of-hours work.

As well as a clinical commitment, the majority of consultants maintain an active interest in

research, frequently in both clinical and molecular genetics. The rapid development of genetics requires constant updating of information and so regular attendance at local, national and international meetings is a necessity. Currently, about two-thirds of consultant posts are within the NHS, with the remainder being academic posts.

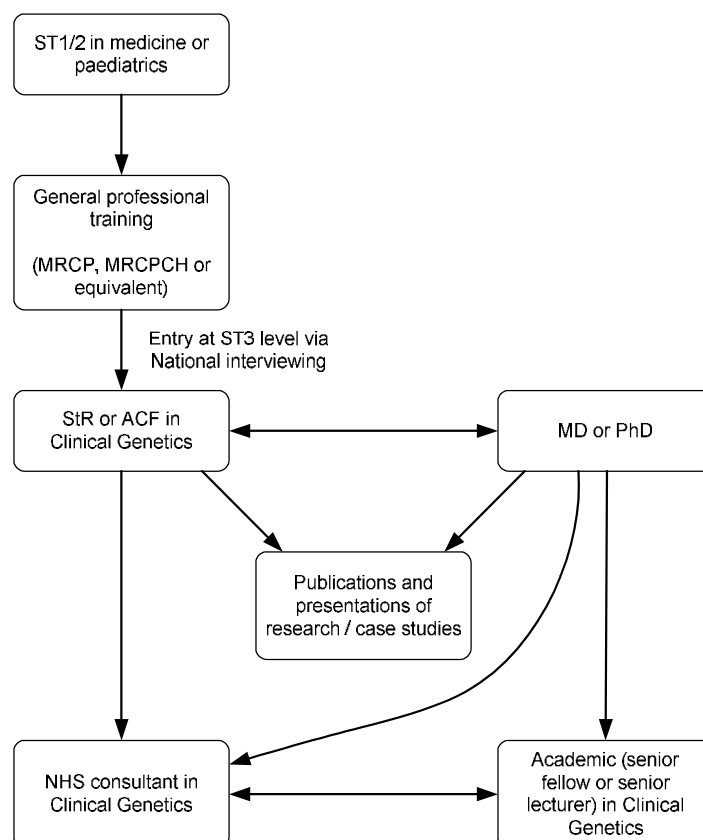
Training

Entry to the specialty is at ST3 level. Prior to MMC, entry followed basic training that preferably included both adult and paediatric medicine. Now, satisfactory completion of both foundation year competencies and those in core medical training (CMT1 and 2) or level 1 paediatrics is essential. A broad range of experience in acute general medicine or paediatrics, with exposure to unselected medical take, is essential. In 2008 appointment was by national interview, though for 2009 only it is possible that some deaneries will insist that posts are included in those allocated to run-through training. National entry requires either adult or paediatric MRCP. For run-through training in medicine, part 1 MRCP is essential. Part 2 is desirable, although realistically it would be difficult to obtain part 2 once started in genetics.

At interview you will need to demonstrate evidence of a commitment to clinical genetics, such as research experience or audit and attendance at genetic clinics or national meetings. Experience as an LAT in clinical genetics would be an advantage.

Training Pathway

This is the usual pathway; more general experience is an advantage.



The training programme is over 4 years, of which at least three years must be clinical training. It is generally expected that research will be undertaken during the training period, ideally aiming for an MD or PhD. Most trainees take time out of the programme for a postgraduate degree, for which a year is accredited toward the CCT.

The training programmes for StRs obviously vary with the regional centre, with major differences mainly due to the size of the centre. For example, training in a centre with four StRs and eight consultants is a very different experience to being the only trainee in a centre with two consultants. However, broadly speaking, training involves dysmorphology, neurogenetics (muscular dystrophies, Huntington's, dementias, spinal muscular atrophies, etc.), cancer genetics, ophthalmic genetics, metabolic genetics, prenatal diagnosis and fetal dysmorphology as well as experience in biochemical genetics, cytogenetics and molecular genetics.

Academic Clinical Fellowships (Walport posts) offer the option of a three-year attachment with 25% devoted to academic pursuits, aiming to obtain funding to enter a PhD programme at the end of the three years. ACFs will then need to obtain a clinical lecturer post in order to complete training for CCT.

Given the huge number of genetic conditions (over 19,000 entries in OMIM – Online Mendelian Inheritance in Man) StRs are expected to be able to use information technology to gather information about a given condition (rather than keeping it all in their heads) and to then be able to interpret and convey the appropriate information to patients and families. The use of digital imaging systems for clinical photographs of dysmorphic children will also ensure that trainees become au fait with information technology.

Most centres do not offer a registrar-led out-of-hours service, although a handful of SpRs in some centres carry a bleep during the day to deal with queries from GPs, ward (usually neonatal) referrals, scan referrals or fetal examinations. This does mean that life is very civilised (unlike the majority of medical jobs); rather than spending evenings and weekends on call, we write MDs, PhDs, book chapters, papers, reviews...

The Future of Genetics

The future *is* genetics! Given the advances expected from the human genome project, technological advances in SNP array, pharmacogenetics and the prospects for non-invasive prenatal diagnosis from peripheral blood sampling, someone will need to be at the interface between scientists and patients. That's us! Another very exciting development, still in its infancy, is the specific treatment of genetic disease with drugs targeted to the molecular pathways affected by the mutation. Drugs are also being developed to treat certain types of mutation which may have much wider application than to a single disease that may, in itself, be very rare. The role of geneticist will involve not only the clinical management of patients and families, but also the education of other professionals. It is expected that genetics will permeate all branches of medicine. An integrated view of clinical genetics² suggests that regional genetics centres will play a lead role in linking the needs of patients at primary care level with specialized care at secondary and tertiary level. Primary care trusts will therefore need input ranging from referral guidance to support for those dealing with patients with genetic issues in the community.

Genetics enjoyed a big expansion in recent years with the government having allocated large amounts of money to the specialty³. The expansion has slowed considerably but workforce planning indicates that the need for clinical geneticists will continue to increase for many years

yet. Clinical genetics is still a relatively young specialty with only around 12% of the workforce over 55 years of age. This means that consultant posts are likely to become available predominantly by the creation of new posts. As with all specialties the current funding climate makes it difficult to predict the job situation ahead, but being a rapidly advancing field means that we may fare better than many specialties. It is worth considering that the majority of expansion is likely to be in posts for those interested in adult genetics (for example, cancer genetics and cardiac genetics) whilst the majority of those in post at the moment are interested in paediatric genetics.

So what does a clinical geneticist do? Our job involves counselling and spending time with patients, assessing the risk of genetic conditions and facilitating decision making regarding management of a problem, in conjunction with the intellectual stimulation of the most rapidly advancing field of medicine, all within the framework of civilised hours. We can't think of a better combination.

References

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June 2000.
2. Donnai D and Elles R.
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BMJ, 2001;322:1048-1051
3. <http://www.doh.gov.uk/genetics/whitepaper.htm>

Further information

British Society of Human Genetics website: www.bshg.org.uk

Clinical Genetics Society website: www.clingensoc.org