Role of the Clinical Geneticist

CGS members were invited to comment on the following document (approved by CGS council on March 15th, 2000) before its submission to the Joint Committee on Genetic Services. Date for Final Submissions was June 30th, 2000.

Report from Council to the membership of the Clinical Genetics Society, and to the Joint Committee on Genetic Services , submitted by members of CGS Council: *Helen Hughes (President), Diane Donnai (Vice President), Trevor Cole (Treasurer),Cyril Chapman (Secretary), Layla Al-Jader, John Dean, Gareth Evans, Peter Farndon, Susan Huson, Bob Mueller, Mary Porteous, Annie Procter (SpR rep), Elizabeth Sweeney (SpR rep), Karen Temple, and four co-opted members: Louise Brueton, Jill Clayton-Smith, Frances Flinter, Judith Goodship.*

Clinical Geneticists are physicians who have undergone speciality training in genetics after general professional training in Medicine or Paediatrics (and occasionally other disciplines, such as psychiatry, obstetrics and gynaecology, ophthalmology). The speciality training covers a broad range of sub-specialities such as the genetics of adult and paediatric disorders, cancer, dysmorphology, and neuro-psychiatry. Training also includes basic theoretical genetics, counselling theory and practice, laboratory experience and research.

Clinical Geneticists work within the **Regional Genetic Centres** and, together with genetic counsellors and scientists, provide genetic services to the population of a defined geographical region. These genetic services are delivered through a network of central, joint and district clinics. In any particular region, strong links are formed with genetic laboratories, oncology and fetal medicine centres, and with community and primary care teams.

Following discussions at the CGS Council in September 1999, the following list was developed in order to document the responsibilities of a Clinical Geneticist:

- **diagnosis of genetic disorders affecting all ages and body systems, birth defects and developmental disorders**
- **investigation and genetic risk assessment**
- **genetic counselling**
- **predictive testing for late onset disorders using agreed protocols**
- **where appropriate, follow-up and support and co-ordination of health surveillance for specific genetic conditions**
- **where appropriate, the offer of genetic services to extended families**
- **where appropriate and where sufficient resources exist, maintain genetic family register services**
- **liaison with genetic laboratories**
- **participation in national networks (e.g. Dysmorphology club, Cancer Genetics Group, UK Huntington's Consortium)**
- **education and training of genetic professionals and other health care professionals**
- **resource of expertise and information for other specialists, primary care doctors and other health professionals**
- **audit and participation in clinical governance**
- **continuing professional development**
- **interactions with patient groups**
- **research – clinical, biomedical, psychosocial and service related**
- **management and service development**
- **work in support of local Trust and Universtiy**
- **representing the speciality through local and national committees**
- **development of service guidelines and standards, advise commissioners of service and advise those establishing screening programmes**

The CGS Council felt that several areas of responsibility (**highlighted above**) merited further discussion and debate because they represent areas in which there was concern that there could be significant **variability of practice** between Clinical Geneticists in the UK. Consequently, the Council, together with 4 co-opted practising clinical genetic members, met to discuss and record their views on current practice in these particular areas.

It was not the aim of this meeting to provide a comprehensive and detailed account of these activities, rather to elucidate the main principles of Clinical Genetic practice in the UK at the present time.. Eighteen UK Clinical Geneticists (see above) were given the list of responsibilities and were asked to discuss them with their colleagues in their respective Genetic Departments prior to the meeting. Particular emphasis was placed on the need to discuss the 4/5 areas highlighted above in order to ensure that the conclusions drawn from the debate could be presented as a fair reflection of the body of opinion within the current UK workforce.

Follow-up, support, co-ordination of health surveillance and services to extended families

As in any other medical speciality, there is strong consensus of opinion that clinical geneticists will choose to follow those affected individuals for whom we believe that we may significantly influence their diagnosis and management. This is good medical practice. In addition, and possibly in contrast to other tertiary care physicians, clinical geneticists clearly have a responsibility to offer services to extended family members who may be at risk (or who have concerns about their risk) of developing a disorder or of transmitting it to their children. This is reflected in our tendency to think in terms of “families” rather than “patients”. Such family involvement is the essence of the service which geneticists provide. Contact with relatives usually is made only with the consent and co-operation of the index patient/family.

It is within the role of the clinical geneticist to be available to support families at times of distress and also to facilitate their decision making processes. However, geneticists should guard against serving as substitutes for other more focused supportive community services. It is a constant challenge to recognise the boundaries of professional involvement. Geneticists strongly agree with the concept of health surveillance and care pathways and are fully prepared to participate in these activities. Some geneticists would wish to undertake a co-ordinating role, especially if linked to a special service or research interest in a specific genetic disorder. Distinction needs to be made between the main role of the geneticist as provider of clinical genetic services, and the circumstances when specially qualified geneticists may chose to act as providers of medical care to those with particular genetic disorders.

Education

Clinical geneticists are conscious of an ever increasing demand on their time to provide talks / lectures / seminars to a variety of health professionals. The need for genetic knowledge and information permeate almost all branches and professional levels of medicine, not to mention other disciplines such as ethics, social science, the law, media, insurance etc. Access to the internet exposes patient groups and the lay public to genetic information which can serve to confuse as well as to inform. Clarification of misinformation can be extremely demanding

of time. Clinical geneticists recognise that they have responsibilities in the area of education and readily accept that, together with other genetic health workers, i.e. genetic nurse specialists and associates, their position may be best suited to translate some of the complex scientific concepts inherent to genetics into more understandable information. This is relevant when discussing how developments in the molecular area, in particular, are applicable to patient care at any one moment, - a role which geneticists would regard as one their core tasks as health professionals. It is now proving almost impossible to respond to all requests, and therefore clinical geneticists must agree with service Commissioners as to how best to prioritise the educational demands made upon them and, together with genetic nurse specialists / associate colleagues in respective Departments, agree how to respond.

Many Departments are actively involved in developing courses for different consumer groups and this collective approach will help to make the most efficient use of limited time and resources and also will allow development of specific skills in those best suited to the task.

Genetic registers

Genetic registers were first established in the UK in the late 60's / early 70's. Their stated aims were to achieve complete ascertainment of particular diseases and, by proactive contact, to have a role in their prevention. Registers often evolved from a specific research interest. In most centres where registers currently exist, they have developed to be a tool that facilitates review and re-contact , and are particularly focused on the time at which individuals at potential risk may benefit most from information that can influence medical management and decisions with regard to family planning. In some centres there appears to be a blurring between the function of a clinical review system, organised surveillance and the use of genetic registers. It is recognised that where registers exist, they must be resourced appropriately so that data accuracy can be maintained, consent for inclusion and confidentiality ensured, and the process of re-contact made explicit. Amongst registers most commonly in use in the UK are those for Huntington disease, the muscular dystrophies, fragile X, and some cancers e.g. familial polyposis. Decisions about the financial support of registers currently in use and the addition of new disorders must be taken in conjunction with Commissioners of genetic services.

Research

In a rapidly developing speciality where there are far more questions than answers, research forms an inherent part of the clinical geneticist's role. There is unanimity amongst practitioners that participation in research is essential to good clinical practice and that research activity should be broadly defined and not restricted to grant funded or laboratory projects. Legitimate research activities include clinical delineation and study of natural history, biomedical, psychosocial, service delivery etc. and can all be performed either individually or in collaboration.

Given the value that Clinical Geneticists place on research activities, they feel that Commissioners of services, as well as employers, whose aim it is to offer optimum care to families, need to ensure that consultant contracts and work plans reflect and acknowledge research demands. There is a danger that current staffing and funding levels (see below), as well as the limitation of time in the current 4 year training programme for specialists registrars, could lead to the sacrifice of this essential activity to the detriment of patient care.

Conclusion

The 18 Clinical Geneticists (16 consultants and 2 specialist registrar representatives) who met to discuss their roles, were drawn from 12 UK genetic centres and were felt to be representative of the current UK workforce. Detailed discussion was confined to specific areas where it was felt that there could be variability of practice. However what emerged from the debate was a remarkable consensus of opinion regarding the role of a clinical geneticist. The variability between centres and individuals, when it occurred, seem to result from differences in manpower and financial resources. The last manpower survey of all personnel in UK Genetics Centres took place in 1995 (RCP report). Although the number of consultant clinical geneticists has increased steadily over the last 5 years (expansion of approximately 8%/ year), the gap between need (3 whole time equivalent consultant clinical geneticist including those practising in cancer genetics / million population) and availability of personnel (including trainees, genetic nurse specialists / genetic associates) still remains significant, as does the variability in manpower between centres. Therefore it is inevitable that clinical geneticists have to prioritise their activities. It is recognised that not all clinical geneticists can fulfil all roles to the same degree or even with the same expertise. Thus, as the speciality continues to expand, members of Departments / Directorates inevitably will need to work in a collective manner so that, between them, most of the functions relating to patient care can be delivered to the population that they serve.

There remains a great deal of ignorance and mythology amongst many of our medical colleagues, let alone administrators and commissioners, regarding the exact role and function of a Clinical Geneticist within the health care system. Geneticists therefore have to continue to explain to the uninitiated what our responsibilities are and what training is required to perform the varying and challenging roles in this particular speciality in the year 2000. It is hoped that this document can serve not only as a summary record of the activities of clinical geneticists at this time in the UK, but also as a basis for designing job plans, manpower planning, and discussion with commissioners and government in this rapidly expanding speciality.