

gig today

summer 2007

On the road again! GIG abroad.

The international importance of genetics in healthcare is becoming ever more apparent. The recognition of the need to involve patients and families in all stages of the development and delivery of high quality services and support to individuals and families is also growing rapidly and GIG is being called on to provide this input more and more. This means that GIG's staff are on the road more both in the UK and further afield.

Much genetic research is carried out by international consortia rather than by individuals or teams based in one university or academic hospital. Such projects usually cost tens of millions of pounds (dollars or euros) and they are directed by steering committees representing the partner institutions and the other interested bodies. One of these is a coordinated project studying gene therapy known as "Clinigene". I am a member of the International Advisory Board for this project, with a particular remit for communication to patients and families and the general public. At a recent committee meeting (in Seattle) this was a major agenda item and as a result more information about what is actually happening in the field of gene therapy (and

there has been considerable progress recently) should be coming out in the near future.

At the other end of the spectrum, families with genetic diseases from poorer countries in the developing world need genetic services just as much as those in the developed world do. A major international meeting in Rio de Janeiro brought together national governments, major international bodies such as the US Centre for Disease Control and the National Institutes for Health as well as scientists, doctors and patient groups to plan for better services. GIG was asked to play a leadership role in this, offering support to patient groups from throughout South America to develop this role and boost their impact on behalf of their membership.

Back at home cost and clinical effectiveness is an issue for the NHS and for all other European Health Care systems. Planners need to know what works, for who and can they afford it! Health Technology Assessment (HTA) is a growing academic discipline increasingly used by bodies like National Institute for Health and Clinical Excellence (NICE) to make recommendations about new interventions in healthcare. For these to be sound it is

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(l-r) Maggie Ponder, Chair, Dee Heaps and Alastair Kent, Director

Founding Member and trustee of GIG retires

Dee Heaps, from the Tay Sachs Disease Association was one of the founding members of the Genetic Interest Group and has worked tirelessly as a trustee ever since. Although it was as a member of the Tay Sachs association that she came to GIG her experience went far beyond. In particular she had spent many years working in the field of mental health and in this area her knowledge was invaluable to us. Dee has supported GIG in many ways throughout her time with us and we are extremely grateful for all her kind words of encouragement and support. Dee was very sad to go but said that she has met some fantastic people here at GIG and that she will remain in touch through our mailings.

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On the Road again!

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essential that patients and families are able to express their views as to what is important, and what they want to see happen with regard to new treatment opportunities. GIG has been working with other bodies on how to value the outcomes from genetic services (especially where there is no cure for a condition) so their impact can be properly appreciated. This work was presented to the HTA community at their annual conference in Barcelona, where it provided an important extra dimension to more narrowly focused measures of cost and clinical effectiveness (used by health service planners to decide where resources should be spent). The more that planners can be helped to understand the impact of genetic conditions from the families point of view, the better decisions they are likely to make, with consequent benefit in the form of improved services and support for individuals and families.

Perhaps the most tangible impact of working internationally came out at the end of May when after a vigorous lobbying campaign by GIG and other European Patient Organisations, including Eurordis and the European Genetic Alliances Network the Advanced Medicinal Therapy Regulations were adopted by the European Parliament at the Council of Ministers in the face of opposition from the Rapporteur and a group of Christian Democrat and Green MEP's. These regulations create a Europe-wide framework for

licensing novel therapies such as gene therapy and those using stem cells and will be a great encouragement for researchers in this fast moving field. Without a coordinated patient and family campaign it is doubtful that these regulations would have been adopted so quickly or in a form that has so much potential benefit for developing and introducing novel therapies.

Despite our global perspective, GIG remains firmly rooted in responding to the issues facing UK families, and much work has been going on here at home – on policy issues such as the review of the in-vitro fertilisation legislation, or practical matters of service delivery and the provision of information to all citizens across our multicultural society and in raising awareness in the media and elsewhere of the importance of genetics and of the needs and expectations of those affected by or at risk from genetic disease. Many of these aspects of our work are highlighted elsewhere in this edition of GIG Today.

I should also like to take the opportunity to wish you all a well deserved summer break. The support we get from our members is invaluable and we appreciate the efforts you make to work with us greatly.

Alastair Kent

European Society of Human Genetics Nice, France.



GIG had a strong presence at the European Society of Human Genetics conference this year. Both Anna Allford, myself and Celine Lewis had poster abstracts accepted on the developments within our projects - the Family Route Map Project www.gig.org.uk/familyroutemap.htm and the Eurogentest project www.eurogentest.org.

The ESHG had over 1000 delegates from all over Europe and further afield talking on a diverse range of subjects from "The future of genetic counselling and of genetic services" to "Cytogenetics" and "New technological advances in genomics"

Many of the sessions were very useful to the work that we carry out here at GIG and have given us plenty of issues to discuss and review now we are back in the office.

GIG was also part of the European Genetic Alliance Network (EGAN) stand where we distributed patient information leaflets and spoke to delegates about the work that GIG and EGAN do on behalf of patients affected by genetic disorders.

The delegates were from a variety of backgrounds mainly clinical and molecular genetics. Many of the healthcare professionals from other European countries were very interested to see how patient groups are run and set up in the UK and the Netherlands and were keen to help their patients in setting up their own support networks, which was extremely encouraging.

Lobbying on behalf of patients

In the last issue of GIG Today, we explained some of the lobbying work that we've been doing in the UK and in the EU. We're delighted to be able to write in this issue about our success on a couple of these issues.

Advanced Therapy Medicinal Products Regulation

Briefly (this a highly technical and complicated regulation), this regulation was conceived as a framework for the legislation of advanced therapies and medicinal products (ATMPs). Regulation of ATMPs, which are tissue therapy, gene therapy, cell therapy, and other complex treatments, would be performed centrally by the EU. This would mean a high Europe-wide standard of safety, and a normalised licensing procedure. This common landscape across the European Union would encourage investment in the new technologies required to deliver ATMPs, as companies can be sure of a large market once they obtain an EU marketing authorisation for their product. The end result of this concept would be an environment in which investment in ATMPs is encouraged and their safety assured. (For more information check your GIG today back-issues!)

The fly in the ointment is a familiar one. Some ATMPs are likely to be derived from, or containing stem cells of some kind. This is a familiar debate for the Genetic Interest Group, and the reason we took a strong lobbying stand point on this issue. At the worst point in the progress of the regulation through the EU system, the regulation had been turned on its head, with the possibility of a ban on centrally licensing ATMPs derived from or containing stem cells. This would have been worse than not having the regulation in the first place.

Our response was a strong coordinated patient lobby, aiming to explain the value that patients of incurable or intractable conditions place in progress toward better therapies. Last issue we were awaiting the plenary stage, after two years of progress. This is the stage at which all MEPs can vote to accept the bill or send it back for revision. The odds were stacked against us: it was the first

reading of this bill, the MEP in charge of it was anti-stem cell research, and there was a lot of controversy surrounding the bill from other MEPs. The argument as we presented it was essentially that the lives of patients who live with intractable conditions should supersede our opponent's ethical concerns regarding the use of stem cells. That no potentially fruitful avenue of research should be closed before it has been explored fully.

Emotions ran high. Table manners were ignored as an opposing Green MEP Hiltrud Breyer, stormed out of a dinner debate organised by our colleagues and friends EPPOSI. A press conference, organised by Eurordis, the European Organisation for Rare Disorders, and a collaborator on the lobby, was high-jacked by another opposing MEP. In retrospect, perhaps this reflected our progress, because the regulation was adopted in its first reading, with a landslide acceptance by MEPs.

Human Tissue and Embryology (Draft) Bill

As the last issue of GIG Today explained, the Genetic Interest Group and the Association for Medical Research Charities (AMRC) organised a letter to the Prime Minister regarding our concern at the possibility of a ban on cytoplasmic-hybrid embryo research; a technology that has potential to improve the availability of embryonic stem cell lines for research. Our letter was not ignored, combined with a letter to The Times from the scientific community and the results of the House of Commons' Science & Technology Committee's findings, the Department of Health has indicated a change of position in the draft bill.

Both of these lobbies by GIG have had to deal with the controversial argument for and against the use of embryonic stem cells in research. It is a difficult argument to win and to be involved in. However, we will continue to make the argument for research and work to ensure a regulatory environment that encourages and supports research.

Nick Meade

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Patients' Experiences to Inform Health Professionals

‘Telling Stories: Understanding Real Life Genetics’ has been launched. This website will help health professionals, in particular those in the nursing professions to better understand peoples’ experiences of living with a genetic condition so that it can improve their practice.

Freely available on the Internet at: www.geneticseducation.nhs.uk/tellingstories it is a web-based teaching and learning tool



Jon Snow who launched the website, discusses the website with contributors and Telling Stories team members

developed by a project team led by the University of Glamorgan with GIG, the Wales Gene Park, University of Plymouth and the NHS National Genetics Education & Development Centre. It was funded by the Wellcome Trust.

The site is a collection of stories, mostly from family members but with some health professionals and cover a range of conditions. The website relates all of the stories to key skills which nurses, midwives and health visitors must know by the time that they register. Using narrative to communicate key skills for professional practice is an effective way for students to learn, understand and appreciate

issues which are important to families. The resource is now being linked to many undergraduate nursing courses and is also being promoted for those who already qualified.

Mick Mason, founder of FAPgene.org.uk and a storyteller himself commented: “When I was first diagnosed with familial adenomatous polyposis (FAP) I was told about the threat to my children. It made me think how many people out there are in the same position? I wanted to tell my story to pass on my own experiences to help raise awareness of genetic issues to doctors and nurses”. Currently, there are 95 individual stories on the website. GIG members in particular have generously shared their stories and can be seen on the website. If you would like to share your story through this website, or would like to learn more about the resource, please contact Dr Emma Tonkin, University of



Mick Mason, a Storyteller talking to Dr Heather Skirton, a member of the team, about his contribution to the website file

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The views and opinions in this newsletter are not necessarily those of the Genetic Interest Group.

Scot-Gen Project

I am just over half-way through my six months of working for GIG, and the time is flying by. One of my main focuses in the past few weeks, has involved contributing to the Public Health and Society module of the ScotGEN e-learning project, currently under construction in Scotland.

ScotGEN (The Scottish Genetics Education Network) is a network of individuals in Scotland with an interest in genetics education. It was established in 2005 as collaboration between the four Scottish genetics centres and Napier and Robert Gordon Universities. One of the recommendations of the Calman Report (2006) and the Review of Genetics in Scotland (2006) was the provision of genetic education across Scotland with the emphasis on the commissioning of a Scottish MSc in Genetic Counselling.

It is hoped that the ScotGEN education resources would reach out beyond undergraduates and masters audiences and would engage with other professional and public groups that share an interest in genetic health issues. On the completion of the project a website will be available.

Rebecca Skillen

Involving community is the key to better translation

The current gold standard for translating information is to 'back-translate' into English, to check the sense. However the Genetic Interest Group (GIG) and the London IDEAS Genetics Knowledge Park have been developing a new protocol. This forms part of the Translation Project which aims to provide accurate and understandable genetic information for patients in eight minority ethnic languages – Arabic, Farsi, Gujarati, Punjabi, Somali, Sylheti, Turkish and Urdu. Bilingual lay groups have been involved in the Translation Project from the very beginning to evaluate the English version of the leaflets that were created as well as to look at the translated text. They have provided vital feedback to the project that is not always available through back-translation alone.

It is vital when providing complex information on important and sensitive issues, that it is as clear and as accurate as possible. When GIG held focus groups with members of each community they highlighted areas that could be improved which we could not have detected through back-translation alone. There were instances where a correct word had been used by the professional translators but those from the community were not as familiar with it and in some cases did not recognise the word. The level of the translated language was in places also felt to be too "high brow". Other amendments that came out of the focus groups held were also key to the accuracy and understandability of the information, for example in some of the languages there is no word for "parent" only the plural "parents" and when explaining inheritance patterns it was felt to be important that the words Mother and Father were used with diagrams rather than the plural word for parents. Another comment that was noted was that there was only one word for infant, toddler and child - the distinctions that exist in English don't exist in that particular language

There are more than 300 languages spoken in London, and the city receives the largest proportion

of new immigrants to the UK. This project identified the need for patients to have access to accurate information in their mother tongue within genetics and it has now translated 37 leaflets, forms and letters into the languages mentioned, with an individual tailored glossary for each leaflet. The leaflets range from information about specific genetic conditions to general genetic concepts such as inheritance patterns. The leaflets are freely available and we hope they will be widely used within clinical genetics departments around the UK as well as by patients and patient organisations. The master glossary of genetic terms that has been created has already been received warmly by interpreters who work within hospitals. This glossary along with leaflets containing images to help in the description and understanding of inheritance patterns and will be valuable tools to help interpreters.

The first 4 languages (Gujarati, Punjabi, Urdu, Turkish) are now available on www.londonideas.org/translations for the rest of 2007. In the longer term they will be provided from the GIG website www.gig.org.uk

All languages will be available within a few months online once final amendments and adjustments have been made.

Sylheti and Somali will be available as audio files, as this reflects the need of the community

Many of the translations are the only resource of their kind, in a particular language, within the UK, and we are delighted to be able to fill such a needed gap in service provision.

With over 300 languages spoken in London alone GIG has continued work to do in helping patients to access up to date, accurate and accessible information on genetics in order to help them in the decision making process.

Melissa Winter

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Have you heard of the Compact and

The Compact is an agreement between the Government and the voluntary and community sector made in November 1998 which details how the two should work together. It aims to improve the relationship between the two sectors for mutual advantage. The Compact is made up of five codes of good practice, these can be downloaded in full from <http://www.thecompact.org.uk>. These documents act like a legal agreement with clear points that outline both government undertakings and undertakings by the voluntary and community sector. The five Compact Codes of Good Practice are:

Funding and Procurement

The Funding Code was originally published in 2000, and revised in 2005.

The Code seeks to improve funding and procurement relationships, to the mutual advantage of the funder and funded, and safeguard the sector's independence. The Code is for everyone in England involved in distributing, seeking or receiving public funds.

Consultation and Policy Appraisal

The Consultation and Policy Appraisal Code was published in 2000. The Code is a tool for ensuring effective consultation and better policy outcomes both for the voluntary and community sector and government.

The Code states that wherever possible a 12 week consultation should be made available for replies to allow for different working practice across the voluntary and community sector. Where this isn't possible consultation documents should specify why a shorter time has been set. Consultations should always be fully publicised to target audiences taking into account specific needs, and interests.

Black and Minority Ethnic Groups

The BME Code was published in 2001. It recognises the significant role that the BME voluntary and community sector plays in building stronger communities. The BME sector also represents a broad range of faith groups, refugee and asylum seeker organisations.

The Compact has significant benefits for the BME sector, including better use of limited resources and linked services. Sharing information within partnerships can also lead to a more inclusive community plan and better positioning for BME organisations and groups when it comes to delivering services. The Compact Working Group has urged the BME sector to use the BME code and laws such as the Race Relations Amendment Act

2000 to demand seats at decision making tables and drive forward change.

Volunteering

The Volunteering Code was published in 2001 and revised in 2004.

The Volunteering Code identifies four key principles fundamental to volunteering:

- Choice – Volunteering must be a choice freely made by each individual.
- Diversity – Volunteering should be open to all no matter what their background age, race, sexual orientation, faith, etc.
- Reciprocity – Volunteers' offer their contribution unwaged but should benefit in other ways in return for their contribution to wider social objectives.
- Recognition – Explicit recognition of the value of what volunteers' contribute to the organisation, to the community, to the social economy and to the wider social objectives is fundamental.

Community Groups

The Community Groups Code was launched in 2003. The Code seeks to provide understanding of the community sector and community groups including recognition that this sector has particular characteristics including greater autonomy. Community Groups also provide services not delivered by statutory agencies.

The Code stresses the need to include faith groups as partners and recipients of funding as faith groups reflect broad ethnic diversity.

Local areas are now producing local codes of good practice as part of Local Compact development work to better reflect local priorities. There are compact regions and they have each drawn up their own codes of practice based on ones mentioned above

The Compact Advocacy Programme

The Compact Advocacy Programme was set up, and is run, by the National Council for Voluntary Organisations (NCVO), it is a scheme that provides practical support and wider campaigning to the sector in cases where the Government has breached the Compact.

Since 2002, the Compact Advocacy Programme has been instrumental in ensuring compliance with the Compact at a national level through advocacy, campaigning and lobbying government departments on behalf of the sector. The NCVO works with organisations to design and implement a campaign

how it could help you?

in order to change the way a government department or local authority works. The Programmes work at both national and local level and have shown that with the help of an advocacy service Compacts can be given "teeth" and can transform the way the voluntary and community sector and government interact with each other.

Local Compact Advocacy

The Compact Advocacy Programme has secured funding from the Big Lottery Fund to expand its work. The latest phase of the programme includes a positive drive to implement local Compacts and bring about a major change in the relationship between the voluntary and community sector and local government bodies. By taking up individual instances of Compact breaches and engaging in wider campaigning the Compact Advocacy Programme provides a mechanism to ensure local Compacts work.

How Can Compact Advocacy Help You?

Voluntary and community organisations who believe that their local council or other local public bodies have breached the Compact should contact the Compact Advocacy Programme at the NCVO.

GIG's members using the Compact.

How can the compact help you? One of GIG's members, Kay Parkinson from Alstrom Syndrome UK has recently had cause to use the compact and her story was told in the Third Sector earlier this year and is reproduced with their permission on this page.

More Information

For more information on the Advocacy work that the NCVO do for voluntary organisations involved in The Compact please look on the NCVO website at <http://www.ncvo-vol.org.uk/thecomact.asp> or contact Saskia Daggett on 020 7520 2581. For more information on The Compact see <http://www.thecomact.org.uk/> **Melissa Winter**

Compact in action: Alstrom Syndrome UK and the Department of Health

A tiny charity for sick children has used the Compact to secure its share of public funding.

The Compact can help the smallest of charities in their battles with large state bodies. Kay Parkinson set up Alstrom Syndrome UK a decade ago after her two children were diagnosed with the condition, which leads to childhood blindness and obesity because of insulin-resistant diabetes. It is extremely rare: fewer than 400 people worldwide are diagnosed with it.

The charity, of which Parkinson is the only full-time employee, set up the first database of UK families affected by the condition (there are 30) and raised money to establish specialist surgeries at Birmingham Children's Hospital and Torbay Hospital.

In April 2006, the agency formerly known as the National Specialist Commissioning Advisory Group, which oversees the commissioning of specialist health services in the UK, awarded each hospital's

Alstrom clinic roughly £130,000. But the terms of the grants specified that the money could not be spent on voluntary organisations.

"We weren't happy," Parkinson says. "We collected the only UK database on Alstrom Syndrome, yet the hospitals were getting paid while I was expected to provide surgeries and clinics for free. We are delivering a public service: they can't do it without us - they haven't got the mechanism to bring in families."

Then Parkinson heard about the Compact. "The more I read about it, the more I thought that just about everything in the agreement was being ignored," she says. "I was amazed to find that no one at the NSCAG or at two hospitals that I spoke to knew about it."

She contacted the NCVO, and in January its Compact advocacy programme wrote to the NSCAG

explaining that the grants had breached the Compact by failing to recognise the value of working in partnership with the voluntary sector.

A meeting followed in February. Parkinson then received a letter from the Department of Health offering a one-off payment of £46,000 and a pledge that her organisation could be included in future NSCAG funding applications. The charity and the hospitals have since submitted a joint £120,000 bid.

"I don't know how we would have managed this year without the Compact," Parkinson says.

John Plummer,
Third Sector, 6 June 2007

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Eurogenguide - the first few months

European genetics are some of the most advanced in the world. As the technology advances, it will be possible to treat an increasing number of disorders. The continent possesses enormous potential to treat an ever increasing range of conditions. In view of this, there are an increasing number of people for whom testing or screening will become an option over the course of their lives. In order to progress, it is crucial that the public participates in research via the donation of DNA samples to biobanks. However, the network that supplies information about genetics is less advanced than the technology that it supports. The EuroGenGuide project has been devised in order to help close the gap. The project began in January 2007 and will last for three years. I am employed at the Genetic Interest Group as the project officer, collaborating with twelve partner organisations around Europe. Each organisation and representative was approached because of the high level professional knowledge and expertise that they can bring to the project.

The guide will have two parts – one providing information for patients and the public explaining what is involved in testing, screening and research, the other containing educational guidelines for health professionals, outlining the principles of good clinical practice in respect of new genetic treatments and procedures, such that patients are able to make an informed decision about what is available, and whether or not to take advantage of them. This is crucial, as a necessary condition of achieving autonomy in decisions about treatment is the means for patients to make an informed choice based on high quality, up-to-date information. Another reason for the creation of the EuroGenGuide project is to help increase the amount of DNA samples donated to biobanks, via raising the standards of information

available to the public. These two aims are complementary and hence if the EuroGenGuide project helps in achieving these aims, the resulting enhancements in the clinical delivery of genetic technology will help to improve the lives of those with serious and debilitating genetic conditions.

This June I attended the ESHG annual conference in Nice, where there was a short progress meeting with some of the EuroGenGuide partners. The conference was a great opportunity more generally to raise the profile of, and publicise the project and its aims. Discussions focused on work carried out over the last three months, since the first meeting in late March. Since then two draft

protocols – for gender issues in European genetic testing, and informed consent guidelines – have been written and will be reviewed in light of what is said in Nice. Another major development has been the first and second drafts for the design and layout of the EuroGenGuide website. The project has a wide remit and hence the website must fulfil a great many demands for a great range of people, patients, the public and health professionals alike. In view of this it is a great challenge to be

involved developing something that, developed successful, will be so useful to so many. I will be presenting the designs and, once again, will move on to the next stage of development in view of the feedback I receive.

More recently I was asked to write an article about the EuroGenGuide to feature in the European Parkinson's Disease Association newsletter, 'EPDA Plus'. This is a great opportunity once again to raise the profile of the project, and I hope that the second half of the year proves to be as fast-moving and successful as the first six months.

Alex McKeown

Eurogenguide Partners

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 michael livingston - heart europe
 raluca nica - gamian, romania
 annetter dumas - alzheimer europe
 avril daly - fighting blindness, ireland
 george veckmans & genevieve pierquin - rare
 disorders belgium, belgium
 ysbrand poortman - wanda, netherlands
 christina funnell - health coalition initiative, uk

Family Route Map Project

It has been a busy time since the publication to the GIG website of the first Report for this project which details the series of six Focus Groups held in 2006 for patients, their families and carers www.gig.org.uk/docs/FocusGroupReport_final_colour.pdf

Based on the findings we have moved forward developing a framework for a generic Family Route Map and commenced the first three of the six condition-specific Family Route Maps. We are currently working on those for Barth Syndrome, Gorlin Syndrome and Multiple Endocrine Neoplasia (MEN) and work will commence on the remaining 3 conditions; Myotonic Dystrophy, Nail Patella Syndrome and Syndromes without a name, in the second half of the year. The next stage is to ask patients and others with experience and an interest in each condition to review the draft version. So as to assess the Route Maps we are asking people to complete a short Quality Survey looking at the layout and content. If you are interested in helping with the project by being a reviewer for any of the six conditions we should be most grateful for your help, please contact either Anna or Melissa at GIG anna@gig.org.uk or melissa@gig.org.uk or phone 020 7704 3141.

The data from the on-line survey which widened the participation for those affected by the conditions, their relatives and carers beyond the focus groups, has been analysed with the help of our Volunteer, Nicki Taverner. This has confirmed the issues and concerns found in the focus groups as being more wide-spread and interestingly, no further themes were identified. The new information fitted into the seven themes already established through the analysis of the focus groups; Information, Communication, Treatment/Surveillance, Diagnosis, Education in genetics for healthcare professionals, Empowering patients, parents & carers, and Ethical, legal, & social issues. In order to gain the views of General Practitioners (GPs) regarding these seven themes we recently held a focus group to explore some of the issues raised by patients, their families and carers in relation to primary care services. This GP focus group demonstrated a large degree of congruence between what patients want from primary care and how GPs wish to deal with their concerns and issues. Albeit, there remains a need for further information about rare genetic conditions to be easily accessible by primary care practitioners, for example, medical websites where healthcare professionals can learn a little about the condition and types of surveillance or treatment available, especially as there may be no absolute proven (evidence based) treatment for many of these conditions. 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. The full report can be

found on the GIG website at http://www.gig.org.uk/gig/docs/GPFocusGroupReport_final.pdf

One possible future project that could help address some of the needs for consensus around treatment and surveillance of patients with rare genetic conditions involves a pilot for Networks of healthcare professionals with an interest in a particular genetic condition or group of conditions. To determine if our Members would be interested in these Networks and find out more information about existing such networks, we undertook a rapid appraisal (on-line survey) and gathered the views of our Member organisations and their Advisors during May and June. We have looked at the information you sent us and believe that it supports a project if we can get funding for it. Thank you to all who responded to this survey and completed the questionnaire or sent us their comments.

Mike Knapton, GIG Trustee and one of our Advisory Group, referred us to an important Department of Health Consultation 'Commissioning Framework for Health and Well-being' at the meeting of the Family Route Map Project Advisory Group in May. Although the deadline for a response to the consultation was extremely tight we decided it was very important to ensure that Commissioners understood the needs of patients with rare genetic conditions and included this in their proposed Joint Strategic Needs Assessment. The overall effects on health and well-being for the population as a whole will without doubt benefit from many of the changes outlined in this consultation paper, however, if Commissioners seek only 'local' views then patients with rare genetic conditions will remain invisible. If you wish to have our full response, the document is freely available to download from the GIG website www.gig.org.uk/latest-news.htm

We have also spent some time in the last couple of months investigating how we can spread awareness and disseminate some of our results to date. Articles for publication have been prepared and we are continuing to write further reviews and clinical papers. Our achievements so far include:

- An article accepted for publication by the Association of Genetic Nurses and Counsellors;
- A Poster Presentation at the European Conference of Human Genetics in June 2007; and
- A Poster has also been accepted for the Conference of the British Society of Human Genetics taking place in September 2007.

If you would like further information about the project or would be interested in getting involved with reviewing the draft versions of the Family Route Maps please contact Anna Allford anna@gig.org.uk or Melissa Winter melissa@gig.org.uk or phone 0207 7043141.

Anna Allford and Melissa Winter

Summer 07

PHG Foundation

In May this year GIG visited the PHG Foundation in Cambridge to find out what they were doing and what they planned to do over the coming years. GIG met with its Director Ron Zimmern, who explained the background to the organisation and where he hoped it would go in the future.

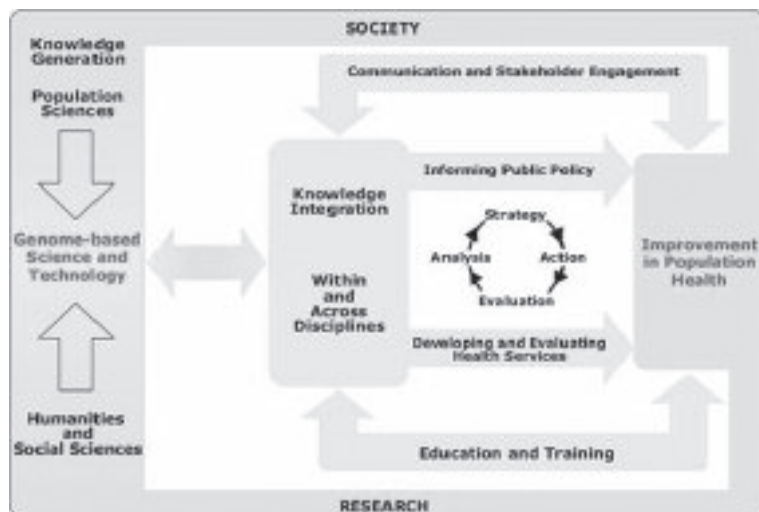
The origins of the charitable PHG Foundation lie with the Public Health Genetics Unit (PHGU) in Cambridge. Founded in 1997, the PHGU later formed the core of the Cambridge Genetics Knowledge Park, one of five such parks funded by the Department of Health and the Department of Trade and Industry. In April 2007, the PHG Foundation was established as an international charity, with the vision of realising the maximal health benefits from advances in biomedical science; this mission builds on the ground-breaking work in public health genomics, but also expands beyond previous experience to embrace a still wider range of opportunities to improve population health. Having already secured funding for 5 years, Executive Director Ron Zimmern is already looking ahead to ensure the ongoing work of the PHG Foundation the foreseeable future.

Public Health Genomics as a discipline

So what is public health genomics, and what will the new Foundation be doing? Public health genomics a recently emergent discipline pioneered in the UK by the Cambridge specialists, has been defined as 'The responsible and effective translation of genome-based knowledge for the benefit of population health'. Genomics refers to the whole spectrum of scientific and clinical knowledge and

Diagram taken from the Graph-Int website
<http://www.graphint.org/>

understanding that is emerging following the sequencing of the human



genome. What does this amount to in practical terms? Ron Zimmern explained "We are agents of change; if there is something that needs to be done, we try to make things happen", Much of this is achieved by working through existing structures such as the NHS and regulatory systems. Practitioners of public health genomics are more than academics, confined to a narrow field of interest; rather, the PHG Foundation supports the improvement of human health through all potential avenues, such as the legal system, or social customs. "We focus on areas of policy and frameworks for health" said Ron Zimmern, "rather than looking at just the genetics itself".

To work effectively, the PHG Foundation needs to understand not only genome-based science and medicine, but also population sciences such as epidemiology, bio-statistics, information and management science, along with the humanities and social science too. Knowledge integration lies at the core of public health genomics, and so the Foundation integrates the new understanding derived from research and shapes it within four distinct areas, listed below and also in the diagram:

- 1) Communication and stakeholder involvement
- 2) Informing public policy
- 3) Developing and evaluating our services
- 4) Education and training.

Public health genomics is often said to involve translational research; however, Ron Zimmern felt that this can be a confusing term, as it means different things to different people. In fact, there are two different forms of translation:

Type 1 translation - sometimes referred to as 'from bench to bedside', this focuses on the transition from basic research to clinical applications.

Type 2 translation -this goes beyond the bench to bedside transition, to anticipate and develop services and policy that will allow clinical implementation of new products and interventions.

The importance of type 2 translation was highlighted in the recent Cooksey Review (A Review of UK Health Research Funding, December 2006), which looked at publicly funded health research in the UK; Ron Zimmern observes that the PHG Foundation is ideally placed to perform and

advise on the measures involved in making this final step from new clinical applications into actual health care practice.

Work of the PHG Foundation

The strategic objectives of the PHG Foundation are:

1. To identify the potential of biomedical science to benefit health and to disseminate that knowledge for public benefit
2. To contribute to the integration of biomedical science into mainstream clinical and public health services
3. To foster a social and regulatory environment receptive to the application of biomedical science for health
4. To promote the development of systems and policies for the evaluation of technologies that derive from biomedical science
5. To work with partners to provide education and training to support the responsible application of biomedical science for health.

Ron Zimmern explained: "It's a very exciting time, bringing people together and working with public

health colleagues all over the UK and further afield".

For the first year, the PHG Foundation will focus on three key areas. The first is the evaluation of genetic tests and other diagnostics and biomarkers. The second is cardiovascular genetics, where work will focus on moving genetics into mainstream medicine by bringing together stakeholders to consider the role of genetics in diagnosis, prevention and treatment of cardiovascular disease. This will include both rare single gene disorders and more common forms of heart disease. The third area the Foundation will focus on is the

use of fetal DNA present in the mother's blood for genetic screening and diagnosis; this novel technique allows early and non-invasive testing.

GIG and other voluntary organisations will play a role in helping to disseminate the work of the PHG Foundation. The PHG Foundation is also working with colleagues in Europe through the Public Health Genomics European Network (PHGEN), and (EuroGentest the European Network of Excellence concerned with laboratory genetic testing services) and across the world as part of GRaPH-Int (Genome based Research and Population Health International network).



Cavernomas The Angioma Alliance UK

When we broke for lunch at the 2007 AGM for the Long-term Conditions Alliance, someone approached me who had the misfortune of seeing me at another meeting. She remarked, "You are the person representing that rare brain condition." I gave her a leaflet about cavernous angiomas. I was "pleased" by her response "Oh, it's not that rare is it?"

Cavernous angioma, cavernoma, cerebral cavernous malformation (CCM) or cavernous haemangioma, the condition has all these names, occurs in approximately 0.5% of the population or 1 in 100-200 individuals. (According to the Parkinson's Disease Society of

the United Kingdom, Parkinson's Disease affects 1 in 500 people).

Why have you not heard of cavernomas? Because, unfortunately, some misdiagnosis still occurs. Also, fortunately, many cavernomas do not bleed remaining asymptomatic to the individual meaning their discovery only occurs in post-mortems and are not the cause of death.

Cavernomas are clusters of abnormal blood vessels in the brain or spinal cord. Rarely cavernomas can occur elsewhere in the body. Cavernomas can cause haemorrhages, seizures, headaches and significant/permanent neurological deficits, namely imbalance, slurred speech, double vision and

imbalance. Cavernomas look somewhat like raspberries; they consist of many little bubbles (caverns) of various sizes filled with blood and lined by a special layer of cells (endothelium). These cells are similar to those that line normal blood vessels, but they lack the other layers of protection found in normal blood vessels, and the bubble-like structures of cavernomas sometimes ooze blood.

Depending on their location, cavernomas that haemorrhage can cause seizures and other neurological deficits, including numbness and weakness in arms and/or legs, ocular difficulties, speech and balance problems.

A type of surgery called craniotomy (or opening of the

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skull) may be used on some cerebral cavernomas. Other techniques exist such as radiosurgery using the Gamma Knife Radiosurgery. But this remains a controversial treatment.

Cavernomas are of two types: hereditary (familial) and spontaneous. In the familial form, the illness is passed from generation to generation in an "autosomal dominant" fashion. This means that each child of an affected person will have a 50% chance of having the disease her/himself. In the sporadic form, the child of an affected individual has no greater chance of having the illness than anyone in the general public.

Three separate genes have been implicated in familial form: CCM1, CCM2, and CCM3. A mutation on any one of the three

can cause the illness.

Individuals can be screened genetically for mutations on these genes. Before beginning the clinical genetic testing process, one must know that genetic testing by sequencing the C[erebral] C[avernous] M[alformation] genes can only rule a mutation in; it cannot rule one out. A negative result does not necessarily mean that a genetic mutation does not exist. There are two reasons for this. First, there are ways that a gene can mutate that are not picked up by sequencing – follow up testing would be performed if this were suspected. Second, it appears that not all of the genes that can cause hereditary cavernomas have been discovered at this time.

Having said that, if you wish to

have genetic testing and a genetic mutation is identified, this can make it easier for other family members to be screened for the mutation. Rather than going through an MRI, other family members can submit blood, or in some cases simple cheek swabs, for "CCM Known Mutation Detection" (see below).

Gene testing for cavernomas is now available through Dr. Jonathan Berg, University of Dundee and Dr. Eric Johnson at PreventionGenetics in the USA.

For information on the genetic testing that is currently available do contact the Angioma Alliance.

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EVENTS and NEWS

The Second International Symposium of the Lowe Syndrome Trust - 'Molecular and Clinical Advances in Lowe Syndrome' Royal Society, London 7th December 2007

This event will bring together the various UK research groups funded by the Lowe Syndrome Trust, the Lowe Syndrome Scientific Advisory Board and other national and international medical professionals and scientists involved in Lowe syndrome. The programme includes 2 guest lectures and a series of short basic and clinical talks from leading UK, European and US scientists and clinicians.

Lowe syndrome is a genetic disorder caused by a missing enzyme and this can affect the brain (with seizures, mental retardation, impaired speech and developmental delay), the kidneys (with loss of important salts and nutrients, and eventual kidney failure), the eyes (with cataracts), the bones (with deformity and arthritis), and the muscles (with weakness). Unfortunately, few children survive beyond their late teens or early adulthood.

The cost of registration is £30 (£10 for PhD students and research fellows) and includes a buffet luncheon, tea and coffee. To register, please contact Lorraine Thomas on 0208 458 6791 and visit www.lowetrust.com for more information. CME accreditation has been applied for.

New Research and Policy Manager at GIG from September.

GIG is delighted to announce that we have appointed a new Research and Policy Manager, Dr Amy Hunter, who begins with GIG in September.

Amy has a PhD in molecular biology and some training in clinical genetics. She has experience in the private sector and has worked for the patient charity RNID. Most recently Amy has been the manager at the London IDEAS Genetics Knowledge Park.

Amy will be working closely with Alastair, Nick and Melissa to ensure that the work that GIG undertakes in the Policy and Research fields is robust, of high quality and will benefit those we support.