Clinical genetics services into the 21st century

SUMMARY OF A REPORT OF THE CLINICAL GENETICS COMMITTEE OF THE ROYAL COLLEGE OF PHYSICIANS

The shift of emphasis from rare genetic disorders, traditionally the field of the clinical geneticist, towards common conditions central to all medical specialties, is likely to continue and accelerate, as recognition of specific genes and the new understanding begins to be translated into possibilities for prevention and therapy. By the beginning of the 21st century, all fields of medicine will be utilising genetic advances in their practice, as well as in research. It is thus necessary to look at the specialty of clinical genetics not only in terms of its own activities, but also in relation to other specialties and to medicine overall. This report attempts to do this and to define the role that clinical genetics will and should play in this broader scene.

The growing clinical importance of genetics is recognised by two recent reports from the Department of Health [1,2]; the first documents the advances in practical applications of genetics at the individual family and population level and emphasises the need for strengthening and developing existing NHS structures for genetics services. The second examines the recent progress in identifying the genetic component of common diseases and the implications for wider clinical practice; this report stresses the need for further research and for rigorous evaluation to avoid premature or inappropriate applications.

The increased appreciation of, and demand for genetics services is not only reflected in professional views, but directly by the public at large, as seen in the high media interest in new advances and in the services provided (or lacking). Lay societies involved with genetic disorders are playing a major role, particularly by their concerted action through federations such as the Genetics Interest Group (GIG), whose recent report [3] strongly supports the value of genetics services in a wide range of disabling chronic disorders. Further support has come from a recent House of Commons Select Committee report on human genetics [4], which has made a number of important recommendations regarding clinical practice, research and wider issues in this field.

The current report is based on a survey of all individual genetics centres in early 1995. It examines specifically three areas of importance:

i the current manpower data on clinical genetics, giving present numbers and likely future trends;

ii the present role of the clinical geneticist and how this may change in the light of current and future developments;

iii the relationship between clinical genetics and other specialties, as genetics increasingly becomes an integral part of medicine.

The report was written against the background of major structural changes in the UK National Health Service—changes which have not been made with clinical genetics in mind but which profoundly affect the delivery of genetics services. Appropriate models for contracting for specialist services such as medical genetics have been outlined in a Department of Health document [5]. We have, however, looked beyond these immediate issues to the type of service that will meet the advances and challenges likely to be present at the start of the 21st century, now less than five years away. We have also tried to present the report in an international context, mindful of the close links between clinical geneticists in different countries, of the increasing convergence of health care in Europe and the influence of previous College reports in helping other countries to plan services.

Manpower in clinical genetics: current situation and trends

Senior career grade (consultant) posts

The number of consultants in clinical genetics has increased steadily since the earlier surveys were carried out (Fig 1a) and now stands at 79 (58.4 whole time equivalents (wte)). However, this still falls far short of the target figure of two wte per million population recommended by the 1991 report of the Royal College of Physicians [6], the current level being approximately half this figure (Table 1).

Figure 1a shows that around 40% of consultant posts are academically based, a proportion that has increased since 1990, which emphasises both the predominant location of genetics services in teaching centres and the major role of clinical geneticists in research and teaching.

Training posts

At the time of preparation of the report, the senior registrar and registrar grades had not yet been merged into a single unified training grade as recommended

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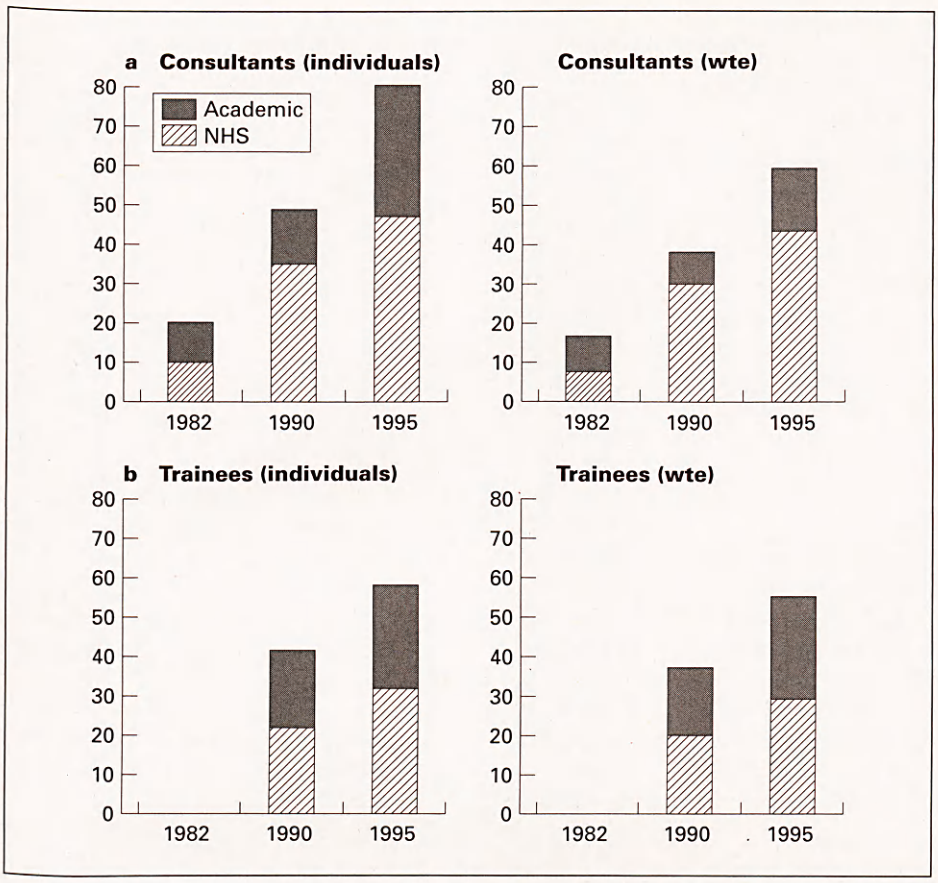

Fig 1. Manpower trends in clinical genetics: numbers and source of funding (all UK)

in the Calman proposals, but this imminent change, planned for late 1996, makes it particularly important to keep training numbers closely adjusted to new opportunities for permanent positions. Figure 1b shows that these training posts have continued to grow since 1990, and that posts funded from outside the NHS continue to account for close to half the number.

Genetics nurse specialists and other co-workers

Co-workers, principally specialist nursing staff, are involved with providing clinical genetics services alongside clinical geneticists. Table 2 shows that there are now 108.8 wte co-workers, a figure exceeding that of consultant clinical geneticist posts (58.4 wte) but falling far short of the four per million target given in the 1991 report [6]. It can also be seen that the distribution of co-workers is less uniform than that of medical staff, reflecting in part the different approaches of service delivery in specific centres.

The aims of medical genetics and the role of the clinical geneticist

A widely accepted aim of medical genetics is 'to help those families with a genetic disadvantage to live and reproduce as normally as possible' [7]. The activities contributing to the achievement of this include the clinical fields of genetic diagnosis, counselling, support and management, and the laboratory based services of molecular genetics and cytogenetic analysis.

The role of the consultant clinical geneticist has evolved over the past 20 years into a number of activities which reflect the aim of the service (Table 3); all are largely clinical in nature, but differ from most other specialties in that they are family based, usually involving more than one member, and frequently including individuals who are themselves healthy. The clinical geneticist's role is described in more detail in the full report.

The clinical geneticists are also likely to act as a valuable safeguard against premature, inappropriate or excessive application of new genetic developments and for alerting colleagues in other specialties to the potential dangers as well as benefits of these developments, which are likely to continue and increase into the 21st century.

Clinical genetics and other medical specialties

The most profound and extensive change to be expected in clinical genetics by the beginning of the next century will be that all clinical specialties will use genetic techniques and approaches in their practice, and that clinical genetics will no longer be the only specialty with major involvement. This changing

relationship is relevant both to the role and the manpower requirements for clinical geneticists, and it is therefore important to examine the possible situations that might arise. These are, in summary:

1. Other specialties might increasingly take on the genetic aspects of their own fields, including those activities currently undertaken by clinical geneticists, with the result that fewer clinical geneticist posts would be needed in the future.
2. System specialists and primary care physicians may feel unable to handle genetic issues and developments and may expect clinical geneticists to do this for them, giving a need for greatly increased numbers.
3. Some balance may be reached between the two situations above.

Present developments already suggest that the first possibility is unlikely; even if clinicians incorporate some genetic advances into their practice, the pace of new developments is such that they will be equalled or exceeded by others that require the involvement of clinical geneticists. The second option is a serious possibility that could threaten to overwhelm existing genetics services if not recognised and checked. Signs of this are appearing in referrals for familial cancers.

Clinical genetics and familial cancers

Until recently, neither cancer research nor clinical oncology practice related closely to clinical genetics, but the development of specific registers and preventive programmes for the rare but important series of single gene-determined tumours (eg familial adenomatous polyposis coli) has led to a convergence that has been sharply accelerated by the identification of the specific genes involved and the consequent availability of molecular diagnostic testing. Involvement of clinical geneticists is being further stimulated by the finding of mendelian subsets of patients in common cancers, such as breast cancer. Even though such categories may be only a small proportion of the total, they represent considerable numbers by comparison with the rare disorders traditionally seen by clinical geneticists, while many more families are now referred because of the possibility that they might fall into a genetically determined subset. In future years, this situation is likely to be repeated for other common cancers. Cancer related referrals now make up over 20% of the total workload in several of the genetics centres.

Clinical genetics and common diseases

The example of familial cancers is likely to be the first of a series of major developments in the isolation of genes for common disorders, each of which will involve, to some degree, clinical genetics as a specialty.

Table 1. Clinical genetics consultant staff in the UK: March 1995

Region†	Population (millions)*	Consultants (wte)	Additional need (wte)
Northern	3.102	2.9	3.3
Yorkshire	3.708	3.0	4.4
Trent	4.765	–	4.7
Nottingham	–	1.6	–
Leicester	–	1.6	–
Sheffield	–	1.6	–
E. Anglia	2.094	3.2	1.0
NW Thames	3.520	3.0	4.0
NE Thames	3.812	–	4.8
Inst. Ch. Health	–	2.1	–
Royal Free Hosp	–	0.7	–
SE Thames	3.717	2.7	4.7
SW Thames	2.999	1.6	4.4
Wessex	2.496[a]	2.5	2.5
Oxford	2.837[b]	3.2	2.5
S Western	3.744[c]	–	5.5
Bristol	–	1.0	–
Exeter	–	1.0	–
W Midlands	5.289	4.3	6.3
Mersey	2.413	1.8	3.0
N Western	4.031[d]	–	1.3
St Mary's Hosp	–	5.2	–
RMCH	–	1.6	–
Wales	2.906	3.8	2.0
N Ireland	1.632	1.6	1.7
Scotland	5.120	8.4	1.8
All UK	58.191	58.4	57.9

* Source: OPCS Population data 1993 (estimated)

† The term 'region' in this table and the rest of the report is used to indicate the UK health regions as organised up to 1993, with populations mostly between 2–5 million, and usually centred on a single university medical school. Wales and Northern Ireland function as single health regions, while some of the Scottish regions serve smaller populations

a Wessex region excluding Bath & Swindon

b Oxford region including Swindon

c S Western region including Bath

d N Western region (two centres serve approx 5.5m including an additional 1.5m from neighbouring regions)

It is clear that system specialists are already requesting the help of clinical geneticist colleagues as they increasingly identify the genetic component of the major disorders in their practice. Cardiology provides an example of a specialty where genetics involvement is just starting but which by the beginning of the next century may be as extensive as is currently the case for cancers. Genetic subsets can now be identified in important groups of congenital heart disease due to deletions of chromosome 22; comparable situations

Table 2. Overall clinical genetics staff in the UK: March 1995 (NHS and other funding)
Numbers in parenthesis include whole time equivalents (wte)

Region	Consultants	Assoc. spec/ staff grade	Trainees (SR+R)	Other medical sessions (wte)	Co-workers (wte)
Northern	5 (2.9)	1 (0.4)	6.0	0.2	7.5
Yorkshire	3 (3.0)	1 (1.0)	2.0	—	7.0
Trent	—	—	2.0	0.8	12.6
Nottingham	2 (1.6)	—	—	—	—
Leicester	2 (1.6)	—	—	—	—
Sheffield	2 (1.6)	—	—	—	—
E Anglia	7 (3.2)	—	3.0	—	3.3
NW Thames	3 (3.0)	—	2.0	—	2.0
NE Thames	—	—	—	—	3.7
Inst. Ch. Health	4 (2.1)	—	3.5	—	—
Royal Free Hosp*	1 (0.7)	—	—	—	—
SE Thames	3 (2.7)	—	3.0	—	3.8
SW Thames	3 (1.6)	—	4.0	—	2.0
Wessex	3 (2.5)	1 (0.6)	1.5	—	5.4
Oxford	5 (3.2)	—	4.0	0.6	5.8
S Western	—	—	—	—	4.7
Bristol*	1 (1.0)	—	—	—	—
Exeter*	1 (1.0)	—	—	—	—
W Midlands	5 (4.3)	—	2.0	2.0	8.2
Mersey	3 (1.8)	—	—	—	3.8
N Western	—	—	—	—	11.5
St Mary's Hosp	6 (5.2)	—	6.0	2.30	—
RMCH*	2 (1.6)	—	—	0.9	—
Wales	5 (3.8)	—	3.0	2.3	15.3
N Ireland	2 (1.6)	—	2.0	0.5	1.0
Scotland					
Aberdeen	2 (1.6)	1 (1.0)	2.5	—	0.7
Dundee	1 (1.0)	—		—	1.0
Edinburgh	4 (2.6)	—	2.0	—	3.0
Glasgow	4 (3.2)	—	6.0	—	4.5
All UK	79 (58.4)[a]	4 (3.0)	54.5[b]	9.6	106.8[c]

* Centres in England without either funding or manpower approval for full-time training posts

a Individuals (wte)

b 58 individuals. NHS part-time training scheme counts as 0.5 wte (6 individuals in England and Wales, 1 in Scotland). Includes 20 honorary SR/R in England & Wales and 6 in Scotland—not all with manpower approval for training

c NHS and research/other funded. Proportion of total involved in non-clinical activities not known

are being recognised in cardiomyopathies of childhood and early adult life due to specific gene mutations; while familial hypercholesterolaemia is now known to be the commonest mendelian disorder in Northern Europe and to account for a significant proportion of early coronary heart disease.

At present, most of those common disorders where a series of interacting genes is involved, such as diabetes, hypertension and most vascular disease, do not promise an easy resolution of the various factors or a simple application of molecular approaches in risk estimation. As indicated in the Department of Health report [2] individual genes may, in these cases, contribute only a small portion of the total variance and their role will vary from family to family. It seems likely that if such applications do become feasible, they will become the province of the particular specialist involved rather than of the clinical geneticist, but the clinical geneticist will have a valuable role in advising on how new tests may most appropriately be applied in a family context.

If some balance is to be reached in the use of clinical genetics services this will inevitably demand a selective approach for referrals to clinical geneticists, and clinicians will have to learn to recognise both those areas that they can handle themselves, and the

Table 3. Principal activities of the clinical geneticist

Genetic diagnosis	—	particularly rare familial disorders and malformation syndromes
Genetic counselling	—	for patient and family members at potential risk
Genetic registers	—	identification of genetic and health risks for extended family members, especially in late onset or X-linked disorders
Screening and population based programmes	—	close links with those responsible; direct involvement with genetic counselling for those found to be at high risk
Research	—	essential in a rapidly evolving field
Education	—	undergraduate and postgraduate
Links with genetics laboratories	—	often essential if misinterpretation is to be avoided
Expert advisory role	—	in relation to planning and purchasing of services

important minority of cases where potential problems or complexities make referral the wiser course.

Joint appointments

Although joint clinics between clinical genetics and other specialties are well established, joint appointments have been rare. With the growth of genetic applications in specific fields, such joint appointments now offer particular opportunities that may allow the necessary clinical specialist and genetics expertise to be combined. Joint appointments between oncology and genetics centres are an example that has evolved in several regions, but by the beginning of the new century this pattern might be seen in other specialties. Training programmes in genetics already allow for such a possibility, but the training demands for accreditation in some other specialties have proved less flexible and may need adapting.

Genetics and primary care

General practitioners and others in primary care are in a particularly favourable position to recognise genetic problems and to utilise new developments in genetics. They, like clinical geneticists, are commonly concerned with the family as a unit, while their existing involvement with community based preventive measures, such as immunisations and cervical cancer screening, makes the primary care setting a logical one for genetic screening programmes, such as cystic fibrosis carrier screening. The need for appropriate training in genetic issues of those in primary care, and for links between genetics specialties and primary care practitioners, is increasingly recognised [8] and represents a major challenge.

Planning for the future: recommendations and predictions

Planning for the 21st century must start now, since those entering training today will not emerge as consultants until the turn of the century, even with the shortened training period envisaged in the unified training grade. Our recommendations and predictions are contained in responses to the following questions.

1. What will be the role of the clinical geneticist in the 21st century?

a The role of the clinical geneticist, as summarised in Table 3, is now well established; genetic diagnosis and counselling will continue to form the clinical foundations of the geneticist's work.

b Special interests will become considerably more developed than at present; the existence of a team of 3–5 consultant clinical geneticists in each centre will allow this development. We do not consider that a consultant's work should be confined to a particular area, except in the case of joint appointments in a specialty.

c The increased use of co-workers (genetics nurses and allied counsellors), working in close conjunction with clinical geneticists, should be considered in areas where diagnostic and other specifically medical skills are less necessary.

d As developments in genetics become increasingly applied to common disorders, other clinical specialties and those in primary care will need to take on the greater part of the involvement, with clinical geneticists handling the minority of more complex situations.

e The educational role of clinical geneticists will become especially important; only when all clinicians have satisfactory basic undergraduate education and postgraduate training in medical genetics, will they feel confident in applying new genetic approaches in their own clinical work. Clinical geneticists, especially those with academic appointments should remain the focal point of these educational activities.

f Population screening and gene therapy are two areas which are likely to be more widely applied in the next century. While not central to the work of the clinical geneticist, both will require involvement, especially in the co-ordination and implementation of programmes.

2. How many clinical geneticists will be needed?

a It is likely that the current increase in demand will continue, even with more selective referrals and

the greater involvement of other specialties, and of co-workers.

b The current referral rate of around 400 new families per million population per year can be regarded as the baseline for general clinical genetics services, on which staffing plans can be made. New developments are likely to raise this baseline.

c The 1991 recommendation of two wte consultant clinical geneticists per million population has yet to be met in any centre. This now should be the *minimum* number to allow the range of activities outlined in this report to be undertaken.

d Joint appointments (eg between cancer services and genetics) may be a suitable approach to meeting need in some specialist areas.

3. How many training and other medical posts will be required?

a The current expansion rate of consultant clinical geneticist posts (currently 11% per year) means that there is a shortfall of trained individuals which, unless corrected, will cause a lack of suitably trained consultants.

b An increase from the present 43 to around 56 unified training posts in England and Wales, as suggested by both DoH and RCP data, is needed to maintain an adequate number of fully trained new consultants; and should be implemented without delay.

c Close monitoring of trainee numbers will be needed to avoid imbalance. In this respect the specialty has a creditable record, but accurate data must continue to be collected.

d We do not recommend any general increase in associate specialist and staff posts; these grades are of value in using the skills of particular individuals available locally, but are likely to remain few in number and should not form a major part of general service provision in the specialty.

4. What should be the organisational basis of clinical genetics in the new century?

a The concept of regional centres, each serving 2–5 million people, is of great value and should be preserved and strengthened. This will be essential if a clinical genetics team, with well developed and complementary special skills, is to be created.

b Locally based services should progressively increase, with centrally based clinics concentrating mainly on specialist and joint clinical developments.

c Scope for joint appointments with other specialties should be created (eg in the field of familial cancers).

d Genetics laboratories should continue to be closely associated, both in location and function, with clinical genetics services.

e Future changes in patterns of Health Service contracting must allow for the family based nature of clinical genetics, for the frequent crossing of geographical boundaries, and for the need to incorporate new developments. Recent changes in NHS structures have seriously endangered the regionally based structure of existing services, and the unique features of the specialty must be recognised.

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