



The Family Route Map Project

Finding a way through the maze



Report of the development of a tool to help individuals and families with genetic conditions together with their carers and healthcare professionals to find current services and information

Project Team:

Anna Allford Genetic Interest Group
Melissa Hillier Genetic Interest Group

Advisory Group:

Dr Paul Brennan, Clinical Geneticist, Northern Genetics Service, Newcastle upon Tyne.

Dr Ian Ellis, Clinical Geneticist, Clinical Genetics Service, Liverpool Women's Hospital.

Dr Mike Knapton, GP, Cambridge and Clinical Director for The British Heart Foundation, London.

Steve Potter, Therapy Liaison Manager, Genzyme Therapeutics Ltd.

Acknowledgements

We would like to thank all those who took part in the focus groups and interviews and for the help and support of AMEND (Association for Multiple Endocrine Neoplasia Disorders), Barth Syndrome Trust, Gorlin Syndrome Group, Myotonic Dystrophy Support Group, Nail Patella Syndrome (UK), and Syndromes without a name (SWAN).

Special thanks to our Volunteer, Nicki Taverner, Genetic Counsellor, West Midlands Regional Genetics Service, who analysed the data from the online questionnaire.

The Family Route Maps project is funded by the charity Jeans for Genes and an unrestricted educational grant from Genzyme Therapeutics Ltd.

All photographs courtesy of FLICKR www.flickr.com under the Creative Commons License Agreement
Front cover image www.flickr.com/photos/sidelong/234192777

First published February 2008
Genetic Interest Group.

Copies available from: www.gig.org.uk/familyroutemap.htm

Genetic Interest Group (GIG)
Unit 4D, Leroy House
436 Essex Road, London, N1 3QP
Phone: 0207 7043141
www.gig.org.uk
Registered Charity No 1114195
Company Registered by Guarantee 05772999

Contents

Foreword		5
Executive Summary		6
Chapter 1	Introduction	11
	1.1 Background	11
	1.2 Project aims	11
	1.3 Methods	13
Chapter 2	Results	14
	2.1 Information	17
	2.2 Communication	18
	2.3 Diagnosis	19
	2.4 Treatment and Surveillance	20
	2.5 Education of Healthcare Professionals	23
	2.6 Ethical, Legal & Social Issues	25
	2.7 Empowerment of patients, parents and carers	26
Chapter 3	Conclusions	28
Chapter 4	Recommendations	30
Chapter 5	Evaluation	31
References		36
Addendum		37
List of Tables		
Figure 1	Schematic representation of the seven emergent themes	9
Figure 2	Objectives for the Family Route Map Project	12
Figure 3	Stages of the Project	13
Figure 4	Topics explored in the patient focus groups	14
Figure 5	Emergent Themes from Patient Focus Groups	16
Figure 6	Causes of Low Patient/Parent Satisfaction	17
Figure 7	Current surveillance of the conditions - online survey results	22
Appendices	1. Summary Protocol Flow Chart for the Family Route Map Project	37
	2. GP focus group vignettes	38
	3. Contact details for Patient Support Organisations	39
Glossary		40
Family Route Maps	Multiple Endocrine Neoplasia Disorders Barth Syndrome Gorlin Syndrome Myotonic Dystrophy Nail Patella Syndrome SWAN (Syndromes without a name)	43



Photo: www.flickr.com/photos/incendiarymind/459989142

Foreword

This is the Internet Age with 24 hour Broadband at our fingertips. We should be able to gain instant Wi-Fi access to information anywhere and on an unprecedented scale. Yet all too often we still hear of the difficulties that people with rare conditions experience in getting access to expert advice and specialist treatment. Of course if you know the right questions to ask, you can 'Google' your way through the many pitfalls that families report to their specialist support groups. But that person has to be resourceful and motivated. Donald Rumsfeld's comments in 2002 have never been truer; "...we know there are some things we do not know. But there are also unknown unknowns - the ones we don't know we don't know."

The Genetic Interest Group (GIG) is a national alliance of patient organisations with a membership of over 130 charities that supports children, families and individuals affected by genetic disorders. As part of their continuing work to promote awareness and understanding of genetic disorders, GIG has undertaken the Family Route Map Project. This project has sought the personal experience of families with a number of rare disorders as they navigated through the maze of information and services. Not surprisingly they found that families with rare (and often genetic) conditions struggle to find an appropriate expert or specialist team to assess and advise them. There are recurrent themes: *why did it take us so long to get here?; why didn't we meet someone who knew about us earlier?; why have we been left and lost in the system for so long?* Too often, the story is of people falling at the many hurdles and being diverted into the many byways that people encounter in a complex system like the NHS.

These conditions might be rare individually but collectively there are many hundreds. Together they are common across the population. It may be years or even a lifetime before a generalist clinician encounters some of these conditions. They may be rare - and therefore only seldom seen by healthcare professionals - but they affect patients and families living with them. The greatest challenge with rare disorders is recognising clues. All healthcare professionals should at least know where to refer a person. This is often through a Clinical Genetics Service with the necessary skills to access expertise and specialist clinical care at a national or international level. The model emerging is a hub and spoke approach with local clinicians and primary care supported by a national expert or specialist centre. This GIG project has produced sample Route Maps for selected conditions that indicate general themes that can be applied to all rare disorders. This template is now being made widely available by GIG.

It remains to be determined how much information and support should come from primary care and how much should come from the expert in the specialist clinic. We also need to think about how to recognise expertise. Many patients are unsure of what treatment and surveillance they should have. Perhaps this is due to reluctance on the part of some healthcare professionals to ask for the advice of experts as they may want to manage the condition themselves, or a mistaken concern to protect the family from a long distance journey. People will travel for expert advice, but how far and how often needs a flexible approach taking account of how many experts there are around the country. The patient may be managed in a satellite clinic, empowered by the Route Map and their own *Personal Passport* for that condition in a national form of shared care that we are so familiar with in the ante-natal clinic setting.

The Route Map Project does not advocate the perceived American model that says that more is always better. Rather, GIG argues for tailored care with evidence-based clinical pathways and above all it emphasizes the need to empower the patient and family. Recurring themes are highlighted in the outcome of the Route Maps project: financial help, psychological help, information, education etc. A conclusion to draw might be the creation of a new medical specialty of Genetic Medicine. It might not be offered in all hospitals, but it would benefit from being designated as a National Service Framework (NSFs). This could develop from such Route Maps, perhaps with its own National Clinical Director for Genetics Services. In this way genetic diagnosis and initial counselling could be better linked to ongoing support, treatment and screening for the increasing number of genetic conditions for which intervention can be offered.

Despite all the technology available, key emergent themes remain: good communication; sympathetic and timely listening; people acknowledging their limitations and not being afraid to refer to other clinicians despite the increasing complexity of commissioning in the current NHS. We should use all the technology to empower patients and support them on their journey which hopefully will become shorter and more direct.

Advisory Group

Executive Summary

Background

With advances in genetics knowledge, increasing numbers of rare genetic conditions are being identified. Currently there are about 4000 known genetic disorders with more being discovered. 1 in 18 people have a disease with a major genetic component and if congenital anomalies were included then 79 per 1000 live born individuals have been identified as having a genetic condition before the age of 25 years¹. Genetic conditions may manifest throughout life and often affect a number of systems within the body, so patients need to see healthcare professionals from several different specialties during their lifespan. The clinical management of these conditions may be quite complicated and there is no standardisation of management within the National Health Service (NHS), with different specialties dealing with different genetic conditions in different ways.

“These very rare diseases’ make patients and their families particularly isolated and vulnerable.”

Rare Diseases: Understanding this Public Health Priority. Eurordis, November 2005

The Genetic Interest Group (GIG) is a national alliance of patient organisations with a membership of over 130 charities which support children, families and individuals affected by genetic disorders. GIG's primary goal is to promote awareness and understanding of genetic disorders so that high quality services are developed and made available to all who need them. In 2006 GIG undertook a survey of its members in response to the call for feedback from the Department of Health (DH) on the effects of services for patients since the introduction of the White Paper “Our Inheritance, Our Future”². GIG analysed the findings and highlighted 10 key points to the DH³, three of these are of particular relevance to this project:

- Barriers to effective communication between healthcare professionals within the NHS, and with other agencies was considered detrimental to the care of patients with genetic conditions.
- There is a need for a more coordinated and integrated approach to care provided by the NHS for these patients, their families and carers.
- Primary Care is still by and large unaware of the impact of genetic disease on families.

The need for clear 'signposting' to information and services and a resource that could be shared with healthcare professionals strengthened the project's goal and underpinned the discussions with patients, families

and carers, and clinical staff with expertise and experience in six specific conditions; Barth syndrome, Gorlin syndrome, Multiple Endocrine Neoplasia Disorders (MEN), Myotonic Dystrophy, Nail Patella syndrome and Syndromes Without A Nameⁱ.

Some of these six conditions may present in infants or children, whereas for others the onset is adolescence or adulthood and many patients are seen by a number of specialties before the diagnosis is made. In primary care the majority of GPs feel comfortable with their gatekeeper role and only a few are willing to become more involved in genetic risk assessment and counselling, however most have received no formal education in genetics and many are unaware of what services are provided locally⁴. This makes it difficult for patients with rare genetic disorders to receive information, support and care, and many find similar issues in secondary and even tertiary care with only a few clinicians having experience and expertise in their condition.

Project details

Developing Family Route Maps as a Tool to help families with genetic conditions to access appropriate information and services in the UK was the primary objective for this groundbreaking and unique project. It was agreed that listening to the concerns of patients, families and carers was essential. Workshops were held for each condition with the aim to explore information and services currently available to these families as the first stage in the development of the Route Maps and also to ask them what they would like to see included. Additionally, an on-line questionnaire was made available through the GIG website with links to the websites of the six Patient Support Organisations, and interviews with clinicians and other healthcare professionals with expertise and experience in these conditions were undertaken. In primary care in the UK we know very little, from the point of view of the patient, about caring for patients with rare genetic disorders and therefore primary care services were talked about in all of the patient focus groups. A focus group with General Practitioners (GPs) discussed the concerns and issues raised in the patient focus groups through a series of vignettes presented for discussion. From this collective information a template for a Family Route Map that could be used generically by other Patient Support Organisations was also created.

Seven important over-arching themes were identified from the qualitative data with a number of sub-categories and these are discussed in this report in relation to

ⁱ Syndromes without a name is a term used by the charity to describe infants and children with complex multi system congenital disorders which remain undiagnosed.

current care and future possible developments. The seven themes are:-

- Information;
- Communication;
- Diagnosis;
- Treatment and Surveillance;
- Education of Healthcare Professionals;
- Ethical, Legal and Social Issues; and
- Empowerment of patients, parents and carers.

Overcoming barriers

The patients, parents and carers involved in this project were by and large well informed as many had already been in touch with Patient Support Organisations and some were themselves involved in providing support to others within one of the six charities representing the chosen conditions. However, it should be noted that during the course of the focus group discussions, people were actively exchanging relevant information and peer-learning took place. Newly diagnosed patients, the less articulate, less forthright and less resilient patients have an incredibly difficult time trying to get what they need or in some cases remain ignorant of the care they should be receiving because the current system of health and care services doesn't proactively seek to provide information in a timely and comprehensible fashion. Instead many people need to ask the right questions or for some finding relevant information is left to chance.

"It was only by chance that we came across the [Support] Group...it's really a case of who you know."
(Gorlin Syndrome focus group)

Findings from this project suggest that patients with rare genetic disorders are not given sufficient information about their condition, services are considered 'patchy' and some families are still not aware of, or accessing, NHS Clinical Genetic Services. Many patients report: delays in being diagnosed; difficulties accessing the treatment and surveillance they need due to a lack of knowledge about rare genetic conditions in the medical profession and to an absence of coordination and continuity of care; and little psychosocial support leaving patients and families frustrated and 'stuck' in the system.

"For the first year of his life [referring to son] I felt really isolated." (SWAN focus group)

Discussions between healthcare professionals, and individuals and their families about genetics-related information crucial for informed decision-making are often ad-hoc or absent, as those affected by genetic conditions move through their life-stages. Information through Patient Support Groups and the media (including the internet) fulfilled some of their needs and helped them ask questions of professionals in order to make

informed choice which is core to service delivery in the NHS^{5,6}.

The development of Family Route Maps will give patients access to information that they may otherwise have had to wait months or even years for, due to the rarity of the conditions and the inconsistent information that is available. The Route Maps will also help the health professionals who work with these families to explain all the treatment options, where to go for further information and which other disciplines the condition requires contact with. We anticipate that the Family Route Maps will also help to educate patients and professionals and will raise awareness of the complexity of these conditions and what patients may need to know.

Medical issues

A recent European Commission Public Consultation Communication 'Rare Diseases: Europe's Challenges' highlighted that *"The lack of specific health policies for rare diseases and the scarcity of the expertise, translate into delayed diagnosis and difficult access to care."* Patients do not complain about their genes as they might their joints or chest for example. They complain of certain signs and symptoms that need to be recognised as part of a rare disorder and patterns need to be drawn across the family to recognise its inherited or familial basis. In this project only 2 people out of 57 patient focus group participants and 3 phone interviewees, said they were satisfied with the treatment and surveillance they or their affected family member(s) received. Even where guidelines and consensus documents currently exist, for example, guidelines for Multiple Endocrine Neoplasia Disorders Type 1 and 2, Myotonic Dystrophy Care Card and Care Pathway, and Gorlin syndrome NICE Guidelines, there are still patients who are not receiving surveillance or treatment in accordance with those guidelines. The low incidence of each of these conditions means that there are few people living in a local geographical community at any one time. The majority of people in this project said they already travel, or are prepared to travel, to see clinicians who are experts in the field.

"Thankfully I have now found an oral surgeon and dermatologist who have a broad knowledge of the condition [Gorlin syndrome]. I would gladly travel 100+ miles to see them." (Online Survey respondent)

Many people with rare genetic conditions feel they receive fragmented care for their condition and would prefer to have one lead clinician acting in a coordinating role, responsible for their care, a recent finding also of the NHS National Genetics Education and Development Centre⁷. Those patients and their families or carers involved in the Family Route Map project mentioned the desire for a 'champion'; someone with the necessary knowledge and skills who could coordinate their care, preferably within a 'Centre of Excellence'. Due to the

lack of knowledge about the condition and to the necessity of being seen by several different specialties, patients and/or their relatives often have to coordinate their care, ensuring that they are referred to all of the appropriate specialists for the necessary surveillance and treatment. This is a significant undertaking in terms of time and effort, especially as patients are unlikely to be able to self-refer, and it would be helpful for families if they had a Champion who could help. This could be a clinician with knowledge of the condition who acts as a central reference point, or could equally be an advocate who is not necessarily involved in care or knowledgeable about the condition, but could fill this coordinating role.

"I think we all need an advocate." (Myotonic Dystrophy focus group)

Importantly, managing the psychological aspect of having a long-term genetic condition was often felt to be neglected and many people wanted support and guidance to access appropriate services. In 2007 a report by Picker Institute⁸ concluded that there is a lack of effective signposting to information about health and social care services. Raising awareness and disseminating the findings of the project together with the Family Route Maps will be key to improvements for patients, their families and carers. The six condition-specific Family Route Maps for individuals identified with or at risk of those conditions will be available from the Patient Support Groups, and those clinicians with an interest plus Regional Genetics Services. Helping to educate all healthcare professionals about these conditions is no less important but it is recognised that this will require additional targeted resources from within the medical, nursing and associated healthcare professions to ensure genetics is firmly embedded in their practice.

Primary Care

The Healthcare Commission in 2006 found that patients in England with long standing conditions affecting their health or those who have a disability respond much more negatively when asked about their experience of primary care services than people who describe themselves as being in reasonably good health⁹. In this project, patients with these six complex rare genetic conditions and their relatives feel that primary care currently does not adequately meet their needs. GPs were felt to lack relevant knowledge and skills to manage these patients as little is still known about most of these conditions. It was agreed by participants in the patient/family focus groups that consensus documents for clinical management developed by experts and made available to GPs and other healthcare professionals would be useful. Some participants felt that the support of their GPs was helpful and others would have liked more support from them, however, it was also felt that it is asking too much of GPs to manage rare genetic

conditions due to their already heavy workload. Practice nurses were perceived as friendly and supportive by patients and families and some patient focus group participants felt that with enhanced training they could offer a vital link at GP surgeries and practice-based clinics.

Participants in the GP focus group were well informed of the risk factors and protocols for referral to a Clinical Genetics Unit for more common genetic cancers but for rare genetic disorders it appears less clear. They demonstrated a large degree of congruence between what patients want from primary care and how doctors wish to deal with their concerns and issues, albeit, there remains a need for further information about rare genetic conditions to be easily accessible by primary care practitioners. GPs confirmed that practice nurses already help to identify the needs of individuals and families with chronic illness on an ad-hoc basis and also through the clinics they hold but time constraints currently prohibit more support. Primary care occupies a unique position in the community whereby healthcare professionals often see other family members and as such could facilitate early detection of these less common conditions.

Conclusion

The overwhelming message from people was that they want relevant up-to-date information and more support to enable them to understand their condition and actively be involved in their care. Healthcare professionals with expertise and experience in these rare genetic conditions seek to facilitate this and many are enthusiastically in an advisory capacity with the relevant patient support groups.

The project team postulates that the ability of the patient, their parents or carers, to feel empowered and actively take a role in managing their condition and be involved in shared decision-making is dependent on satisfactorily fulfilling their need for open discussion and the resolution of the issues and concerns expressed in this report, namely:

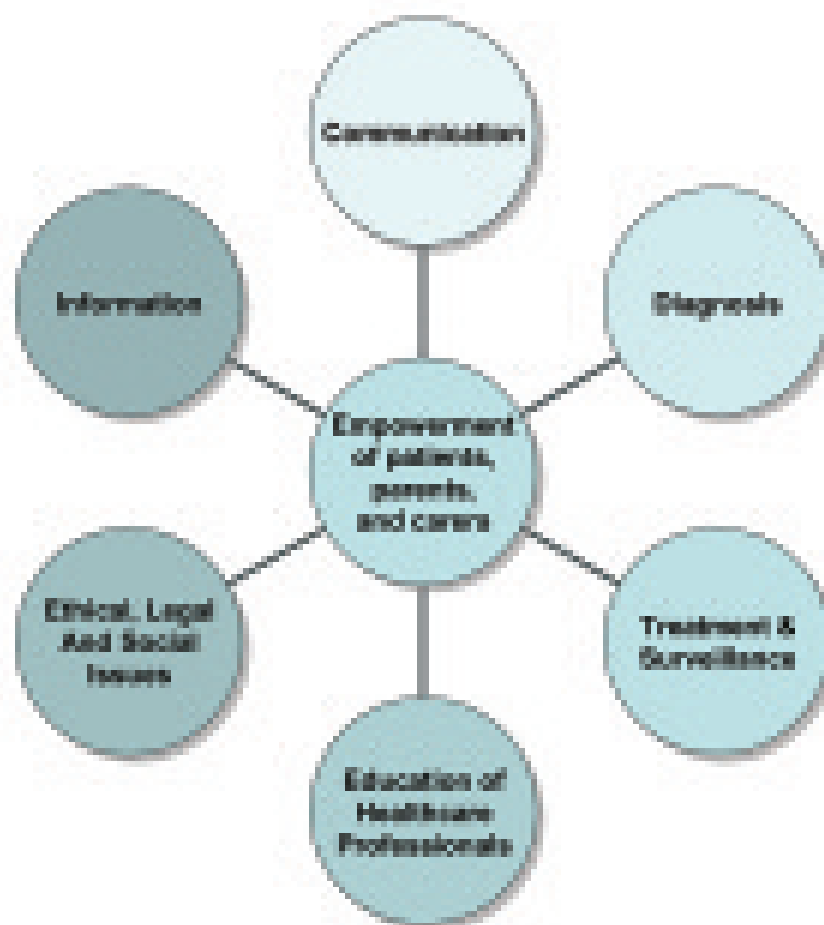
- People want to receive up-to date quality information;
- Good communication between patients and their healthcare professionals and also between professionals involved in their care is vital;
- Early and correct diagnosis could potentially offer improved health and quality of life for people living with long-term genetic conditions;
- Appropriate treatment and surveillance is essential for patients and families with genetic conditions;
- Healthcare professionals require more training in genetics and its implications for families and

- should be willing to learn jointly with patients about their specific condition;
- Knowledge and understanding of the ethical, legal and social issues of living with a genetic condition are crucial for people to make decisions.

It may be useful to conceptualize the seven themes identified in this report using a diagram (Figure 1) which represents 'empowerment of patients and their families together with parents and carers' at the centre of a hub. Patients and families require all of the above conditions to be met if they are to feel empowered otherwise they become frustrated with the system and feel 'stuck'. Fulfilling the need for discussion and resolution of the issues and concerns expressed in this report will hopefully enable people to be truly empowered and actively involved in their care with an appropriate degree of responsibility for shared decision-making with their professionals.

Family Route Maps could provide the basis for on-going dialogue and shared decision-making, and serve as a platform for facilitating better coordination and continuity

Figure 1. Schematic representation of the seven emergent themes



of care. Different approaches to provision of care, including clearer guidelines about clinical management and a clinical champion are discussed in this report. Greater awareness of rare genetic conditions and the service provided by clinical genetics departments within the medical profession was called for and some participants felt the role of clinical genetics could in some cases be extended to that of 'champion', coordinating care.

This project has not only helped patients with the six condition groups, their families and carers but it will have a much wider impact as this report provides guidance for other patient support groups to use to develop their own Route Maps should they wish to carry out a similar project. A generic template is also available from GIG as a Microsoft Word or Publisher document.

It is anticipated that this report and the findings of the project will be of value to healthcare professionals and those providing training to them as well as individuals with rare genetic conditions and the Patient Support Organisations representing them. It is also anticipated that it will be a resource for Commissioners of services, policy makers and others interested in the views and experiences of patients, their families and carers based on the health and care services they receive.

Recommendations

- Up-to-date training and education in genetics for all healthcare professionals should be a requirement for their continuing professional development.
- Greater awareness of rare genetic conditions amongst the medical profession could lead to earlier diagnosis for patients with rare genetic conditions.
- Better communication between patients and professionals and also between professionals involved in their care would enable patients to make informed choice.
- A referral to the Clinical Genetics Service should be offered to all patients.
- Clearer guidelines for treatment and surveillance of rare genetic conditions are required.
- Coordinated care within a multidisciplinary team approach model of care preferably at a 'Centre of Excellence' is called for by patients to counter fragmentation across organisational boundaries.
- Resources should be made available for Centres of Excellence or Networks of Expertise to offer services to patients within rare condition groups in order to ensure equitable access to care.
- Provision of counselling and psychosocial care should be made available to all who request it.
- In primary care, practice nurses could be a resource for reliable sources of information for patients and families and provide continuity of care with much needed psychological support.



www.flickr.com/photos/97445131@N00/250736277

Introduction

Chapter One

1.1 Background

*"...rare diseases are characterised by the large number and broad diversity of disorders and symptoms that vary not only from disease to disease, but also within the same disease. The same condition can have very different clinical manifestations from one person affected to the other. For many disorders, there is a broad diversity of subtypes of the same disease."*¹⁰

The Family Route Map project commenced in April 2006 working with Patient Support Groups representing the following six genetic conditions together with patients, their families and carers who have experience of one of these conditions;

- Barth Syndrome,
- Gorlin Syndrome,
- Multiple Endocrine Neoplasia Disorders,
- Myotonic Dystrophy,
- Nail Patella Syndrome, and
- Syndromes without a name.

The GIG member charities invited to take part were selected based on their size, the rarity of the condition and the fact that the majority have scarce resources with many being run by volunteers in their own homes. Consideration was also given to the need for a sustainable continued working partnership over the project period of two years between GIG and the Patient Support Groups and expressions of interest were invited from a number that fitted the criteria.

AMEND, the national charity committed to supporting individuals and families affected by Multiple Endocrine Neoplasia Disorders (MEN) in which several endocrine glands develop non-cancerous (benign) or cancerous (malignant) tumours or grow excessively without forming tumours, has about 90 patients as members. The Barth Syndrome Trust in the UK supports the families of boys affected by this very rare condition characterized in males only by; cardiomyopathy, neutropaenia, muscle weakness and general fatigue, and growth delay. The charity has 12 families as members. The Gorlin Support Group, supporting the needs of people with Nevoid Basal Cell Carcinoma Syndrome which gives rise to multiple jaw cysts, and /or basal cell carcinomas, has around 380 families as members. The Myotonic Dystrophy Support Group, with around 460 members, serves patients with this most common and variable form of muscular dystrophy, together with their families and carers. Nail Patella Syndrome UK, registered as a charity in 2003 to support people with this condition affecting nails, knees and elbows plus other body systems such as kidneys and eyes, has around 160 families as members. The

charity SWAN, with around 1000 member families in the UK, supports families of children who have congenital conditions requiring care for their complex needs but these children do not yet have a diagnosis. It is therefore possible that these 1000 families may potentially represent 1000 separate rare conditions. It should be noted that it is likely that many clinicians will seldom or never have seen any of the above rare genetic conditions.

1.2 Project aims

The primary aim of the Family Route Map Project is to help patients and families to access services and information that is currently available, in partnership with the relevant Patient Support Organisations and healthcare professionals with expertise and experience in each of the six conditions (Figure 2). Key to this has been gathering and collating information and the experiences of patients and families or carers, and health professionals. Thereby, uncovering what people with long-term genetic conditions find important and need to aid discussion around decision-making and to be involved in managing their condition.

Secondary to this it was the requirement to develop a template for a generic Family Route Map and publish and disseminate the findings, materials, and learning at the end of the project. The generic template is available free from GIG for any patient support organisation to use to help develop a Family Route Map for the condition(s) within their own membership. A Family Route Map has been produced for each of the six conditions in this project and these serve as examples of how the relevant issues for each of the conditions have been identified and addressed within the seven main themes that emerged from the data.

Additionally, creating opportunities to raise awareness amongst healthcare professionals has been core to the project. The project team have written articles and Abstracts for publication and given spoken presentations.

- Association of Genetic Nurses & Counsellors - article published in the Newsletter of the British Society for Human Genetics, June 2007

Poster presentations;

- The ESHG 2007 Conference in Nice - June 07
- British Society for Human Genetics Conference in York - September 07
- Royal College of General Practitioners in Edinburgh - October 07 (2 posters)
- European Conference on Rare Diseases, Lisbon - November 07

Spoken presentations;

- Eurogentest Conference, Oporto September 06
- Eurogentest Conference, Milan October 06

Figure 2 Objectives for the Family Route Map Project

Objective	Main criteria	Detailed Criteria
<p>Objective 1:</p> <p>For GIG and contacts of Patient Support Organisations to work in partnership to determine current care and information and what patients, families and carers would like to see included in a Family Route Map.</p> <p>This was to be assessed by considering:</p> <ul style="list-style-type: none"> - the range of people / organisations involved, and - the extent to which the process enabled those involved to work in partnership. 	<p>Range of people:</p> <ul style="list-style-type: none"> - Reached the number of participants / organisations as specified [target numbers set] - Achieved a sample to provide findings representative of general patient group opinion - Involved groups likely to be disproportionately affected by changes (e.g. those with poor health, less articulate, poor access to health services, heavy users of services, carers). <p>Working in partnership:</p> <ul style="list-style-type: none"> - Transparency and lack of bias in determining what issues and concerns are of importance to people. - Iteration to allow development of ideas from people to influence the design of the tool and project findings - Integration of results from patients, families and carers together with healthcare professionals processes (including appropriate timing for assimilation of results). 	<p>Range of people:</p> <p>The process was expected to be:</p> <ul style="list-style-type: none"> - inclusive - involve groups disproportionately affected - representative <p>Working in partnership:</p> <ul style="list-style-type: none"> - Process to be open and transparent - Participants' belief that the results of the exercise will be influential - Participants' belief that the results reflect the discussions held - Those involved have a shared understanding of the task they are engaged in - Participants have the opportunity to learn from each other - Participants have sufficient information or resources to enter into the partnership.
<p>Objective 2:</p> <p>To produce a tool to help signpost patients and families with, or at risk of, the six genetic conditions to current information and services plus a generic template for other Patient Support Organisations and a report to help them develop their own Family Route Map if they wish.</p> <p>This was to be assessed by considering:</p> <ul style="list-style-type: none"> - completion of the targets for developing and reviewing the Family Route Maps, and - measuring the level of satisfaction with each Route Map by the relevant patient/family and clinical Reviewers. 	<p>Development and review:</p> <ul style="list-style-type: none"> - The generic template was to be developed based on the results of data gathered. - Targets were set to roll out the development of each condition specific Route Map in stages, based on the generic template. - Reviews were planned for each of the stages. <p>Measuring satisfaction with the tool:</p> <p>No criteria were specified initially, but questions were later developed for surveys which explored these issues, as follows:</p> <ul style="list-style-type: none"> - Assessment of the quality of the content of the Family Route Maps. - Assessment of the relevance and usefulness of the tool. 	<p>Development and review:</p> <p>Regular reports and meetings were used to advise of activity against targets.</p> <ul style="list-style-type: none"> - Monthly reports to GIG. - Articles in GIG Today the quarterly magazine of GIG. - Reports to, and six monthly meetings with the Advisory Group. - Regular reports to funders. <p>Measuring satisfaction with the tool:</p> <p>Data was obtained through surveys.</p>
<p>Implicit Objective:</p> <p>To publish the six condition-specific Route Maps and widely disseminate the findings of the Family Route Map Project together with a generic template and report.</p>		<p>In practice, the communication activities focused less on gaining general coverage but rather to:</p> <ul style="list-style-type: none"> - develop and disseminate consistent messages to those with an interest in the areas of health and healthcare education - widening the reach of the Final Report by officially launching it at an event to reach the target groups; patients, Support Groups, health and care professionals, government and policy makers. <p>These therefore became the criteria against which activities and achievements were assessed.</p>

1.3 Methods

Three stages for the project were planned; Gathering the information, Developing the Family Route Maps, and Disseminating the information and learning. These are summarized in Figure 3 and discussed further under the relevant section headings. Qualitative research methods in healthcare are used widely to provide an iterative

approach and allow for novel theories to be formulated from the interpretation of the findings¹¹. Focus groups and supplemental interviews to generate data were identified as providing the in-depth and rich narrative that would determine peoples' perceptions of their own experiences with patients belonging to the six member support groups of GIG.

Figure 3 - Stages of the Project

Stage One Gathering the Information
Visits to Patient Support Group contacts and leaflet produced for each group about the project
Publicise the project and how to get involved to Support Group members through the leaflet and regular articles in GIG Today the quarterly magazine to all member organisations
Interviews with healthcare professionals
Focus groups organised with patients & families for each condition
Invitations sent out through the Support Groups for the patient focus groups
Patient/families focus groups and supplemental phone interviews held plus results analysed
Online survey developed with the help of the support groups to widen participation
Survey posted to GIG website with a link to Support Group websites and results analysed
A focus group for General Practitioners (GPs) organised in response to issues raised by patient focus group participants
Focus group for GPs held and results analysed
Stage Two Developing the Family Route Maps
A generic Family Route Map framework developed using emerging themes from data analysis.
Six condition-specific Family Route Maps developed with the Support Groups
First draft of each condition-specific Route Map sent to Reviewers with Quality questionnaire
Reports of patient/families and GP focus groups plus online survey published to GIG website
Results of Review of draft Route Maps analysed and revisions made to Route Maps
Final draft of each condition-specific Route Map sent to Reviewers with Pilot questionnaire
Final amendments made to Family Route Maps and printed
Stage Three Disseminating the Information and Learning
Articles written for BSHG, GIG Today newsletter, Support Group newsletters and abstracts presented on the project as Posters at UK and European Conferences
First draft of final Report written and circulated to Project Team
Revisions made to first draft and Final Report completed and printed
Invitations designed and sent out for the official launch and workshop of the project findings arranged at The Wellcome Conference Centre, London
Family Route Maps and Final Report launched and disseminated to healthcare professionals, Patients Support Groups, Department of Health, and NHS and Care Services
Articles proposed for medical journals and popular press to raise awareness of project

Results

Chapter Two

Gathering the information:

There were four components to gathering views and opinion:

1. Patient/family focus groups and interviews.
2. Patient/family on-line survey.
3. Healthcare professional discussions.
4. GP focus group.

A summary protocol flowchart for the project was produced (see Appendix 1) and a decision taken to gather data from the Patient Support Groups regarding the genetic condition, number of members, patient information, and clinicians' involvement in the group etc. It was important to identify early on 'supporters' for the project, in particular those health care professionals with an interest in the disorder, in order to ensure that the Family Route Maps, once completed, would be of use to patients and professionals and fulfill their function. Therefore the project leads arranged a series of visits to each contact for the chosen Support Groups and collated additional background information found on the Contact-A-Family website or the Support Group's own website.

A full-colour leaflet was created using a double sided A4 sheet folded into 3, to send to members of the Support Groups with further information about the project. This was designed so that an inside page could be personalised by each charity to say why they wanted to be involved. The leaflet invited patients and their families to get involved in the project in a number of ways, as follows;

- Complete a short questionnaire either online or by post
- Tell us your experiences of accessing services
- Take part in a focus group
- Help us to pilot and develop the Family Route Maps
- Provide us with copies of information leaflets that have helped you.

Additionally, people were asked to help raise awareness for the project through the Jeans for Genes Campaign which provides this benefit as part of their commitment to help fund the project. This could be through;

- Speaking to newspapers and magazines. Jeans for Genes has an annual coverage in both national and local papers, telling stories of fundraising events and stories of families affected by genetic disorders
- Speaking to radio and TV - each year Jeans for Genes events appear on local news stories as

- well as on radio stations around the UK
- Speaking at local schools with Jeans for Genes presentation packs.

All of the Support Groups were extremely proactive recruiting families for the Jeans for Genes media campaign and generating awareness of the conditions in the news, locally and nationally. They were also invited to publish articles about the project written by the project leads, in their newsletters and to send out the project leaflets to their members wherever practicable. In support of this partnership and to ensure that the groups had resources to do so GIG provided a donation to each Support Group. Participants were recruited for the focus groups during this awareness phase and where possible the meetings were held during each Support Group's Conference or Family Days, or where there were none scheduled then for each remaining group it was arranged that they should meet in the GIG office in London during the weekend and volunteer travel expenses were provided.

Information about the project was posted to the GIG website with a link to a questionnaire. The survey was developed with the help of the Support Groups and it was hoped that through open access on the internet affected patients and their families who may not belong to these Support Groups would also contribute to the project, thereby widening participation. Questionnaires could either be completed on-line or sent by post and a Freepost address licence was purchased.

Focus groups were planned for patients, their families and carers, to gather qualitative data around three themes: patient experience; patient information; and what a Family Route Map might include. Clinical staff were also invited to have an input separately through discussions and a General Practitioner (GP) focus group was held in response to the issues raised by patients regarding primary care.

Patient Focus Groups:

Information about taking part in these focus groups was circulated to people who had either agreed to attend or requested further information following the distribution of the Support Group newsletters and personalized project leaflets. Discussion in the focus groups lasted between 60 and 90 minutes. Due to the symptoms of fatigue that prevail in people with Myotonic Dystrophy there was a great deal of flexibility around the needs of each participant in that focus group and rest breaks were provided in between topic discussions.

Figure 4 Topics explored in the patient focus groups

- Information provided to patients, their families and carers about the condition.
- Information about genetics and how the condition could affect other family members.
- What types of treatment and/or surveillance patients and their families receive and whether they receive these locally or at specialist centres.
- Information to inform the development of a route map for patients and families with the condition.
- What healthcare professionals have been helpful/unhelpful.
- Access to benefits or other services outside of the National Health Service for patients and families with the condition

Consent in each focus group was given by all participants immediately before commencing the discussion which included permission to audiotape the meetings. The Facilitator followed a semi-structured question guide of the topics for the focus group meetings (Figure 4). The topic guide highlighted general areas to be covered but also allowed people to talk about issues that are important to them.

The discussion focused on what people with hereditary conditions say about care and the way in which they receive treatment or are given information. A Co-Facilitator was present in all groups to take notes and observe group dynamics. Notes from the discussion for each meeting were anonymised and circulated to those participants who had wished to review them, amendments were made accordingly and final notes together with quotations were generated after which all tape recordings were destroyed.

Only one meeting required supplemental telephone interviews, the Syndromes without a name (SWAN) focus group was only attended by four participants due in part to the difficulties faced in traveling by many parents caring for children with complex needs. A number of other parents had expressed an interest but were unable to attend, therefore they were contacted and 3 respondents consented to a phone interview lasting between 30 and 60 minutes. The interviews were designed to increase the amount of information collected for SWAN and were semi-structured so that parents could comment and provide their own ideas on the phone. The questions were based around the discussion that took place in the SWAN focus group.

Analysis of the focus group discussions and interviews identified common themes and reliability was ensured through regular meetings between the main analyst and the other researcher to discuss all analytical notes. No new themes emerged during analysis of the final two

focus groups at which point it was considered that saturation had been achieved.

Online Survey:

Patient Support Groups advertised the online questionnaire to their members inviting them to take part in the study and it was also made available through the GIG website to all patients and their families in the UK with one of the six conditions to widen participation. Respondents answered a number of questions about where they or their relative are treated and how they access information about the condition, and were then asked about difficulties they had experienced, suggestions for improvement in services and any other comments they wished to make. These free-text answers corresponded to the themes which emerged in the focus groups and no new themes were evident.

GP Focus Group:

A further focus group was undertaken in Primary Care with GPs from a single large Health Centre because at the start of the project there had been no plans made to approach GPs in this way. This focus group was held in response to what patients with or at risk of a genetic condition together with their relatives and carers from the six previous focus groups told the project team. In order to explore these areas participants in the GP focus group were invited to discuss each of the resultant emergent themes around a vignette describing a hypothetical consultation with a patient presenting at their surgery (Appendix 2).

Developing the Family Route Maps:

The seven emergent themes were used to develop the framework for the generic Family Route Map and a format was chosen for the layout of the leaflet. Meetings to discuss the findings, both general and specific to each of the conditions, were set up with the contacts from each of the Patient Support Organisations. Draft versions of Route Maps were developed as a result and circulated to people who had agreed to be Reviewers within each condition area with the exception of Barth syndrome, as this Patient Support Group undertook a Workshop with families to agree the content and develop it further. Reviewers included patients and families, clinical staff and others with an interest. They were invited to complete a short survey about the quality and content of the condition-specific Family Route Map which contained a likert scale and some free-text questions allowing them to add any suggestions for further information they wished to include. The results of this survey were analysed and Route Maps were revised and a further draft version sent to the Reviewers for each of the conditions (including Barth syndrome) for the Pilot phase. The request this time was that the Reviewers



Photo by Jay Wilkie

might like to invite other patients, colleagues and healthcare professionals with an interest to comment on this resource, again a survey was included to record their responses. This collective data contributed towards the evaluation of the Family Route Maps.

Final amendments were made to each Family Route Map and these were then professionally printed. A link has also been supplied for the websites of the Patient Support Groups so that the leaflet can also be downloaded from the internet. It is intended that future revisions and updates can easily be undertaken by the Support organisations and further Route Maps printed by them whenever necessary.

Disseminating the information and learning:

These data from the focus groups, interviews and discussions with clinicians were collated into a series of reports and published on the GIG website. These can be freely downloaded from the website www.gig.org.uk/familyroutemap.htm

Throughout this project preliminary findings were reported at national and international conferences and through publications, raising awareness of the importance of the issues and concerns from the patient's point of view within the specialties of genetics and general practitioners and the European community for rare diseases.

Regular bulletins and updates were provided through articles written by the project team and published in GIG Today (quarterly magazine of the Genetic Interest Group) and the Patient Support Groups involved in the project. Reports to the project Advisory Group, Jeans for Genes and Genzyme sought to make available relevant information relating to timescales and budgets in addition to the important findings from the project.

The final stage of the project included the writing and publishing of this report and the dissemination of the findings which will be of value to healthcare

professionals and those providing training to them as well as individuals with rare genetic conditions and the Patient Support Organisations representing them. It is also anticipated that it will be a resource for Commissioners of services, policy makers and others interested in the patient views and experiences of, health and care services.

Results:

All patient focus group participants and interviewees said they had found it to be a very positive experience for them and common themes were identified (Figure 5). In total 57 participants attended 6 patient focus groups and the number of participants for each group was between 4 and 12. Sixty-four respondents completed the on-line questionnaire, with some individuals from each condition group. These respondents were affected by the condition and/or a relative/carer of an affected individual. Recruitment of participants for the project through the Patient Support Groups does provide a particular perspective on the provision of support from such groups that may not be true of all people with genetic conditions, for example, the emphasis on the need for access to support groups. Although the genetic conditions in this project are very different from each other, analogous difficulties have been reported by individuals with each of the conditions in terms of impact and the management required, indicating that these are likely to be universal for most, if not all, genetic conditions.

Figure 5 Emergent Themes from Patient Focus Groups

- Information.
- Communicating with healthcare professionals.
- Diagnosis of rare genetic disorders.
- Treatment/surveillance for rare genetic conditions.
- Education in genetics for healthcare professionals.
- Empowering patients, families and carers.

Participants in the patient focus groups and telephone interviews and those who completed the online questionnaire described difficulties at all stages and the majority felt they had lacked the necessary information or support to enable them to feel empowered to share in the decision-making and choices regarding care. As these conditions are rare most participants commented that neither the primary care healthcare professional nor the individual or family affected has heard of their condition before it was diagnosed. This makes accessing the appropriate information, treatment and services extremely hard for both parties and means that patients, their families and carers have difficulty getting appropriate support.

Many people felt dissatisfied with care in general and primary care services; low quality of care, poor support and feeling low priority, were identified as contributing to such feelings (Figure 6).

"As a family with a disabled child you're just bottom of the pile." (SWAN phone interviewee)

The main concerns that emerged from the discussions, survey and interviews with all participants, including patients, families, carers and healthcare professionals, are around difficulties that impact on clinical management; Diagnosis, Coordination of care, Continuity of care, and Psychosocial Impact, and these are discussed under the seven emergent themes.

Figure 6: Causes of Low Patient/Parent Satisfaction

2.1 Information

The patient focus groups and interviews have provided an insight into the way in which patients and their families with these particular conditions access currently available information. Problems identified by people include: concern around the general lack of information about these rare genetic conditions, both for patients and healthcare professionals; the variability of quality for available information; the reliability of available information; and who controls the information flow.

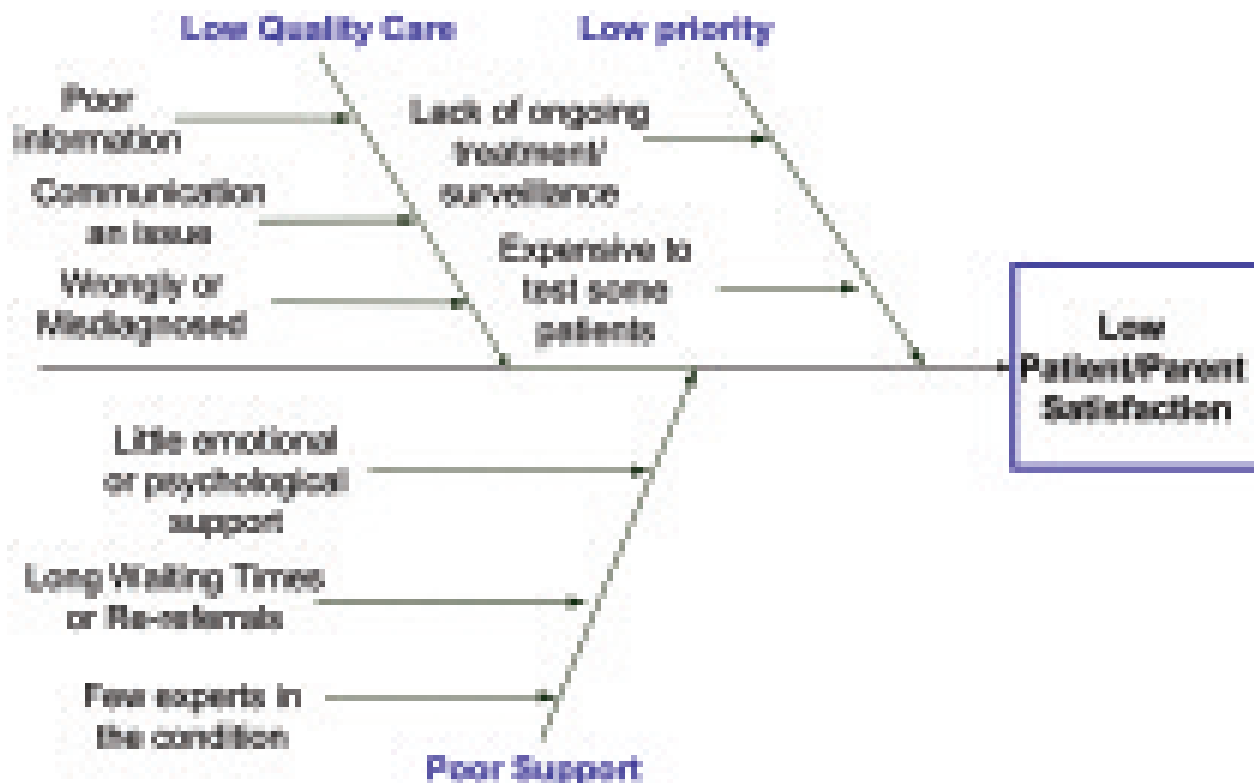
The timing of information and the amount of information (too much or too little) provided were discussed and a participant in one patient focus group said she was 7 months pregnant when she was told that her husband's condition is hereditary and *"it was a bit too late then."*

"Information-wise, there is a lot of information on the internet but being given information by professionals is practically zilch!" (Myotonic Dystrophy focus group)

Summary letters following an appointment at the Clinical Genetics Unit (CGU) were said to be helpful by participants in most focus groups, however, it was noted that these letters did not mention all the points discussed and some people felt that it was difficult to take in everything as patients or parents at these consultations.

"It's a lot of information to relay back to the family if you don't understand it." (SWAN focus group)

Causes of Low Patient/Parent Satisfaction



Quality of information was raised as a concern as some participants had found information unreliable and even slightly negative and wanted factual information in layman terms to enable people to make informed decisions. Some patients felt upset or shocked by the information they had been given or read.

"..it was out of the text book which is horrific" (Gorlin syndrome focus group)

"Now I've read all that information [about the condition] that has really shocked me, the fact that I could end up having renal failure!" (NPS focus group)

When discussing information from clinicians, many participants in the patient and family focus groups suggested that it varied a great deal both in the accuracy and amount and some felt that they had not been given much information at all.

"It felt like you were on your own!" (Myotonic Dystrophy focus group)

A comment from a SWAN phone interviewee illustrated how stressful it can be for parents receiving information, she stated *"x [child's name] was 3 when I was told she had lifelong learning disability - it was just so dramatic!"* The respondent said she did not expect to be told this but the doctor did tape the interview so that she could listen back to the salient points.

For parents of children with a genetic condition there is also an issue of how much information to give, and when, to their children; this needs to be addressed thoughtfully and in conjunction with parents and the patient directly. Some groups agreed that peoples' need for information changes according to their different life-stages, for example, childhood, adolescence and adult.

"...for our son who has got it [the genetic condition] you have got to limit the amount of information that you give him." (Gorlin syndrome focus group)

Entitlement to benefits was mentioned in all patient and family focus groups and phone interviews. Many participants and interviewees felt they had not been informed that they could claim certain benefits and it was suggested that more sharing of information is a solution.

"Nobody told us about DLA (Disability Living Allowance) for x [daughter's name]... What's annoyed me is that these people must know that you are eligible for it but they don't tell you. You have to push for it - you have to ask for it!" (Myotonic Dystrophy focus group)

Knowing where to acquire knowledge about these conditions was however, expected of the clinical staff by patients, parents and carers. Information for healthcare

professionals about these rare genetic conditions may also be scant or indeed some of the disorders may have as yet been little researched. Participants in the GP focus group suggested that it is extremely challenging to recommend websites to patients where they can find reliable, up-to-date sources of information for rare genetic conditions.

"It's difficult because I don't want to recommend a site if I haven't got the faintest idea about the site...and because there are a lot of these sites I wouldn't necessarily know...which to recommend." (GP focus group)

A discussion took place in the GP focus group about how GPs and primary care health professionals can help patients to understand information they have found on the internet about their condition. It was agreed that explanation of medical terms could be one way of helping.

"Sometimes all they want is a translation." (GP focus group)

2.2 Communication



Photo: www.flickr.com/photos/chefranden/54663534

The way in which clinicians communicate with patients was felt to be extremely vital to enable the patient or their family to be involved in their care and shared decision-making, even when a clinician may not have all of the information about a condition that a patient may want to know.

"We've met somebody [a clinician] who is real, who is able to communicate directly and easily." (Multiple Endocrine Neoplasia focus group)

Communication with health care professionals was

highlighted as a problem in all patient focus groups, people said they are not being listened to. A participant in the Barth syndrome focus group explained that even after her baby had been diagnosed as having cardiomyopathy, whenever she returned to the hospital with him certain that he was in heart failure then she would go through 3 or 4 doctors who, when she told them she thought he was in heart failure, only told her *"no, we are treating it as a viral thing."*

"We need more doctors to listen to parents." (Barth syndrome focus group)

Lack of inter-professional communication, with medical notes not available to all clinicians and little evidence of discussion between the medical specialties impacts on the patient, who often feels they have to describe in detail their condition over and over again or occasionally defend their position as some healthcare professionals do not believe their needs for particular care or treatment. One participant in a patient and family focus group commented that he has a more positive experience when seeing a clinician who is an expert in the field of Gorlin Syndrome and said *"You don't have that explaining to people every time."*

"...it takes them months to get notes from another hospital." (Myotonic Dystrophy focus group)

Additionally, it was suggested that regular feedback is also required.

"What is missing in all this is about feedback. Somebody to keep you up-to-date with what is happening." (SWAN focus group)

2.3 Diagnosis

Many participants in the patient focus groups had experienced delays in diagnosis, largely due to the lack of knowledge of these rare conditions even within clinical specialties. Some had been misdiagnosed with another condition with related symptoms, whilst others had undergone an extended period of uncertainty before being eventually diagnosed. This period of time had been very difficult for participants, and the delay had often affected subsequent prognosis. A participant in the MEN focus group commented that her previous, older GP, had misdiagnosed her as having 'irritable bowel' for a number of years, but she added *"my GP now is a younger one and he is terrific."* Furthermore, one participant in the Barth syndrome focus group explained that her son had a heart transplant in 1998 but that it wasn't until three years ago when her sister also had a son born with dilated cardiomyopathy that a doctor identified the condition in the family. It was felt by participants in all patient and family focus groups that without knowledge of risk factors for future pregnancies, further children may be at risk and likewise other family

members may remain uninformed regarding possible risks to themselves or their own children.

"I didn't realise my daughter could be a carrier". (Barth Syndrome focus group)

Therefore, referral to an NHS Clinical Genetics Department at an early stage around diagnosis was seen as an essential part of the care package, either for assistance with diagnosis (for example for children remaining without a diagnosis) or for providing information once diagnosis had been made by another medical specialty.

"I wish I'd had access to some kind of geneticist. It was only when I had my little girl that we were referred to a geneticist and it does put it all into perspective and explains a lot of things." (Gorlin syndrome focus group)

It was acknowledged by participants in the GP focus group that patients with rare genetic conditions may face difficulties in getting a diagnosis and have limited choices for clinicians whom they see and where they receive treatment and surveillance. Additionally, consideration was given by them to cases where children with complex needs do not have a diagnosis, for example, Syndromes without a name. A suggestion was made by one participant for GPs to help parents of such children.

"...one might do a letter on their behalf asking paediatricians if they would review the case and possibly instigate a genetic referral." (GP focus group)

Participants in the GP focus group were well informed of the risk factors and protocols for referral to a Clinical Genetics Unit for more common genetic cancers but for rare genetic disorders it appears less clear.

"[My GP asked] 'why do you want to see a geneticist, you know you've got the syndrome'." (Gorlin Syndrome focus group)

GPs in this project's focus group identified that with a growing and active local Clinical Genetics Unit it would be easy to assume that someone with a rare genetic condition would have already been seen by the service but as this is not always case, it should be considered as a further referral option.



2.4 Treatment and Surveillance

This project has revealed difficulties in clinical management which are reported by patients with each of the genetic conditions. For optimum management of these genetic conditions, it is important that surveillance and treatment is established after diagnosis. Patients who took part in this project are seeing various medical specialties for surveillance of all of the body systems affected by the condition. The appointments are generally not coordinated, and around half of them have to visit more than one hospital to see these specialists. Out of 57 focus group participants and 3 phone interviewees only 2 people said they were satisfied with the treatment and surveillance they or their affected family member received.

Respondents who completed the questionnaire described the surveillance they are currently accessing, and there was wide variation between individuals with the same condition, with many not receiving surveillance at all (Figure 7). This demonstrates that there is a problem where guidelines and consensus documents exist as they are not always being followed, for example; MEN CONSENSUS Guidelines, Myotonic Dystrophy Care Card and Care Pathway, and Gorlin syndrome NICE Guidelines, are available. Lack of treatment protocols and care pathways for many other patients affected by rare genetic conditions also reflects the difficulties GPs may have when selecting a specialty to refer patients to, when they seek a second opinion. It was acknowledged by GPs that for many rare genetic conditions there may only be a few Consultants in the UK with expertise in a particular condition. It was agreed that best practice would be for a Consultant to inform a GP of other clinicians with a specialist interest in order that a patient may have a choice of whom to consult for a second opinion.

"Yes, but if you are talking about rare conditions and there is only one other, or two other [consultants] then it is the Consultant's job to point that out to the GP." (GP focus group)

Those affected by these conditions appreciate that it is not possible for healthcare professionals to know about

www.flickr.com/photos/thisparticulargreg/362937046

every condition, but feel they should have the option to be treated by an expert, even if this involves travelling outside of their Primary Care Trust (PCT). People said that they would be prepared to travel in order to see an expert in the condition or to be cared for by a Centre of Excellence.

"Patients should be under the care of a specialist centre." (MEN focus group)

Access to services is variable across the country and many patients feel there should be access to the same care for all.

"It seems to be a lottery to me!" (MEN focus group)

The majority of the questionnaire respondents travel up to 20 miles to access their surveillance, though some travel further to see certain specialists. People feel that they should have the option of either accessing surveillance locally or travelling to see the specialist that they choose, but this is not currently available.

"I would much rather travel 3 hours in the car to... see a specialist that listens to me because all of x [child's name] conditions are rare." (SWAN focus group)

It is apparent that not all patients with the same condition are receiving the same surveillance and/or treatment. This may be appropriate due to different manifestations of the same condition, but patients feel unsure about whether they are receiving the most appropriate care, as many healthcare professionals do not have knowledge and experience of these rare genetic conditions.

"...in terms of eyes, kidneys [surveillance]...I have nothing whatsoever." (Nail Patella Syndrome focus group)

It was felt that patients needed someone who could ensure they were accessing the appropriate surveillance and treatment, coordinate these appointments and ensure that the various clinicians were communicating their findings to each other. However, this does not

appear to be happening, and it is unclear who could take on responsibility for this co-ordinating role. Ideally, patients would like a multidisciplinary team approach for clinical management, with a central coordinator as a champion or advocate. This has been the situation for some paediatric patients, with the paediatrician as coordinator, but this is lost with the transfer to adult services.

"I need to know...there is a central person who has clinical knowledge, which I don't have, and who will say...you should go and see him [referral to a specialist]." (Gorlin Syndrome focus group)

In addition to the lack of co-ordination of care, people also experience difficulty with continuity of care. There are also difficulties due to seeing different clinicians within the same specialty, who may have different amounts of knowledge and understanding. Patients are generally referred to a team, rather than an individual doctor, and many felt that they see a different clinician each time they attend.

"There is no continuity. I've been going to see a kidney specialist for 19, 20 years plus now and I don't think I've seen the same man [doctor] twice!" (NPS focus group)

The difficulties for patients with life-long chronic conditions requiring to be re-referred to a consultant for

each treatment episode was highlighted by many participants of the patient and family focus groups. Although these individuals require ongoing screening over a period of many years, some have been discharged by certain medical specialties as they only keep patients on their register for a year.

"I can only keep you on my books for a year and that's it!" (NPS focus group)

This means that those who need annual surveillance have to keep going back to their family practitioner to be re-referred each year, and there appears to be no mechanism for ongoing care.

"they keep trying to discharge me...they keep trying to get rid of me." (Gorlin Syndrome focus group)

People felt that psychological support was really important but most people had not been offered any and one patient focus group participant suggested that "as partners and parents we need a little bit of guidance as well". A couple of participants with these rarer genetic cancer syndromes stated that they had found help and emotional support through a Clinical Psychologist at a Cancer Centre and a Macmillan Nurse.



<http://www.flickr.com/photos/andy-germany/2089214849>

Figure 7. Current surveillance received by online survey respondents with each of the conditions

Surveillance by Condition	BS		GS		MEN		MD		NPS		SWAN	
	No.	%	No.	%	No.	%	No.	%	No.	%	No.	%
Renal	1	25	1	8	1	20	-	-	18	72	-	-
Ophthalmic	-	-	2	15	-	-	1	10	19	76	-	-
Orthopaedic	-	-	-	-	-	-	1	10	2	8	-	-
Paediatrician	-	-	-	-	-	-	-	-	1	4	-	-
Scans (CT/MRI/USS)	-	-	1	8	2	40	-	-	1	4	1	14
X - rays	-	-	4	31	-	-	-	-	-	-	1	14
Dermatological	-	-	9	69	-	-	-	-	-	-	-	-
Neurological	-	-	-	-	-	-	2	20	-	-	-	-
Cardiac (ECG)	1	25	-	-	-	-	8	80	-	-	-	-
Dental screening	-	-	1	8	-	-	-	-	-	-	-	-
Physiotherapy	-	-	-	-	-	-	-	-	4	16	1	14
Lung Function	-	-	-	-	-	-	2	20	-	-	-	-
Muscle strength	-	-	-	-	-	-	2	20	-	-	-	-
Tumour screening	-	-	-	-	1	20	-	-	-	-	-	-
Blood tests (various)	1	25	-	-	4	80	1	10	-	-	1	14
Other	1	25	2	15	-	-	2	20	1	4	1	14
None	2	50	3	23	1	20	2	20	5	20	6	86

Key

BS = Barth Syndrome

GS = Gorlin Syndrome

MEN - Multiple Endocrine Neoplasia

MD = Myotonic Dystrophy

NPS = Nail Patella Syndrome

SWAN = Syndromes Without a Name



Photo: www.flickr.com/photos/janamills/145601495

2.5 Education of Healthcare Professionals

In this project participants reported that they would welcome the openness of healthcare professionals to learn together with them about their condition and be valued for their contribution of experience of living with a rare condition, an attribute that people with genetic conditions in their family were found to value highly in a recent large study regarding providers' knowledge of genetics¹².

Some patients in this project report that they have been seen by doctors who have not read their medical notes, and find themselves explaining why they are there, having to convince the doctor of the surveillance they need. Participants talked repeatedly about the frustration of having to explain their condition to different healthcare

professionals, and having to fight for what they need using terms such as "difficult" or "hard" to describe this experience. There was an appreciation that healthcare professionals could not know everything about every condition, but it was felt that they should be taking more responsibility for reading back through the notes and finding out what the patient required.

"If they [doctors] have a client on their case load then they need to learn about it [Myotonic Dystrophy]."
(Myotonic Dystrophy focus group)

Many of the difficulties in accessing appropriate treatment are due to a lack of knowledge about the conditions amongst healthcare professionals and a lack of communication between healthcare professionals. It was suggested that guidelines for the clinical management of these conditions should be drawn up by

the experts within the field, so that patients and their healthcare professionals are aware of the appropriate management. This has happened for some genetic conditions, such as Neurofibromatosis Type 1 (NF1), where Members of the United Kingdom Neurofibromatosis Association Clinical Advisory Board collaborated to produce a consensus statement for the diagnosis and management of NF1¹³, and it would be helpful if this could also be undertaken for these rarer conditions.

Findings from the patient focus groups revealed dissatisfaction with primary care services, particularly when the role of primary care was discussed in relation to complex genetic conditions affecting multiple body systems. GPs were thought to lack relevant knowledge and skills to manage these patients.

"Even after we did have a diagnosis and when he was ill there were occasions when I didn't even bother with my GP, I'd go straight to the children's unit." (Barth Syndrome focus group)

Participants in the GP focus group demonstrated a large degree of congruence between what patients want from

primary care and how GPs wish to deal with their concerns and issues, albeit, there remains a need for further information about rare genetic conditions to be easily accessible by primary care practitioners, for example, medical websites where healthcare professionals can learn a little about the condition and types of surveillance or treatment available, especially as there may be no absolute proven (evidence based) treatment for many of these conditions.

Importantly, even when lack of knowledge of these rare conditions by healthcare professionals was openly expressed to patients in a supportive way, participants felt there was an opportunity to learn together, valuing each others experience. Some people felt it important to actively become involved in teaching sessions for healthcare professionals, however, one parent of a young child with a rare genetic condition commented that although he agreed for the junior doctors to be present during a hospital appointment he was left with "a sense of being on display" because MEN Type 2b is so rare. People generally felt that when they are admitted to hospital, doctors are keen to learn from patients but this may be done in a negative way if the patient appears to be an 'interesting case'.

Four Generations of a Family

Photo: www.flickr.com/photos/homegrownskinny/544437327



2.6 Ethical, Legal and Social Issues

Ethical issues arising from genetic conditions were discussed by participants in the patient focus groups, including the notion of informing other family members of the diagnosis and whose responsibility it is to share genetic information. Equity of access for benefits and care, genetic testing, diagnosis, treatment and surveillance, transfer of affected children to adult services, NHS contractual arrangements and perceived financial restrictions were debated, and legal issues such as applying for insurance and holding a driving licence were mentioned. Difficulties around living with a genetic condition in the family impacted on education for affected children, the ability of patients or parents and carers to maintain paid employment, and the social stigma of feeling 'different', all of which contribute to feelings of being isolated without the necessary appropriate counselling and psychosocial support.

Finding information about benefits and care entitlements is extremely difficult and many people additionally felt they needed support and help from people who understood the process for applying and had experience of completing the associated complicated forms.

"You don't know what you are entitled to so how can you find out?" (Myotonic Dystrophy focus group)

SWAN phone interviewees when asked what would improve things for you and your child stated difficulties around claiming DLA especially for long-term conditions. One interviewee felt that in a situation where there is a severe disability in childhood there should be a visit arranged in the home and parents should not have to fill in complicated forms every year. This interviewee has particular problems because of the need to claim DLA for 2 of their children and the benefits department won't look at both in relation to each other within the family, the interviewee has to phone back to discuss each claim separately. This interviewee stated that the *"..system for claiming needs tidying up"* and related how they felt benefits are hindered when there is no diagnosis because it is only this year that they got DLA indefinitely for their children because each of their hospital Consultants (7 in total) had agreed to write letters individually to support their claim.

One participant in a patient and family focus group felt expensive tests were only being offered to those with life-threatening conditions because they had read about some new way of testing and wondered if their child could benefit from this diagnostic procedure.

"It makes me feel a bit like I've got to fight for everything I need" and added "...who makes that decision and is it down to money?" (SWAN focus group)

Some people felt under pressure to undergo genetic testing either for themselves or their children, it was considered a difficult decision to make and any advantages needed to be weighed against the disadvantages. In addition to the health and psychosocial implications, testing has an impact financially, with a confirmed diagnosis improving access to benefits but possibly having a negative impact on the cost of, or obtaining insurance and financial services. One participant in a patient and family focus group felt "lucky" to have been given information about insurance policies even though the negative news was not what he wanted to hear. It was felt in general that such tests would lead to future higher premiums for most types of personal insurance including, life/accident policies, travel insurance and mortgage insurance.

It was agreed that having a diagnosis was essential, as illustrated by the difficulties faced by parents with undiagnosed children. People saw a diagnostic label as necessary for access to services, appropriate surveillance, and benefits, as well as to support. One parent said that when contacting the education department they ask for a diagnosis but she admits her child doesn't have one.

"If you could say something like 'Autism' it'll suddenly unlock all these doors, won't it. You feel like making one up!" (SWAN focus group)

However, such problems are not isolated to those without a diagnosis but include individuals in whom the condition has been diagnosed but lack the support of healthcare professionals with the understanding of the medical and psychosocial impact of that genetic condition on the family¹⁴. Many participants expressed frustration at the lack of co-ordination and continuity in their care, and at having to constantly explain the condition and fight for what they need. They describe themselves as 'slipping through the net' with no-one taking responsibility for them, as everyone thinks that someone else is caring for them. A further area of concern was raised by participants regarding the difficulty of transferring into adult services from paediatrics when their child reached that age. In order to meet their needs, patients need an advocate who can provide support and also assist with the coordination of their care.

"I think we all need an advocate." (Myotonic Dystrophy focus group)

People felt the new funding and contractual arrangements between Primary Care Trusts and NHS Hospital Trusts could lead to limitations on what tests and treatment is offered them, similarly some patients with cystic fibrosis, a complex genetic condition requiring monitoring and treatment throughout life, consider

themselves expensive for the NHS and feel that the resource implications may have an effect on their care¹⁵.

"...we need a degree of honesty. If funding is a problem we need to know." (NPS focus group)

Issues around education emerged in the childhood years for some children with genetic conditions including: Statement of Special Educational Needs or IEP (it was suggested that an Annual Review of the Statement should be undertaken with the Special Needs Coordinator); Nursery care for children with complex conditions; Respite care; and Mobility and access (especially if in a wheelchair). Furthermore, children at the age of 16 also have additional considerations, including: access to College/Higher Education (support and advice regarding careers can be found through the Connexions service www.connexions.gov.uk); Transport requirements; financial aspects as Care Allowances are no longer available after 16; and the fact that some children will be dependent and living at home.

In addition to the medical management of patients in terms of surveillance and treatment, there is also a need for psychosocial support as some people experience feelings of isolation. The burden on patients, families and carers where there is a genetic condition in the family was expressed by many people. Patients, parents and carers sometimes find it difficult to find or maintain paid employment due to their own health needs or the needs of caring for affecting family members especially as this may require taking time off work for appointments or to have treatment with a period of recovery.

"[as a working parent] I feel really penalised by all the appointments...they never fit them around you working". (SWAN focus group)

Moreover, the social stigma of having disorders where physical appearance may be involved was described in some instances as leading to bullying.

"I really had the 'mick' taken out of me at school, I was bullied...just because I'm different." (NPS focus group)

"I didn't get bullied by other school friends but I had problems with the teachers. Gym teachers, PE teachers putting me up against the wall, (telling me) 'put your arms out straight in front of you!' I have never forgotten that feeling of being totally different." (NPS focus group)

There is a feeling that people with disabilities are seen as "bottom of the pile" in society, which can be very demoralizing. Participants reported being unable to access psychological support, although some had very supportive family practitioners it was generally felt there was a lack of mechanisms for support or when provided it was not helpful.

"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). When x was born x should have been diagnosed at birth. x spent a whole year in hospital without coming home...they kept saying I was neurotic". (SWAN focus group)

Many participants in the SWAN focus group had received most information and support from Support Groups and other families in the same situation, as without a diagnosis parents experienced difficulties and frustration in gaining the collaboration of health and care professionals to identify and manage appropriately their child's complex needs. In general, some participants from the focus groups told us that the support of their GPs was helpful and others would have liked more support from them, however, it was also felt that it is asking too much of GPs to manage rare genetic conditions due to their already heavy workload.

"We know how overworked GPs are anyway." (MEN focus group)

2.7 Empowerment of patients, parents and carers

These data indicate that the needs of these patients and families are not currently being met, and a number of suggestions were made as to how the situation could be improved. Support groups are seen as very important sources of information and support, filling a need that is not met within the health service. Information through support groups and the media empowered people and helped them make informed choice which is core to service delivery in the NHS^{5, 6}. It was felt support groups enabled them: to have a "stronger voice" collectively as a group of patients and families with a rare genetic condition; share experiences and information with others; and receive emotional support from people who understand what it is like to live with the condition.

"Everything I've found out, and that's education, not just health, social stuff, everything is from other mums!" (SWAN focus group)

Access to genetic information is seen as particularly important, with many patients and their relatives or carers using the media, especially the internet, to find out as much as they can about the condition¹². It can be difficult to find information about these rare conditions, but support groups have patient information written in language that is easy to understand, which can be accessed via the internet or through contacting the group directly. It was suggested that all patients should be provided with information and details of the appropriate support group at diagnosis, because they fill such an important role as part of ongoing care. Some participants in the patient and family focus groups said that they had been given the contact details of the

national patient support group for the condition by their Clinical Genetics Department. It was suggested by participants that it would be beneficial for healthcare professionals to point families to the support group at diagnosis, for individuals to access whatever they need, whether that is information and/or support.

"The AMEND (Association of Multiple Endocrine Neoplasia Disorders) site [website] was probably the best thing for us...it would have been good if he [the clinician] had known about the AMEND site to direct us to it." (MEN focus group)

Practice nurses were perceived as friendly and supportive by patients and families and some participants from one of the patient focus groups felt that with enhanced training they could offer a vital link at GP surgeries and practice-based clinics. However, reservations were expressed about how Practice Nurses would receive training to do this. GPs confirmed that practice nurses already help to identify the needs of individuals and families with chronic illness on an ad-hoc basis and also through the clinics they hold but time constraints currently prohibit more support.

"(Yes) a sympathetic listening ear and the knowledge of where to go to get help...but there are a huge number of patients with chronic diseases that go through their [Practice Nurses] clinics. We've got 2,500 people on our chronic disease list...developing relationships with them? It's a huge list!" (GP focus group)

In the GP focus group it was agreed that empowering patients with sources of information and support is part of the GPs role, however, this must be gauged individually

and supported by explanation and translation of medical/scientific material together with emotional support where required. Software is available for GPs and health professionals in primary care to help patients find websites offering support; however, the group was not confident that such software lists information relating to very rare conditions.

Many participants felt that everyone needed help and guidance within families affected by genetic conditions.

"...We have had to take our lad [with Gorlin syndrome] out of school. He's 15 and he was in and out of hospital having the treatment and then there was the recovery period. He's now doing GCSEs and because he got so far behind with the work there is no support at all. We had to fight our corner because they were actually going to prosecute us for him not being at school". (Gorlin syndrome focus group)

The majority of the people who took part in the project have been recruited through the relevant Patient Support Groups and as such may be biased in favour of those who benefit from being involved with a support group. However, with the lack of information available from other sources, it is likely that the information provided by them is valuable for all those affected by the conditions. Whilst patient support groups in the UK aid their members, their role is not to give advice and therefore effective partnerships with healthcare professionals with experience and expertise in the condition, could further enhance their role in providing the most up-to date information to individuals and their families or carers.



Photo: www.flickr.com/photos/20181006@N00/118969357

Conclusions

Chapter Three

Qualitative research techniques can provide powerful tools to understand in more depth opinion in health related care¹⁶. This is one of the first projects to consider individual perspectives and involve patients with rare genetic conditions, their families and carers in designing their own holistic route through health and care systems. An holistic approach was adopted in this project, seeking to inform and add to the dearth of current knowledge of how people living with rare genetic conditions find relevant information and access NHS and other services. It was always the intention of the project team to focus on the development of a practical tool to help people rather than academic research. However, data from the focus groups and interviews have provided an insight into the way in which patients and their families with these particular conditions access currently available information and services.

For some families, it only became evident that the condition was genetic when another person was diagnosed, and others had to fight to be referred to the Clinical Genetics Department. In addition to medical services, they also have a need for wider support; psychosocial support, help with accessing benefits, care and special education were all described as difficult to gain access to by respondents. Crucially, it was agreed that having a diagnosis opens doors to patients and their families to access financial benefits, social care and appropriate support. Many of the needs of these patients do not differ from other patients with rare or chronic diseases: the protracted time in being diagnosed; the search for an expert or specialist with an understanding of the condition; and the need for coordinated, multidisciplinary care, are all common features but the extra facet of genetics is that genetic conditions may affect other family members and healthcare professionals do need to ask '*Could this be genetic?*'¹⁷. The revised Curriculum for the foundation years in postgraduate medical education has included the ability to take a focused family history and construct and interpret a family tree as a requirement¹⁸. This should facilitate health care professionals in asking the right questions to illicit any indication of a heritable condition.

Although this study focuses on six specific conditions, it is an important source of information about the patient experience of clinical management and commonalities such as the need for clinical guidelines are likely to apply to the majority of genetic conditions.^{12,15} This project has found that patients' needs are not being met and has generated suggestions for possible strategies that could be used to provide better care. A possibility favoured by patients is the option to attend a specialist centre, as this

would ensure they are receiving the appropriate treatment and surveillance and allow for coordination and continuity of care. Patients currently find it difficult to identify experts within the field, so require more signposting, and experience difficulties with getting referral to these experts. A qualitative study investigating the experience of MEN1 patients who attend a specialist ward at a Swedish University hospital found that they were satisfied with their medical care, feeling well cared for by healthcare professionals who they trust.¹⁹ They attend once or several times a year, depending on individual need, and feel a close relationship with the clinic staff. Although their quality of life is affected by physical, psychological and social limitations, the majority have adjusted to their situation, describing themselves as healthy. However, they do still report a lack of medical and genetic information, indicating that genetic counselling might be appropriate to fully meet their needs as is the case for any genetic condition. This suggests a further role for clinical genetics to be part of the multidisciplinary care for patients with rare genetic conditions, providing information, coordinating care, and acting as a point of contact for any queries or concerns as called for by some respondents.

Some departments do fill this role for patients with certain conditions. For example, the North West Regional Genetic Family Register service in Manchester holds the details of patients with 11 genetic conditions, including MEN, and carries out systematic detailed annual review of files by a genetic counsellor.^{20,21} This ensures that all patients are receiving appropriate surveillance and treatment, and also provides them with a point of contact for any questions or concerns that they have. The majority of patients welcomed the ongoing contact and open access provided by the register service, and also felt able to contact them for emotional support.²² Therefore, the genetics service is meeting client needs for provision of information and support and for coordination and continuity of care. However, this may not be appropriate for all genetic conditions as it may be more appropriate for coordination of care to be overseen by a different medical specialty for certain conditions and this was also expressed by a few respondents.

Patients strongly want a patient centred approach to their consultation with primary care professionals, especially in the domains of communication, partnership and health promotion, wanting them to be interested, sympathetic and involve them in decisions, giving them sufficient time and attention and also providing them with advice on health promotion and self care.²³ A suggestion from some participants in one of the patient focus groups is

that practice nurses in primary care could carry out this role of champion, coordinating care to ensure appropriate surveillance and treatment, and providing support to patients. It was felt that they did not need to have extensive knowledge of the condition, provided they knew what surveillance should be taking place, as they would be able to follow guidelines that had been put in place by experts. They are viewed as friendly and supportive, and it was felt they were ideally placed to be an advocate for patients who feel that their voices are often not heard. This provision of guidance, support and a listening ear fits the paradigm that nurses need to identify the needs of families with chronic illness and facilitate access to appropriate support.²⁴ GPs have asserted the importance of the role practice nurses could have in collecting routine family history information in the context of other primary care clinic services, particularly as concerns about familial disorders may be related to them during clinics.⁴ Practice nurses support the idea of them playing a role in genetics in primary care, although they feel that they need more education to do so.²⁵

However, patients who seek help from primary care to decide their options and choices relating to treatment may find they need to consult more specialised services to be able to make informed choices.

"I have to ask him [the patient] if he's asking the wrong person really. There is no way that I could give an opinion on that obviously, for their treatment...and is it about a second opinion?" (GP focus group)

The government provided funding through the genetics white paper in 2005 for projects involving ten GPs with a Special Interest in genetics (GPwSI) to explore and develop provision of genetic-based healthcare in primary care. The DH website (www.dh.gov.uk) lists further details of these projects and the NHS National Genetics Education and Development Centre (www.geneticseducation.nhs.uk) has been working with those GPs to propose a genetics competence framework for GPwSI.

The six genetic conditions in this report serve as case studies for the issues and provide a template for contextualizing the project findings, however, each of the seven themes that have emerged can be identified in the case studies and doctors' perspective of another genetic condition Ehlers Danlos Syndrome,²⁶ a rare genetic disorder associated with joint hypermobility, pain and disability and in its rarest form, death from arterial, bowel or uterine rupture. This lends further support to our conclusion that the issues and concerns highlighted in this report are widespread.

More research is required to explore ways of providing joined-up services for people, without which they will remain lacking in information and unaware of services and care that could increase the quality of the lives of

themselves and their families and potentially improve the prognosis for the expected course of their disease. This project has provided evidence that people with rare and life-long genetic disorders experience difficulties in accessing optimal care, and can be stigmatized in areas such as employment, insurance and eligibility to benefits. In 2006 the government's white paper *Our Health, Our Care, Our Say* identified that there is a need for high quality information in order to help people choose and access services but our findings suggest that there is insufficient information available for people living with these long-term genetic conditions to do so. The Family Route Maps developed during this project provide signposts to information, support and services for people living with six rare genetic conditions and a generic template is available for other patient support organisations to help design their own. These resources are available from www.gig.org.uk/familyroutemap.htm

Recommendations

Chapter Four

For individuals with rare genetic conditions:

- In primary care, practice nurses could be a resource for reliable sources of information for patients and families and provide continuity of care with much needed psychological support.
- Provision of counselling and psychosocial care should be made available to all who request it.
- A referral to the Clinical Genetics Service should be offered to all patients.

For health and social service providers:

- Primary care nursing is set to expand as a discipline²⁷ and a suggestion for practice nurses to act as an advocate for patients may provide the 'listening ear' that they require but usually only after a diagnosis has already been made and as an aide to patients managing their long-term condition. General practice is in a unique position to understand the impact or threat of illness in a patient's life.²⁸ Open dialogue and further wider discussion needs to take place to engender debate of the proposed role of practice nurses acting as Advocates for patients with rare genetic disorders.
- Up-to-date training and education in genetics for all healthcare professionals should be a requirement for their continuing professional development.
- Greater awareness of rare genetic conditions amongst the medical profession could lead to earlier diagnosis for patients with rare genetic conditions.
- Better communication between patients and professionals and also between professionals involved in their care would enable patients to make informed choice.
- Clearer guidelines for treatment and surveillance of rare genetic conditions are required.
- Coordinated care within a multidisciplinary team approach model of care preferably at a 'Centre of Excellence' is called for by patients to counter fragmentation through organisational boundaries.

For government and other public bodies:

- Resources should be made available for Centres of Excellence or Networks of Expertise to offer services to patients within rare condition groups in order to ensure equitable access to care.

For GIG:

- There is a need for additional work to investigate the appropriateness and effectiveness of multidisciplinary clinical networks, either operating 'virtually' to coordinate care or providing services at a 'Centre of Excellence'.

For further research:

- A further qualitative study involving patients with rare genetic conditions together with primary care teams to investigate the role of primary care in the diagnosis, treatment and surveillance of these and many other genetic disorders would provide a platform for future discussion and planning for the growing numbers being identified at risk of genetic conditions.

Evaluation

Chapter Five

There were two main parts to the Family Route Map Project's evaluation:

- criteria for success and basic data collection and analysis (focus groups, interviews and questionnaires) were planned by the project team at the start
- successful delivery of a tool to help people with one of the six conditions to access current information and services, together with a generic template and report to enable other Patient Support Organisations to design their own if they wish, were dependent on the findings from the data and was an iterative process.

Consideration has been given to the extent to which each objective was met (including the implicit objective), which includes coverage of levels of involvement, who was involved in the different processes, methods used and outcomes. Lessons for the future including what worked well, what worked less well, and specific lessons on levels of engagement, representation, commitment and integrity, costs, timing, trust, iteration and integration of the results of the engagement are also important aspects.

The commitment of each Patient Support Group was paramount to the success of this user-led project and partnership working. Their help, advice and hard work at each step ensured the outcomes would be delivered. It should be noted that this was a huge undertaking for these particular groups as the majority had no paid staff, or office to support them, instead many worked voluntarily from their home and often were themselves either affected or a carer of an affected individual, or both. The integrity of all those involved in this project together with the project team helped ensure that the final tool was developed through the mechanisms in place to involve users.

Specific components of the project that worked well and were essential include the engagement of patients and families at risk of, or living with the six genetic conditions through focus group meetings and the discussions with clinical staff who have an interest or expertise in the six conditions. Their help and cooperation was vital to successfully deliver a tool that would be useful to patients, families and healthcare professionals. These face to face discussions proved to be informative and helped build the relationships and trust necessary to complete the project effectively. Feedback from the patient/family focus groups was extremely positive and

people enjoyed the opportunity of meeting with others and exchanging information. Importantly the seven overarching themes that emerged from the focus group meetings, online questionnaire and meetings with healthcare professionals underpinned the integration of the information into the development of the generic and later, each condition-specific, Family Route Map.

As this project was unique and innovative in its design it was difficult to conceptualize the precise details of what a Family Route Map might look like or the information it might contain at the beginning of the project and explanations to the members of the support groups that the project team met with may have appeared sketchy. This was largely due to the fact that the process was designed to be an iterative one, whereby information that resulted from each discussion influenced the next stage of the development of both the project and the Family Route Maps. The aims and objectives of the project however, were presented and reinforced at all opportunities but it may have been better to have created a webpage on the GIG website from the outset where these could be readily accessed by individuals and organisations. This would have served as a reminder to all involved and improved the overall transparency.

Timescales and targets for the project were met and numbers of participants considered to be appropriate for focus groups were achieved in all but one (the SWAN focus group) of the patient/family meetings. Parents of children without a diagnosis who had expressed an interest in being involved were subsequently contacted and invited to complete a questionnaire on the telephone based on what people in the SWAN focus group had told us.

The additional GP focus group was decided by the project team to be necessary as a result of the discussions around primary care in the patient/family focus groups and a separate topic sheet using vignettes to discuss possible scenarios in the primary care setting was used. Suitable numbers were achieved in this focus group but due to the limited time and budget restrictions it was not possible to arrange a further focus group in primary care at an inner city practice. Therefore the results may be skewed toward a practice in an urban area with access to a locally based Regional Clinical Genetics Service. Use of large-scale postal surveys in primary care would not have been suitable for the in-depth discussions we needed to have to explore peoples ideas and experience and would have required a great deal of time, resources and funding to achieve which were not built into the initial project design.

Although the number of completed online questionnaires was relatively low the project team had always felt it was important to widen the participation as not everyone chooses to join Patient Support Groups. These data added to and supported the information collected during the focus groups. It also provided a mechanism to involve people in the project in other ways as respondents were given the opportunity to give us their contact details if they wished to have further involvement or be kept updated. Contact information was used to build a list of individuals who would become Reviewers for the development stages of the Route Maps.

Issues of inclusiveness and diversity within Patient Support Groups could not be resolved within the remit of this project and many people from the Support Groups felt they did not have good access to under-represented groups within the community. Although some individuals from these harder to reach groups were involved in this project, the communities were not actively engaged and ethnicity monitoring could have better gauged the extent of this. GIG will take this into account when working on projects with other Patient Support Groups in the future and determine ways to achieve greater participation by representation from all sections of the community.

Budgets were monitored regularly by the Advisory Group and the project team had to constantly adjust spending and the way in which developments were planned in response to the information gathered to keep within budget. A small grant was later awarded from the ABPI (Association of British Pharmaceutical Industries) for the cost of the Launch Meeting and Workshop as the original budget did not include this but the project team felt it was important to publicise the project findings with stakeholders in this way.

The project team aimed for transparency throughout the project and maintained a lack of bias in determining what issues and concerns are important to people allowing for iteration as people developed ideas to influence the design of the Family Route Maps and project findings. Importantly, when working in partnership with smaller Patient Support Organisations additional time must be built in to the project to ensure that information can be disseminated to their members and that the communication flow loop is completed. In this way information can flow laterally across each charity and also back up to the project team. A significant proportion of time was also given over to the final stage of development of the six condition-specific Family Route Maps in order to reach consensus agreement on content.

Initial evaluation of the Family Route Maps took place as part of their development. Clinical and patient/family Reviewers were sent the condition-specific Route Maps at two stages and invited to complete a survey incorporating a likert scale and some free text questions for comments and further suggestions. Satisfaction was

measured and revisions made as a result. Use of internet for the Quality and Pilot surveys would have been helpful as suggested by one Reviewer who has difficulty writing due to muscle weakness associated with Myotonic Dystrophy. This Reviewer would have liked the option of using their computer keyboard to fill in an online questionnaire. Although not initially provided as online questionnaires, following this feedback this method was introduced before sending out the remaining Pilot Survey packs for Myotonic Dystrophy, Gorlin Syndrome and SWAN.

Reviewers who completed the Quality Survey were very satisfied with the overall quality of the information included in the Family Route Maps both in terms of its aims and clarity (Figures i and ii). In response to the question *Does the Family Route Map achieve its aims?*, the combined scores for the category 'Very Good' and 'Excellent' reveal that 74% of Reviewers confirmed that the six condition-specific Family Route Maps achieved their aims. The design also scored extremely high with 79% of Reviewers rating it as 'Very Good' or 'Excellent'. Reviewers felt that the Family Route Maps provided support for shared decision making (Figure iii) and it was also agreed that they are a resource for additional sources of information and support (Figure iv), as the majority of Reviewers rated this aspect as 'Excellent'(58%) and 'Very Good' (31%).

Figure i. Clarity of information in the Quality Survey



Figure ii. Satisfaction with the clarity of the aims for the Route Maps in the Quality Survey

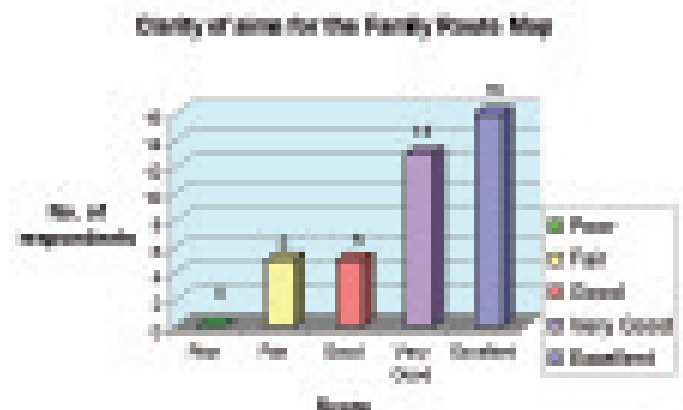


Figure iii. The Family Route Map as a Tool for Shared Decision Making (Quality Survey)

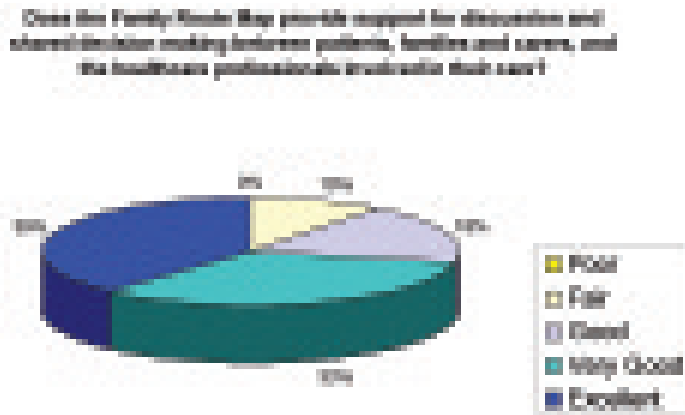
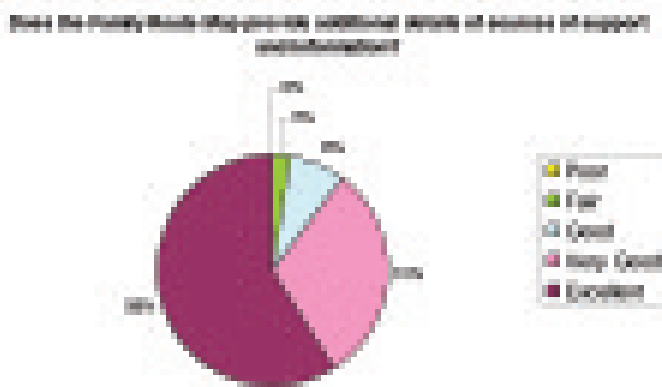


Figure iv. Provision of additional sources of support and information (Quality Survey)



Results of the Pilot Survey revealed that these condition-specific Family Route Maps would be useful for newly diagnosed patients, their families or carers (Figures v and vi) with the majority of the Reviewers choosing to describe their function in this way as 'Excellent' or 'Very Good'.

Figure v. Relevance of information to newly diagnosed patients (Pilot survey)

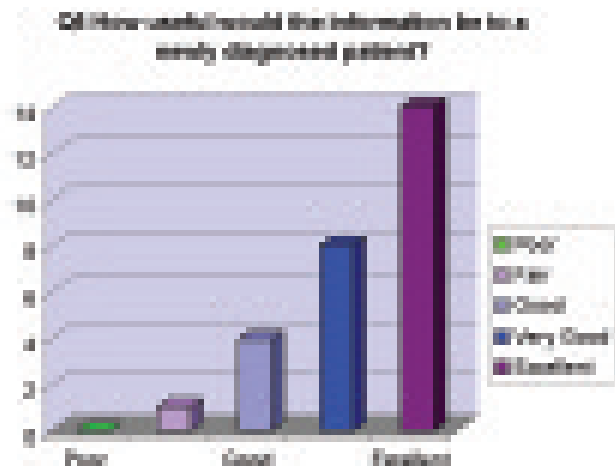
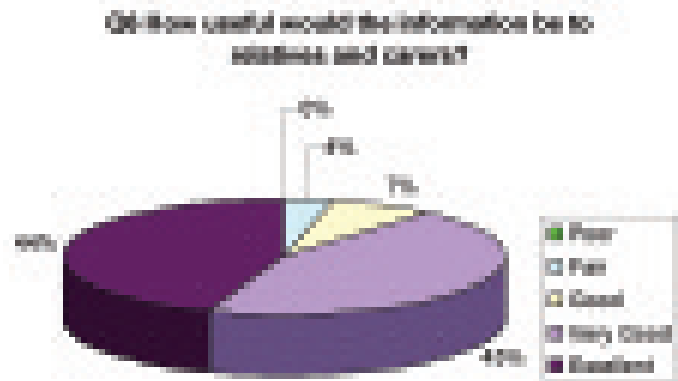


Figure vi. Usefulness to relatives and carers (Pilot survey)



"The leaflet is an excellent source for the most important support networks available for parents/carers for children with 'genetic conditions'. It is brief but encapsulates the resources that are able to assist and advise further for support. I have used all of these sources for information for myself." (SWAN Pilot Survey Reviewer)

Additionally, 92% of responses to the question "How useful would the information be to non-genetic healthcare professionals?" showed it to be either 'Excellent' or 'Very Good' (Figure vii). This was an essential facet of the criteria for success of the Family Route Maps as many patients reported difficulties communicating with healthcare professionals who had never heard of, or knew little about, their condition.

"I would find this useful when attending medical appointments, where they are unfamiliar with the syndrome." (Gorlin syndrome Pilot Survey Reviewer)

The overall rating of the content of the Family Route Maps as a source of information and services for the genetic condition demonstrates that Reviewers concluded the information to be extremely significant with minimal shortcomings (Figure viii). The collective results of the Quality and Pilot surveys indicate that the production of a tool to help signpost patients and families with or at risk of these six genetic conditions (Objective 2) for the Family Route Map project have been successfully achieved. The generic Route Map which uses the seven emergent themes as its basis for including information relevant to any genetic condition should likewise prove germane.

Dissemination and publication of the findings from the project together with the six condition-specific Route Maps (Implicit Objective) has been achieved. Throughout the project, findings were reported to stakeholders: healthcare professionals with an interest; Patient Support Organisations; GIG's membership; other health related organisations; funders; and the public; via Conferences, articles and reports. Additionally, a briefing

meeting on this project took place in February 2008 at the House of Commons, Westminster, prior to the formal launch as part of the 1st European Rare Diseases Day. It is intended to undertake a further evaluation exercise at the meeting and workshop to launch the Family Route Maps and this report in March 2008. Those attending

will be invited to give their feedback on both the event itself and also the content and quality of the condition-specific Family Route Maps, and the applicability of the Report and its findings. The Genetic Interest Group will later report on this.

Figure vii. Usefulness to non-genetic healthcare professionals (Pilot survey)

Q7 How useful would the information be to non-genetic healthcare professionals?

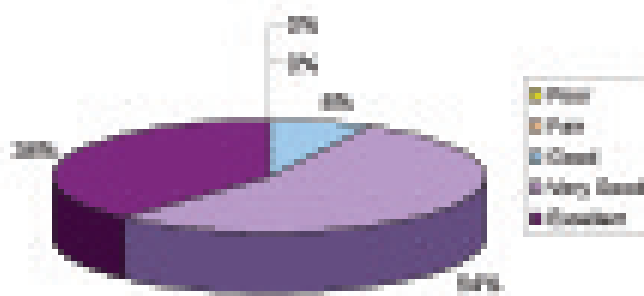
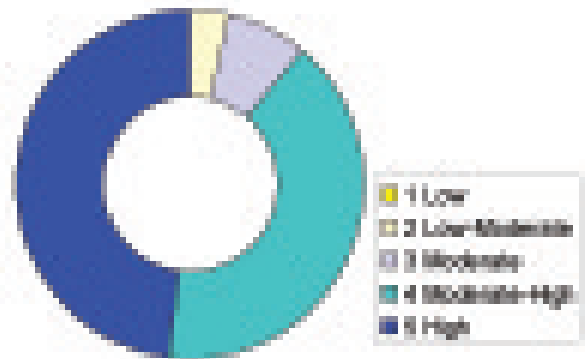


Figure viii. Results of the rating as a source of information and services for the genetic condition in the Pilot Survey

Results represented as the percentage split between the five scores: low-high



<http://www.flickr.com/photos/hamedmasoumi/871579644/sizes/o/in/set-72157600318256992/>



Photo: www.flickr.com/photos/yolandafenwick/321865394

References

1. Baird, P.A. et al. Genetic Disorders in Children and Young Adults: A Population Study. *Am J Hum Genet* 1988;42:677-693.
2. Department of Health. (2003) *Our Inheritance, Our Future* Realising the potential of genetics in the NHS. London, DH.
3. Response to Department of Health Survey on the White Paper "Our Inheritance, Our Future". Genetic Interest Group. Unpublished. 2006
4. Watson, E.K., Shickle, D., Qureshi, N., Emery, J. et al. The 'new genetics' and primary care: GPs' views on their role and their education needs. *Fam Pract* 1999;16:420-425.
5. Department of Health. (2003) *Building on the Best: Choice, Responsiveness and Equity in the NHS*. London: DH.
6. Department of Health (2004) *Better Information, Better Choices, Better Health: Putting Information at the Centre of Health*. London: DH.
7. Burke, S. et al. The experiences and preferences of people receiving genetic information from healthcare professionals. NHS National Genetics Education and Development Centre. September 2007. (http://www.geneticseducation.nhs.uk/downloads/genetics_experiences_report.pdf accessed 11/01/08)
8. Swain, D. et al. *Accessing information about health and social care services*. Picker Institute Europe. April 2007
9. Healthcare Commission. (2006) *Variations in the experiences of patients using the NHS services in England. Analysis of the Healthcare Commission's 2004/2005 survey of patients*. Healthcare Commission.
10. *Rare Diseases: Understanding this Public Health Priority*. Eurordis, November 2005 www.eurordis.org
11. Mays, N., Pope, C. Qualitative research in health care: Assessing quality in qualitative research. *BMJ* January 2000; 320:50-52.
12. Harvey, E.K., Fogel, C.E., Peyrot, M., Christensen, K. D. et al. Providers' knowledge of genetics: A survey of 5915 individuals and families with genetic conditions. *Genet Med* 2007;9(5):259-267.
13. Ferner, R.E. et al. Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *J Med Genet* 2007;44:81-88.
14. Marini, F., Falchetti, A., Del Monte, F., Carbonell Sala, S. et al. Multiple endocrine neoplasia type 2. *Orph J Rare Dis* 2006;1:article 45.
15. Lowton, K., Ballard, K.D. Adult cystic fibrosis patients' experiences of primary care consultations: a qualitative study. *British Journal of General Practice* 2006;56:518-525
16. Barbour, R.S. The newfound credibility of qualitative research? *Tales of technical essentialism and co-option*. *Qual Health Res* 2003;13:1019-1027.
17. Burton, H. *Addressing genetics, delivering health: a strategy for advancing the dissemination and application of genetics knowledge throughout our health professions*. Public Health Genetics Unit, Cambridge. 2000
18. *The Foundation Programme Curriculum 2007* (http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_081918.pdf accessed on 24/01/08).
19. Stromsvik, N., Nordin, K., Berglund, G., Engebretsen, L.F. et al. Living with Multiple Endocrine Neoplasia Type 1: Decent Care - Insufficient Medical and Genetic Information. *J Genet Couns* 2007;16:105-117.
20. Evans, D.G.R., Maher, E.R. and Macleod, R. Uptake of genetic testing for cancer predisposition. *J Med Genet* 1997;34:746-748.
21. Wright, C., Kerzin-Storrar, L., Williamson, P.R., Fryer, A. et al. Comparison of genetic services with and without genetic registers: knowledge, adjustment and attitudes about genetic counselling among probands referred to three genetic clinics. *J Med Genet* 2002;39:e84.
22. Kerzin-Storrar, L., Khan, A.A., Watters, E.A., Kingston, H. et al. A Regional Genetic Family Register: opportunity for active - rather than reactive - genetic counselling. *Am J Hum Genet Suppl* 1991;49:42.
23. Little, P., Everitt, H., Williamson, I., Warner, G. et al. Preferences of patients for patient centred approach to consultation in primary care: observational study. *BMJ* 2001; 322:468-72
24. Newby, N.M. Chronic illness and the family lifecycle. *J Adv Nurs* 1996;23:786-791.
25. Bankhead, C., Emery, J., Qureshi, N., Campbell, H. et al. New developments in genetics - knowledge, attitudes and information needs of practice nurses. *Fam Pract* 2001;18:475-486.
26. PRACTICE: A Patient's Journey Ehlers-Danlos syndrome, *BMJ*, Sep 2007; 335:448-450
27. Illiffe, S. Nursing and the future of primary care Handmaidens or agents for managed care? Editorials. *BMJ*; April 2000; 320: 1020-1021
28. Kumar, S., Gantley, M., Elywyn, G., Iredale, R. Integrating genetics into primary care in practice. In *Letters*. *BMJ*; December 2001; 323: 1367

Appendix 1

Summary Protocol Flow Chart for the Family Route Map Project

Participants

The families with the following six genetic conditions will be included as these conditions are wide-ranging and include some which are life-threatening to babies, two different cancer syndromes and some which cause pain and mobility difficulties for children and adults:

Barth Syndrome;
 Gorlin Syndrome;
 Multiple Endocrine Neoplasia Disorders (MEN);
 Myotonic Dystrophy;
 Nail Patella Syndrome (NPS);
 Syndromes without a name (SWAN).

We will also interview healthcare professionals who have an interest in these rare genetic conditions plus a group of GPs.

Overview of protocol

GIG Project Officer and Communications Manager attend meetings with Patient Support Group members relevant to the conditions to explain the project and prepare leaflet to be sent to individuals and families.
▼
Leaflets detailing information about the project and an invitation to contact GIG team if they would like to be involved will be distributed through the Support Groups together with articles/adverts in Support Group Newsletters and GIG Today Newsletter.
▼
Individuals and families contact the GIG team by phone, email or post. Team establish willingness to discuss their experiences of information and services for their condition at a focus group meeting or by taking part in a survey.
▼
Arrangements are made for attendance at a focus group or interview for those who wish to participate.
▼
Written consent is reviewed at the commencement of the interview and the team also get consent to audiotape the focus group discussions. Interviews and discussions take place with time allowed for debriefing.
▼
The comments made during the focus group discussions to be content-analysed for common themes.
▼
On-line patient and family questionnaires and interviews plus clinical discussions and/or focus group will be used to gather further opinion and information.
▼
The resulting data will be analysed and compared to the previous results for new/converging common themes.
▼
The report of the findings will form the basis for a generic template of a Family Route Map and this will be used to develop the six condition-specific Family Route Maps.
▼
Dissemination of the findings through healthcare journals, newsletters and conferences will be sought.
▼
Each Family Route Map will be further developed and piloted with individuals, families and carers together with healthcare professionals.
▼
Family Route Maps and Final Report will be published and launched to patients, the public and healthcare professionals and be freely available on the GIG website.

Appendix 2

GP focus group vignettes

Case 1

Information

A lady of 30 presents because she thinks she may be at risk of breast cancer as it is in her family. What sorts of information could you provide her with?

Case 2

Communication

A mother says her young baby has been diagnosed with a genetic condition and wants to see a consultant who specialises in that condition for a second opinion. How do you approach this request?

Case 3

Diagnosis

A patient who has symptoms of irritable bowel says a relative has been diagnosed with a genetic cancer syndrome and wonders if he could also have it. How would you manage this concern?

Case 4

Education of healthcare professionals

A patient brings in lots of information after searching the internet because they have found out that they have a genetic condition in the family and need help to understand the information. How do you proceed?

Case 5

Empowering patients, parents and carers

A carer of another patient with a genetic condition asks for help to get more support for themselves in managing the care of their spouse. What sort of assistance can you provide?

Case 6

Treatment/Surveillance

A patient wants to know more about treatment options, including specialist surgery for a genetic cancer syndrome. How can you help this patient?

Case 7

Ethical, Legal and Social Issues

A parent with a young child who has developmental delay and difficulties in nursery wants to discuss genetic testing for their child and also education needs. What would you do?

Appendix 3

Patient Support Groups

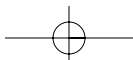
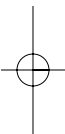
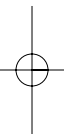
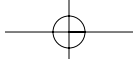
<p>AMEND Website: http://www.amend.org.uk</p> <p>Contacts:</p> <p>MEN1: Liz Dent Email: liz.dent@amend.org.uk Tel: 01423 780594 (9am-8pm only) Fax: 01423 780959</p>	<p>Mrs Jo Grey AMEND Chairperson PO Box 89, Tunbridge Wells, Kent, TN2 9GL</p> <p>MEN2/Sporadics: Jo Grey Email: jo.grey@amend.org.uk Tel: 01892 525308 (9am-8pm only)</p>
<p>Barth Syndrome Trust (UK and Europe) Website: http://www.barthsyndrome.org.uk</p> <p>Contact: Email: info@barthsyndrome.org.uk</p>	<p>1 The Vikings Romsey Hampshire, SO51 5RG</p>
<p>Gorlin Syndrome Group Website: http://www.gorlinggroup.co.uk</p> <p>Contact Email: info@gorlinggroup.co.uk</p>	<p>11 Blackberry Way Penwortham Preston PR1 9LQ Telephone helpline:</p>
<p>Myotonic Dystrophy Support Group Website: http://www.mdsguk.org</p> <p>Contact Email: mdsg@tesco.net</p>	<p>Mrs. M. A. Bowler S.R.N. S.C.M. National Coordinator Myotonic Dystrophy Support Group, 35a Carlton Hill, Carlton, Nottingham, NG4 1BG Office Tel No: 0115 9875869</p> <p>Telephone helpline: 0115 9870080</p>
<p>Nail Patella Syndrome (UK) Website: http://www.npsuk.org</p> <p>Contact: Email: npsuk_info@yahoo.co.uk</p>	<p>PO Box 26415 East Kilbride Glasgow G74 1YW</p> <p>Telephone No: 0800 1218298</p>
<p>Syndromes without a name Website: http://www.undiagnosed.org.uk</p> <p>Contact Email: info@undiagnosed.org.uk</p>	<p>Telephone/Fax: 01922 701234</p>

Glossary

Autosomes	Are chromosomes other than the sex chromosomes. Hence autosomal refers to inheritance through those chromosomes that are not the sex chromosomes.
Barth Syndrome	A rarely diagnosed genetic disorder that affects males. It is caused by a recessive X-linked gene resulting in an inborn error of metabolism. When a mother is a carrier of the gene for Barth Syndrome there is a 50% chance in any pregnancy that she will pass the X-chromosome with the faulty gene onto the child. If the child is a boy and inherits the gene he will develop the condition or if a girl inherits the gene she will be a carrier. The main symptoms include: Cardiomyopathy; Neutropaenia; Skeletal myopathy; and Growth delay.
Basal cell carcinoma (BCC)	Is the most common skin cancer.
Cardiomyopathy	Is a disease of the heart (cardio) muscle (myo) disease (pathy): Dilated Cardiomyopathy is a disease of the heart muscle that leads to enlargement of the heart's chambers, robbing the heart of its pumping ability.
Carrier	A person who has one faulty and one working copy of the gene for a recessive genetic disorder or for a characteristic
Chromosome	A thread-like structure made of DNA which is found in the nucleus of animal and plant cells. Most human cells contain 46 chromosomes (23 pairs), but eggs and sperm (the sex cells) contain only 23 unpaired chromosomes.
Clinical Geneticist	Clinical geneticists are physicians who have undergone higher specialty training in genetics after general professional training, usually in medicine or paediatrics (sometimes in other specialties such as psychiatry, obstetrics and gynaecology, ophthalmology and general practice)
Clinical Genetics Unit or Clinical Genetics Service	A service provided under the NHS for patients and their families with genetic conditions. Clinicians and Genetic Counsellors discuss with individuals and families their risk from inherited genetic conditions and the choices for them.
DNA Deoxyribonucleic acid	Chromosomes are made up of long strands of DNA and genes are segments of this DNA.
Dominant	If a faulty gene is dominant, it will show an effect even though there is a working copy of the gene on the other chromosome. A person only needs to inherit a faulty gene from one parent to develop the condition.
ECG (electrocardiogram)	Is a graphic produced by an electrocardiograph, which records the electrical activity of the heart over time.
Enzyme	A protein which helps chemical reactions to take place in cells.
Embryo	A developing organism. In humans the word embryo is used until about eight weeks after fertilisation.
Fertilisation	When a sperm cell penetrates the outer layer of an egg cell and joins with it to form a new life.
Fetus	In humans the developing embryo in the womb from about eight weeks after fertilization until birth.

Focus Group	Is a group discussion on a particular topic and explores the experiences, opinions, feelings, attitudes and suggestions of the people taking part. The aim of a focus group is to talk about issues that are important to the participants.
Gene	A segment of DNA which carries coded instructions for amino acids, the building blocks from which proteins are made. Passed from one generation to the next in the chromosomes, genes are responsible for determining our inherited characteristics.
Genetic Condition or Genetic Disorder	A condition resulting from the genetic make-up of that individual. The word 'disorder' implies unwanted consequences (for example disability or disease) arising from the genes which one has inherited.
Genetic Counselling	Genetic counselling is not primarily "counselling" in the psychological sense, it is practiced in a <i>non-directive</i> manner. This means that you will not be "directed" or told what decision you should make. Genetic Counsellors try to explain the facts as clearly as possible, giving the person or family accurate information on their options in a way which they can understand, and helping them to make up their own minds.
Genetic Counsellors	Genetic Associates, Nurses, Counsellors and other non-medical staff working within Clinical Genetics.
Gorlin Syndrome	The three main components of the Gorlin (naevoid basal cell carcinoma) syndrome are multiple basal cell carcinomata, recurrent jaw cysts and non-progressive skeletal abnormalities. It has a dominant pattern of inheritance but is extremely variable in individuals and even families.
GP (General Practitioner)	A NHS doctor often referred to as the 'family doctor'.
In vitro fertilisation (IVF)	In vitro fertilisation (IVF) is a technique in which egg cells are fertilised by sperm outside the woman's womb. The fertilised egg (zygote) is then transferred to the patient's uterus with the intent to establish a successful pregnancy.
Metabolic disorder	When a person is born with a vital enzyme missing or not working properly and a metabolic blockage occurs. The blockage usually means that chemicals are unable to get through to where they are needed and so build up abnormally on one side of the blockage, which can have very serious consequences for health.
Multiple Endocrine Neoplasia Disorders	Are rare conditions in which several endocrine glands develop noncancerous (benign) or cancerous (malignant) tumours or grow excessively without forming tumours. MEN1 and MEN2 are hereditary syndromes which show autosomal dominant inheritance. MEN2 has at least three distinct variants. The tumours associated with these conditions produce excessive amounts of various hormones, which in turn can lead to different medical problems.
Myotonic Dystrophy	Is the most common muscular dystrophy of adult life and can also affect other organs in the body. Age of onset can be from birth (congenital myotonic dystrophy) to late adult onset. It can affect either sex and the affected parent has a 50% risk of passing the condition on with each pregnancy.
NHS	National Health Service
Nail Patella Syndrome	Is an autosomal dominant condition affecting the nails, skeletal system, kidneys, and eyes. Absent or partially missing nails and knee caps (patella) represent the common findings. Kidney involvement may lead to renal failure and there is also a risk of glaucoma (an eye disease due to raised pressure in the eye).

Neurological	A neurological genetic condition is an inherited disorder affecting the nervous system.
Neutropaenia	An abnormally low level of neutrophils in the blood.
Neutrophils	The main white blood cells for fighting or preventing bacterial or fungal infections.
Preimplantation Genetic Diagnosis (PGD)	Tests early-stage embryos produced through in vitro fertilization (IVF) for the presence of a variety of conditions. Embryos free of conditions that would cause serious disease can be implanted in a woman's uterus and allowed to develop into a child.
Prenatal diagnosis	Prenatal diagnosis is offered to women with pregnancies at increased risk of chromosome abnormality. Indications for prenatal diagnosis include increased maternal age, an increased risk from serum screening, an abnormal finding at ultrasound scan or a family history of chromosome abnormality for example.
Prenatal Tests	Tests carried out when a woman is pregnant to check if the fetus is developing normally.
Recessive	If a faulty gene is recessive, it will usually show little or no effect unless the same recessive gene is faulty in both of a pair of chromosomes. A person who inherits two faulty copies of a gene will have the condition.
Screening	Genetic screening is when doctors test everyone within a population or subset of that population to see if they have a particular gene or predisposition towards a specific genetic disorder.
Sex chromosomes	These are responsible for determining the sex of an individual. In humans they are known as the X and Y chromosomes, females have two X chromosomes in most body cells, while males have an X and a Y chromosome.
Syndromes without a name	There remain children with complex illness or disability for whom the diagnosis at present is unclear. More tests to discover rare, hitherto unidentified, disorders may become available. Without a specific diagnosis inheritance patterns may be unclear although specialists can sometimes advise, based on general genetic principles.
Testing	Genetic testing is when doctors test an individual with a known family history of a particular genetic disorder to see whether that person has, or does not have, a copy of the faulty gene associated with that disorder.
Ultrasound Scan	An ultrasound scan builds up pictures of organs and areas inside the body using sound waves. These sound waves have a frequency beyond human hearing. An ultrasound scan is often used during pregnancy to obtain pictures of a baby in the womb.
X-linked	Refers to the inheritance of a particular characteristic or disorder from a gene carried on the X (or female sex) chromosome. Also known as sex-linked inheritance.





Genetic Interest Group (GIG)
Unit 4D, Leroy House
436 Essex Road, London, N1 3QP
Phone: 0207 7043141
www.gig.org.uk
Registered Charity No 1114195
Company Registered by Guarantee 05772999

