Genetic Interest Group

Working to benefit all people affected by genetic disorders

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by guarantee (No 05772999)

Report of the Family Route Map Project
General Practitioners Focus Group

Held on 17th May 2007 in Cambridge
Facilitator: Anna Lane, Co-Facilitator: Melissa Winter

Introduction

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
- 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.

This focus group was held in response to what patients with or at risk of a genetic condition together with their relatives and carers told the Genetic Interest Group (GIG) at a series of six focus groups designed to explore: the information needs; experience of service provision; and possible content, for specific-condition *Family Route Map'* plus a Template that could be used generically by other Support Groups.

Method

Eight General Practitioners (GPs) at a Primary Care practice in Cambridge participated in a discussion around the emergent themes from the earlier patient focus groups. The seven identified themes are:

- **Information**
- **Communication**
- **Diagnosis**
- **Treatment/Surveillance**
- **Education of healthcare professionals**
- **Ethical, Legal & Social issues**
- **Patients, and parents/carers being empowered.**

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by guarantee (No 05772999)

In order to explore these areas participants were invited to discuss each theme around a vignette describing a hypothetical consultation with a patient presenting at their surgery (see Appendix 1).

Results

Diagnosis, Treatment and Surveillance

GPs described the need to uncover patients' expectations and match these to the current information regarding rare genetic conditions and services available to them or their families and carers. Participants discussed the risk factors for more common inherited cancers and referral guidelines to NHS Clinical Genetic Services, including the need to take a Family Tree (Pedigree) in the primary care setting.

> *"They do have protocols and there was also a useful flow chart in one of the medical journals about genetic cancer so with 90% of people you are never going to get past the first stage, they do not fit the criteria."*

It was agreed that some patients with a family history of cancer however, could require reassessment and one participant stated,

> "*With that information I think we ought to go back and re-visit their file and start again."*

Participants accepted that delays in diagnosis or patients being mistakenly diagnosed with another condition can sometimes happen when patients have rare genetic disorders. However, this was set in context by the need to balance over-investigation of people with the low incidence of these conditions in the general population.

> *"The problem is that if it is the first thing that springs to mind we would be investigating a large number of people who don't have such a rare syndrome and there are consequential adverse effects for the patients in being over-investigated. And so I think every doctor has enormous sympathy for those who have experience of delays in diagnosis whether it is somebody's fault or just the nature of the condition but there are going to be cases that take a long time to reach diagnosis and that's until a screening tool can be developed to facilitate early diagnosis in a non-intrusive and non-damaging sort of way…"*

Consideration was given by the group to cases where children with complex needs do not have a diagnosis, for example, Syndromes without a name.

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by guarantee (No 05772999)

> *"For some of these conditions there is no diagnostic test and genetics might seem like 'the test'…so it is as said earlier, about dealing with the expectations and understanding that it is uncertainty and doubt that they are living with…"*

A suggestion was made by one participant for GPs to help parents of such children.

> *"…reflect it back to the specialists on their behalf…and one might do a letter on their behalf asking paediatricians if they would review the case and possibly instigate a genetic referral."*

Possible reasons for requests from patients who have already been diagnosed with a rare genetic condition were discussed by the group and whilst it was generally agreed to support such a request there wasn't agreement on how to establish the mechanism for doing so. Some participants felt it would be appropriate to approach the Consultant who had first given the patient their diagnosis but it was recognised that this could be difficult if there had been a disagreement between the Consultant and the patient.

> *"I don't see this as a big issue…unless this is hiding some problem with the first Consultant, which might be a personality clash or that he's not very good at communicating…"*

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

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

It was acknowledged by the group that for many rare genetic conditions there may only be a few Consultants in the UK with expertise in a particular condition. It was agreed that best practice would be for a Consultant to inform a GP of other clinicians with a specialist interest in order that a patient may have a choice of whom to consult for a second opinion.

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

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

Genetic Interest Group

Working to benefit all people affected by genetic disorders

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by guarantee (No 05772999)

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

> *"Sometimes all they want is a translation."*

Further discussion followed about the types of information that can be found on websites and whether or not it is reliable. Some patients in the previous focus groups had said they found quite frightening information by searching the internet themselves and GPs also expressed concern.

> *"As soon as you make the choice to search for information, nobody can protect you from finding things that you might find distressing…"*

Recommendation of websites to patients for reliable, up-to-date sources of information was felt by the group to be extremely challenging.

> *"It's difficult because I don't want to recommend a site if I haven't got the faintest idea about the site…and because there are a lot of these sites I wouldn't necessarily know…which to recommend. Isn't there a genetic website that brings a lot of these links together?"*

A discussion followed regarding websites which provide links to Patient Support Groups for rare genetic disorders and the GIG website www.gig.org.uk was highlighted together with the charity, Contact A Family www.cafamily.org.uk by the Facilitator as examples.

Software is available for GPs and health professionals in primary care to help patients find websites offering support, however, the group was not confident that such software lists information relating to very rare conditions.

> *"Part of our job is signposting and we do have software for self-help groups and also I've checked we do have, recently, patient directories in the waiting room for information on those national self help-groups."*

Ethical, legal and social issues
The Facilitator asked the group to consider if there is a role for Practice Nurses to be a 'listening ear' and provide emotional support for patients with rare genetic disorders following a suggestion that came from one of the patient Focus Group meetings. One participant agreed that although services for children with chronic conditions are provided,

> *"…(there) is not much for chronically ill younger people."*

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity No 1114195
A company limited by guarantee (No 05772999)

However, it was felt that the role of the Practice Nurse encompasses elements of supporting patients in the ways suggested.

> *"But I think practice nurses do, do that, but not in a sort of comprehensive way… I think there are various Practice Nurses who do feel they are a 'listening ear.'"*

Patients, families and carers had also expressed a need for psychological support and suggested these services are scant and not easily accessible and one participant commented.

> *"There will be some practices and Practice Nurses who will form a helpful bond with that particular family but it doesn't mean to say that a practice would like to have a service because that would potentially leave our nurse doing very little else other than talking to these families with chronic health concerns…and I think what you're talking about is forming ad hoc support links wherever they can; patient support groups, their family and friends. Hopefully they find a core of people who they can lean on and the Practice Nurse and the GP may be a part of that but not necessarily in every case."*

However, it was recognised by the group that the number of existing patients living with chronic health conditions is significant and Practice Nurses already see a great many so the amount of support they can offer is limited.

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

Summary

Data from the six patient focus groups showed many similarities in the way in which patients, their families or carers, whatever their genetic condition, described the availability of information and services, the way in which these are used, and where there are gaps in services or a lack of information. Some patients, relatives and carers with rare genetic conditions consider the level of support they receive from primary care to be helpful while others would like more. GPs acknowledge that these patients may face difficulties in getting a diagnosis and have limited choices for clinicians whom they see and where they receive treatment and surveillance. GPs in this focus group are well informed of the risk factors and protocols for referral to a Clinical Genetics Unit for genetic cancers but for rare genetic disorders it is appears less clear.

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by
guarantee (No 05772999)

Participants from earlier focus groups identified referral to NHS genetics services as essential, perceiving it as helpful for patients and families with or at risk of genetic conditions and valued the further information that can often be gained from consulting with Clinical Geneticists and Genetic Counsellors. In this focus group it was agreed that empowering patients with sources of information and support is part of the GPs role, however, this must be gauged individually and supported by explanation and translation of medical/scientific material together with emotional support where required. Importantly, engaging patients in a discussion to establish their expectations is seen as paramount in order to be able to match them to currently available information and services. Some patient focus group participants had insightfully expressed this as a need for 'honesty' from healthcare professionals.

Conclusion

This focus group demonstrated a large degree of congruence between what patients want from primary care and how GPs wish to deal with their concerns and issues. Albeit, there remains a need for further information about rare genetic conditions to be easily accessible by primary care practitioners, for example, medical websites where healthcare professionals can learn a little about the condition and types of surveillance or treatment available, especially as there may be no absolute proven (evidence based) treatment for many of these conditions. This lack of treatment protocols and care pathways for patients affected by rare genetic conditions also reflects the difficulties GPs may have when selecting a speciality to refer patients to, when seeking a second opinion.

Many patients use information technology to look up more about their condition but often would like help in finding sources of reliable information together with help in understanding it. Additionally, patients, relatives and carers want support to utilise this information. Presently acquiring this help is on an ad-hoc basis with no consistency for patients, and their families or carers. Participants in this focus group preferred to remain non-judgemental regarding sources of information and in particular websites, however this presents a dichotomy as there is clearly a need for reliable information which calls for a value judgement on the part of primary care health professionals when approached to provide information.

Discussion

How could primary care and Practice Nurses in particular, have a greater role in the care of patients with long-term genetic conditions and be a resource for reliable sources of information for patients and families to provide greater continuity of care with much needed psychological support?

Further Information

Working to benefit all people affected by genetic disorders

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity
No 1114195
A company limited by guarantee (No 05772999)

The full report of the patient focus groups **Family Route Map Project: report of a series of six Focus Groups** can be freely downloaded from the website of the Genetic Interest Group http://www.gig.org.uk/docs/FocusGroupReport_final_colour.pdf
or for further information please contact the authors Anna Allford anna@gig.org.uk or Melissa Winter melissa@gig.org.uk or phone 0207 7043141

Acknowledgements
We would like to thank all those GPs who took part for their help and willingness to be involved in this patient-led project.

Genetic Interest Group

Working to benefit all people affected by genetic disorders

Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Tel: 020 7704 3141
Fax: 020 7359 1447
email: mail@gig.org.uk

Registered Charity No 1114195
A company limited by guarantee (No 05772999)

Appendix 1

Case 1
Information
A lady of 30 presents because she thinks she may be at risk of breast cancer as it is in her family. What sorts of information could you provide her with?

Case 2
Communication
A mother says her young baby has been diagnosed with a genetic condition and wants to see a consultant who specialises in that condition for a second opinion. How do you approach this request?

Case 3
Diagnosis
A patient who has symptoms of irritable bowel says a relative has been diagnosed with a genetic cancer syndrome and wonders if he could also have it. How would you manage this concern?

Case 4
Education of healthcare professionals
A patient brings in lots of information after searching the internet because they have found out that they have a genetic condition in the family and need help to understand the information. How do you proceed?

Case 5
Empowering patients, parents and carers
A carer of another patient with a genetic condition asks for help to get more support for themselves in managing the care of their spouse. What sort of assistance can you provide?

Case 6
Treatment/Surveillance
A patient wants to know more about treatment options, including specialist surgery for a genetic cancer syndrome. How can you help this patient?

Case 7
ELSI
A parent with a young child who has developmental delay and difficulties in nursery wants to discuss genetic testing for their child and also education needs. What would you do?