



Quality, and Meeting the Expectations of Patients and Families

**A report regarding the meeting of GIG's stakeholders in
Scotland that took place at the MRC Human Genetics Unit,
Edinburgh on Monday 24th April 2006**

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INTRODUCTION

The Genetic Interest Group is an organisation well placed to help facilitate partnerships between genetics patients and health professionals allowing patients and their families opportunities to directly influence service developments and so achieve change in this aspect of health care, planning and delivery. To this end, GIG hosted a meeting of its key stakeholders in Scotland. The purpose of the meeting was to discuss issues around genetics services in Scotland and how these impact on genetics patients and their families.

The meeting was by invitation only. Those present consisted of patients and their families, health professionals and representatives of other relevant groups such as commissioners, researchers and public health physicians.

A fruitful and productive meeting took place in a very positive and constructive atmosphere. It was clear that GIG's presence in Scotland is very welcome and that there are very important roles for GIG to play in Scotland. The meeting provided the basis for an action plan for the role that GIG can play in leading the development of good practice regarding how listening to patients and responding to their needs, desires and aspirations can be introduced into health service delivery in Scotland.

The meeting, chaired by Alastair Kent, Director of the Genetic Interest Group, was held under Chatham House rules. This means that the discussion in this report is not attributable, and it inevitably reflects the author's interpretation. While every effort has been made to report this in an even-handed manner, responsibility rests with the author alone.

Thomas Barclay
Assistant Director
Genetic Interest Group

ATTENDING

Maggie Ponder	GIG
Alastair Kent	GIG
Thomas Barclay	GIG
Gillian Scott	GIG
Anna Lane	GIG
Buddug Williams	GIG
Dr. Wayne Lam	Consultant in Clinical Genetics, Western General Hospital, Edinburgh
Dr. Zosia Miedzybrozka	Head of Service and Clinical Genetics Consultant, Aberdeen
Dr. Jonathon Berg	Clinical Genetics Consultant, Dundee
Margaretha van Mourik	Genetic Counsellor, Glasgow
Dierdre Evans	NHS Scotland National Services Division, Edinburgh
Lynn Hutchinson	Project Manager, Pregnancy & Newborn Screening Programmes, Edinburgh
Dr. Harden Carter	Public Health Consultant, Lothian NHS Board
Pippa Millican	DEBRA Scotland
Arlene Smyth	Turner Syndrome Support Society
Anne Wilson	Contact a Family
Jessie Roberts	PAMIS
Maureen Phillips	PAMIS
Brenda Garrard	PAMIS
Oonagh Wilson	Campaigner, Breakthrough Breast Cancer
Helen Macfarlane	Hirschsprung's & Gut Motility Disorders
Dr. Gill Haddow	Innogen
Dr. Maureen Boxer	Former Lead Scientist, UKGTN
Alix Groom	Genetic Support Groups, Northern Region, Newcastle upon Tyne

Following a brief welcome and introduction from Maggie Ponder, Chair of the Genetic Interest Group, Alastair Kent gave an overview of the Genetic Interest Group, its history and *raison d'être*.

Founded in 1990, the Genetic Interest Group (GIG) has grown to become a UK-wide alliance of organisations with a membership of approximately 140 charities supporting families and individuals affected by genetic disorders. Its primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them. GIG also aims to promote the transfer of scientific advances into products and services quickly, and to combat adverse discrimination arising from the abuse of genetic data.

GIG aims to work and cooperate in partnership with other people and organisations, seeking common ground where possible. It seeks to avoid being at loggerheads with anyone, but is also unafraid of controversy whenever it feels it is appropriate: GIG is pro-research and endorses such issues as stem cell research, the use of animals in research and the rights of individuals to make personal choices on the basis of information and knowledge and free of pressures to select one 'expected' outcome over any other. GIG is the only UK organisation of its kind.

The organisation is very much involved not only with its member organisations, but also very actively and directly with and on behalf of people and families who are, or could be, affected by genetic conditions and disorders. These include groups who might otherwise have no voice or influence on government policy and the provision of the medical and care services on which so many of them depend.

Much of the work GIG carries out enables groups to have a voice where otherwise they may have had none. For example, GIG responds on behalf of genetics patients to government consultations and similar exercises carried out by other public and private sector bodies. Through GIG, genetics patients can influence government, policy makers and opinion formers and ensure that their voice is heard.

GIG's strength arises from continuous liaison and interaction with its members, with health professionals, patients and families, the media, the pharmaceutical industry, academia, the NHS, politicians and the public at large. Its knowledge, experience and expertise has developed its reputation as an organisation able to speak from the viewpoint of individuals and families affected by genetic disorders, and as an

organisation with a significant and respected ability to address issues that fall within its remit.

GIG's Board of Trustees consists of people representing a cross-section of its membership, and includes patients, carers and family members. Member organisations nominate people to stand for election at the Annual General Meeting. This ensures that Trustees are familiar with and involved in the issues pertinent to GIG. They are people who have experience, directly and indirectly, of the health service.

GIG is very much a democratic organisation, and this is reflected in the 'one member one vote' system at our AGM, and also when our membership is consulted on key issues.

GIG has access to a wide network of contacts in both the lay and professional communities. These people provide advice, support and specific inputs to the activities of the organisation as and when the need arises.

GIG set up *GIG in Scotland* not only to fulfil its remit of being a truly UK national organisation and to allow those affected by genetic disorders and their families in Scotland to enjoy the same opportunities of representation already enjoyed by their counterparts in England and Wales, but to also to establish a presence and prominence in Scotland to enable it to be in a position to directly assist and support individuals and families affected by genetic disorders in Scotland. Scotland is in many ways a different country from England and Wales with its own institutions, structures and agendas. Since devolution these differences have become greater, and continue to do so. But the desires and expectations of patients are similar and are independent of geographical location.

Since her appointment as GIG's Development Officer - Scotland last September, Gillian Scott has already contributed significantly to the genetics community here. In arranging this meeting to consult with its stakeholders in Scotland, GIG seeks to listen and learn and to form alliances, and to define its role and strategy in Scotland to maximise the influence of patients and families to the genetics debate and ensure our limited resources (but unlimited energy!) are used to best advantage.

Gillian indicated that she had spent time and effort since her appointment in getting to know (and be known by) support groups in Scotland and finding out what are the key issues with respect to genetics services in Scotland. She is seeking to engage with the media to promote genetics in Scotland; she has contributed to issues around familial breast cancer and

also stem cell research; she is getting involved with public engagement and health professional education through engagement with, for example, Generation Scotland and ScotGen. She hoped to get involved with the development of a user-group forum (possibly involving e-mail) and the measurement and monitoring of quality of genetics services in Scotland.

The NHS Scotland National Services Division is keen to have close involvement with patients and patient groups. The molecular genetics consortium of 4 laboratories has worked together collaboratively and successfully. A 'user group' of clinical geneticists meet every 6 months or so and monitor quality standards, and provide advice with regard to which genetic tests should be prioritised.

Quality Improvement Scotland (QIS), an organisation akin to NICE in England, set standards for clinical services. Genetic staff and GIG intend to approach this is the organisation to request that standards are set for genetic services in Scotland, but the timescale for this initiative is as yet unknown.

The view was expressed that patients and health professionals may share similar expectations as to what a genetics service should provide. However, this is not always so, and any quality standards programme will need to address the divergent and, occasionally mutually exclusive, expectations of patients and families and clinicians. This will require patients and families to be consulted.

QIS's inspection teams are likely to consist of people connected with QIS, People First and patient and family representatives. The issue as to whether patient and family participants in these inspection teams will be paid was raised. This seemed likely.

It was pointed out that Contact a Family have been established in Scotland for a number of years, and so GIG should ensure that it works in partnership with CaF to avoid unnecessary duplication. Contact a Family is a UK-wide charity providing advice, information and support to the parents of all disabled children - no matter what their health condition. It also enable parents to get in contact with other families, both on a local and national basis. The information they provide is also useful to health professionals. Well known for its directory, its helpline is also very important. Its support pack for health professionals, *Working With Families Affected by a Disability or Health Condition*, establishes standards of best practice.

The opinion was expressed that, similarly to experiences in England, many patients and families referred to genetics services have little idea as to why they have been referred. (However, the situation in North East Scotland may be a little different as most patients there will have had an opportunity for a preliminary discussion with a genetic counsellor before meeting a clinical geneticist.) The opinion was also expressed that there may well be ignorance amongst non-specialist clinicians as to why they are referring (or why they should refer) patients to a genetics service, and also what a genetics service is and what it can provide.

The discussion identified that, as well as a need for public education regarding genetics and genetic conditions, there is also a need to raise professional awareness regarding these as well as genetics services. This is important to minimise the dissemination of poor information and advice to patients and their families by non-specialists in clinical genetics.

A need for information in the form of leaflets, DVDs, websites and other forms of electronic communication (e.g., e-mail, text messages, etc.) was identified. This would assist with keeping patients 'in the loop' regarding their conditions, the tests involved and their treatments. Interim reports regarding the progress of genetic tests, the timescales involved (and why these may be weeks or months or even years), and their results need to be provided in meaningful and patient-friendly ways and with greater clarity as to what test results mean (negative as well as positive results; what phrases such as 'in the clear' mean; what the consequences are of being a 'carrier'; etc.). Patient-friendly wording is required that allows patients to feel that they are kept in touch with developments and that they and their test have not been lost in the system. This could be along the lines of 'Testing has continued up to this point. Testing continues.'

Response time targets for some genetics tests have been agreed, and these should be published in a manner that is accessible to patients. Patient support groups could assist disseminating this information to patients.

This kind of information could also benefit non-specialist professionals. It was agreed that there is a need for information for professionals, and for this to be disseminated in the form of courses and workshops, magazine articles and websites in a manner coordinated with the programmes and outputs of other bodies, such as the Royal College of General Practitioners.

A general need to distinguish between 'genomics' and 'genetics services' was identified.

Health professionals also require advice and protocols regarding the coordinated management of patients with complex genetic conditions.

Patients and their carers are a potential training resource for health professionals which has never been adequately appreciated or exploited.

Issues around the consequences of the geography of Scotland were identified. There are many rural and remote areas, and there is a need to ensure equity of access to genetics services, and also to the kind of information discussed earlier, that is independent of post-code.

Major research is relatively well-funded in Scotland by organisations such as the MRC and the Wellcome Trust. However, it is not so easy to obtain funding for the small projects (of the order of perhaps a few thousands of pounds) that are essential to bridge the gap between the laboratory development of a test or procedure and its introduction to 'mainstream' clinical practice. The profile of these small but important projects needs to be raised with potential funders.

In England and Wales GIG has produced training materials in the form of booklets and workshops for members of research ethics committees, and there certainly seems to be scope to replicate this in Scotland.

Finally, it was suggested that Gillian could map out the services that were currently available to genetics patients and families in Scotland, assess what works well and what doesn't work quite so well (if at all) and use this to identify and prioritise needs.

SUMMARY OF OUTCOMES

Several inter-related and overlapping central themes in which GIG could/should be involved were identified by the meeting:

COMMUNICATION – It was essential to get this right between the various stakeholders represented at the meeting. Some very useful direct and practical steps were identified, and these were particularly aimed at helping patients and families to receive the information they need to know and in appropriate forms.

For example, information in the form of leaflets, DVDs, websites and other forms of electronic communication (e.g., e-mail, text messages, etc.) could be employed to help keep patients ‘in the loop’ regarding their conditions and treatments. Interim reports regarding the progress of tests and test results could be provided in a meaningful and patient-friendly way and with greater clarity as to what test results mean (negative as well as positive results; what does ‘in the clear’ mean; consequences of being a ‘carrier’; etc.).

Some of this would also benefit non-specialist professionals as well as patients. GIG could participate in efforts to ensure that information, and the dissemination of information in the form of courses and workshops, magazine articles and websites fits in with the programmes and outputs of other bodies, such as the Royal College of General Practitioners.

Tweaking and adjusting what communication is already being done is one way forward in ways that would not overload or slow down the system. It was discussed that some patients could possibly receive information directly from laboratories at the same time as their GPs and consultants (e.g. common sex chromosome abnormality leaflets).

The guidelines for GPs produced by Contact a Family (and available on CaF’s website - www.cafamily.org.uk) were identified as a good model.

Furthermore, a need to develop communications between professionals (clinicians and researchers) as well as directly between these professionals and patients and families was identified.

Patients and carers are a training resource for health professionals, and GIG could be instrumental in coordinating their contribution to this.

GIG could help devise and test relevant documents ('forms of words') that tell the things that need to be said in universally understandable ways.

QUALITY STANDARDS – An intention to have these in Scotland was clearly stated, but what they will be, how they are to be determined and monitored and how and when they are to be implemented is still to be determined. GIG can help with this.

PROFESSIONAL AWARENESS – Awareness among professionals regarding clinical genetics is patchy. How this can be remedied is not yet clear, but perhaps carefully selecting targets (e.g., midwives, ante-natal clinicians,) for awareness training opportunities is one possibility.

PUBLIC AWARENESS & SUPPORT – This could involve other initiatives such as Generation Scotland and ScotGEN. Also, GIG could partner other organisations and become involved in their events (e.g., science fairs).

EQUITY OF ACCESS – There is a need to map what already exists (and where it exists) in the provision of genetics services in Scotland. This would allow disenfranchised and geographically remote groups to be identified and appropriately targeted.

ETHICAL ISSUES – GIG could participate in the provision of appropriate support and education for ethics committees and their members in Scotland in the manner already provided in England and Wales.

RESEARCH – There is a need to identify, and provide much needed support for, those matters that relate to pre-service delivery phases of genetics services development that require relatively modest funding and that are essential, but are difficult to obtain funding for. There is a role for GIG to participate in the leverage of funding from, for example, appropriate government departments.