

# Clinical Genetics Society

## Administrative Office:

Clinical Genetics Unit  
Birmingham Women's Hospital  
Edgbaston, Birmingham. B15 2TG  
Tel: 0121 627 2634  
Fax: 0121 623 6971  
Email: [cgs@bshg.org.uk](mailto:cgs@bshg.org.uk)  
website: [www.clingensoc.org](http://www.clingensoc.org)  
Registered Charity No. 274959



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## Roles of the Clinical Geneticist

**This statement represents a consensus reached by the Council of the Clinical Genetics Society (CGS) in 2011 and is an updated revision of an earlier document, entitled 'Role of the Clinical Geneticist' (Ref. 1), drawn up by the CGS Council and four co-opted Clinical Geneticists, in 2000. Whilst the core duties, roles and responsibilities of Clinical Geneticists remain essentially unchanged, the huge growth in molecular genetic tests and introduction of new technologies into NHS laboratory diagnostics has expanded the Clinical Geneticist's role, prompting a re-emphasis of certain areas of responsibility. Furthermore, the landscape of delivering genetic services has evolved, and will continue to do so, which has implications for consultant job planning.**

Clinical Geneticists are physicians who have undergone speciality training in genetics after core medical training in either Medicine or Paediatrics leading to attainment of MRCP or MRCPCH, or equivalent (previous routes of entry, e.g. via obstetrics and gynaecology, are no longer possible, though experience in this, and other disciplines, is welcomed). The curriculum for speciality training, which underwent major revision in 2010 (Ref. 2), covers a broad range of clinical experience, knowledge, skills, and behaviours, including genetic and inherited disease in adult and paediatric medicine, congenital malformations and non-genetic birth defects, dysmorphology, cancer genetics, neurogenetics, prenatal and preimplantation genetic diagnosis, cardiac genetics, and the genetics of other common diseases. Training also includes basic genetic science and theory, counselling theory and practice, the ethical and societal implications of medical genetics, laboratory experience and research.

Clinical Geneticists work within Regional Genetic Centres and, together with genetic counsellors and scientists, provide comprehensive genetic services to the population of a defined geographical region, ranging from approximately 1 million to approximately 5 million. Some centres provide a national service for certain rare diseases upon designation of national specialist commissioning status. Much of the day-to-day clinical workload is complex as the specialty deals with several thousand different genetic conditions which,

collectively, affect at least 5% of the population. Rare Disease UK has highlighted the fact that for children and older patients with rare conditions there may be a 4-5 year delay in obtaining a correct diagnosis, implying that improvement is possible with early involvement of the best clinical and laboratory expertise .

Clinical genetic services are usually delivered regionally in a hub-and-spoke model of central and peripheral clinics, as well as combined clinics in conjunction with a variety of other disciplines, e.g. ophthalmology and cardiology. Strong links are formed with local and national genetic laboratories, and Clinical Geneticists are valued members of local and regional clinical networks, such as oncology groups, fetal medicine and cardiology.

**Clinical Geneticists consider the following activities to be part of their normal duties, roles and responsibilities. In compliance with current job planning terminology, these are here divided into activities classed as ‘Direct Clinical Care’ (DCC) and ‘Supporting Programmed Activities’ (SPAs). Where the impact of new technologies demands emphasis of a particular role, this is described in *italics*.**

### **Direct Clinical Care**

- Diagnosis of inherited disease and genetic disorders (many individually rare, but collectively common) affecting all ages (including before birth), that affect any body system, birth defects (genetic and non-genetic), and developmental disorders.
- Investigation, using both specialised genetic and conventional non-genetic tests, for the purposes of diagnosis, risk assessment and management.
- Delineation and communication of the natural history, complications and appropriate management of inherited disease, genetic disorders and birth defects, and communication of this to relevant professionals.
- These activities are likely to involve review of medical records, clinical assessment, as well as appropriate investigation of relatives of the index case or proband.
- Genetic counselling and effective communication of appropriate genetic information by a variety means (e.g. letters, telephone, information leaflets).
- Predictive genetic testing of at-risk relatives for conditions where a familial cause has been identified using agreed protocols where available.
- Where appropriate, follow-up, support and coordination of health surveillance / screening for specific genetic conditions.

- Identification of genetic risk to the wider family and, where appropriate, the offer of genetic services to extended family members.
- Supervision of trainee clinical geneticists and Genetic Counsellors in relation to their clinical caseloads.
- Maintenance of paper records and/or an electronic clinical management system and patient database, and genetic family register(s) (where funding is available).
- Liaison with genetic laboratories, including discussion and interpretation of complex results and data of uncertain significance.
- Participation in local and national clinical networks, e.g., oncology, neurology, fetal medicine, cardiology; Dysmorphology meeting, Cancer Genetics Group, UK Huntington Disease Consortium and GenEthics meeting.
- Provide a resource of expertise and information for other specialists, primary care doctors and other health professionals, including interpretation of laboratory reports conveying complex genetic results and data, both pathogenic and those of uncertain significance.
- Recruitment of suitable patients to national research studies, screening studies and, where appropriate, clinical trials.

### **Supporting Activities**

Includes a wide range of non-direct patient contact activities in which many Clinical Geneticists participate

- Departmental management.
- Service development and liaison with other disciplines across the healthcare organisations of the Regional Genetic Centre's catchment area, including advising regional screening services co-ordinators and specialist commissioners.
- Development of service guidelines and standards.
- Liaison with genetics laboratories in relation to the clinical implications pertaining to the introduction of new technologies.
- Participation in clinical governance and clinical audit.
- Continuing professional development (genetics is a rapidly advancing science, thus making this particularly important).

- Appraisal of fellow consultants and other genetic professionals, as necessary.
- Involvement in research – clinical, biomedical, psychosocial and service related; active recruitment of patients to research projects nationally and internationally.
- Training, educational supervision, clinical supervision, and appraisal of specialist registrars in clinical genetics.
- Education and training of other medical and health care professionals, sometimes through multidisciplinary team meetings, especially in relation to rapidly evolving knowledge in genetics; for example, the introduction of high throughput technologies and generation of complex data from laboratory genetic tests.
- Work in support of local Trust and/or University.
- Teaching of clinical genetics, and the broader subject of human genetics, to medical (and sometimes other) undergraduates, especially in relation to the clinical application of new technologies.
- Representing the speciality through local and national (sometimes international) committees or working groups.
- Interaction with patient support groups, attendance of MDT and clinical network meetings, and availability for advice (in areas of special interest).

### **Staffing, Job Plans, and the Future**

In the year 2000, at the time of the last document, there were 96 whole time equivalent (WTE) NHS Consultant Clinical Geneticists. In 2008 the specialty was estimated to be expanding at a steady rate of approximately 8% per annum, according to the RCP (*Ref. 3*), a rate nearly twice that of all other disciplines, on average. In 2010 there were approximately 140 WTE NHS posts (data collected by CGS). Since then economic pressures have begun to affect the NHS, in common with the general situation. New full time (FT) NHS posts are now limited to 10 Programmed Activities (PAs), which affects the time available for Direct Clinical Care (DCC) compared with previously. At the same time, there is a trend for hospital Trusts to increase the proportion of DCC PAs in new job plans at the expense of SPAs.

The CGS Council strongly supports the retention of one additional DCC per half day clinic for preparation and follow-up work directly related to the care of the patient and their family. This has always been justified on the basis of the complex nature of the workload which, far from diminishing, has increased due to the complexity of interpretation of genetic tests.

The Council also strongly supports the allocation of at least 0.25 SPA for Educational Supervisors, who frequently give more time to this role. New trainees have to adjust to forms of practice that are very different from what they are used to and close supervision is necessary.

In addition, adequate SPA time must be preserved within job plans for Consultant Clinical Geneticists in order that essential supportive roles can be fulfilled, including the following:

- Consideration must be given to the education and resource role demands of a Clinical Geneticist. The relevance of genetics and genomics in all branches of medicine has been rising rapidly over the last 20 years and will continue to do so. Despite this, the level of required expertise in clinical genetics in the medical profession, outside the specialty itself, is very limited. Measures are being taken to improve this situation through the National Genetics Education and Development Centre in Birmingham but changes are slow. Clinical Geneticists, therefore, require time to educate, inform and provide an expert resource to the wider medical community on genetic matters for the foreseeable future. This will ensure that patients in mainstream specialties obtain the maximum benefit from the increasing availability of genetic tests in a patient pathway outside clinical genetics services (sometimes referred to as 'Mainstreaming Genetics'). Rapidly changing technologies are transforming diagnostic opportunities and Clinical Geneticists play a key role in interpretation of results, both to medical colleagues and patients alike. Clinical Geneticists receive referrals from virtually all hospital specialities and primary care, and often function as a resource for general information and expertise in relation to clinical matters.
- Advancing genetic technologies mean that new or refined diagnoses may be possible for patients seen in the past and the scale on which this will be possible is likely to increase exponentially. There are significant resource implications for routine re-contact of families seen many years in the past, notwithstanding ethical issues in knowing whether such contact would be welcomed. Whilst it will be appropriate to offer further appointments to those individuals where evidence based interventions might now be beneficial, resource constraints in most regional services will mean that this can only be offered on an ad hoc basis and such future contact should, where possible, be discussed with patients at their initial appointment. Patients can be invited to make contact with a service at some point in the future should they wish to investigate whether scientific progress has opened new diagnostic or therapeutic possibilities. Such activity would count as DCC.

- Clinical Geneticists will continue to play a key role in recruiting their patients to research, including surveillance studies and clinical trials, to improve the evidence basis for future recommendations and to inform medical practice and health economic assessments.

CGS recommends that these important and special roles of the Clinical Geneticist should be recognised and protected as formal components of job plans.

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## **References**

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