

Genetics in the reformed Health Service

Changes for the better?

ABSTRACT—The practical value of medical genetics, in particular the development of molecular genetics complemented by clinical diagnosis and counselling, is widely recognised. There is strong independent support from government and patient organisations for augmenting genetics services in all health regions; this support gives much reason for optimism. But there appears to be a hiatus following the reform of the Health Service: no genetics centre has, as yet, adequate resources and there has been no increase in clinical genetic manpower in the last two years. Even worse, Wales and at least one English region have devolved genetic services to districts, which appears to be contrary to government policy for genetic services.

These factors have inevitably limited the implementation of many opportunities for improved patient care and the prevention of genetic disease. However, medical geneticists, assisted by the Royal College of Physicians and others, want to respond positively to the changes in the Health Service. Recommendations are made for strategies which promise to maintain integrated regional clinical and laboratory services and to achieve well evaluated developments.

Background

Exactly two years ago data were collected for the Royal College of Physicians of London (RCP) report [1] on progress towards a comprehensive national network of regional genetics services. There were then great inequalities between regions and nowhere was manpower adequate for the clinical applications of the advances in molecular genetics. Recommendations were therefore made to strengthen the regional organisation of genetics services. They included an increase in the number of consultant clinical geneticists* by 77

* Although often used synonymously, the term 'clinical geneticists' is used to describe medically qualified doctors in direct contact with patients and "medical geneticists" for all geneticists in health care including clinical geneticists, clinical cytogeneticists, clinical molecular geneticists and clinical genetic co-workers (genetic nurses, social workers and associates).

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to achieve a minimum of two posts per million of population.

Why should clinical and laboratory genetics services be integrated and organised regionally?

In addition to individual patients, geneticists care for whole families who do not always live conveniently within a single district. Genetic registers, which provide the essential source of support for families afflicted by genetic disease, are thus best organised at the level of regions which are the largest population grouping allowing clinical contact. Clinical geneticists and genetics laboratories are unable separately to provide an efficient service because many genetic tests require planned family investigations and precise clinical diagnosis. Some genetic tests are too rare to be efficiently organised in each district where throughput would be too scanty to maintain quality and allow adequate training. Moreover, because of the unprecedented rate of new discoveries in genetics, research and development are crucial and require a 'critical mass' which is not achievable at individual district level. The intensity and speed of laboratory research and development have generated a new cohort of scientists in the Health Service whose role has evolved as being complementary to that of clinical geneticists with whom they work closely.

Independent confirmation of the value of genetics services

Influential confirmation of the value of regional genetics services comes from reports [1,2] from the RCP, a Joint Statement from the medical royal colleges, correspondence between the Clinical Genetics Society and Mrs Bottomley, the minister for health (now Secretary of State), the House of Commons Health Committee [3], Government [4] and lay organisations [5]. Most recently [3], the House of Commons Health Committee took advice from the Clinical Genetics Society and stated in its report:

... we believe that the weight of evidence is sufficient to indicate the need for improved genetics services in the United Kingdom at the primary and community health care level backed up by a sufficient regional clinical genetics service incorporating the required laboratory services, pathology services, counselling services, clinical laborato-

ry and counselling expertise and the setting up and running of genetics registers. We recommend that the Department of Health, as a matter of urgency, instruct each Regional Health Authority to review its genetics services. This review should determine the extent to which regional genetics services are able to provide for the needs of their population. (para 133) . . . We also recommend that the Department of Health provide guidance for relevant health personnel on genetic disorders, their prevention and the specialist services that are available' (para 134).

In its reply [4] the Government said that it

' . . . agrees that there is a need for Regional and District Health Authorities to review their genetic services. Guidance on the scope and timing of such reviews will be prepared by the NHS Management Executive in the light of its priorities for the NHS. As a first step, the Chief Medical Officer will write shortly to all Regional and District Directors of Public Health on the subject. The letter will invite Directors of Public Health to consider the needs of the local population in planning for these services, whether provision meets those needs, and quality standards' (para 2.59).

'In December 1991 the Department of Health announced that it will fund a confidential enquiry into counselling for genetic disorders. The enquiry is being conducted by the Department of Medical Genetics at the University of Manchester and the research unit of the Royal College of Physicians. The purpose of the enquiry is to discover how well the needs of patients and families who are threatened by genetic disorders are met. The results of the enquiry should lead to recommendations to improve further the quality of care given to these patients' (para 2.60).

' . . . the Department of Health recognises the importance for health professionals of maintaining an up-to-date awareness and understanding of genetic disorders and is considering how this can best be achieved. The forthcoming letter from the CMO outlining the nature and purpose of services for genetic disorders will serve as a catalyst for promoting awareness among health professionals of the importance of genetic disorders and their sensitive management.' (para 2.61).

The Genetic Interest Group (GIG), which represents more than 60 charities for patients with genetic diseases, reported [5] on replies GIG had received from regional health authorities about proposals for genetics services. A wide disparity was noted between different RHA proposals with uncertain district purchasing plans for genetics. GIG believes that

' . . . currently, molecular biology has more to offer in the prevention of chronic disease than any other field of medical research . . . '

' . . . it is difficult to see how districts who are concerned to purchase existing techniques of disease treatment will quickly acknowledge and fund such necessary new tests and techniques.'

GIG representing patients' needs wants genetics services to remain

' . . . a coherent package of clinical investigation, laboratory tests and specialised counselling . . . '

These services should

' . . . be distributed equitably . . . (and) . . . genetic services should become a special initiative for each district guided by advice from the Department of Health . . . '

GIG suggests that more resources might be devoted to the needs of genetic patients and families, perhaps using the Government sponsored AIDS programme with ring fenced central funds as a model.

' . . . the current total of 5,000 AIDS sufferers within the U.K. (should be compared) with the birth every year in the U.K. of at least 30,000 babies with disorders with a genetic component from which they will suffer life long disability or death by early adulthood.'

The reform of the Health Service

In the light of this strong support from government and consumers it is interesting to see how the reform of the Health Service is affecting genetic services. The reforms were received constructively by medical geneticists [6] and by the Royal College of Physicians of London (RCP) which published and distributed the 1990 report on genetics services [1] and guidelines for purchasers of genetics services [7] to every district health authority in UK. The RCP also established a working party (Chairman Dr Robin Winter) to advise on collecting the clinical data necessary for service agreements. This working party has identified a basic data set and has suggested how genetics referrals might be costed.

The survey

The present report examines the changes in *clinical* genetics services which have occurred during the last two years, including the first year of the reforms. (The Association of Clinical Cytogeneticists (ACC) and the Clinical Molecular Genetics Society (CMGS) are responsible for collecting laboratory data.) Clinical data were collected by sending to clinical geneticists in every Health Service region copies of their 1989 data for updating. Information about the effects of the

reforms was also requested using a semi-structured questionnaire. All centres replied including one from each of the 15 regions in England (two each from Trent and South Western sub-centres) and Wales, from Northern Ireland and from the four centres in Scotland. Changes in clinical manpower between 1989 and 1991 are documented in Table 1 and, with the views of respondents about the effects of the reforms, are summarised below.

Overall manpower

There has been no increase in total manpower since 1989 in clinical genetics with the equivalent of 166 whole time personnel (WTE) in 1989 and 1991. Some regions have suffered considerable reductions, most notably East Anglia (11.4 to 7.7 WTE) and NE Thames (8.8 to 2.8 WTE); others had smaller reductions including Northern, NW Thames, SE Thames, and N Ireland. These reductions are potentially very damaging because they represent a large proportion of a small base. However, nine regions (counting Scotland as a single region) have had small to moderate increases (see Table 1).

Manpower according to grade type

There has been a net increase of 6.34 WTE consultants with nine new consultant clinical geneticist posts (Northern, Yorkshire, Oxford, N Western (2), Wales, Scotland (3)) and three retirements and moves (Trent, S Western and W Midlands). Senior registrar and registrar numbers have fallen slightly from 39 to 37 posts (34.4 to 30.2 WTE). There has been a slight increase in other medical posts from 39 to 42 but this has had little effect in WTE (17.5 to 16.9 WTE). Clinical co-worker posts have also risen slightly in number (from 80 to 82) but remaining at 69.8 WTE.

Thus, at a time of unprecedented scientific advances during which virtually every major human disease gene has been mapped, there has been no overall increase in professional manpower trained to care for the affected families. This situation is extremely worrying but at least there is now one or more consultant clinical geneticist in every health service region (except Tayside in Scotland) qualified to provide a minimal genetics service.

Comments from respondents

Although a majority of respondents expressed doubts and anxieties, an appreciable number of clinical geneticists reported that they see opportunities for improvements attributable to the reforms, most notably greater scope for planned growth and development. However, even the most optimistic currently regard the changes as being like '... the proverbial curate's egg. . . becoming a clinical directorate gives greater scope for careful budgeting. . . but this is

negated by repeated cost-improvement schemes . . .'. In practice, there seems to have been little real progress towards an internal market for genetics and because genetic services have so far not generally been devolved to districts, in the majority of regions direct negotiations are not yet required with each individual district. However, in two English regions and in Wales regionally purchased genetics services have been devolved and there seems at present no mechanism for funding new developments. This is a major step backwards and one which the Department of Health does not recommend. The danger is real of administrative confusion, and potential damage to genetics services is shown by the situation in Wales which has abolished all regional services: '... Region will cease to act as a broker and all funding will be by individual districts on a 'club subscription basis'. Active negotiations are in progress with individual DHAs and there is great uncertainty over the funding basis with no apparent mechanism, central or otherwise, for new developments . . .'.

The delay in fully implementing the proposed NHS changes has allowed the strangeness to wear off and several centres hope that a well organised clinical directorate, with a business manager actively engaged in lobbying purchasers, will be helpful. This will be important as purchasers may not yet have understood how the clinical management of families (rather than simply individual patients) complicates costing. Purchasers will need advice about the resources required to buy genetics services for rare genetic disorders and to appreciate the clinical and community work necessary on large families scattered across several districts or regions.

Some posts appear to have been frozen (two senior registrar and a registrar post) although it is not clear that these are the consequence of the reforms.

Extra-contractual referrals (ECRs) are also seen as major potential problems. ECRs are essential for rare diagnoses which it would be hopelessly inefficient to attempt to provide in every region (or district) and arrangements between genetic centres are already in place for sharing these rare diagnoses within consortia. There is widespread reluctance to accept the need for charging colleagues although this may become inevitable.

Surprisingly few anxieties were expressed about the effects of competition between genetics centres, probably because clinicians were not directly involved. However there are concerns that genetic counselling may not always be available for patients diagnosed in private cytogenetics laboratories.

A pervasive problem relates to the need to deal with a burgeoning management bureaucracy without any extra provision for the genetics centre of clerical or management support. The need for clinical data for general resource management is widely recognised and the Royal College of Physicians circulated to all clinical geneticists a report from a working party

Table 1. Clinical (non-laboratory) genetic manpower 1989–1991 whole time equivalents (WTE). (Number of individuals in parenthesis)

NHS Region	Consultant		Junior medical		Other medical		Clinical co-workers		Non-clinical academics		Total	
	1989	1991	1989	1991	1989	1991	1989	1991	1989	1991	1989	1991
01 Northern	1.70(2)	2.20(3)	3.00(3)	2.60(4)	1.60(2)	0.40(1)	3.60(6)	3.10(5)	0.25(3)	0.15(2)	10.15(16)	8.45(15)
02 Yorkshire	1.82(2)	2.82(3)	0.00(0)	2.00(2)	0.40(1)	0.40(1)	3.30(4)	4.00(4)	0.00(0)	0.00(0)	5.52(7)	9.22(10)
03 Trent	3.93(5)	3.10(4)	1.00(1)	1.50(2)	1.80(5)	2.10(4)	9.50(10)	10.00(11)	0.25(1)	0.25(1)	16.48(22)	16.95(22)
04 E Anglia	2.80(4)	2.40(4)	3.00(3)	2.20(3)	0.00(0)	0.00(0)	5.50(6)	3.00(3)	0.10(1)	0.10(1)	11.40(14)	7.70(11)
05 NW Thames	2.00(3)	2.00(3)	2.00(2)	1.00(1)	0.00(0)	0.00(0)	2.00(2)	2.00(2)	0.00(0)	0.00(0)	6.00(7)	5.00(6)
06 NE Thames	2.20(3)	2.20(3)	5.10(7)	0.50(1)	0.00(0)	0.10(1)	1.50(1)	0.00(0)	0.00(0)	0.00(0)	8.80(12)	2.80(5)
07 SE Thames	2.20(3)	2.70(3)	3.60(4)	2.60(3)	0.00(0)	1.10(3)	3.00(3)	3.30(3)	1.35(4)	0.65(2)	10.15(14)	9.55(14)
08 SW Thames	0.73(1)	0.70(1)	1.00(1)	1.50(2)	0.00(0)	0.00(0)	2.00(2)	1.00(1)	1.00(1)	1.00(1)	4.73(5)	4.20(5)
09 Wessex	1.60(2)	1.60(2)	0.50(1)	0.50(1)	0.60(1)	0.60(1)	2.50(3)	2.70(4)	0.00(0)	0.00(0)	5.20(7)	5.40(8)
10 Oxford	2.60(3)	3.60(4)	1.00(1)	2.00(2)	0.86(6)	1.09(5)	2.00(2)	4.00(4)	0.00(0)	0.00(0)	6.46(12)	10.69(15)
11 S Western	2.10(3)	1.80(2)	0.00(0)	1.00(1)	0.05(1)	0.20(2)	2.40(3)	2.00(3)	0.00(0)	0.00(0)	4.55(7)	5.00(8)
12 W Midlands	2.80(4)	2.30(3)	1.00(1)	2.00(2)	0.05(1)	1.00(2)	4.10(6)	4.50(6)	0.40(2)	0.40(2)	8.80(14)	10.20(15)
13 Mersey	1.00(1)	1.00(1)	0.00(0)	0.00(0)	0.20(1)	0.20(1)	1.80(2)	2.80(3)	0.00(0)	0.00(0)	3.00(4)	4.00(5)
14 N Western	3.60(4)	4.90(6)	2.00(2)	3.00(3)	5.10(9)	4.70(10)	10.20(11)	10.90(14)	1.20(3)	0.60(2)	22.10(29)	24.10(35)
15 Wales	2.60(3)	3.60(4)	5.50(6)	3.10(4)	1.20(3)	2.40(5)	8.50(9)	8.20(9)	0.15(1)	1.00(1)	17.95(22)	18.30(23)
16 N Ireland	0.50(1)	0.50(1)	0.00(0)	0.00(0)	2.00(3)	1.30(3)	1.00(1)	1.00(1)	0.05(1)	0.00(0)	3.55(6)	2.80(5)
17 Edinburgh	0.00(0)	1.60(2)	0.00(0)	0.00(0)	2.10(4)	1.20(2)	1.40(2)	1.40(2)	0.90(2)	0.90(2)	4.40(8)	5.10(8)
18 Aberdeen	1.60(2)	2.20(3)	2.10(3)	1.50(2)	0.00(0)	0.00(0)	0.50(1)	0.20(1)	0.05(1)	0.05(1)	4.25(7)	3.95(7)
19 Dundee	0.00(0)	0.00(0)	0.00(0)	0.00(0)	0.10(1)	0.10(1)	0.00(0)	0.00(0)	1.25(2)	1.25(2)	1.35(3)	1.35(3)
20 Glasgow	1.60(2)	2.60(3)	3.60(4)	3.60(4)	1.00(1)	0.00(0)	5.00(5)	6.00(6)	0.00(1)	0.00(1)	11.20(13)	12.20(14)
Total	37.38(48)	43.72(55)	34.40(39)	30.20(37)	17.51(39)	16.89(42)	69.80(80)	69.80(82)	6.95(23)	6.35(18)	166.04(229)	166.96(234)

(chairman Robin Winter) suggesting a list of basic information necessary for setting and monitoring service agreements. Only ten of the 22 respondents would currently be able to collect this essential information. The reasons given include lack of investment in computer hardware, unsuitable software and poor clerical support.

Successful strategies

There are often new opportunities during times of rapid change, and this certainly applies to the Health Service. Clinical genetics responded quickly to the current changes [1,6,7] and as one respondent commented: '... the ethos of running and managing clinical genetic services as an efficient and well-managed and audited business is actually very appealing...'. A number of promising strategies for maintaining and developing genetics services are evolving, although it is still too early to judge their success. Clinical geneticists working with managers are identifying ways to exploit the potential advantages of the reforms, and respondents draw attention to the value of the RCP 'Purchasers Guidelines' [7] in these negotiations.

With the election of a Conservative government for a fourth term, it seems certain that the purchaser-provider separation will continue and the distribution of NHS funding will depend increasingly upon local decisions about the health needs of district populations. In line with an explicit NHS research and development strategy, there will also be greater emphasis on evaluation of new clinical procedures before their widespread introduction. Medical geneticists will inevitably increasingly be involved in management if they wish to maintain existing and introduce new genetic services. The following recommendations are designed to help them and reflect the progress being made in several regional genetic services.

Recommendations

1. All regions are strongly urged to implement previous recommendations to increase clinical genetics manpower with adequate supporting staff, including those necessary for data collection. Advice on the needs for laboratory genetics should be sought from the Royal College of Pathologists and the relevant professional societies.

2. Providers of genetic services need to influence those who are charged with advising health authorities on the health needs of resident populations. In this they will be assisted by quoting reports from the Royal College of Physicians [1,7], independent support from charitable organisations and the imminent letter to health authorities from the Chief Medical Officer. This dialogue will allow a mutual education process between public health doctors and hospital based geneticists. There is a particular urgency in discussions with districts where funding for regional services has been devolved, but all medical geneticists are advised to establish these links because even where genetic services are currently regionally purchased, future devolution of funding decisions to districts appears inevitable.

3. Medical geneticists must make themselves familiar with local arrangements for funding to avail themselves of the rapid advances in genetics. The most time-consuming will involve piecemeal funding by many districts, but specific funds may be available for service developments (for proven procedures) and medical innovation schemes (for procedures requiring further clinical evaluation). All regions will now have a Regional Research and Development Committee whose chairman should be a good source of advice.

4. Because of the rapidly increasing workloads and the applications of genetics throughout medicine, medical geneticists must encourage understanding of genetics amongst other specialists and general practitioners and in this they will be helped by the confidential enquiry into counselling for genetic disorders [8].

5. Urgent consideration has to be given to the ways of introducing population screening for carriers of genetic disorders into community genetics to ensure that it is carried out efficiently but protects the individual's right to accept or reject screening.

6. To bring all these aspects into a single comprehensible form for purchasers, the genetics service should provide, and revise as necessary, a business plan which states the following:

- what the aims of genetics services are ('mission statement')
- how genetics fits into the overall provision of health care
- how the genetics service is currently provided
- the strategy for achieving policy objectives

- gaps in present service provision
- how and when these gaps might be closed
- what innovations are required.

7. Each region should have a genetics committee whose membership includes genetics providers and purchasers. This will be a source of trusted impartial advice on the development, organisation and quality of services. The committee should meet regularly with district directors of purchasing to discuss recent developments, progress with service agreements, and to form a consensus on the needs of district populations, balancing geneticists' priorities with competing demands on precious resources.

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