

Introduction

The Genetic Interest Group (GIG) is a UK-wide alliance of organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders. Each disorder is individually rare, but taken together, one in fifty of the population of the UK are affected by genetic conditions. This equates to approximately 120,000 Scottish citizens who are currently affected and many more family members who are at risk. GIG's primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them. GIG is the only UK organisation of its kind.

Our response to this consultation is focussed upon the needs of our member groups and the patients and families that they represent.

1. Improving your experience of care

Localisation

GIG appreciates the logic behind the proposal to decentralise and localise services, however many of our members with rare long-term genetic conditions require care that can sometimes only be delivered by a specialist service. GIG would be extremely cautious about rare genetic conditions being dealt with solely at a local level, and therefore welcomes the proposal to invest in a transport system for patients who need to travel to specialist centres.

Integrated care for rare genetic disorders

Through our own research with patients¹ GIG has found that one of the major barriers to receiving good care is the lack of communication between various healthcare professionals who may not be based in the same hospital. The issue of having to "continually repeat yourself" to different professionals (who often know less about the rare condition than the patient) is a real one that has an impact on the care patients receive. GIG therefore proposes that integrated care should be provided for patients of complex conditions that require care from more than one of the traditional specialisms of healthcare provision in Scotland.

Integrated care for rare genetic disorders, *why it is necessary?*

Many rare genetic disorders are complex, affecting many of the body's systems. They also can display a wide variation in their presentation, although the vast majority result in severe, chronic ill health, often progressive disability and sometimes (very) premature death.

Families affected by these rare conditions often experience additional problems arising from the organisation and delivery of health services that make access to appropriate health care and support more difficult to secure than it needs to be. As a consequence of the small numbers affected by any one disorder, expertise in the diagnosis and

management of a specific condition is scarce. This factor could be more acute for patients in remote areas who live long distances from centres of expertise. Normal community-based services are often not able to provide speedy, accurate diagnosis or an integrated and appropriate package of intervention and support that will give the best chance for the patient.

Patients with complex multi-system genetic disorders are frequently heavy users of the NHS. For example, patients with Alström's Syndrome (about 35 families known in the UK) require support from ophthalmologists, audiologists, cardiologists, diabetologists, dieticians, endocrinologists and others. At a more common end of the spectrum patients with Neurofibromatosis 1 (NF1) (2100 in Scotland) may need neurologists, ophthalmologists, orthopaedic surgery, cranio-facial surgery, dermatologists, plastic surgery, psychiatry, oncology and educational psychology.

If the services provided are not integrated, optimising the outcome for the patients can be at best difficult – for example, in the case of Alström's Syndrome the proper management of patients' unusual type of diabetes has an impact on the cardiological options. In the case of NF the optic gliomas that sometimes occur behave differently from sporadic optic gliomas in patients without NF, and thus need different management. (The more usual drug treatment does not work. If it is given in error, it is deleterious to the patient's condition and a waste of resources.) Many other similar examples could be provided for other conditions.

Integrated care for rare genetic disorders, *how it can be provided*

As a reflection of the difficulties experienced by families with "their" condition, many patient support groups have invested considerable sums of money (derived from the voluntary fundraising efforts of their members, most of whom are directly affected) in providing a range of services and support. At the most basic level this can be a volunteer helpline dispensing practical advice and information. Other groups have sophisticated networks and employ professional staff. This investment, built on voluntary income, is increasingly proving unsustainable, and many charitably funded services are proving increasingly frail.

Depending on the nature and population frequency of the conditions, integrated care could be provided as follows:

1. **Specialist centres / multidisciplinary clinics** with a variety of expertise to allow patients to see all the specialists relevant to their condition at a single site, on a single day. This system would be mutually beneficial to patients, who will receive a high quality of care on a convenient basis; and to health professionals, especially those with a research interest in the condition. The centres or clinics could be grouped by conditions that require similar specialisms for the rarest of disorders, rather than focusing on one particular condition per clinic.
2. **Genetic Care Coordinators** who would take a key role in helping patients access care e.g. NF, Muscular Dystrophy (MD) and ataxia. The specialist advisor would work closely with specialist healthcare professionals to ensure that patients get appropriate care, and offer emotional and practical support on social and educational issues associated with the disorder. They would become the patient's first port of call, reducing the burden on GPs and health visitors; and they would act as a source of information for healthcare professionals. This system should be funded by the NHS, but in the past patient organisations have stepped in to fill the gap; both the NF Association and the MD Campaign were forced to cut the number of specialist advisers for the UK in the past due to financial difficulty.

3. **Genetic Disease Registries** where a health professional is in charge of recalling patients once (or possibly twice) a year to the genetic centre or multidisciplinary clinic to undertake a similar role to the Genetic Care Coordinators mentioned in point two. Registries are established successfully in various areas of the UK in genetic centres.

No single model will fit all disorders. For some the specialist multi-disciplinary clinic, together with a Genetic Care Coordinator is a good approach. For others this is not practical and the employment only of a Genetic Care Coordinator is more appropriate.

Many of the charitable support groups offer several of the care models. All find funding difficult. The rarity of these disorders, and hence the smallness of each of the charities, presents particular challenges in maintaining continuity and quality of services.

Integrated care for rare genetic disorders, *Evidence from patients and professionals for the benefit of multidisciplinary clinics and specialist advisors from the Usher multidisciplinary clinic*

Usher syndrome is a genetic condition characterised by congenital deafness combined with progressive blindness from childhood or adolescence. Balance problems are also common. It is clearly a severely disabling condition, and as yet there are no cures. Patients need ongoing care and monitoring from audiology, vestibular and ophthalmology specialists. In addition geneticists provide diagnostic and carrier testing, and counselling for patients and their families.

A survey of members of the charity Sense, demonstrated strong support for the establishment of a multidisciplinary clinic, where patients could see all the specialists needed for their condition in a single site, on a single day. The first 'Dual Sensory Clinic' was held in July 2007 in London. This is an excerpt from a patient's account (reproduced with kind permission by Sense):

"Previously, I had undergone vision tests and tests for hearing, and balance separately at different hospitals. ... My first session [at the new clinic] was with Dr Maria Bitner, who is a clinical geneticist. It was at this meeting, which I had been waiting for with much anticipation, that I received the conclusions of the test results and a precise diagnosis of my type of Usher. A discussion then followed into areas of clinical research, support and even possible treatment in the future.

If Dr Bitner did not have an answer to my enquiry, she was happy to refer me immediately to Professor Linda Luxon [audiologist], who was able to provide me with the information that I was seeking. This was a very satisfying and reassuring outcome for me.

Furthermore, Mr Andrew Webster, Head of Retinitis Pigmentosa Clinic, Moorfields, carried out more eye tests, using the latest specialist equipment. I was even offered state-of-the-art hearing and balance tests, which I undertook, to check if any changes had occurred.

I found the medical professionals very helpful, patient and understanding of my needs. They advised me they would contact me for an appointment in a year's time. I found this arrangement very useful, as the thorough ongoing monitoring tests would only take a morning to be completed. All the results and findings will be kept securely in one clinic."

Integrated care for rare genetic disorders, *role of primary care*

While some patients feel that the support they receive from their GPs is helpful, and others would like more support from them, overwhelmingly patients feel that GPs do not currently have the knowledge or expertise to diagnose or manage their conditions¹. One patient (with Gorlin's syndrome), having asked his GP for a referral to clinical genetics, was told "Why do you want to see a geneticist, you know you've got the syndrome". Many people with genetic conditions experience this. It is becoming a more common complaint, as more becomes known about the genetic causes of common conditions, such as heart disease, and cancer. Clinical genetics could have been very useful to the patient in a variety of ways e.g. addressing concerns about the progression of the condition, about other members of the family, including supporting communication within the family, and discussing risks to his potential future children.

GIG members have proposed practice nurses taking on a support role to help navigate patients through the maze of services (not unlike the Genetic Care Coordinator role outlined above) but GPs expressed (understandable) concern about the burden this would place on the already full schedules of practice nurses.

If primary care is to have a greater role in the management of such conditions, there is a clear need for training to support the development of adequate skills, and for capacity building to allow better integration between primary and specialist care.

Integrated care for rare genetic disorders, *supporting care for rare conditions through national networks*

National networks of health professionals with an interest in particular genetic conditions is a vital way for up to date information to be shared between professionals and patients. By involving patient support groups such networks can make a real difference in standards and access to care. GIG sees the encouragement of such networks across Scotland and the UK (and Europe) as a priority, and welcomes the Scottish Executive Health Department (SEHD) funding for the Scottish Muscle Network (SMN). In addition, the SEHD is funding a two-year project, run by GIG, to establish better patient engagement with health services in Scotland. The remit of this project will include supporting and facilitating the SMN and the development of similar networks for other genetic conditions.

Integrated care for rare genetic disorders, *the way forward*

As is clear from work carried out by GIG and other patient groups, that families with rare genetic disorders are already making (entirely legitimate) extensive demands on health services. It is also clear that without coordination and proper commissioning that there is a significant risk that a substantial proportion of the resources being expended will be used at less than optimal effectiveness, with significant health gains for the patient and the family being foregone as a result.

Furthermore, the efforts of patient support groups to help the families they represent to navigate the system to secure the services they need is increasingly vulnerable, making it more likely that in the future the deployment of NHS resources will be less effective than it need be.

We acknowledge important steps taken by the SEHD such as funding the existing Scottish Muscle Network and a two-year award to GIG to promote patient engagement and service development in Scotland. However, to fully address the issues there needs to be a

strategic investment in an appropriate range of models to provide integrated specialist services for families with rare disorders. Such a mechanism would:

- a) deliver integrated, effective and appropriate care to patients.
- b) legitimise trans-speciality collaboration and logical ordering of interventions and support.
- c) incorporate and strengthen appropriate patient support group developed services, thereby enhancing quality and securing continuity.
- d) make good use of scarce NHS expertise and encourage research.
- e) provide baseline robust models for extension to other disease areas.
- f) probably be cost neutral. At worst the extra cost of delivering services properly would be marginal. However it is likely that money would be saved as demands would be met in a logical, timely and coherent manner, which would make best use of professional time and expertise. The mechanism would also properly recognise and value patient time and their input to management of their own condition and incorporate and consolidate the work of patient support groups in robust and accountable structures.

To complement these proposals, and empower patients and their families, GIG is producing a series of “Family Route Maps” to help individuals find the relevant information and services they need.

2. Best Value

In the field of genetic healthcare in Scotland, GIG is concerned about the career advancement opportunities accessible to genetic counsellors. There should be the opportunity for safe, good quality training structures for genetic counsellors. There should also be career development through the establishment of Principal, Consultant and Manager Genetic Counsellor posts. These posts would lead genetic counselling training, education and research to integrate best practice genetic care throughout the Scottish healthcare professions. This allows the best available staff to plan their career, and more importantly stay in the field of genetic healthcare. Without opportunities such as these, Scottish genetic healthcare will lose the resources spent training genetic counsellors as they move to other locations or specialities that offer a more complete career plan.

4. Tackling Health Inequalities

Health inequalities have been a major concern of GIG's for a number of years. The term can be interpreted in a number of ways, but before these are discussed individually it should be noted that patients of rare and/or inherited conditions are dealing with inequalities even before they receive their diagnosis. Rare conditions are not as well understood as more common conditions; patients can often face a battle to get a diagnosis. The complex interacting symptoms presented by rare multifactorial conditions need integrated care, coordinated by a professional who understands the condition. This can often be difficult or impossible to obtain. These problems need to be addressed before patients of rare conditions can possibly receive equitable care. This issue of the difficulty of receiving care of as high a quality as that for common conditions can be relieved by the uptake of multidisciplinary clinics as described in section one of this document.

Inadequate care for patients of rare conditions

GIG would welcome the approach to look at the wider health needs of those with genetic conditions. In recent research¹ carried out by GIG it was extremely clear that patients' psychological needs were often not being met, or even considered as part of their healthcare plan. One participant at the workshop with AMEND (The Association for Multiple Endocrine Neoplasia Disorders) felt that so many family members had been positively diagnosed that it left the participant feeling like the whole family needed counselling, but they were not provided with the psychological counselling that she had hoped to receive.

Difficulties in accessing the benefit system for families with a rare condition

Many focus group participants had enormous difficulties in accessing social care, including Disability Living Allowance (DLA) and this was of major concern. A Mum from one of GIG's telephone interviews said "As a family with a disabled child you're just bottom of the pile". Patients felt let down that they were not given any assistance in finding social support and many of the forms needed to be completed were extremely difficult and many eventually found local voluntary groups to help them complete them in the required format. Having more signposts and awareness of the support that patients can receive would help enormously in tackling some of the health inequalities that are currently present.

Inequalities in health for members of ethnic minority communities

This issue has been a particular concern for GIG and we have undertaken a number of projects to assist ethnic minority communities' access to genetic healthcare.

One of GIG's major projects over the last five years has been The Translation Project, a joint venture between GIG and the London IDEAS Genetic Knowledge Park to translate 38 leaflets into 8 languages. The information that was translated is to be used in genetic clinics. GIG has developed a protocol for the translation of health information for patients which can be used outside the genetic context². Having information in a person's mother tongue is always an advantage when discussing health information as much of the information can be difficult to explain and is complicated to understand, especially when under stress. The guidelines that GIG has produced could be used when translating information in other areas of healthcare in Scotland.

It is important that it is just as easy for members of minority ethnic communities to access healthcare as the rest of the population. The first step in understanding how well an ethnic minority community is accessing healthcare is to record the ethnic origin of patients. GIG has carried out two projects looking at ethnic monitoring in the field of clinical genetics, focussing on facilitating ethnic monitoring and making sure that it is relevant to genetic healthcare delivery. Once ethnic monitoring data is collected however, it should be acted upon. Initiatives should be taken to include ethnic minority communities that are not successfully accessing healthcare.

5. Anticipatory Care and Long Term Conditions

GIG welcomes the development of individual care plans. This would help patients access services, and stipulate how often they should receive them. However, GIG has some reservations regarding the creation of such care plans.

1. What information would be held within such a care plan?
2. Who would have responsibility for monitoring the care plan and updating it?
3. Who would be responsible for the creation of the care plan? Would this be done in conjunction with patients and those with expertise in the field of the condition in question?

Care plans need to be done in a meaningful way, so that they are properly used and are useful to patients and health professionals. The NHS needs to be realistic when producing such care plans and should involve service users and voluntary organisations.

Working with patient support groups is key to identifying the issues that are on the horizon, and that the NHS can plan for and be proactive in tackling. GIG has a part-time Development Officer (gscott@hgu.mrc.ac.uk) based in Edinburgh and would welcome the opportunity to work with the NHS in helping to identify the issues our members face when accessing health and social care services.

Greater capacity needs to be built (including training of staff and expansion of services) for cognitive and psychological therapies. As highlighted in the previous question, GIG has found that the psychological impact of patients diagnosed with genetic conditions are often over-looked or ignored. A phone interviewee for the Family Route Map Project said that “[h]e (the psychiatrist) is horrified by how we have been let down and how alone I am on my own with x (child’s name) ... I am so traumatised by everything I’ve been through with x (child’s name) that’s why I’ve not had any more (children)”

A key theme that has come out of work recently carried out by GIG which could be a way to improve the management of genetic conditions, is for the creation of a “Genetic Care Coordinator” who can help patients co-ordinate their care. With so many appointments and so many hospitals, it is often overwhelming for patients. Concern has been expressed by patients with long-term genetic conditions around the practice of discharge from hospital Out-Patient clinics. Continued surveillance and assessment of the condition is vital for early intervention, halting disease progression and maintaining quality of life. One focus group participant commented “they keep trying to discharge me ... they keep trying to get rid of me.” For many genetic conditions developments in treatment and surveillance is changing and improving rapidly and it can be very difficult for patients to ensure they are receiving the most up to date care. The role of such a Genetic Care Coordinator would be extremely beneficial in improving long term care and treatment for patients.

Another key finding is that patients value the creation of multidisciplinary clinics and find them extremely beneficial to the level of care they receive. The creation of such clinics would have a dual role of reducing the number of appointments patients need to have, and enabling healthcare professionals to exchange and develop knowledge and expertise inter-professionally. Patients care can also be monitored by the specialists on a more regular and co-ordinated basis.

7. Continuous improvement in healthcare

Whole journey time

GIG welcomes reductions in waiting times for patients. However, the 18-week “referral to treatment” measurement is a difficult concept to fit to clinical genetics services which are specialised and most often not part of a pathway leading to definitive treatment. This is still an issue under negotiation between genetic services in England and the Department of Health (DH). A recently published “frequently asked questions” document from DH notes:

“The time taken [between referral and first appointment] is dependent on the patient to supply information and consent of relatives followed by enquiries to other agencies such as other hospitals looking after relatives, death certificates etc. The time taken to appointment is not in the control of the clinical genetics professional and it is not until after assessment that it is known whether a referral is indeed appropriate.

“In general, genetic services are most often not part of a pathway leading to definitive treatment. It will be in contact with the service for a lengthy period, often for years. In this respect, the service is most often like those for chronic conditions. By definition, these are life-long conditions/risks, although contact with the genetic service may be required at specific points only.

“Receipt of first definitive advice from a consultant geneticist may reasonably stop the clock if treatment by the genetic service (e.g. counselling) is not required and if the original referral was direct to the consultant geneticist; however, this rule will be kept under review as genetics services and treatments develop.

“Consultant referrals for new conditions identified as the result of a genetic test will start a new 18-week clock.”

With any waiting time target, whatever the limit is set at, it is important to keep in mind the familial nature of genetic conditions and that once a diagnosis is confirmed in an individual, it may well then be necessary to consult with other family members. Also it is important to consider the tests that need to be carried out and the time it takes for this to happen. Unlike other areas of medicine, genetic care often involves a prolonged period of time of investigation. As laboratory diagnostics improve, so the time needed lessens, however it can still be some time before results return to the clinician.

A concern that GIG would have with regarding to waiting times is that patients may be rushed through the consultation with a clinician who is not familiar with their condition, rather than allowing patients to wait slightly longer to see someone with expertise and experience in particular conditions who would be able to provide more appropriate information and help.

In general GIG believes that there should be parity between the health services in the UK. All services should strive to reach their best possible whole-journey-time/waiting-time target; appropriate quality standards should be developed and introduced for Scottish genetic services to ensure improvements continue to be made.

GIG will be happy to comment further, in person, or by correspondence, on any part of this consultation.



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¹ "Family Route Map Project – a report of a series of six focus groups, March 2007"
http://www.gig.org.uk/docs/FocusGroupReport_final_colour.pdf

² <http://www.gig.org.uk/docs/promotingaccess.pdf>