

Family Route Map Project Report of a series of six Focus Groups



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Executive Summary

Six Focus Groups and three telephone interviews provided the Genetic Interest Group (GIG) with views of patients together with their families and carers who have experience of one of six rare genetic conditions.

A series of Focus Groups and one set of supplemental telephone interviews took place between July and December 2006 for patients with one of six rare genetic conditions, and their families and carers. Participants were recruited through Support Groups for the following conditions; Barth Syndrome, Gorlin Syndrome, Multiple Endocrine Neoplasia Disorders, Myotonic Dystrophy, Nail Patella Syndrome, and Syndromes without a name. The aim was to gather information, views and peoples' own perceptions of currently available information and services to inform the development of Family Route Maps designed to signpost information and guide patients, families and carers through the available services within the UK.

All Focus Group participants and interviewees said they had found it to be a very positive experience for them and common themes were identified. Seven clear categories emerged: Information; Communication; Education of healthcare professionals; Diagnosis of rare genetic disorders; Empowering patients and parents/carers; Ethical, Legal and Social issues; and, Treatment & Surveillance of patients and families with rare genetic disorders.

Findings 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. Additionally, many feel they receive sub-optimal treatment/surveillance for their condition and would prefer to have a Centre of Excellence responsible for their care with one lead Clinician acting in a co-ordinating role.

These Focus Groups have provided an insight into the way in which patients and their families with these particular conditions access currently available information and services. It was clear from the discussions that information for healthcare professionals about these rare genetic conditions may be scant or indeed some of the disorders may

have as yet been little researched. Additionally, healthcare professionals aren't thinking "*Could this be genetic?*" Ideally, patients want to have all the options available explained to them in order to make an informed decision and also have their expertise and experience valued. Focus Group participants felt they were sometimes given inaccurate information and for parents of children with a genetic condition there is also an issue of how much information to give, and when; this needs to be addressed thoughtfully and in conjunction with parents and the patient directly. Currently, by far the most important source of information for people with rare genetic conditions was felt to be Support Groups/Charities and other patients or parents of an affected child. Crucially, it

was agreed that having a diagnosis opens doors to patients and their families to access benefits and social care.

They told GIG that healthcare professionals are not thinking "*Could this be genetic?*"

The seven themes highlighted in this report will be developed further with input from patients, their families and carers together with healthcare professionals involved in their care. A Family Route Map is planned for each of the conditions and a generic template will also be developed for other Patient Support Organisations to use as the basis for their own Family Route Map.

Here are some of their concerns:

- They were not given enough information about their condition.
- Health and care services were not of a consistent standard throughout the UK.
- Some families are still not being referred to the Clinical Genetics Service.
- Many patients believe they are not receiving optimal care and monitoring.
- Most patients want one doctor who is an expert in their rare disorder to co-ordinate their treatment and surveillance.
- All patients would like to have a 'Centre of Excellence' responsible for their clinical care.

Introduction

The six genetic conditions that this project will produce a Family Route Map for are:

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

The Family Route Map project commenced in April 2006 working with 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.

Developing Family Route Maps as a Tool to help access appropriate information and services in the UK for families with genetic conditions is the primary objective for the project. The aim of this series of Focus Groups was to explore information and services currently available to these families as the first stage in the development of the six condition-specific Family Route Maps and also to ask them what they would like to see included. From this information a template that could be used generically by other Support Groups will also be created.

Focus Groups and supplemental interviews with patients belonging to the six Support Groups were identified as providing the in-depth and rich narrative that would determine peoples' perceptions of their own experiences. We produced a topic guide for the Focus Groups which gave us general areas about what we wanted but also allowed people to talk about issues that are important to them. We are interested in what people with hereditary conditions say about care and the way in which they receive treatment or are given information. Only one meeting required supplemental telephone interviews, the Syndromes without a name (SWAN) Focus Group was held in London but was only attended by four participants due in part to the difficulties faced by many parents caring for children with

complex needs. A number of other parents had expressed an interest but were unable to attend, they were contacted and 3 respondents took part in 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 give us their own ideas on the phone. The questions were based around what people had told us in the SWAN Focus Group.

AMEND (Association of Multiple Endocrine Neoplasia Disorders), which has about 90 patients as members, was considered to be the Support Group in the best position to arrange the first patient/family Focus Group and this took place on July 15th 2006. GIG gave a presentation about the Family Route Map project at the May 2006 AMEND Patient Conference and their June newsletter carried an article about the project together with the personalized project leaflet inviting people to get involved. The remaining Focus Group meetings were arranged similarly through the other patient Support Groups and where possible arrangements were made for the Focus Group to take place during existing patient meetings. All Focus Group meetings had taken place by December 2006.

Information about taking part in these Focus Groups was circulated to people who had either agreed to attend or requested further information. With the permission of participants Focus Group meetings were recorded and notes taken by the Facilitator and Co-Facilitator, these were later used to analyse the data for emergent themes. As a result of further themes being identified in later Focus Groups data were re-analysed to cross-check for them being present in the findings of the earlier meetings for ; Barth Syndrome, Gorlin Syndrome, Multiple Endocrine Neoplasia Disorders, and Myotonic Dystrophy.

A template will also be freely made available for other Support Groups to use to base their own Family Route Map on.

Focus Group Themes

Seven themes emerged that were common to all six Focus Group meetings:

- Information;
- Communication;
- Education of healthcare professionals;
- Diagnosis of rare genetic disorders;
- Empowering patients and parents/carers;
- Ethical, Legal and Social issues; and
- Treatment & Surveillance of patients and families with rare genetic disorders.

All Focus Group participants and interviewees said they had found it to be a very positive experience for them and common themes were identified. Seven clear categories emerged: Information; Communication; Education of healthcare professionals; Diagnosis of rare genetic disorders; Empowering patients and parents/carers; Ethical, Legal and Social issues; and, Treatment & Surveillance of patients and families with rare genetic disorders.

Each theme is outlined below with illustrations of the way the participants and interviewees described their experiences and where possible, how this might be improved for others. Each of the above themes is not separate and there is an overlap between many of these areas for each of the quite different conditions. Where emergent findings also reflected those of previous groups this was considered during subsequent discussions.

Information

Issues identified by participants in all Focus Groups and phone interviews included: 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. Some groups agreed that peoples' need for information changes according to their different life-stages, for example, childhood, adolescence and adult. Participants in the Barth Syndrome Focus Group were even more specific and broke this further

down to several age groups within the pre-16 year olds and a post-16 years category. One group also noted that in addition to patients receiving Genetic Counselling and having access to information and advice to help with family planning and Preimplantation Genetic Diagnosis (PGD) there should be advice about living wills and end of life plans. All groups agreed that a SIGNPOST to Support Groups should be included as one of the first items in the Family Route Maps.

Participants in the Barth Syndrome Focus Group described the lack of information available to them (and also healthcare professionals) due to the fact that little is still known about it. One participant said *"I had been to see a Geneticist to try to find out what was going on but there was very little information there."* Quality of information was also raised as an issue as some participants had found information unreliable and even slightly negative and wanted factual information in layman terms to enable parents to make informed decisions. This preference was also highlighted in discussion by some other groups.

When discussing information from clinicians, participants in the Myotonic Dystrophy Focus Group suggested that it varied a great deal both in the amount and accuracy. Some participants felt that they had not been given much information at all and one commented *"It felt like you were on your own!"* Another participant said

Lack of information was an issue that was discussed in all Focus Groups.

"I was under the impression until just recently that it was something in just my family and my mum has only just found out that she has it." Participant in the NPS Focus Group

"Nobody told us about Disability Living Allowance (DLA)." Participant in the Myotonic Dystrophy Focus Group

“Information-wise, there is a lot of information on the internet but being given information by professionals is practically zilch!” One of the participants had been given a poor prognosis and outlook for the future of her baby born with Congenital Myotonic Dystrophy but felt this information later proved to be unfounded, however, she said she had been glad to know the worst as she considered everything else a bonus when the worst didn’t arise. It was felt by one participant that healthcare professionals over-emphasized the negative aspects of affected children and future generations being worse than the affected parent. Another suggested that families were not given the help because *“it is not a trendy illness... it has not got a specific cure.”*

Participants said they wanted factual information, in layman terms, to enable them to make informed decisions. One group wanted their Family Route Map to include a SIGNPOST to ‘Choices’ providing background information regarding the options available and relevant questions to ask healthcare professionals about these options.

The timing of information and the amount of information (too much or too little) given were also discussed. One participant in the Gorlin Syndrome Focus Group said she was 7 months pregnant when she was told that her husband’s condition is hereditary and she felt *“it was a bit too late then.”* Another participant said he’d received information from the Clinical Genetics Unit (CGU) and it was very basic but *“it was out of the text book which is horrific.”* He added that his wife (who has the condition) was very concerned by what information they’d been given and said *“...for our son who has got it you have got to limit the amount of information that you give him.”* A further participant felt the information to be good but said it could be overwhelming, describing the amount of information as *“...a bit of an overload”*. This participant said *“I’ve got all the information I need”*.

A participant in the Nail Patella Syndrome Focus Group said he is one of 5 siblings and they all have Nail Patella Syndrome (NPS). He stated *“I*

was under the impression until just recently that it was something in just my family and my mum has only just found out that she has it. We knew that my Grandma had glaucoma but didn’t know that it was part of it (NPS).” This participant added *“It was only recently that we realised it (NPS) was something other people had...Now I’ve read all that information (about NPS) that has really shocked me, the fact that I could end up having renal failure!”*

It was agreed in the AMEND Focus Group that patients need to be told what options are available regarding treatment and testing when first diagnosed. There was an agreement

that an early part of the Family Route Map should include a SIGNPOST

to Choices providing background information regarding the options available and relevant questions to ask healthcare professionals about these options. The group stated it should be the clinical staff who first diagnose Multiple Endocrine Neoplasia Disorders (MEN) that give new patients the AMEND Family Route Map.

Information given by healthcare professionals to families where a child has no diagnosis was felt to be lacking and many such children have complex health and other needs. One parent in the Syndromes without a name (SWAN) Focus Group said *“The information that we have is that his symptoms have been caused by something that we don’t know about.”* SWAN telephone interviewees were asked to rate how useful information is from; the internet, other parents, and Support Groups or Charities. The responses seemed to favour other parents and Support Groups/Charities as providing the most useful information the majority of the time. Additionally, a Focus Group participant

stated *"Everything I've found out, and that's education, not just health, social stuff, everything is from other mums!"*

Entitlement to benefits was mentioned in all Focus Groups and during phone interviews. Many participants and interviewees felt they had not been informed that they could claim certain benefits. One participant in the Myotonic Dystrophy Focus Group stated *"Nobody told us about Disability Living Allowance (DLA) for x (his 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!"* A further participant agreed and commented *"You don't know what you are entitled to so how can you find out?"* More sharing of information was suggested as a solution.

Communication

One of the difficulties encountered by people in the Gorlin Syndrome Focus Group was mentioned as being the need to explain to clinicians all about their condition. A participant 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."* and importantly, this sentiment was echoed by participants in all Focus Groups. Additionally, the way in which clinicians communicate with patients was felt to be extremely vital, even when a clinician may not have all of the information about a condition that a patient may want to know. One participant in the AMEND Focus Group commented *"We've met somebody (a clinician) who is real, who is able to communicate directly and easily."* This participant also raised the point that finding information regarding the long-term aspect of having MEN 2b was very difficult and a discussion around clinicians reluctance to offer too much information about this may be due to the fact that this condition remains rare

and relatively little researched even today. Indeed a further participant who has MEN had been told by his clinician that *"there is just not enough information."* A participant in the Myotonic Dystrophy Focus Group described how she had been advised not to read a lot of information about the condition which could frighten her, she stated *"I was told not to read that."*

It was agreed in the Myotonic Dystrophy Focus Group that having the confidence to speak up can be difficult and depends on the individual.

Communication with health care professionals was highlighted as a problem in all Focus Groups. One participant in the Barth Syndrome Focus Group said *"We need more doctors to listen to parents."* Another participant added *"It's a journey, at first you aren't this person, you do think doctors know more, but you soon learn."* A participant in the SWAN Focus Group said *"I have really bad issues with my local hospital, really seriously bad issues. They don't listen to anything at all and x (child's name) has nearly died several times. They are not listening! I would much rather travel 3 hours in the car to Great Ormond Street Hospital for Sick Children (GOSH) to see a specialist that listens to me because all of x conditions are rare...and yet it's a big hospital, our local hospital, you just expect them to have an interest really."* Another participant commented *"What is missing in all this is about feedback. Somebody to keep you up-to-date with what is happening."*

Participants in the Gorlin Syndrome Focus Group stated that there is a lack of inter-professional communication amongst medical professionals about their condition which is evident when patients are seen by their clinicians, they commented that it is noticeable

Patients want improvements in communications with healthcare professionals and also between them.

"We've met somebody (a clinician) who is real, who is able to communicate directly and easily."

Participant in the MEN Focus Group

"We need more doctors to listen to parents."

Participant in the Barth Syndrome Focus Group

"Everything is quite separate...the hospitals never speak amongst themselves either." Participant in the SWAN Focus Group

"What is missing in all this is about feedback."

Somebody to keep you up-to-date with what is happening." Participant in the SWAN Focus Group

Being able to discuss their genetic condition with healthcare professionals who understood their needs and conveyed important information to them was paramount.

"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." Participant in the Gorlin Syndrome Focus Group

by; their lack of knowledge about the patient, their medical history and the condition itself. One participant in the Barth Syndrome Focus Group expressed concern that healthcare professionals treating their son wouldn't even phone a specialist in Bristol known to have an interest in Barth Syndrome to find out more information about this very rare condition. Another participant in the SWAN Focus Group stated that for her child *"Everything is quite separate...the hospitals never speak amongst themselves either."* A participant in the SWAN Focus Group wondered if because GOSH is a specialist centre that *"...there is more co-ordination, more link-up there?"* A participant in the Myotonic Dystrophy Focus Group also said *"it takes them months to get notes from another hospital"*.

Participants in the Gorlin Syndrome Focus Group described their experience of being seen by a Consultant in Clinical Genetics. One participant stated *"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."* One participant in the SWAN Focus Group explained that it was only when she got an appointment to see the CGU that other doctors began listening to her. A participant in the AMEND Focus Group said that in order to get to see the CGU he had to keep reminding his Consultant to refer him. This participant commented *"You have to keep being proactive all the time."* Summary letters following an appointment at the 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 participants in a few of the Focus Groups felt that it was difficult to take in everything as patients at these consultations. One participant in the SWAN Focus Group stated *"It's a lot of*

information to relay back to the family if you don't understand it." and a further participant added *"You take your child with you (to the appointment), which obviously you've got to take your child with you but then last time we went x (child's name) was turning the room upside down, and I was trying to listen to what he (the Consultant) was saying and x was running round the room!"*

A discussion took place in the Nail Patella Syndrome (NPS) Focus Group about the way in which Clinicians need to be certain of their facts when communicating information to patients with NPS, their families or carers. Additionally, they felt the way in which healthcare professionals deliver information could be improved. One participant was told by a doctor *"You'll be in a wheelchair by the time you are 21 or thereabouts."*, referring to the information verbally provided by the doctor she commented *"I don't think it was evidence, it was uneducated guessing!"* A further participant said that when their child was three months of age they had been told *"this child will never walk!"* by a Consultant in charge of her care and at that time they had sought a second opinion from GOSH. A comment from a SWAN phone interviewee illustrated how stressful it can be for parents receiving information, she stated *"x was 3 when I was told she had lifelong learning disability – it was just so dramatic!"* However, the interviewee 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.

Many participants felt that it depended on where you live as to whether you are seen by doctors who have expert knowledge in these rare conditions.

Education of Healthcare professionals

Many participants in all of the Focus Groups felt that expertise in these rare genetic conditions is confined to either a few specialist centres or limited to those clinicians who have an interest and as a result where a family lives plays a significant role in the services provided, however, the benefits to patients of being seen by a specialist centre were considered paramount. One participant stated *"My little boy went into heart failure when he was 10 days old and we were lucky to be in Bristol"* because a few doctors based in Bristol have a specialist interest and hold an annual Barth Syndrome Clinic. A further participant felt that it was only because the Newcastle Genetics Unit was involved with his nephew's care that his own son was diagnosed with Barth Syndrome as where he lives he didn't think his son was being properly investigated. A SWAN Focus Group participant said *"We go into A&E and say 'these are the drugs x (child's name) needs to be on, x needs a drip, you must do this now' and recently the doctor said 'No, I disagree, take her home' and x became very ill."* This participant commented *"I actually say at our local hospital 'this is rare' and I've got to prove it about x, which is disgusting I think... I shouldn't have to tell the doctors what to do – it should be the other way round!"* Moreover, this participant stated *"I don't feel like that at GOSH though because they are so specialist in their field and I've only got to say 'this is x and what is wrong and it's dealt with but if I go to another hospital they don't understand what x's conditions are."*

Participants in the Gorlin Syndrome Focus Group generally felt that healthcare professionals need further information about the condition to help them diagnose and treat patients. One participant said *"They (the medical profession) have not known enough about the condition really, have they?"* Another participant recalls being diagnosed with Gorlin Syndrome

in Leeds by a doctor whilst having skin grafts and at the time neither the participant himself nor the other doctors present knew anything about the condition. A further participant added *"It's like a learning curve for them and a learning curve for us."* Some participants felt that lack of knowledge on the part of their clinicians had led to a delay in diagnosis, a participant commented that she was concerned that their young daughter had blisters all over her skin but one of the doctors told them to *"get bacterial soap from the supermarket as that might sort it out"*. However, they did not entertain that idea and 15 months later the family got the blood test result confirming she had Gorlin Syndrome. A participant in the AMEND Focus Group said the Paediatric Consultant admitted he didn't know everything about MEN 2b but stated *"he was our first port of call and was very helpful. He ultimately led us to the Endocrinologist who made the diagnosis."* Participants in the Muscular Dystrophy Focus Group suggested implications for heart complications associated with Myotonic Dystrophy were not well known in this country and although most Neuromuscular Consultants would know, Cardiologists may not be aware.

All Focus Groups were asked about the role of GPs (General Practitioners) and Primary Care Teams with respect to caring for patients with these six rare genetic conditions and most participants felt their GPs did not have the knowledge or expertise to diagnose or manage these conditions, however, some felt that their support was helpful and others would have liked more support from them. One participant in the Gorlin Syndrome Focus Group stated that last year he had asked his GP if he could see a geneticist but the GP's reply was *"Why do you want to see a geneticist, you know you've got the syndrome (Gorlin Syndrome)."* Another participant commented that his family are *"under*

Patients generally feel that healthcare professionals need to know more information about these rare conditions.

"If they (GPs) have a client on their case load then they need to learn about it." Participant in the Myotonic Dystrophy Focus Group

"(The) Consultant Paediatrician is a great doctor but he knows nothing about it ...and we've both learned together. We've provided him with articles." Participant in the MEN Focus Group

Some patients feel they are not being referred to the Clinical Genetics Service because GPs don't understand enough about genetics.

"Why do you want to see a geneticist, you know you've got the syndrome (Gorlin Syndrome)." Participant in the Gorlin Syndrome Focus Group

5 different hospitals ... although it's a lot of travelling it works and our GP has been educated by us - we're quite lucky in that respect. I think the central point is the GP". One participant in the Myotonic Dystrophy Focus Group stated *"If they (GPs) have a client on their case load then they need to learn about it."*

Additionally, a participant mentioned that their GP *"didn't know about the condition and he had to look it up."* and another participant mentioned the lack of information and knowledge that their GP had about Myotonic Dystrophy and its consequences for affected people. A further participant described how her nephew who was 13 years old had recently died from cardiac complications of Myotonic Dystrophy, she stated that he had experienced dizzy spells and been seen by the GP 3 months before his death but no cardiac function or exercise tolerance tests had been done. Despite already having a diagnosis of Myotonic Dystrophy his GP suggested it was because his tongue was too big for his mouth, it was his age and he wasn't breathing properly when he was running. A participant in the AMEND Focus Group said that he doesn't see the point in seeing his GP about his condition because he considers that his GP knows nothing about MEN. It was mentioned that the information for GPs which they can access through the internet needs to include MEN and The AMEND Support Group. It was also noted that Consensus documents and protocols could be developed by clinicians with an interest in MEN and that these would be available to GPs to inform them about treatment and surveillance needs of MEN patients.

A discussion took place in the NPS Focus Group around patients and Support Groups getting involved in medical education for healthcare professionals regarding this rare condition and how newly qualified doctors have previously been invited to the NPS Annual Conference to help inform them. One participant in the

Myotonic Dystrophy Focus Group said *"we must all spread awareness as best we can but there is a willingness by some doctors and unwillingness by others..."* A participant in the AMEND Focus Group, whose son's diagnosis was very recent, said their *"Consultant Paediatrician is a great doctor but he knows nothing about it ...and we've both learned together. We've provided him with articles, thank goodness for your (AMEND) website"*. Additionally, another participant in the Barth Syndrome Focus Group felt she had been lucky that her son was in the care of a doctor who had just read a medical paper about Barth Syndrome. Some participants in other Focus Groups felt that if doctors write papers for medical journals about their condition this can also spread awareness and educate other doctors.

Diagnosis of rare genetic disorders

Many participants in all of the Focus Groups had experienced delays in diagnosis of these rare genetic conditions and some had been misdiagnosed. One reason for delays was possibly felt to be that very often patients under the care of a Consultant within a hospital see only junior doctors. Participants in the Barth Syndrome Focus Group had directly experienced delays in their young sons being diagnosed even after they had been found to have Dilated Cardiomyopathy. One participant commented that her little boy had a heart

transplant at 17 weeks of age in February 2000 but was told it was not genetic. However, it was diagnosed eventually as Barth

Delay in being diagnosed or being wrongly diagnosed could lead to serious illness or a poorer quality of life for patients.

"It was really a story of delays as far as we are concerned but we eventually got the right result." Participant in the Gorlin Syndrome Focus Group

Another participant in the MEN Focus Group said her GP had misdiagnosed her as having "irritable bowel" for a number of years.

Syndrome in August 2003 because her sister had a son born with Dilated Cardiomyopathy. Another participant said “x (name of son) was nearly five before we found out he’d been diagnosed with it (Barth Syndrome)...he’d had a (heart)

looking back over the medical history there were clear signs of the condition. He said that it took about a year to get all the blood tests done and that his daughter is the first presentation in their family, he commented, “it was really a story of delays as far as we

- Having a diagnosis could help ensure patients, their families and carers receive:
- Appropriate medical care and monitoring
 - Information to help with benefits, equipment and adaptations
 - Appropriate nursery and schooling for an affected child
 - Supportive therapies such as Physiotherapy, Speech and Occupational therapy

transplant at GOSH but there was clearly something else going on.”

One participant in the AMEND Focus Group who has MEN had been contacted by the Consultant of her daughter who had been clinically diagnosed with MEN. However, this participant also had a father and sister who had been diagnosed with the condition and a former enquiry with the GP about it on the recommendation of her sister’s Consultant, had been dismissed and no testing or referral offered (this was 6 years ago). A further participant has a son aged 4 with MEN 2b and his diagnosis was not made by a lead Consultant Endocrinologist who saw him in Scotland but by another Endocrinologist in his team who was concerned and had seen this very rare condition once before. The first that another participant heard of MEN was through her mother who had done an internet search based on the participant’s pattern of tumours and illness.

Many participants described difficulties in being diagnosed with Gorlin Syndrome, either in themselves or other family members. One participant described how his daughter had massive jaw cysts and the surgeon thought it might be a genetic condition and referred the family to the CGU. However, he felt that

are concerned but we eventually got the right result.” Another participant said “My wife had signs of it (Gorlin Syndrome) when she was younger but she was never diagnosed. Then we had both children, our oldest one had hydrocephalus and our daughter’s got hydrocephalus.” He described how they had only been referred to a CGU because they wanted a third child but was surprised by what the Consultant told them when they arrived at the appointment; this participant commented “As soon as my wife walked through the door he said ‘I know what you’ve got’ (Gorlin Syndrome).”

A participant in the NPS Focus Group said that after the birth of their daughter “...they (doctors) didn’t really know what was wrong with x so they thought x had Down’s Syndrome. The Geneticist who came and tested for Down’s said it was not Down’s (Syndrome) but he didn’t say what it was.” This participant said the Geneticist eventually gave a diagnosis of NPS when x was a year old. Although this participant has got NPS herself it was not diagnosed until after her daughter had been given her diagnosis. Another participant said “my mother had it (NPS)” but a doctor

Patients should receive a diagnosis or test result face to face in order to be able to get the necessary support and information at the time, according to their responses and needs.

only noticed the appearance of this participants arms and legs when the participant went into hospital to have an

operation for a strangulated hernia at the age of 18.

A missed diagnosis or being misdiagnosed was discussed in the AMEND Focus Group. One participant felt that any differential diagnosis around constipation in an infant should include MEN as a possibility. Another participant agreed and felt that her previous, older GP, had misdiagnosed her as having 'irritable bowel' for a number of years, and a further participant outlined how he had also been diagnosed with Irritable Bowel Syndrome (IBS) by his GP but was eventually admitted to hospital and found to have stomach ulcers (one of the signs/symptoms of MEN). His diagnosis of MEN came from the Consultant and since then he hasn't been back to his GP so he doesn't even know if his GP is aware of his rare condition. One participant in the Myotonic Dystrophy Focus Group described how the 22 year old daughter of a sister of his wife had been diagnosed with Myotonic Dystrophy about 5 years ago and this led to his wife and also their own daughter being diagnosed because the family was told it is a genetic condition which can be passed on to other members of the family.

It was agreed by participants of the SWAN Focus Group that although difficult and stressful, trying to establish a diagnosis for their child was essential for a number of reasons including;

- Access to health care services
- Access to benefits
- Access to appropriate nursery provision and education
- Access to support including emotional/psychological.

The Myotonic Dystrophy Focus Group participants also noted that once a diagnosis had been given then access to help at school, social care, Disability Living Allowance (DLA) and equipment/adaptations was made

possible. One participant mentioned that DLA could only be back-dated to the time of receiving the diagnosis even though it was evident that her daughter had Myotonic Dystrophy from very early on, she said *"We had to carry her until she was 2 years old because she couldn't walk!"*

One SWAN participant said *"we go annually"* to see the Geneticist but as yet there is no genetic diagnosis and she feels her child *"will never be diagnosed..."* she continued *"(we've) lost hope!"* A further participant commented *"We're not motivated to go back (to the CGU) ...and a Paediatric Neurologist (said) there was no point in us pursuing a genetic route. He said 'often these kids turn out to never have a diagnosis' "*. By contrast, a further participant said that at GOSH *"They tell me every time we go, 'we might never have a diagnosis, well we might in 20 years but we will keep looking for you'."* One participant said that when contacting the education department they ask for a diagnosis but she admits her child doesn't have one and commented *"If you could say something like 'Autism' it'll suddenly unlock all these doors, won't it. You feel like making one up!"* Another participant agreed that now having had a diagnosis of Autism for her child since September things have improved *"It's really changed...I feel more confident... and proud to say it."* Furthermore it was stated by a participant that *"we do need labels it would be so much easier to say he's got (for example) cerebral palsy because then I wouldn't have to explain it all over again to people."*

Receiving a diagnosis or genetic test result by phone was felt by the Myotonic Dystrophy Focus Group to be inappropriate in most circumstances unless it had been pre-arranged with the patient and their family. One participant said that they had thought they were strong and had agreed to receive the test result on the phone, the participant commented *"we*

Everyone felt that Patient Support Groups are very important.

"For the first time in my life I've met other people, apart from Mum, who have got it (NPS) and I think that's what you need." Participant in the NPS Focus Group

"We had a lot of support from them (Myotonic Dystrophy Support Group)." Participant in the Myotonic Dystrophy Focus Group

"That's where your stronger voice is...that's where your strength grows." Participant in the SWAN Focus Group

knew that she (their daughter) had the condition but still I think that maybe we should have been in the room (with the CGU staff member)". It was therefore suggested that patients should be seen face-to-face to get their results in order to be able to get the necessary support and information at the time according to their responses and needs.

Empowering patients and parents/carers

Many participants in all of the six Focus Groups together with all three of the phone interviewees reported that they had found information through Support Groups and the media (including the internet) to empower them and help make informed choice. Throughout the NPS Focus Group meeting participants referred to the wealth of information they had been given after contacting the NPS (UK) Support Group. One participant emphasised the importance to her of being in the Support Group, she stated, "For the first time in my life I've met other people, apart from Mum, who have got it (NPS) and I think that's what you need." Other Focus Groups reported similar positive benefits of contacting condition-specific Support Groups.

Participants of the Gorlin Syndrome Focus Group felt that one of the main sources of support and reliable information was through contact with the Support Group and its members. One participant said "It wasn't until I saw the 'Bitter Inheritance' programme (BBC Documentary broadcast 31/01/02) that I... (knew) what Gorlin Syndrome was all about." As a result of seeing the programme he then contacted the Gorlin Syndrome Group and got information about the condition. One participant in the Myotonic Dystrophy Focus Group recalled "we had a lot of support from them (Myotonic Dystrophy Support Group)." She further commented on the Conferences that the Myotonic

Dystropy Support Group (MDSG) provide for members "It is very worthwhile, you get so much different professional input in it."

All participants in the AMEND Focus Group felt that having the help of AMEND was vital and one participant recalled that before it started there was only American stuff on the internet but her family also sought help from Cancer Bacup for information so there was some information about the condition before going for genetic testing. It was agreed by participants in the AMEND Focus Group that new patients going for testing would find a 'MEN checklist for Genetic Testing' useful that detailed the sorts of issues that might arise and the types of questions they may like to ask to obtain the information in order to make informed choice regarding testing for themselves and for their children. One participant commented that "by the time you get to see genetic centres...and you get into what would be the (Family) Route Map, I think people are finding they have to find out about this before they get there."

Participants in the SWAN Focus Group discussed finding people who can help parents and carers and one Focus Group participant suggested "...the Champion might be a professional or an advocate or someone who at least is a skilled parent who wishes to take on that role, who could be that co-ordinator." Further discussion about the role of Patient Support Groups which had grown out of a need to provide information and also raise awareness of the need for better services continued and one participant stated "that's where your stronger voice is...that's where your strength grows." Support from other parents and groups was agreed to be essential for families and one participant said that after many years of not knowing any other mums who have children with Special Needs

Some participants who have a child affected by a genetic condition felt that they needed a 'champion' to help them, while others felt they as parents were their own child's champion.

"You (the parent) have to learn about it (the condition) and you have to champion your child." Participant in the Barth Syndrome Focus Group

"The Champion might be a professional or an advocate or someone who at least is a skilled parent who wishes to take on that role, who could be that co-ordinator." Participant in the SWAN Focus Group

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

"I think we all need an advocate." Participant in the Myotonic Dystrophy Focus Group

"As partners and parents we need a little bit of guidance as well". Participant in the Gorlin Syndrome Focus Group

Some participants described the social difficulties they encountered:

"For the first year of his (son) life I felt really isolated." Participant in the SWAN Focus Group

"I really had the 'mick' taken out of me at school, I was bullied...just because I'm different." Participant in the NPS Focus Group

"As a family with a disabled child you're just bottom of the pile." SWAN Interviewee

"Now I'm starting to meet other parents as well." One participant commented *"What you're finding is outside those specialist areas the expertise comes from parents."* and everyone agreed. Another participant said *"You have to do all the work yourself, don't you"*.

The Barth Syndrome Focus Group discussed how important it is for the parent to co-ordinate the care of their child and keep copies of all letters and test results. One participant said *"You (the parent) have to learn about it (the condition) and you have to champion your child."* Another participant commented that *"as Mums we do know a lot more (about the condition) than most doctors do."* and a further participant added *"It's a journey, at first you aren't this person, you do think doctors know more, but you soon learn."*

There was an agreement in the AMEND Focus Group that an early part of the Family Route Map should include a signpost to CHOICES providing background information regarding the options available and relevant questions to ask healthcare professionals about these options. The Myotonic Dystrophy Focus Group participants felt that it is important that patients and their families keep asking healthcare professionals to review their condition and care. It was recognised that having the confidence to speak up can be difficult and depends on the individual. A participant in the Myotonic Dystrophy Focus Group commented *"I think we all need an advocate."* It was also suggested that psychological reassurance or a 'listening ear' is required and that an advocate for the patient would be helpful and perhaps this may be a role for their local Practice Nurse. One participant in the Gorlin Syndrome Focus Group stated that they have found help and emotional support through a Clinical Psychologist at a Cancer Centre and a Macmillan Nurse. The group felt that psychological support was really important but most

people present had not been offered any and one participant suggested that *"as partners and parents we need a little bit of guidance as well"*.

Ethical, Legal and Social issues

Participants of the AMEND Focus Group commented on aspects of the condition that affected their lives. One participant felt that so many family members had been positively diagnosed with MEN that it left the participant feeling like the whole family needed counselling but Genetic Counselling did not provide the psychological Counselling that she had hoped to receive. Concerns and issues were expressed by SWAN Focus Group participants around the needs of themselves or their family as well as their SWAN child, one participant stated *"I am so traumatised by everything I've been through with x that's why I've not had any more (children)."* Another participant commented *"For the first year of his (son) life I felt really isolated."* The first participant then described how a new Psychiatrist who has taken an interest has helped arrange a meeting for all of her child's healthcare professionals to discuss a care plan with her and her child's father. This participant said *"He (the Psychiatrist) is horrified by how we have been let down and how alone I am on my own with x."* A phone interviewee also felt there had been no emotional support for her and her family and a further interviewee stated *"As a family with a disabled child you're just bottom of the pile."*

Sharing information in families about genetic

Some participants felt that decisions about the provision of NHS services were unjust for patients with rare conditions in need of specialist services and care and thought it was due to budget restrictions.

"It makes me feel a bit like I've got to fight for everything I need... who makes that decision and is it down to money?" Participant in the SWAN Focus Group

"(My) local GP doesn't want to know because of financial reasons". Participant in the Gorlin Syndrome Focus Group

Participants who had already had an appointment with a Clinical Genetics Unit showed an understanding about the implications for other family members with regard to how they could be affected by the condition.

conditions was discussed in most of the Focus Groups and it was apparent from the discussion in the AMEND Focus Group that within those participants who had already had an appointment with a CGU there was an understanding about the implications for other family members with regard to how they could also be affected by the condition. Some participants, however, felt that there had been pressure to test children in order to manage their condition medically and provide them access to care. A participant of the Gorlin Syndrome Focus Group raised the issue of passing on information about the condition to family members but said that some of his family did not want to know and he therefore felt it could put future generations at risk. He mentioned that if they should develop the condition without anyone realising what could be done to help prevent the development of skin cancers it would be a neglect of their duty to inform these family members.

It was generally felt that future genetic tests would lead to higher premiums for most types of personal insurance but many participants were still unclear as to what the current affects are.

"I could have done with stuff on insurance." Participant in the MEN Focus Group

Another participant in the AMEND Focus Group said they felt that having Genetic Counselling was a chance for the Consultant in Clinical Genetics to review their young son and stated that the Consultant had mentioned about what may happen in the future, including insurance policies. This participant said that the information which had been helpful was that regarding insurance and to consider what may happen when his son marries and has children. A participant stated *"I could have done with stuff on insurance."* The group agreed that he had been "lucky" to have been given information about insurance policies even though the news was not what he wanted to hear. It was felt in general that genetic tests would lead to future higher premiums for most types of personal insurance including,

life/accident policies, travel insurance and mortgage insurance. A similar discussion in the Myotonic Dystrophy Focus Group concluded that the benefits of finding that you do have the condition and then getting treatment/surveillance and being administered anaesthetics correctly, outweigh the disadvantages of being penalised in terms of insurance.

Social care and benefits, including Disability Living Allowance (DLA) were an issue for the majority of participants in this series of Focus Groups and phone interviews. A participant in the Myotonic Dystrophy Focus Group said that in the case of getting benefits and other help *"but you've got to go to them, you've got to push for it!"* A further discussion also took place

about applying for DLA, including the fact that children can get it. It was agreed that some people have difficulty obtaining it and that the forms are complicated. Issues around DLA were discussed in all Focus Group with the exception of AMEND. SWAN

phone interviewees when asked the question "What would improve things for you/your child?" also 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 they are claiming DLA for 2 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.

Financial assistance for patients and families is not easily accessible.

"But you've got to go to them, you've got to push for it!" Participant in the Myotonic Dystrophy Focus Group

"System for claiming needs tidying up" SWAN Interviewee

They got all of their Consultants to write letters (7 in total) individually to support their claim. A further phone interviewee said that the family have to apply every 3 years to claim DLA because x hasn't got a diagnosis. Barth Syndrome Focus Group participants agreed that access to Disability Living Allowance can be difficult and suggested that Citizens Advice Bureaux (CAB) may help or even some hospitals have Advisors.

A further area of concern raised by the Barth Syndrome Focus Group regarded the difficulty of transferring into adult services from paediatrics when their child was at that age. Other issues emerged in the childhood years for boys with Barth Syndrome 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 Co-ordinator); Nursery care for children with complex conditions; Respite care; and Mobility and access (especially if in a wheelchair). Furthermore, boys who are now reaching 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 boys will be dependent and living at home.

A participant in the Myotonic Dystrophy Focus Group mentioned that he had received some financial help with adaptations in his home as he is now almost entirely confined to a wheelchair. It was suggested that you can get adaptations but there might be a limit to the amount for a Grant. The group discussed the availability of a grant for adapting the home and buying equipment but in the case of installing a lift there might not be sufficient money available through this

grant. The problem of long waiting lists for electric wheelchairs was also highlighted and it was later noted that assessment of need outcomes vary according to opinion, e.g. in some areas there is no provision if the patient is unable to go out on their own to do shopping or go to the theatre etc.

A discussion took place regarding the lifelong financial implications of Gorlin Syndrome patients for the NHS. It was generally felt by the group that services or the most appropriate treatment were not always being offered due to budget restrictions by GPs and NHS hospital Trusts. One participant said his *"local GP doesn't want to know because of financial reasons"*. Participants of the SWAN Focus Group discussed the financial implications of genetic testing for the NHS and one participant felt expensive tests were only being offered to those with life-threatening conditions because they had read about some new way of testing on a website www.specialkidsintheuk.co.uk (a Charity providing support, information and contact between families of children with any Special Needs). This participant stated *"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?"*

One NPS participant mentioned the social difficulties experienced as a child with NPS; he stated *"I really had the 'mick' taken out of me at school, I was bullied...just because I'm different."* Another participant mentioned she'd also had problems at school and said *"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."* Now as a parent of a nine year old child with NPS, raising awareness of her child's condition is paramount, she said *"I've told the school, I've told everybody...all his*

friends.”

Treatment & Surveillance of patients and families with rare genetic disorders

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. The majority of participants in all Focus Groups and two of the phone interviewees said they already travel, or are prepared to travel, to see clinicians who are experts in the field. All groups agreed that they would prefer to be under a specialist centre or a ‘Centre of Excellence’ for their care and many participants said they would like a single clinician co-ordinating this care. A participant in the Barth Syndrome Focus Group said that they were struggling to find expertise and another one responded by saying *“It is a case of finding a champion”* and many participants said they had experienced difficulties in accessing services and information, commenting that they had to ‘push’ for these. It was also acknowledged by the group that an explanation was required from professionals about what to expect and that families needed to be guided as to which specialists their son should be seen by. A participant in the Gorlin Syndrome Focus Group summed up the attitude of some clinicians towards treating his skin cancers by saying he felt the message they are giving patients is *“We deal with it and that’s how you are going to be dealt with.”* providing him with no real choice or alternatives to the treatment on offer. This participant suggested that the care for Gorlin Syndrome patients should be multidisciplinary, he stated *“no one doctor really knows the full scope”* and the group generally agreed. He also commented *“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).”*

A participant of the SWAN Focus Group said *“We left the Paediatrician when he (son) was 15 months old without a contact phone number.”* and this led to asking phone interviewees the specific question *“Is there a clinician co-ordinating care of your child?”* One interviewee said there wasn’t and the remaining two said the Community Paediatrician fulfilled this role and one of these interviewees further commented that this clinician is “the lynchpin” and the “key person.” Interviewees were also asked *“Is there a multidisciplinary team (MDT) involved in the care of your child?”* and one said we were *“led into it because (our) children have quite nasty epilepsy.”* Another said there wasn’t and the third said *“Yes, in theory!”* However, *“...in practice hardly ever see anyone.”*

Participants in the Gorlin Syndrome Focus Group discussed which medical professional could co-ordinate the care of a patient or family with Gorlin Syndrome and it was suggested by a participant that this is the role of the GP. However, reservations were expressed about the level of knowledge that GPs have about this rare condition and one participant commented *“Your family GP hasn’t a clue (about Gorlin Syndrome)”*. One participant said *“Would a geneticist be the right...(clinician to see)? I would like to have seen a geneticist and when I finally did, about our pregnancy, it was very much more balanced and informative than just picking up little bits from the dermatologist and the dentist.”* Another participant suggested that *“a geneticist should be your champion”* and added *“GPs, in my experience, have been useless...you never know if you are going to see the same GP for a start”*. This participant also thought that *“maybe the Genetic Counsellor would give you a list of Consultants (experts who could provide treatment and surveillance).”* There was a general consensus in the AMEND Focus Group that GPs did not have

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 family received.

“We left the Paediatrician when he (son) was 15 months old without a contact phone number.” Participant in the SWAN Focus Group

“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).” Participant in the Gorlin Syndrome Focus Group

“(NHS Consultant said) I can only keep you on my books for a year and that’s it!” Participant in the NPS Focus Group

The majority of patients travel some distance to see clinicians who are expert in their conditions and most felt their GP was not the right healthcare professionals to diagnose or manage rare genetic disorders.

“GPs, in my experience, have been useless...you never know if you are going to see the same GP for a start”. Participant in the Gorlin Syndrome Focus Group

Being seen by the Clinical Genetics Service was felt to be extremely important as part of the care for rare genetic conditions.

"Going to see a geneticist should be your first port of call". Participant in the Gorlin Syndrome Focus Group

"I didn't realise my daughter could be a carrier". Participant in the Barth Syndrome Focus Group

the necessary expertise to diagnose or co-ordinate the care of patients and their families with rare disorders, one participant said *"it's asking one person to do too much ...it just doesn't work...not a good idea!"* A participant added *"we know how overworked GPs are anyway."* Another participant remarked on her *"horrendous experience with the GP"* stating *"and I really don't like the sound of them having anything to do with rare disorders or the management or care."*

All participants in all of the Focus Group meetings felt that there should be standard protocols for surveillance and treatment of these rare conditions and although there is an existing protocol for one condition, Myotonic Dystrophy (this can be found on the internet at the following website address

<http://www.gla.ac.uk/centres/muscle/dmcarecard.htm>), patients and their families in the Myotonic Dystrophy Focus Group still believed they were not receiving optimum care as services differed greatly for those present. Continuity of care arose in their discussion and one participant stated that *"I see different Consultants every time!"* Another participant said he has regular check-ups and sees his Consultant once a year and commented *"he knows what he's talking about"*. A further participant said he sees the Neurologist every 6 months at the Neuromuscular Clinic and following a discussion it was agreed that everyone should be seen at a Neuromuscular Clinic. It was suggested that there should be a Neuromuscular Consultant and Network in every area and everyone agreed. It was also suggested that the Neuromuscular Consultant should co-ordinate care and that patients should have at least annual appointments together with an annual ECG for adults or 6 monthly for children. However, it was felt that one clinician co-ordinating care may not work for everyone if that clinician is difficult to see with a long waiting list. It was therefore agreed

that all patients with Myotonic Dystrophy should be seen by a Neuromuscular Network team wherever possible.

One participant in the NPS Focus Group stated how difficult it is to get a referral out of their area to see a specialist who has experience in treating a particular facet of this rare disorder and suggested it might be a funding issue for GPs. Another participant highlighted the problem of trying to identify specialist doctors who understand the needs of patients with NPS as she has found that many Orthopaedic Surgeons only specialise in one part of the anatomy e.g. hips or knees etc. A further participant said that getting to see the right NHS Consultant who specialised in foot problems was very difficult and involved a very long wait of 19 months. This participant did get Physiotherapy and insoles for her shoes but was told by this Consultant *"I can only keep you on my books for a year and that's it!"* She was concerned that she should not have been discharged by her Orthopaedic Surgeon when she was a young teenager and commented *"I did feel a bit neglected."* This participant also said she had no other treatment or surveillance and added *"...in terms of eyes, kidneys...I have nothing whatsoever."*

Barth Syndrome Focus Group participants discussed how their sons are prone to neutropaenia but doctors are still reluctant to give antibiotics. Some participants said that many doctors had mistaken their child's condition for a viral illness. One participant also said even after her son 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."* Another participant stated *"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."

Not all Focus Group participants, or their affected family members, had undergone genetic testing but there was a unanimous agreement that having a Consultation at a CGU is an extremely important and valuable source of information and advice. One participant in the Gorlin Syndrome Focus Group said she was tested early on because it had been identified in several generations of her family, thereby giving her access to the necessary screening and surveillance. Another participant said that after their child had received genetic testing and was confirmed as having Gorlin Syndrome, they as parents had been given the decision to test their other 2 children but they wanted and needed to know and therefore had them tested and were relieved to find *"they were clear"*. Other participants had also felt it was important to be seen by a Clinical Geneticist; one participant felt that one good reason to be seen by a CGU was to enable patients to get counselling and access psychological services as his teenage son had been through a breakdown resulting from having Gorlin Syndrome. Another participant who attends the CGU in Manchester described how if he has any delays in getting treatment or check-ups he contacts the genetics unit and they sort things out. It was noted that at Manchester it is possible to be on a Gorlin Syndrome Register which means that patients are contacted annually to review their treatment and surveillance and where necessary the CGU will directly refer patients to clinicians. One participant commented that the consultant at that CGU *"is taking responsibility, that's the whole idea."* A further discussion followed about the possible role of a geneticist as the co-ordinator of their care and it was considered difficult at some regional CGUs because patients were only ever seen once by a Consultant in Clinical Genetics. A

participant suggested that *"...going to see a geneticist should be your first port of call"*.

A discussion took place in the Barth Syndrome Focus Group about whether or not all immediate family members should have genetic testing for Barth Syndrome and it was agreed that although there exists other diagnostic tests for affected individuals, genetic screening can identify carriers and is therefore helpful. A SIGNPOST for the Clinical Genetics Service in the Family Route Map was agreed as essential by the group in order to find out what testing is available and what the risks of having another affected boy are for a family. One participant said *"I just assumed that I'd got it (the gene) and never got it (the carrier test) done."* Another participant said *"I didn't realise my daughter could be a carrier"*. All other Focus Groups also agreed that this signpost was important and needs to be included in the Family Route Maps.

Finally, other supportive therapies were discussed in several Focus Groups by participants who had found these useful adjuncts to their treatment, they included, Speech Therapy, Physiotherapy and Occupational Therapy. One participant in the Myotonic Dystrophy Focus Group said *"initially we seemed to get blitzed by people, overwhelming at first... Occupational Therapists, Physiotherapists, and they all did their little bit and then went away saying we'll see you in a year. What good is that really?"* An issue was raised in the NPS Focus Group about how difficult it is to go in the daytime for Physiotherapy if you are working, one participant added *"My sister gets it but she pays for it and she goes at night."* Many participants throughout the series of Focus Groups and also the interviewees felt that Physiotherapy was very limited on the NHS and that patients were only offered a set amount of sessions.

Discussion

- Reliable information is relatively difficult to obtain.
- The internet is used as a source of information but again accuracy or relevance cannot be assured.
- The most important source of information for people with rare genetic conditions was felt to be Support Groups/Charities and other patients or parents of an affected child.

Information for patients, their families and carers was considered to be relatively difficult to obtain and its reliability in some cases might be questionable. What participants want is good quality factual information, written in layman terms, produced and endorsed by reliable sources. However, it was clear from the discussions that information for healthcare professionals about these rare genetic conditions may be scant or indeed some of them may have as yet been little researched. This left people feeling frustrated and unable to make informed decisions and choices (where choice was offered) about treatment and care. Many participants had used (and continue to use) the internet as a source of information but accept that sometimes it may be inaccurate or not relevant to their particular condition. A participant in the SWAN Focus Group stated *"The Paediatrician said he (son) had got low muscle tone (hypotonia) and I went straight on the internet and googled it and came up with Prader-Willi (Syndrome)!"* This participant felt that 'hypotonia', as she had been told was the condition her child had, was not a diagnosis for their child but a symptom of an undiagnosed condition and that a better explanation would have helped at the time. However, as a reference source for finding registered websites belonging to Patient Support Groups and Associations the internet is invaluable. By far the most important source of information for people with rare genetic conditions was felt to be Support Groups/Charities and other patients or parents of an affected child.

Services for these six conditions were considered to be a bit of a "post code lottery" and not organised around the needs of patients, their families or

carers. Many participants shared the perception that GPs are not trained to diagnose or manage rare genetic conditions and felt that in some cases were ill-informed as to how the Clinical Genetics Service could help such patients and their families. A participant in the Myotonic Dystrophy Focus Group suggested that Practice Nurses could have a role in the care of patients and their families and this would mean Primary Care Teams being more involved. However, there was a reservation and question mark about how Practice Nurses would receive training to do this. Another participant said "I think we all need an advocate." It was also suggested that psychological reassurance or a 'listening ear' is required and that an advocate for the patient would be helpful and perhaps this may be a role for their local Practice Nurse. Additionally, concern was expressed about the possible budget restrictions placed on patients who want a referral to see specialists 'out of area' i.e. when there is no existing contract between the PCT (Primary Care Trust) and the NHS Hospital where that clinician provides those specialist services.

Additionally, concern was expressed about the possible budget restrictions placed on patients who want a referral to see specialists 'out of area' i.e. when there is no existing contract between the PCT (Primary Care Trust) and the NHS Hospital where that clinician provides those specialist services.

Ideally, what people said they want is to be cared for by a multi-disciplinary team within a 'Centre of Excellence', co-ordinated by a single clinician who is an expert in their field. The proposal

for a 'Centre of Excellence' with a lead Clinician co-ordinating care was largely agreed upon by all who took part in the Focus Groups and when discussed during the phone interviews, parents of SWAN children also felt that this would be best practice.

One participant in the AMEND Focus Group felt that MEN clinics should play a lead role in care of MEN patients and their families but variability, even between these (where they existed), was discussed. Everyone agreed that there should be 'centres of excellence' with experience of MEN and that the key for care for patients with MEN is a 'multi-disciplinary' approach. It was noted that MEN clinics should also have a member of the Patient Support Group there. It was stated that communication between clinicians is so important for patients with MEN and their families. One participant described the care given to his little boy and commented *"I think we have been treated fantastic...just one person overseeing pretty much everything for us and then the communications...the Paediatrician Consultant together with them, they did really, really well."* There was a general consensus that one person (clinician) co-ordinating care is important. It was felt that this person needed to be knowledgeable, approachable and creative. A participant said that having a structured approach to her surveillance made her feel *"fairly happy"*. It was agreed that at present it was a bit of a lottery as to whether or not you were under the care of a Consultant or Centre which co-ordinated your treatment/surveillance. The group suggested that a University Hospital was the next best place if a MEN clinic was not available locally, however, district general hospitals were not perceived as having the necessary expertise for treatment and surveillance. Where best practice already exists it was felt that Patient Support Groups could help make this information available to all patients and their families or carers, to ensure

equity of access and choice. Participants in the AMEND Focus Group also felt that travelling to a multi-disciplinary clinic should be offered to those who wish to choose such an option.

Current treatment and surveillance for these six rare genetic conditions was reported to be sub-optimal by almost all participants and phone interviewees, only two people said they were satisfied with their care and treatment. Many reasons were cited for this perception and the participants gave positive criticism and feedback of the difficulties they have experienced, to help inform development of services. It was agreed that a Protocol is essential for each condition and that Networks of healthcare professionals would help increase provision of services by: disseminating information informed by research to help increase knowledge and awareness for doctors and health service staff involved in their care; and enhancing communication between healthcare professionals involved in their care to help ensure patients get the care they need without feeling they have to 'fight' for it or 'prove' they have a rare condition requiring treatment. Many participants felt that unnecessary delays in treatment increased their risk of developing serious complications or led to a progression of their disorder.

This series of Focus Groups has provided evidence of the way in which patients and their families with these six rare genetic conditions access currently available information and services. The seven themes highlighted in this report will be developed further with input from patients, their families and carers together with healthcare professionals involved in their care. A Family Route Map is planned for each of the conditions and a generic template will also be developed for other Patient Support organisations to use as the basis for their own Family Route Map.

- Services were considered to be a bit of a "post code lottery".
- Possible budget restrictions on services for people with long term genetic conditions were felt to cause difficulties in getting the right treatment or surveillance.
- Patients want a "multi-disciplinary" approach to their care that is co-ordinated by an expert and follows a standard protocol.
- Specialist Networks of healthcare professionals could raise awareness of these rare disorders, spread information to other clinicians and enhance communications between each other.
- practice nurses as part of the primary care team could be a 'listening ear' or an advocate.

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 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.
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.

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