More than 246 mutations involved in *NF1* have been reported by the NF1 Genetic Analysis Consortium up to November 1997, 45% of them deletions.[38] Our intragenic linkage studies pointed to two cases with a deletion, 3% of those investigated. The small sizes of the families and the low number of families containing several generations, the non-clustering of the cases, and the absence of disequilibrium in linkage studies rule out any founder effect for NF1 in northern Finland. Observations in other population based NF studies are similar and confirm the findings of small family size and few generations.[4 6] In the familial cases examined by linkage study here, six out of seven of the first affected subjects in the family had inherited the mutation from the father, a phenomenon which has been shown in 34 out of the 37 published cases (92%) including our data.[37 39 40]

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M POYHONEN*†
S KYTÖLÄ*
J LEISTI*

**Department of Clinical Genetics, Oulu University Hospital, Finland*
†Department of Medical Genetics, Väestöliitto, Family Federation of Finland, Post Box 849, 00101 Helsinki, Finland

Correspondence to: Dr Poyhonen, minna.poyhonen@vaestoliitto.fi

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Genetic registers in clinical practice: a survey of UK clinical geneticists

EDITOR—Genetic registers have now been in use in the United Kingdom for nearly 30 years,[1] although they are not widespread in Europe.[2] They are an integral part of most UK medical genetics services[3] and yet their functions vary from centre to centre. Many registers were originally developed for research purposes, often in connection with one specific inherited disease,[4] while others, designed for service use, may cater for many genetic disorders. The WHO report of 1969 suggested that a list or register of pedigree data should be maintained by each genetic centre,[5] although the purpose of the list was not specified. In its 1972 report,[6] the WHO recommended setting up of family orientated genetic registries as part of a system to provide counselling and diagnostic services, treatment, and long term follow up for patients with genetic disorders. In 1978, the definition of genetic register functions was clarified by Emery *et al*,[1] who suggested five main roles, which are not mutually exclusive. These were the clinical or therapeutic role (follow up and recall), the reference list, to monitor outcomes of service provision, to act as a research tool, and to assist in the prevention of genetic disease through complete ascertainment and family follow up. Since that time, the use of genetic registers for family

follow up at predetermined times has been advocated to inform younger family members of their genetic risks when they reach maturity[3 4] or to carry out interval screening for complications of genetic disorders, such as in Marfan syndrome[7] or the familial cancers.[8] Discussion with colleagues suggested that a diversity of practice in the use of genetic registers in different UK genetic centres has arisen, perhaps because of differing funding priorities. This could raise quality issues in clinical genetics, as members of a single family attending different genetic centres may experience a different service from each centre's genetic register. Expectations raised in one centre may not be translated into service at another. To clarify the use of genetic registers in the UK, and to inform the debate as to whether there should or could be an agreed quality standard for genetic registers, we carried out two questionnaire based surveys. The first was designed to ascertain the nature of registers then in use, and the second to ascertain the views of the UK clinical genetics community about what genetic register services should be provided. In these, we considered issues corresponding to the first, second, and fifth roles of a genetic register of Emery *et al*,[1] but did not consider a register's potential role in monitoring service outcomes, nor its use as a valuable research tool.

The first questionnaire was addressed to each UK genetic centre in 1995 and asked for details of registers in clinical use (purely research registers were excluded), staff employed to support them, and precautions taken to maintain data security. Twenty out of 22 questionnaires were returned (91%). Responses to a second, anonymous questionnaire were sought from all members of the UK Clinical Genetics Society of consultant level or equivalent seniority in 1997. Fifty eight replies were received from a possible total of 77 (75%). Two questions asked for a description of a genetic register and its function, while a further 20 questions took the form of statements about genetic registers. These aimed to elicit opinion on the following issues: (1) what is the purpose of a genetic register, (2) how do patients get onto a genetic register, (3) what data should be stored, (4) should there be separate registers for each genetic disease, (5) what form of consent is required for recording details on a register and, (6) who is responsible for the function of the genetic register. Respondents were asked to grade their opinions of the statements on a five point scale, corresponding to "strongly agree", "agree", "no opinion", "disagree", and "strongly disagree". For issues where there was consensus, the responses "strongly agree" and "agree", and "strongly disagree" and "disagree" were added together to simplify presentation of results. Space was available on the questionnaire for additional comments about some of the statements. A computerised family based register was in general clinical use in 18/20 centres. On average, these registers contained data on 17 700 individual patients (range 3000-48 000) in 6050 families (range 5000-16 000). Sixteen centres maintained disease specific registers (DSR) (table 1). Clinical patient data were integrated with clinical laboratory data in 10 centres. Nine centres employed staff primarily to maintain their registers. In four, the staff were medical (average 27 hours per week), in six nursing (average 34.5 hours per week), and in five secretarial (average of 21.5 hours per week). The majority of this dedicated staff time (80.5%) was within three centres. In four centres, the genetic registers were on stand alone computers and the remainder were on a local area network (LAN). One department's register was part of a general hospital network. None was internet accessible. Twelve used the main database computer for purposes other than running the register. Fourteen felt that access to the computers was physically secure. Thirteen used some form of password protection at machine start up, all used a password at application start up, but only seven changed either password regularly. Only one centre used any form of data encryption. All departments had regular data back up systems although there was considerable variation in the frequency that back ups were carried out.

Table 1 Disease specific registers in the UK

Disease	*No of centres*
Huntington's disease	14
Familial cancers	12
Muscular dystrophies	11
Fragile X syndrome	4
Marfan syndrome	3
Neurofibromatosis	2
Adult polycystic kidney disease	2
Chromosome translocations	1
Other	2

For the overwhelming majority of clinical geneticists responding to our questionnaire, the primary purpose of a genetic register was to facilitate patient management (Emery's "clinical and therapeutic role"), although one out of 52 thought a register should be regarded only as a research tool. The role of a register as a reference list of diagnostic information for relatives was supported by 46/58 (79%), but there was also strong support for the active role of registers in family follow up. A total of 49/58 (84%) thought registers should be used to recall affected patients for interval clinical screening, and 48/56 (86%) supported recall of patients to update them on new developments. A total of 50/56 (89%) supported the recall of children at risk when they reach the age of maturity (16 years in the UK) to offer genetic counselling.

Although there was no consensus in response to specific questions about whether registers should actively attempt complete ascertainment or rely only on referrals to the genetic service (fig 1), responses to other questions suggested that, in practice, most registers rely on referrals.

Fifty two out of 58 respondents (90%) thought that registers should not be restricted to information about affected patients but should also include information about at risk relatives (53/58 or 91%). Most (51/58 or 88%) believed that registers should record laboratory diagnostic information about affected subjects (for example, mutation results, karyotypes), and similar information about carriers of autosomal or X linked recessive disorders and chromosome rearrangements (48/58 or 83%). There was strong support for recording of identifying information about children at risk of developing genetic disorders (52/57 or 91%), but slightly less support for recording children at risk of being a carrier of a recessive disorder or balanced chromosome rearrangement (39/55 or 71%).

Opinions differed concerning disease specific registers, consent, and continuing care of register families (fig 2). There was no consensus as to whether registers should

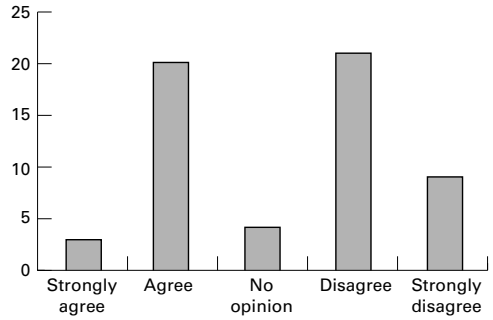

Figure 1 Responses to the statement, "A genetic register should aim for complete ascertainment of genetic disease within the catchment area of the genetic centre".

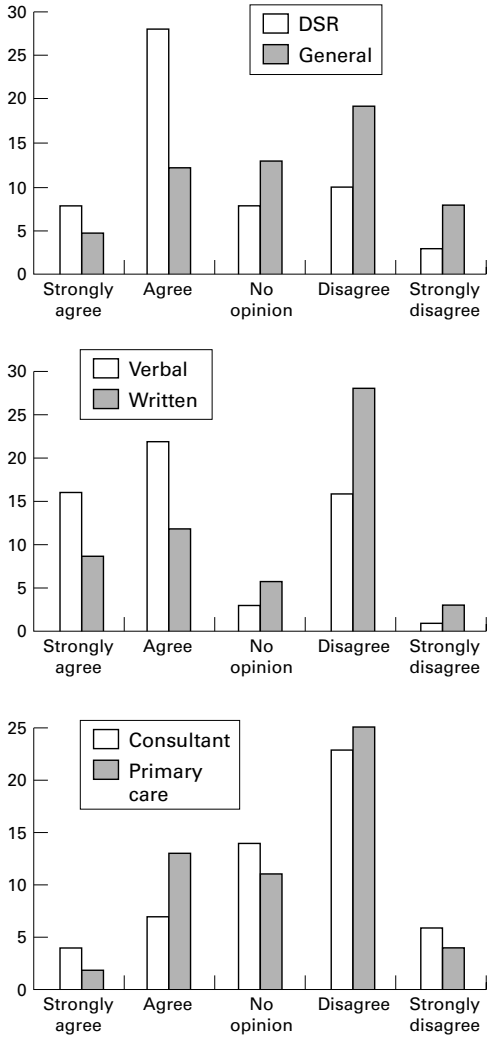

Figure 2 Issues with no consensus. (Top) Should a genetic centre maintain disease specific registers or a more general genetic register? (Middle) Verbal or written consent should be obtained before recording a patient's details on a genetic register. (Bottom) Continuing care of genetic register families should be the responsibility of the clinical genetics consultant or the primary care physician.

record information about one disease only (disease specific registers or DSRs) or whether they should be generic for all genetic disorders referred to the genetics service. There were differences of opinion on the issue of obtaining informed consent. A majority (34/52 or 65%) support seeking verbal consent, but a significant number (16/52 or 31%) oppose this, and the distribution of responses for written consent is more evenly balanced. Having indicated a desire for genetic registers which are involved actively in the management of families with genetic disease, it is interesting to note the dissension from the view that either clinical geneticists or primary care physicians should be mainly responsible for continuing care of such families.

It is clear from the original survey in 1995 that there were at that time considerable differences in resources allocated to operating genetic registers in different centres in the UK, and it seems likely that this would result in different levels of service to patients. The 1997 survey suggests that, in many respects, there is a consensus about genetic register functions, and it seems unlikely that the issues over which there is no agreement (consent, DSRs or generic registers, and responsibility for continuing care) would result in such disparity of resource allocation. The interventionist nature of the genetic register function supported by the UK clinical geneticists raises concerns about the issues for which there is no consensus (such as consent to be included), and in relation to how such functions can reasonably be supported within the resources available to most centres.

From the responses to the second questionnaire in which there was general agreement, it would be quite possible to draw up a specification for a genetic register service that is seen as desirable by the UK clinical genetics community. It is clear that although a genetic register should function as a reference list of family clinical and laboratory information (Emery's second role), it is also thought desirable for it to have a wider function in the organisation of interval review and follow up of family members (Emery's first role). This is to facilitate timely clinical screening and support of those affected by or at risk of genetic disease, to update family members when new information about their family disease becomes available, and to recall children at risk when they reach maturity. This also contributes to Emery's fifth role (prevention of genetic disease). Thus, a genetic register should comprise a list of people affected by, or at risk of genetic disease, linked as families, and linked to a diagnostic index. The register should include facilities to remind clinical genetics staff to consider further contact with the family under a variety of predetermined circumstances, which could include clinical screening protocols, the occurrence of medical advances, or the attainment of a particular age by a family member.

It is not agreed that registers should attempt complete ascertainment (the first part of Emery's fifth role) and it is clear that most registers do not actively pursue this goal in practice. Perhaps this reflects concern about non-directiveness in the application of genetic services, but it may also reflect the way in which genetic services have outgrown their resources in recent years. The issue of whether registers should be disease specific or general is probably relatively unimportant, as it should be possible to devise software which can handle different follow up protocols or review prompts within the same database system, effectively providing disease specific registers within the framework of a general register. The general register approach should reduce the resource implications of genetic registers, as the maintenance of several DSRs can lead to duplication, and reduced efficiency in responding to enquiries, if it is not immediately apparent which DSR might include a particular family's details. The issue of greatest importance is probably that of informed consent, and this is particularly so in the light of the expressed desire to include details of family members (including children) at risk on the genetic register. The problem of responsibility for continuing care follows on from this.

The Nuffield Council Report on genetic screening[8] considered that (living) subjects must give informed consent before their information is stored on a genetic register. We believe that from a legal viewpoint this consent need not be written, although the fact of verbal consent should be recorded for the protection of patient and doctor alike. The need for informed consent has three specific implications. Firstly, to be clinically useful as a reference source of family information, a genetic register should record details of all affected subjects known to the genetic centre. If a person refuses consent to be on a register, or if access to a person to obtain consent is not possible for reasons such as confidentiality, this may create problems in recording information necessary to define follow up arrangements made through a genetic register for his or her relatives. However, it would be unreasonable to expect to obtain consent to record the details of every person mentioned in a pedigree chart which forms part of a conventional paper based genetic record and, by analogy, family history information

which is recorded to inform another person's genetic register record may not require specific consent. Secondly, where a DSR is in use clinically but is derived from a research based register, further consent may be required for this altered use of the recorded data. Thirdly, where a child's details are recorded on the register, a parent or guardian may give informed consent. However, it would seem logical that when a child reaches the age of 16, he or she must give his or her own informed consent for his or her details to remain on the register, unless these details are merely part of the family history information recorded about another person. Therefore, the recording of information about younger family members at risk on the genetic register to facilitate recall when they reach maturity may create a legal obligation to contact these people at the age of 16. It is of course only necessary to take "reasonable steps" to contact the child when he or she reaches maturity, but it would seem important that the information given to parents as part of the consenting process should include details of any intention to make contact when the child reaches 16 years of age. It should be clearly stated that this will be facilitated if the family ensures that the genetic centre is made aware of any change of address.

Informed consent also implies transmission of knowledge about why recall is recommended for a particular disease. Reasons might include the risk of developing the disease in the future, the risk of developing complications (such as cancer in familial cancers, or aortic dilatation in Marfan syndrome), the possibility of preventative screening (for example, mammography, echocardiography), the possibility of predictive genetic testing, or the possibility of transmitting a genetic disorder to offspring. Future genetic and medical interventions might allow the avoidance of some adverse outcomes. The possibilities in all of these areas will vary between disorders, and therefore the case for recall and review may be different for different disorders. Other medical specialties may have active follow up clinics for some disorders in one area of the country, but not in another. It is therefore likely that there will be some variation in the clinical need for this aspect of genetic register function for any particular disorder throughout the country. In order to avoid the problem where some members of a family attend a genetic centre with a register with a review policy, while others from the same family attend a centre whose register does not have such a policy and therefore have false expectations of the service available, it is essential that the information given to the family about the genetic register for the purposes of informed consent should state clearly the intended consequences of recording information on the genetic register, whether follow up is advised, and whether the genetic centre intends to offer active follow up. This could take the form of a supplementary letter to a general information leaflet.

If a genetic register is set up with a review policy for a particular disease, this implies a responsibility for continuing care of a family over time, with the proviso of taking "reasonable steps" to maintain contact as discussed above. Despite the fact that 83-88% of UK clinical geneticists believe that recall of families for updating information or screening is an important function of the genetic register, only 10/35 (29%) believe that this follow up is the responsibility of the clinical geneticist. Interestingly, only a slightly higher proportion believe that it is the responsibility of the general practitioner (15/39 or 38%). North American genetic service providers (physician geneticists, PhD geneticists, and genetic counsellors) expressed a similar opinion in a recent survey, with only 46% agreeing that a "duty to recontact" should be the standard of care.[10] Causing patient anxiety, the burden on staff time, and the fear of litigation were cited as possible burdens of a recontact policy. North American geneticists also considered that primary care physicians could share the responsibility, but might not be very effective. Passing responsibility for recontact to the patient was the most popular option. Unfortunately, our UK questionnaire did not ask as an open question, who should take primary responsibility for maintaining contact. Like our North American colleagues, we believe that some responsibility must devolve on the family. If this is the case, then the issue of proper informed consent becomes even more critical to the satisfactory operation of a genetic register.

The majority of clinical geneticists in the UK regard the provision of recall and review services through a genetic register as an important part of the function of a clinical genetics centre. Provision of services varies throughout the country, partly for historical reasons, and probably partly because of different priorities in the allocation of scarce resources in different regions. Different members of the same family may attend different genetic centres and make false assumptions about clinical genetics services available unless proper information is given about genetic register functions at each centre. This is particularly important

Table 2 Suggested guidelines for the basic operation of a genetic register

1 The genetic register as a reference list	
(a)	The genetic register should contain a reference list of people known to a regional genetics service, linked as families, and linked to a diagnostic index.
(b)	The genetic register should include relevant laboratory information about the families or people recorded (eg mutation or linkage results, karyotypes, biochemical findings). Care must be taken to ensure the accuracy of data recorded.
2 Which family members should be recorded on the genetic register	
(a)	Adults and children affected by disorders with a genetic aetiology.
(b)	Adults at risk of developing a genetic disorder or its complications.
(c)	Children at risk of developing a genetic disorder or its complications.
(d)	Adults who are at risk of transmitting a genetic disorder to their children (eg a carrier of an autosomal or X linked recessive disorder or of a balanced chromosome rearrangement).
(e)	Children who are at risk of transmitting a genetic disorder to their children (eg a carrier of an autosomal or X linked recessive disorder or of a balanced chromosome rearrangement).
3 Review and recall function of the genetic register	
(a)	To prompt recall for review of adults on the genetic register at predetermined intervals, for clinical screening of those at risk of complications of genetic disorders, or to update families on recent medical or scientific developments. The interval set for recall will vary between diseases depending on clinical circumstance and between genetic centres depending on other local service provision.
(b)	To prompt recall for review of children on the genetic register at predetermined intervals, as for adults, or when the child reaches maturity, to offer genetic counselling and further follow up.
4 Informed consent and the genetic register	
(a)	Adults should give informed verbal consent for their details to be recorded on the register. The fact of this consent should be recorded.
(b)	Parents or guardians may give informed consent on behalf of children. Children should be given the opportunity to give or withhold their own consent when they reach maturity.
(c)	The purpose of the register should be explained clearly. It should be made plain whether regular follow up through the genetic register is intended, the frequency of the follow up, and the reason for follow up. It should be made clear how much of the responsibility for facilitating this follow up rests with the family, for example, by informing the genetic centre of changes of address, or by recontacting at defined intervals.
(d)	The use of a printed information sheet is suggested as a reasonable means of fulfilling this part of the process of consent, within the limited resources available to most centres. A tear off consent form could be included as part of this information sheet. A supplementary letter describing aspects of the register specific to the patient and the family disorder may be useful.

where there is an intention to offer genetic counselling to children at risk when they reach maturity. The use of a printed information sheet or letter to facilitate informed consent, including details of services offered for genetic disorders, the follow up intentions of the genetic centre, and the need for family members to keep the genetic register informed of change of address would help to resolve this issue. Based on the findings of our two questionnaires, and consideration of their implications, it is possible to draw up guidance about the minimum genetic register function considered important by UK geneticists and its consequences (table 2). Our questionnaires were addressed only to senior physician geneticists in the UK, but genetic registers are increasingly operated and maintained by genetic nurses or associates, and it would be most important to seek their views on the conclusions and implications of this survey. Further discussions involving physicians, genetic nurses and associates, and the families themselves might help to clarify those issues without consensus. As with all clinical services, genetic register functions should be kept under review as service intentions and practices may change in the light of future clinical and scientific developments.

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JOHN C S DEAN*
DAVID R FITZPATRICK†
PETER A FARNDON‡
HELEN KINGSTON§
DOUGLAS CUSINE¶

**Department of Medical Genetics, Grampian University Hospitals NHS Trust, UK*
†*South East Scotland Clinical Genetics Service, Western General Hospital, Edinburgh, UK*
‡*Clinical Genetics Unit, Birmingham Women's Hospital, Birmingham, UK*
§*Regional Genetics Service, St Mary's Hospital, Manchester, UK*
¶*Department of Law, Aberdeen University, UK*

Correspondence to: Dr Dean, Department of Medical Genetics, Medical School, Foresterhill, Aberdeen AB25 2ZD, UK, j.dean@abdn.ac.uk

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