

Family Route Map Online Questionnaires

This project aims to develop Family Route Maps to help signpost patients with rare genetic conditions to current information and services available to them. The first stage of this project explored the experiences of patients, their families and their carers affected by six rare genetic disorders: Barth Syndrome, Gorlin Syndrome, Multiple Endocrine Neoplasia Disorders (MEN), Myotonic Dystrophy, Nail Patella Syndrome, and Syndromes without a name (SWAN). They were recruited via support groups and asked to attend a series of focus groups, with some supplemental telephone interviews. In addition, a short online questionnaire looking at the same issues was posted to the Genetic Interest Group (GIG) website with links to the condition-specific Patient Support Groups. The data from these questionnaires is presented here.

A total of 64 questionnaires were completed by patients and/or relatives or carers (see tables 1 and 2), who were affected by a range of the six different conditions (see table 3). Respondents were asked about the care that they currently receive in terms of the number of hospitals they visit (see table 4), the distance they travel to these hospitals (see table 5) and the treatment/surveillance that they receive (see table 6). For those who responded to these questions, the majority visit between one and three hospitals, travelling up to 20 miles to their nearest hospital but up to 100 miles to their furthest hospital. Three respondents travel over 150 miles to their further hospital. Four do not attend any hospitals: one who has Myotonic Dystrophy; and three who have Nail Patella Syndrome, one of whom specifies that they have a mild form and another that they are monitored by their GP. It is not known whether the other two respondents receive treatment or surveillance outside of the hospital setting. These responses indicate that most patients are receiving treatment/surveillance for their condition, but about half have to travel to more than one hospital (so care is not co-ordinated in one location), and some have to travel a distance to access this surveillance. Patients with the same condition are receiving different types of surveillance, which may be due to different individuals having different manifestations or different levels of severity. However, it might indicate that patients are not all receiving the optimal surveillance for their condition, and this would suggest that clearer guidelines are required to outline the necessary patient care.

Respondents were asked if they had received information or guidance on the appropriate care for a patient with their condition, and if they had received any helpful patient information (see table 7). Almost three-quarters (47) said they had received guidance on patient care, whilst a few more (51) had received helpful patient information. These numbers may not give a true representation as some

respondents did not appear to make a distinction between these two types of information. However, these questions clearly showed that the main sources of any information were the hospital/medical profession and, in particular, the support group/charity (see table 8). The hospital mainly provided verbal and written information, whilst the support groups provided information and guidance in a range of different formats: verbal, written, website, email/online forum, groups, telephone and conference (see tables 9 and 10). This may mean that the information is more accessible to patients and their carers as they can access it in a variety of ways.

The last three questions on the questionnaire asked respondents what difficulties they had encountered, what improvements they would recommend and for any further comments, all in free text form. These responses were coded according to content, with answers from different respondents outlining similar issues, and the codes were sorted into themes (see page 8). The initial coding process used very specific codes to capture the detail of respondents' answers, and it was found that the codes revealed the same themes as emerged from the focus groups and supplemental telephone interviews. These are information; communication; education; diagnosis; empowering patients; ethical, social and legal issues; and treatment and surveillance.

Information: Respondents talk about a lack of information, with a need for more information for both patients and medical professionals. Many respondents found information on the internet, particularly from support groups, who are seen as important for both information and support. Better signposting is needed, both to appropriate surveillance/treatment and to support groups.

“After living with condition for 65 years, undiagnosed for 30 years, I have learned more about it in the last year from the NPS society than in all that time.”

“I had no access to information until the World Wide Web arrived!”

Communication: Communication between medical professionals and patients can be difficult, especially as patients often have to provide information about the condition so they need medics to be willing to listen and learn with them. There are also difficulties about communication between different medical specialities, with some doctors unwilling to contact the specialists in these rare conditions, and transfer to other specialties (including genetics) can be difficult.

“Contact with medical profession is very difficult, hard to explain needs and get what I need.”

“Easier access between specialists. Ideally someone to take overall responsibility for care so that they could refer to specific specialists when/if the need arose.”

Education: There is a need for education of healthcare professionals as there is little knowledge about these rare conditions and some patients have had their symptoms dismissed by professionals. They often have to educate their doctors about the condition, which can become frustrating, and find that doctors focus on individual symptoms rather than considering all of the effects of the condition.

“A medical professional who is aware of the illness and has a general understanding of medicine rather than needing to constantly explain the illness to different doctors to get 1 symptom treated.”

Diagnosis: Many patients experienced delays in diagnosis, largely due to lack of knowledge of the condition and to health professionals dismissing their concerns. Diagnosis is very important to enable access to services and benefits, leaving those without a diagnosis at a disadvantage.

“In the absence of any diagnosis, it is almost impossible to get any help at all.”

Empowering patients: Some individuals have had to fight for what they need as medical professionals have dismissed their concerns, and they see Patient Support Groups as very important for providing the necessary information and support. They felt that there should be better signposting for patients, both to the support group and in terms of the appropriate treatment and surveillance they need for their condition, particularly as other family members may also need surveillance or testing to determine if they too are affected or at risk of the genetic disorder.

“We as a family have had to battle endlessly to get our daughter any help at all.”
“Support group is invaluable both from an informative viewpoint and support viewpoint.”

Ethical, social and legal issues: Several respondents have found it difficult to access support and a few mention it being a family condition, which indicates some of the psychosocial impact of the condition and may indicate ethical issues. Respondents raised a number of issues around policy and funding, including abolition of prescription charges and inefficient use of NHS resources.

“Access to services is difficult and there are not enough professionals - but it seems to me that resources are not always used efficiently.”

Treatment and surveillance: Many respondents wanted access to a specialist in the condition, preferably working within a multidisciplinary team, and some said they were prepared to travel for this. There is currently insufficient signposting to appropriate treatment, and co-ordination of care is needed as there are many practical difficulties with appointments, such as long waiting lists and having to attend for lots of different appointments. They also wanted equitable access to services for all, as access is dependent on where they live.

“The setting up and publicising of MEN specialist centres with access to an appropriately experienced multi-disciplinary team.”

“A key worker or one person to co-ordinate between services.”

Analysis of these online questionnaires has given an interesting insight into the experiences of patients with rare genetic conditions and their relatives/carers. The quantitative data provides information about patients' access to surveillance and treatment, which indicates a lack of consistency in patient care. The qualitative responses reveal difficulties experienced by patients, which correspond with those identified by the focus groups and supplemental telephone interviews. This correlation validates the identified themes, indicating that they are likely to be an accurate reflection of patient experiences. There is a lack of information and guidelines for patients who have these rare genetic conditions, and it is hoped that the Family Route Maps will help to signpost patients and their families to appropriate information and services so that they find it easier to access the care that they need.

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Table 1: Number of respondents who are patients and who are relatives/carers

	Patients		Relatives/Carers	
	No. of respondents	%	No. of respondents	%
Yes	43	67.19	56	87.5
No	21	32.81	7	10.94
Not answered	0	0	1	1.56
Total	64	100	64	100

Table 2: Number of respondents who are patients and/or relatives/carers

	No. of respondents	%
Patient and relative/carer	35	54.69
Patient, not relative/carer	7	10.94
Relative/carer, not patient	21	32.81
Not answered	1	1.56
Total	64	100

Table 3: Condition affecting respondent

Condition	No. of respondents	%
Barth Syndrome	4	6.25
Gorlin Syndrome	13	20.31
MEN Disorders	5	7.81
Myotonic Dystrophy	10	15.63
Nail Patella Syndrome	25	39.06
SWAN	7	10.94
Total	64	100

Table 4: Number of hospitals attended

Number of hospitals	No. of respondents	%
0	4	6.25
1	24	37.5
2	11	17.19
3	13	20.31
4	3	4.69
5	2	3.13
Not answered	7	10.94
Total	64	100

Table 5: Distance travelled to nearest and further hospitals

Distance to hospital (miles)	Nearest hospital		Furthest hospital	
	No. of respondents	%	No. of respondents	%
0-5	18	28.13	7	10.94
6-10	10	15.63	6	9.38
11-20	13	20.31	11	17.19
21-30	2	3.13	8	12.50
31-50	3	4.69	8	12.50
51-100	6	9.38	7	10.94
101-150	2	3.13	4	6.25
>150	0	0	3	4.69
Unknown	1	1.56	1	1.56
None/not answered	9	14.06	9	14.06
Total	64	100	64	100

Table 6: Surveillance received by patients with the different conditions

Surveillance	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	-	-	-	-	-	-	-	-
Physio	-	-	-	-	-	-	-	-	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

Table 7: Number of patients who have received information and guidance on care for patients with the genetic condition

Received information?	Information about patient care		Helpful patient information	
	No. of respondents	%	No. of respondents	%
Yes	47	73.44	51	79.69
No	16	25.00	13	20.31
Not answered	1	1.56	0	0
Total	64	100	64	100

Table 8: Sources of information about patient care and of helpful patient information

Source of information	Information about patient care		Helpful patient information	
	No. of respondents	%	No. of respondents	%
Hospital	22	34.92	13	20.63
Support Group/ Charity	36	57.14	48	76.19
Contact a Family	0	0	2	3.17
Medical articles	2	3.17	1	1.59
Not specified	1	1.59	0	0

Table 9: Type of information about patient care provided by different sources

Type of information	Hospital	Support Group/ Charity	Contact a Family	Other
Verbal	15	12	0	0
Written	6	19	1	2
Website	0	21	0	0
Email/online forum	0	4	0	0
Group	1	11	0	0
Telephone	0	5	1	0
Conference	0	5	0	0

Table 10: Type of helpful patient information provided by different sources

Type of information	Hospital	Support Group/ Charity	Contact a Family	Other
Verbal	8	15	2	0
Written	5	24	0	1
Website	1	31	1	1
Email/online forum	0	4	0	0
Group	0	1	0	0
Telephone	0	7	0	0
Conference	0	3	0	0

Qualitative Codes Sorted Into Themes

Information

1. Insufficient patient information
2. Insufficient information at diagnosis
3. Internet as an important source of information
4. Importance of support group (information and support)
5. Insufficient signposting, to treatment and/or support group

Communication

7. Dismissal by health professionals, have to fight to fulfil needs
8. Patients have to provide information to health professionals
9. Communication between health professional and patient
11. Transfer between specialties difficult

Education

6. Health professionals have insufficient knowledge/awareness
7. Dismissal by health professionals, have to fight to fulfil needs
8. Patients have to provide information to health professionals
10. Focus on symptoms, not holistic

Diagnosis

2. Insufficient information at diagnosis
12. Delay in diagnosis
13. Lack of diagnosis

Empowering Patients

4. Importance of support group (information and support)
5. Insufficient signposting, to treatment and/or support group
7. Dismissal by health professionals, have to fight to fulfil needs

Ethical, Legal and Social Issues

20. Policy/funding issues (access to benefits, implementation of NICE guidelines, inefficient use of NHS resources, difficulties with insurance, abolition of prescription charges)
21. Family condition
19. Difficult to access support

Treatment and Surveillance

5. Insufficient signposting, to treatment and/or support group
14. Access to specialist/MDT

15. Point of contact to answer questions
16. Co-ordination of care
17. Access to appropriate services available to all
18. Practical difficulties with appointments (waiting lists, number of appointments, distance)