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Chair's Introduction

Measuring the efficacy of an organisation such as the Genetic Interest Group is always difficult. Monetary and numerical factors used as yardsticks for many organisations are limited and most of our projects do not fit conveniently in to the parameters of the financial year. Because of this it is often necessary to assess our progress through identifying 'Tangible Achievements' – defined as *'positive things or events that would not have happened or occurred without the work of GIG'*. During this year GIG provided its membership with a significant number of such achievements. These are all featured in this annual report and they demonstrate the broad scope of the GIG's work on behalf of patients and families whose lives are affected by genetic disorders. There are, however, two Tangible Achievements which deserve special mention.

The first is the significant part that GIG has played in ensuring that the Human Fertilisation and Embryology Bill became law without detriment to the progress of biomedical research and the welfare and prospects of our members and the patients and families they represent. Without strong campaigning from GIG there were dangers that pressure groups would have diverted the legislation away from its intended goals. Pre-Implementation Genetic Diagnosis, Saviour Siblings and Human-Animal Hybrid Embryos were the three main areas in which GIG worked with MPs and Ministers to ensure the needs of our members are met. Although the campaign was successful our work will have to go on to ensure appropriate implementation of the new law.

The second is the part GIG has played in establishing Rare Disease UK. This new organisation is a joint initiative of the Genetic Interest Group, (itself an alliance of over 130 charities supporting children, families and individuals affected by genetic disorders) and other key stakeholders including academics, doctors, healthcare planners and industry who seek an integrated programme of services and co-ordinated use of existing resources within the NHS. The aim is to get the Governments within the UK to support the EU Council Recommendation for creation of national strategies for rare diseases. Collectively rare diseases are not that rare! Some 1 in 17 people in the UK are affected by a rare disease. Currently there is no coherent strategy for integrated care and support for such people. Rare Diseases UK will strive to get such a programme implemented throughout the whole of the Country.

As to the future, the report reviews the ongoing projects which GIG is managing and provides information about new activities being undertaken through 2009 and 2010. These activities again demonstrate the important role that GIG fulfils in helping patients and families in areas where it is difficult for them to manage alone.

One major event to affect GIG this year has been Maggie Ponder's retirement from the post of Chair of the Trustees. In the six years that she has held the post Maggie's contribution has been immense.

In this time GIG has progressed significantly and has fulfilled the role its founders envisaged. Her standing in the fields of genetics and family care, together with her innate skills, have facilitated the establishment of a Trustee Board of exceptional scope and experience and an outstanding team of professional staff carrying out GIG's objectives and meeting its goals.

The immense difficulties that children, parents and families are faced with in coping with rare and little understood disorders are hard for those not affected to understand whether they be health professionals, policy makers or 'just' members of the public. Obtaining a cohesive

strategy within the NHS will be a big challenge but is one that GIG faces with enthusiasm and I believe, with the right policy and personnel. The right access to healthcare, the right to proper communication and the right information have been identified as key patient needs. They are particularly important in relation to all genetic disorders, rare or not so rare. These 'rights' GIG will continue to strive for.

Chris Friend
Chair of the Genetic Interest Group

I. Executive Summary

The benefits to GIG from winning the GSK/King's Fund "Impact" Award for 2008 continue to reverberate for us. The impact on our visibility and the creditability that this has given us has been significant. This is particularly true in respect of those who were unfamiliar with our work but who monitor developments in the voluntary sector. Hopefully it also acts as an indicator to funders (actual and potential) that we are a trustworthy and reliable organisation that will deliver on its promises and commitments effectively.

At the start of the year our Trustees and staff determined to use the opportunity that this award provided to best advantage by making sure that our systems were, as far as possible, fit for purpose and aligned with our strategic priorities. GIG is, and will remain a campaigning organisation that delivers strategic advocacy and targeted interventions in the key policy and political issues of the day. We do this from the perspective of our members, and of the patients and families who live with the daily impact of genetic diseases and disorders. We work on a wide range of issues at every level from the local to the global, depending on where the levers that need to be pulled for the benefit of people with genetic conditions are situated.

2. Patients Influencing Policy

The start of the year covered by this report was busy with the passage of the Human Fertilization and Embryology Bill. This became law on the 13th of November 2008 and is in the process of having its provisions translated into regulations that will guide clinical practice and direct what researchers are allowed to do with human embryos and stem cells. The work on this is ongoing. GIG continues to work alongside scientists, clinicians, other patient groups and politicians to ensure that the Human Fertilization and Embryology Authority puts appropriate and proportionate regulation in place to interpret the provisions of the new Act.

The passing of this Act represents the end of a long policy campaign by GIG. When the Bill was first debated, in the House of Lords, alongside a group of scientists, clinicians, research groups and other patient organisations, we lobbied strongly for a number of changes in the wording of the Bill to reflect our support for ethically sound research on as many fronts as possible. Central to this raft of changes was a reversal in the Government's decision to ban the use of human-animal hybrid embryos, and important technology to speed up research into embryonic stem cells and their possible uses.

Once the Bill reflected our needs, we changed strategy, strongly supporting the Bill as it entered the House of Commons. Central to our communication strategy with MPs were a set of innovative "cartoons" which depicted the controversial processes, such as Preimplantation Genetic Diagnosis (PGD) and the creation of human-animal hybrid embryos, simply to aid MPs' understanding of the concepts.

Our sustained and resourceful campaign to shape the Human Fertilisation and Embryology (HFE) Bill and influence its progress through Parliament attracted significant interest amongst Parliamentarians, the media and the public and led to a dramatic shift in opinion on several controversial aspects of the Bill.

“Your support and invaluable input throughout this process was very much appreciated. It was instrumental in ensuring that the 2008 Act (.....) brings science and society in a way that is fit for the 21st Century.” Rt Hon Dawn Primarolo, Minister of State in the Department of Health. (from June 2007 – June 2009)

Alongside this, GIG has been campaigning in Europe in particular on the issue of animals in medical research due to the revision of the EU Animal Welfare Directive 86/609/EEC. After a six year wait, the draft revision was published on the 5th of November. The idea behind this review is to strengthen and modernise EU law on the treatment of laboratory animals, bringing it into line with the best standards currently required in the EU, and to standardise the law across Europe. However, if the revisions are carried out without proper consideration of their impact, the continuing progress of the high quality biomedical research performed in the EU could be under threat.

GIG as a member of the European Genetic Alliances Network (EGAN) has been a vocal contributor to the debate, ensuring that patient's need for continuing high quality biomedical research is taken into account. The end of this year saw rapid progress in this review, resulting in the acceptance of the Agricultural Committees favourable revisions of the draft being accepted by the European Parliament. The process will continue through next year.

Alongside these campaigning initiatives, GIG continues to contribute our member's views to consultations and debates on issues ranging from education of doctors, to funding of expensive treatments in the NHS, genomic medicine and patients' rights.

GIG is also spear-heading a multi stakeholder consortium (Rare Diseases UK) to generate an integrated pattern of services and support for families with rare conditions. More about both of these features later in this report.

3. Working in Collaboration

Sometimes life at GIG seems to be measured out in meetings and committees. Although this is sometimes be frustrating, the number of committees where we are asked to provide representation in an indication of the success of our strategy of adopting a co-operative rather than a combative approach to those with the power and resources to improve life for those affected by intractable conditions. Without relinquishing our rights and responsibilities we are able to challenge vigorously those whose actions or pronouncements threaten our members' interest, GIG believes in working alongside others in pursuant of shared goals.

With this in mind, the establishment of a new regulatory committee, the Committee of Advanced Therapies (CAT) at the European Medicines Evaluation Agency (EMA), was something we welcomed. This is the statutory committee that will play the major role in licensing new gene, stem cell and tissue therapies in Europe. It is good news that GIG's Director and Policy Analyst have been appointed members and alternate respectively on this committee. We will play a role in ensuring that the regulatory framework for these types of therapy (which hold significant promise of interventions for many of the conditions affecting GIG's members) is appropriate and proportionate to the risks and benefits involved.

GIG's Director was reappointed for a third term to the Human Genetics Commission (HGC), where a major focus this year has been the sale of genetic tests direct to consumers. This is usually via the internet, but a number of companies are offering DNA tests as part of a private medical consultation. GIG, and the HGC, have been concerned to strike the right balance. Direct to consumer tests can play an important role in enabling individuals access to information about their predisposition to common diseases. Some of those currently on offer base the claims they make on limited research, the outcomes of which may be insufficient to support the conclusions that are being drawn. People contemplating buying a DNA test need to be able to understand the limitations on any information that may be revealed, and whether they can trust it to act upon, in order to make an informed decision as to whether or not they want to purchase it.

In a rapidly changing field the scientific goalposts are constantly shifting, so the HGC is working on a Code of Practice for those who make and/or sell genetic tests direct to consumers. This will appear later in 2009, and its publication will, hopefully, help ensure that those purchasing tests from "code compliant" vendors can be reasonably confident in the accuracy and the validity of the service provided - GIG has played an active role in this work, contributing ideas and input to the drafts as they emerged and progress towards finalisation.

4. Working in the Devolved Nations

Health care policy is devolved to the UK's national governments, and we were delighted that our campaign to support the provision of services for families with inherited hypercholesterolaemia (familial hypercholesterolaemia, FH) in Wales was crowned with success, while in Scotland our input to plans for children and young people's specialist services were well received.

Working in Wales

In Wales, GIG's campaign to improve health services for diagnosing inherited high cholesterol will make a real difference. GIG brought together families affected by FH with clinicians, scientists, and policy makers including the Wales Gene Park. Regular briefings and communications increased understanding, raised awareness of the condition and put the case for change and improvements to health services. Twenty-seven months into the campaign, the Welsh Assembly Government announced its commitment to fund a Wales-wide service to identify families at risk of this genetic condition through family cascade screening.

Annual funding for this initiative of £650,000 has now been announced in a partnership between the Welsh Assembly Government and the British Heart Foundation. The programme will help find people in Wales at risk of this condition, diagnose them, and ensure people stay healthy and it will save lives. It is also anticipated that this will serve as a model to promote development of similar services to diagnose FH across England and Scotland, as well as for inherited cardiac conditions in Wales and beyond.

Work in other areas include improvements for neuroscience services in particular for neuromuscular conditions. GIG is an active member of the Wales Neurological Alliance, a successful collaboration between several organisations which has allowed smaller groups representing rarer conditions to contribute to policy development and contribute to the planning of future health services.

Scotland- Getting Patient Views Heard

Engagement

The Patient Engagement Project has been funded by the Scottish Government from April 2008. GIG in Scotland has been busy building links between individuals and groups affected by genetic conditions, and advocacy services such as the Scottish Patients Association. GIG has been recruiting to the GIG Scottish Consultative Virtual Patient Panel and currently has 50 members. The Panel are a sounding board for patient views about genetic developments in healthcare. The panellists regularly feed patient experience into Government and healthcare groups working to improve genetic healthcare in Scotland. GIG aim is to continue to expand the membership to include as many people as possible, with experience of a wide range of conditions, living throughout Scotland including the Borders, Highlands and Islands.

Policy work

A key element of GIG in Scotland's policy work was a consultation about the Scottish Government Patients' Rights Bill in December of 2008. To effectively canvas the views of Scottish people affected by genetic conditions, the GIG patient engagement network brought together the virtual panel and other GIG members and supporters with healthcare professionals, policy makers and researchers. The consultation was organised in conjunction with Gengage (the genetic healthcare public engagement network). A full day event investigated what rights and responsibilities patients, the government and NHS providers should have. Patients and carers who could not attend the day event, also fed in their views by email and by phone. The published report gives a far-reaching description of the needs and barriers that individuals and families with genetic conditions experience.

2009 has seen considerable GIG awareness raising activity in Scotland. GIG has been supporting the European Council's recommendation for action for Rare Diseases, identifying the need for the development of a National UK plan for Rare Disease. GIG members and virtual panellists successfully lobbied their MSPs and MPs and attended a successful launch of Rare Disease UK in the Scottish Parliament on the 3rd March.

GIG reported their consultation of parents' and professionals experiences of the cystic fibrosis (CF) newborn screening programme in Scotland. This focussed on the experiences when the baby's results were 'carrier' or atypical CF. The National Services Division of NHS Scotland are using this to help to mould their service more closely around patient needs.

Committee work

GIG in Scotland are active in numerous forums, informing the political and professional discussions about the services and resources needed to provide high quality genetic healthcare. GIG has been included on several Scottish Newborn Screening committee meetings developing policies and processes for MCADD, haemoglobinopathy and CF newborn screening. GIG has also helped to identify patients for these committees. Furthermore GIG has recently been invited to represent patients' interests in the commissioning of specialist services by National Services Division for NHS Scotland

GIG is also a member of the Scottish Molecular and Cytogenetic Genetics Consortium Users Group meetings, where we raise patient issues about tests as well as the Scottish Clinical Genetics Forum, contributing at regular meetings where clinical genetic service policies and practices are developed.

The 'Calman' Review of Genetics in Relation of Genetic Healthcare in Scotland is leading to improved genetic specialist staffing levels. It has also funded several demonstration projects aimed at providing more comprehensive care across traditional health and social care boundaries. These include Genetic Care Coordinator posts and GPs with a Special Interest

in Genetics, who are each focussing on providing care coordination and multidisciplinary care for single gene disorders. Scotland-wide policies and practices in cardiac, neuromuscular genetic services, learning disability testing and genetic education are also developing at a pace.

GIG has recently joined as active members of the Scottish Cancer Coalition, and the Long Term Conditions Alliance Scotland. These are groupings of charities that are active in giving patient views and feedback on what is required in Scotland. We are of course contributing on behalf of those with genetic conditions.

5. Working at a European Level

Last year's annual report highlighted the fact that, rare diseases, when considered in a case by case basis, or condition basis are difficult for the NHS to take account of in a strategic manner. This is unfortunate, because rare diseases, taken as a whole affect over 3.5 million people in the UK, and are a cause of substantial suffering, consuming many millions of the NHS's budget. Unfortunately much of this expenditure is not well targeted. Families frequently rattle round the system, experiencing wasted consultations and unnecessary investigations as clinicians, keen to help but lacking the support, struggle to find a diagnosis. Even then, once a diagnosis has been revealed, families still find access to services and support more difficult than they need to be because of lack of coordinated planning and difficulties with the integration of different clinical inputs from across the NHS.

This is not a uniquely British problem. Fragmentation, difficulties in communication and a lack of data for evidence based medical decisions are a feature that hampers all European health care systems.

Rare diseases are one of the areas where the European Commission can play a clear role - adding value to the efforts of national health care systems. GIG has been a member of the European Commission's Rare Disease Task Force since its inception, and during the year our efforts were rewarded with the publication of a formal communication from the Commission "Communication from the Commission to the European Parliament, the Council, The European Economic and social committee and the committee of the regions on Rare Diseases; Europe's Challenges which called on Member States to develop National plans or strategies for integrated services and support to be provided to patients and families with rare diseases. The subsequent European Council's Recommendation on an Action in the field of Rare Diseases was adopted unanimously by the European Health Ministers.

Rising to the challenge, GIG has established Rare Disease UK (RDUK) a cross-sectoral alliance of patients, academics, doctors, healthcare planners and industry. RDUK will work with the Government, the Department of Health and the devolved nations to help secure a fair deal for families and individuals with rare diseases, with over 80% of rare conditions being genetic in origin we know this will have a positive impact on our members as well as a positive knock-on effect for genetic conditions that are considered more common. We believe that in the first instance much progress could be made by helping the NHS to work smarter - to spend the money it currently uses supporting patients with rare diseases to better effect. While we reserve the right to call for more investment in this area of health care, our first priority is to ensure that we secure the best health gain from resources currently being invested. Then there will be opportunity to push for more investment in research and development, and in the creation of new ways of working to address currently intractable diseases and disorders.

RDUK has been an initiative waiting to happen. It has received widespread support from all sectors, and it is clear that the time is right for this to happen. Of course the devil will be in

the detail, and our task in the years to come will be to translate this willingness into a detailed plan that helps patients and families secure easy access to integrated services and where novel therapies are developed, to be confident that they will be able to access them if they are safe, effective and worthwhile.

6. Continuing GIG Projects

While policy development and its related campaigning activities are central to GIG's mission we must not underestimate the contribution that our project based work makes to achieving our goals.

Eurogentest and EurogenGuide, two major pan European projects promoting patient access to timely and user friendly genetic information and participation in research projects as active partners, not passive "subjects" continue to deliver solid progress.

EuroGenGuide

In July 2008 the EuroGenGuide website was launched the usage has remained very high with, on average, 20,000 hits per month. The website is viewed from all over the world, not just Europe and we have had usage as far afield as Mexico, Singapore and the United States as well as much of Europe.

The core material for EuroGenGuide was tested and assessed by the project team and members of their organisations during Autumn 2008 and improved according to their recommendations to generate version two of the material. This revised material was published online in January with enhanced core material and new features such as a forum, a mailing list and newsletter system, google translation, a tool providing automatic translation into over 40 languages. Further features will be added later in 2009. The current version of the Guide has been tested by a wide ranging patient user survey all over Europe. 74 patient groups have responded from 17 countries and their comments are being used to make improvements for the final version.

Dissemination of EuroGenGuide has been extremely important this year and the Project Officer has attended various meetings and conferences to support and promote EuroGenGuide, attending the First Eastern European Summit on pre-conception health, Budapest, European Genetic Alliances Network (EGAN) and Roche Annual Workshop, Basel, Central and Eastern European Genetics Network Conference, Prague, as well as making two presentations at the European Society of human Genetics Annual Conference.

EuroGentest

The original eleven patient information leaflets developed as part of the Eurogentest project have now been translated into over 26 languages and we are already in the process of translating an additional four leaflets that relate to the various genetic tests that are currently available through genetic clinics. These leaflets are all available on the Eurogentest website as PDF's to print out or as HTML pages to view online. Feedback about the leaflets has been very encouraging.

"Thank you very much for the Czech leaflets. It is very helpful for me". Geneticist, Czech Republic

"I received a brochure with information for patients on the topic of genetics, genetic counselling and testing, which is the most beautiful thank you". Alenka Vrečar, Slovenia

"I'm very glad to have so useful resource of professional information". Medical Geneticist, Russia

"Hello, this is really beautifully, and we will certainly use it in practice." Medical Geneticist, Croatia

Last month the leaflets and HTML pages were viewed over 8,000 times on the EuroGentest website, with almost 40% of all traffic on the website relating to the patient information pages.

This year one of the main aims has been to promote the availability of this information resource across Europe. This has been done through a number of channels. An article about the leaflets was recently published in the European Society Human Genetics Journal (March 09). In addition, GIG's Project Officer has attended numerous genetic conferences across Europe giving presentations about the leaflets as well as manning the EuroGentest stand where the leaflets in their many languages were on display. Conferences attended included the European Society meeting in Vienna, Austria; The Balkans Genetics Conference in Dubrovnik, Croatia; The Working Day for Human Genetics in Prague, Czech Republic; the Eastern European Rare Diseases Conference in Plovdiv, Bulgaria, and the MediMedGen conference in Ankara, Turkey. Access to the leaflets are now available though the European Society Human Genetics website, and clinicians across Europe are regularly updated about the availability of new leaflets in their language. As this project comes to a close (the project officially ends in December 2009) it is encouraging to know that we have successfully completed our original aim of developing good quality, user friendly information to patients across Europe, whatever country they are in.

7. New Projects

GIG has started three new projects in 2008/9 "Insurance Templates" and "Facilitating Networks" and "Patient Partner" each of which, when finished will make a contribution to ensuring a better deal for patients and families facing the impact of a genetic conditions. We are grateful to BUPA, the Department of Health and the European Commission respectively for the funding that makes this work possible.

Facilitating Networks Project

The first year of the Facilitating Networks project has been a success. Many of the objectives we planned to meet have been achieved; the most significant of these being the level of patient engagement harnessed within this formative stage.

Each of the project's nine patient support groups have been very accommodating in encouraging the patients and families they support to engage in the project's consultation process. Through the use of surveys (both online and postal), focus groups and attending support group meetings, the project's first year has been grounded in the individual and collective experiences of patients and families accessing much needed health and social care.

The information derived from the Family Route Map project produced a substantial body of evidence suggesting potential benefits of clinical networks for individuals affected by rare genetic conditions. Having received over 240 responses to the Facilitating Network's patient survey, the need for this project has been reaffirmed. The recurrent issues expressed by these individuals related to the experience of disjointed services, a general lack of communication amongst health professionals and insufficient awareness of their condition. These shared experiences generated a common ground for the support groups to levy the concerns of their members and created a synergy between grouped conditions which on some levels could have been perceived as disparate. Furthermore, the rich, earnest and

relevant accounts of personal experiences strengthened the rationale for a networked approach to these problems.

The concerted effort to engage with clinicians and researchers who have knowledge and expertise of these conditions has been running alongside the patient consultations. The project employed a robust approach to identify individuals and organisations that could potentially feed into the efforts of the pending networks. Ensuring the patient support groups were at the centre of this communication was key, and in most instances the professionals who were already affiliated with the groups were approached first.

One to one interviews were conducted with professionals where they had the opportunity to volunteer their professional interests. Where clinicians were concerned, we placed an emphasis on their current, and previous experience of managing the patients with the conditions concerned. The need to map where individual and collective expertise would potentially contribute to the network's formation was at the core of all of the discussions held. Furthermore, the talks were vital in opening the dialogue to considering what areas of work the network could focus on. Through this process three clinicians emerged as trailblazers within their fields and have been identified as prospective champions for two of the networks.

The project has also been privy to advice from GIG members who have already taken a lead in developing clinical networks. These groups (PCD, Alstrom, CF Trust and DEBRA) were immensely supportive and open to sharing their respective journeys to achieving centres of excellence. This invaluable information will provide a framework for the imminent networks to evolve whilst following best practice and conserving limited time and resources.

Insurance Templates

GIG has continued its work with the Life Assurance Templates. These were previously developed in order to improve the application process for consumers, ensuring all of the *relevant* information is presented at the initial application stage. We are still working with the Association of British Insurers and various members of the Direct Office Underwriting Group to get these formally launched within the near future. It is important that Life Offices are briefed on the templates and their function within an underwriting process, in order for them to be fully integrated into applications.

GIG is committed to the concerns of our members and as part of our research it has become clear that Travel Insurance is continuing to present problems for many. GIG has been working towards creating an effective dialogue with the travel insurance industry in order to ensure positive patient outcomes. As part of this process it has been important for us to cultivate the views of as many GIG members as possible.

As such GIG developed an online survey which has been very successful and we would like to thank everyone who has taken the time to complete it. We now have well over 100 responses. Patient interviews and workshops with member groups has and will continue to provide further qualitative data which can be used to formulate themes and identify reoccurring problems in order for solutions to be developed. GIG will then be able to use this data by working closely with the industry and trade bodies to demonstrate the nature of the difficulty consumers living with pre-existing medical conditions experience when applying for insurance. As part of GIG's continuing support for our members we will be developing a handy leaflet to ensure people are fully informed about the insurance products they are buying and offer guidance to limit the risk of claims going unpaid.

Patient Partner

Patient Partner is a project which sets out to promote the role of patient organisations in the clinical trials context.

Patient Partner is based on the belief that involving patient organisations as equal partners at all stages of clinical trials contributes to research that is better adjusted to the real needs of patients. It is a three year project within the 7th framework programme funded by the European Commission. The study will take a close look at the part that patient organisations play and are willing to play in clinical trials and will mainly focus its attention on clinical trials with children, the use of biobanks and ethical issues. The Genetic Interest Group is leading on the dissemination work package for the Patient Partner Project.

8. Leading the Patient Voice

In addition to these major pieces of work, GIG has played a role in a number of other projects co-ordinated and led by others.

One of the most worthwhile of these has been “Capability” (Capacity for the transfer of Genetic Knowledge into Practice and Prevention: An International Collaborative Network) where we have been working with European colleagues from Germany and Sweden, and partners in South Africa, Argentina and Egypt. GIG has contributed to creating pilot models for sustainable service development in genetics in the three partner countries. In Egypt this has generated educational materials for couples who have had little or no formal education and who cannot read to enable them to understand the idea of a genetic risk. In Argentina large numbers of primary health care professionals in a remote Northern province have been trained via a series of workshops and seminars, and in South Africa, midwives operating in the community far away from expert help have been given training and support to help identify newborn babies with genetic problems, to treat these appropriately and guide the parents to genetic counselling clinics operated in the District Hospital for that part of their country.

Case study from the Capability Project

If a mother gives birth to a child with a congenital abnormality away from the main centres of population in South Africa, the ante natal care will be provided by a midwife. The training of midwives is such that they often have little awareness of the need to diagnosis birth defects.. There is a risk that the babies may not receive appropriate treatments and there is also a risk that the parents could carry on to conceive another child with a similar condition, if it is genetic.

The Capability Project did a short awareness training programme and provided a group of midwives with a mobile phone that could take reasonably high quality pictures. The idea was that if a baby was born that the midwife felt may not be “normal” they were trained to ask a series of questions and to take specific photographs. The answers to the diagnostic questions and pictures are then sent by SMS to a dedicated number at the clinical genetics laboratory in Johannesburg. There a clinical geneticist could make an informed attempt at diagnosis and call the midwife back to discuss the appropriate course of action. The first time this was used in practice was for a case of Trisomy 18, a devastating and lethal condition. The geneticist was able to advise palliative care and counselling for the parents, this meant that at the rural hospital one of their two valuable ambulances was not tied up for more than a day transporting the baby to the nearest tertiary centre, who would have been unable to do anything further for the baby.

As you can see from the case study the pilots are already paying dividends in health care terms, demonstrating that even a small amount of additional resources, used properly, can make a substantial difference to the lives of children, couples and families at risk.

The UK is at the forefront of developments within clinical genetic services and this project has enabled GIG to share the UK's good practices to those affected by genetic conditions in the developing world.

Another project, closer to home this time, involves the NHS in the North West, the University of Manchester and GIG. The Project "Genetics for Patients" is being undertaken by Dr Marion McAllister. The project aims to develop a new measure of outcome for clinical genetics services. This patient questionnaire could make it possible to evaluate different ways of providing those services, to ensure they were developed to meet patient needs. Because there are often no medicines or operations to offer, it has been difficult to find ways to measure the patient benefits, and services have developed without good research evidence. This project builds on previous research which identified "empowerment" as the primary patient benefit. GIG has helped in recruiting patients to complete the specially designed questionnaire, in order to validate it before it is given to patients before and after attending a clinical genetics service. This validation will be useful in helping to develop services to better meet the needs of the families it serves.

Funding from the Scottish Government enabled us to appoint a full time Patient Engagement Officer based alongside our Development Officer in the Genomics Forum (part of the University of Edinburgh) she has been building networks of patients and carers interested in supporting the improvement of genetic services in Scotland. Following the Calman Review £10 million of new money is to be invested in modernising the genetic service in the NHS Scotland. With GIG's help these developments will be user friendly and relevant to patient and family needs. Funding from the Calman Review enabled GIG to appoint a full time patient engagement officer in Scotland, patient and families able to comment on and contribute to the development of NHS services and other health and research related issues.

Also in Scotland, our Development Officer completed a survey of families taking part in the newborn screening programme for Cystic Fibrosis. The findings from this highlighted training and communication gaps that will need to be addressed for this programme to ensure that parents are well informed and willing to participate.

9. FUNDING

In reviewing the year it is impossible to avoid recognising the growing danger to our activities and, more importantly to the hopes and expectations of the families supported by all our member groups that the “credit crunch” poses. As the long term cost of measures taken to bail out banks and industries begin to hit home, it is clear that GIG, and all its members, face an uncertain future. Generating resources to support our core activities of policy and campaigning has always been challenging and it is likely to become more so. We will need to work hard to cement relationships with our existing funders and to develop awareness of our work amongst new audiences and potential supporters.

Notwithstanding these difficulties, we enter the new financial year optimistically. We have secured funding for our role in a new European Project - Patient Partner. We will be involved in a five year programme funded by the National Institute for Health Research (the NHS's Research and Development arm) called “Reliable, Accurate, Prenatal Non Invasive Diagnosis (RAPID) to manage the introduction of non-invasive pre natal diagnosis (checking the development of the fetus by testing DNA in the mother's blood) into clinical practice. We have also secured funding from the Big Lottery Fund Research Programme for a new three-year research project. The Family Risks, Common Cancers and People from Minority Ethnic Groups project will be led by GIG in partnership with the Division of Primary Care at the University of Nottingham. The project will identify why people from minority ethnic groups with a significant family history of cancer are less likely to access genetics services than the mainstream UK population and will inform the development of interventions to improve access to these groups.

During the last year we have managed to strengthen our relationships with many of our existing funders and two of our existing core grants are to be renewed for a further term. Our concerted effort to engage with new corporate sponsors has also had some success and we have attracted both cash and in-kind donations from several new sources. We will be working hard to develop these relationships in the long term.

2008/09 has been a year in which we have explored new fundraising approaches. In September, we became the first charity partner of the on-line shopping club, KidStart. By using the KidStart website, shoppers can earn savings from over 200 well-known retailers, which they can then transfer to a child's trust fund, a school or one of the five charities listed on the website, including GIG. In February we were selected as one of a group of charities to be supported by the St George's University of London RAG Week and we will receive £4,000 raised through a variety of student charity events. As well as generating income, both these activities have the additional advantage of raising GIG's profile amongst new audiences.

So while the future is challenging, it is not all doom and gloom. We are confident that we are well placed to ride out the worst effects of the credit crunch by exercising due caution in managing our assets, developing our relationships with our allies and exploring new opportunities as they arise.

Useful Links

Report on Patient Rights in Scotland

http://www.gig.org.uk/scotland/docs_scotland/Full_report_of_the_PRB_consultation.pdf

European Genetic Alliances Network

www.egan.eu

EuroGenTest Project

www.eurogentest.org
EuroGenGuide Project
www.eurogenguide.org.uk

A Record of Achievements – 2008 – 2009 (March to April)

Conference Presentations

Gig presented at Various conferences this year, including

- 1000 Genomes (May 2008)
- 1st European summit on pre-conception health and prevention of birth defects, Budapest, August 2008
- Animals in Medical Research (June 2008)
- Ante Natal Results and Choices (ARC) Debate, House of Commons (October 2008)
- Bio Vision (April 2008)
- BioVision
- CEEGN / WANDA conference, Prague, January 2009
- City Bond Insurance conference.
- Education: teaching on module on Cancer genetics (about GIG and cancer patient interest groups) & teaching on Medical Genetics Msc module on GIG and genetic patient needs and experiences
- EGAN / Roche annual workshop, Basel, January 2009
- EPPOSI Chronic Conditions Workshop (Sept 2008)
- EPPOSI Partnership in Rare Diseases Conference (October 2008)
- EuroBio Conference (October 2008)
- EuroGenGuide version 1 launch event – Mayoral reception at York Mansion House, September 2008
- European Society for Gene Therapy (November 2008)
- First Scottish Patient meeting for Alpha 1 Anti-trypsin deficiency (October 2008)
- Genzyme Patient Meeting
- GIG and Gengage launch and Consultation on Patient Rights event in Scotland (Patient and Public Engagement Networks for Genetic Healthcare in Scotland) (Nov 2008)
- House of Lords Enquiry into Genomic Medicine (June 2008)
- ITIJ Travel Insurance conference in Budapest
- Lysosomal Storage Disease Symposium April 2008
- Medical Research Council, Patient Reported Outcomes (January 2009)
- National Cancer Research Network (North East)
- National Services Division (results of GIG CF newborn screening patient and professional experiences investigation)
- Progress Educational Trust Debate (May 2008)
- RNIB –Talking Inequalities
- SAFE Final Dissemination Conference
- Scottish Genetic Counsellor Meeting (CF again)
- Scottish Genetic Education Network (results of GIG CF newborn screening education and information needs)
- South East of England Clinical *Genetic* Network Annual Conference
- Tuberous Sclerosis meeting (Sept 2008)
- Various Patient conferences and meetings including, Alpha 1 Awareness, Cavenoma Alliance, Huntington's Disease Association, Confer Launch event, TSA,

Leaflets and other publications produced

- 'Cystic Fibrosis Newborn Screening: from the Perspectives of Patients and Health Professionals'. Genetic Interest Group.
- 'Your Genes, Your Experiences' leaflet. A patient information leaflet. (GIG Virtual Patient Consultation Panel).
- EuroGenGuide report in EPDA newsletter 'EPDA Plus', Autumn 2008
- GIG insurance leaflet

Consultations responded to

- General Medical Council's "Tomorrow's Doctors" consultation

- HFEA's "Implementing the new Act" consultation
- National Delivery Plan For Children and Young People's Specialist Services In Scotland
- Scottish Government's "Patient Rights Bill" consultation
- Welsh Assembly Government's consultation on the EU Cross-Border Healthcare Initiative
- Department of Health's consultation on regulations to implement the Human Fertilisation & Embryology Act 2008

Legislation, lobbied on

- Campaign around the Human Fertilisation and Embryology Bill/Act
- Campaign around the Revision of EU Directive 86/609 governing animal research
- International Rare Disease Day hosted parliamentary receptions in the Welsh Assembly, Scottish Parliament and Westminster

Website Visitors

- Newly designed GIG website
- Over 40,000 hits per month on the GIG website
- New GIG Forum launched in November 2008
- EuroGenGuide website version 1 went online July 2008, and over the first year has retained a constant average of around 20000 hits per month, lowest around 12000 and highest 33000.
- EuroGenGuide visitors from over 60 countries, from all territories around the world, not just Europe
- EuroGenGuide wide ranging user survey carried out using website's online survey system – 74 patient groups from around Europe in 17 different countries, giving a wealth of feedback to use in improving the guide

Committees

- Association of British Insurers – Genetics Committee
- CESAGEN Advisory Committee
- CliniGene (Advisory Committee and Ethics Committee)
- EGAN/ROCHE Joint Working Group (Director – Co-Chair)
- EuroGenTest Steering Committee
- EuropaBio Patients Advisory Council
- European Genetic Alliances Network (EGAN) (Director – President)
- European Platform for Patient Organisations, Science and Industry (EPPOSI) (Director – Chair)
- GenCAG
- GSK Health Advisory Board
- Human Genetics Commission
- I C B D D W Advisory Board
- INNOGEN Advisory Committee
- National Genetics Education Centre Steering Group
- National Newborn Screening Committee – supporting patient representative on the committee to discuss the addition of a test for MCADD to the heel prick test
- P3G Executive (Steering Committee and Director - Chair of Ethics and Governance)
- Rare Disease Task Force (European Commission)
- Rare Disease UK (Steering Committee – Director – Chair)
- Scottish Cancer Coalition
- Scottish Clinical Genetics Forum
- Scottish Cytogenetic and Molecular laboratory Consortium Users Group –
- Scottish Genetics Review Group
- U A R Board
- UK Genetic Testing Network (Steering Committee and Communications Committee)
- UKFGI (UK Forum for Genetics and Insurance) meetings

Joint working with member groups

- Continuous liaison with patient support groups representatives and consultation with their members as part of the Facilitating Networks Project.
- Contributing articles for member's newsletters (Ectodermal Dysplasia Society, HITS, Ataxia UK, Neurofibromatosis Association, Rett Syndrome Association, Tuberous Sclerosis Association)
- Advising Ataxia UK focus group session for their Exeter Regional Conference.

Fundraising Achievements

- GIG was selected as one of only 10 winners of a prestigious GlaxoSmithKline IMPACT Award 2008. The Awards recognise small to medium charities that are doing excellent work to improve people's health." (April 2008)
- GIG received a donation from Genzyme to re-design website (July 2008)
- Started working in partnership with the on-line shopping club KidStart. (September 08)
- Secured approximately £300,000 in funding from the Big Lottery Fund for the *Family Risks, Common Cancers and People from Minority Ethnic Groups* project (September 08).
- GIG was accepted by St George's University of London as one of the charities to be supported by RAG week (November 08)

Financial Overview/Comments on Audit

Treasurer's Report

There is no doubt that 2008/9 was a difficult year - particularly for the economy but also for GIG. It was the first year of our new company status, intended to provide greater security for the charity & it's Trustees and timely too given the wavering banks, currencies and charity income.

Data from the Charities Commission shows that organisations of our size have found it much more difficult to grow revenue in real terms than the very large charities who have been able to continue to persuade consumers to continue to give. Income for GIG however comes from two main sources, the major donors who have contributed rather less to us this year although there is a timing issue that suggests donations are flat rather than declining. The second source includes the various organisations who commission us to produce reports on their behalf. This latter group has shown considerable growth, unfortunately not enough to fill the gap created by declining Grants but welcome just the same. In such a challenging environment our fundraiser Helen Par deserves praise for her commitment, persistence and unstinting efforts

Our deficit this year is 5.2% of income and at £33k is the lowest for the last three years. We believe if concentrate on managing costs we can aim to achieve break even in our new financial year.

We have adequate reserves to continue and the determination to focus on our new campaign activities. Moving into our new financial year we are actively reviewing our financial strategy to improve our cost management and refine our Project activities. It will be essential to ensure we manage our financial responsibilities with great care and initial forecasts are looking more positive.

Review of Financial Position

Income for 2008/09 increased by 22% over 2007/08 levels (2008/09 - £574,017, 2007/08 - £469,156). The reason for this increase was entirely due to the receipt of additional grants to fund specific projects (restricted funds). Grant income for specific projects rose by 48% (£440,305 compared to £296,869 for the previous year). Unfortunately unrestricted income went down from £202,131 last year to £133,712 for 2008/09. Grant income now accounts for

77% of total revenues (up from 67% last year). Donations comprise approximately 14% and subscriptions from members comprise 7%.

Expenditure for the year was £604,063 (2007/08 - £511,307) resulting in a deficit for the year of £30,046. The costs of running the charity dropped by nearly £10,000 over last year; hence allowing a significant increase in expenditure devoted to charitable activities and total expenditure on charitable activities rose from £420,464 last year to £529,657 for 2008/09. This sum represents 88% of GIG's total expenditure (2007/2008 – 82%).

Reserves Policy

The charity has free reserves of £129,134 which represents over 9 months of unrestricted fund expenditure on all activities. In the event of a future shortfall in funds raised the charity needs to hold minimum reserves of £67,000 to cover the costs of closing the organisation down. This figure represents the current estimate that would be required to pay the staff who would be redundant and other commitments which would need to be met.

The trustees therefore consider that the reserves held are sufficient to meet current and ongoing needs.

Accounts

Please see the full audited accounts

GIG Team

Director

Alastair Kent

The GIG team

Claire Cotterill	Patient Engagement Project Officer
Benjamin Francis	Insurance Templates Project Officer
Melissa Hillier	Communications Manager (promoted to Assistant Director in April 09)
Amy Hunter	Senior Research Manager
Krystle Kontoh	Facilitating Networks Project Officer
Heather Law	Office Administrator
Celine Lewis	Eurogentest Project Officer
Alex Mckeown	Eurogenguide Project Officer
Nick Meade	Policy Officer
Helen Parr	Fundraiser
Gillian Scott	Development Officer Scotland
Buddug Williams	Development Officer Wales/Cymru

Trustees

Christopher Friend	(Chair)
Joannie Dimavicius	(Vice Chair)
Christopher Goard	(Hon. Treasurer)
Maggie Ponder	
John Dart	
John Dodge	
Kit Farrow	
Jane Fisher	
Fiona Hemsley	
Anne Jolly	
Mike Knapton	
Liz Nelson	
Marita Pohlschmidt	
Ruth Sands	

Cooptees

Jerry Brown
Elaine Miller

Observers

Dr Hilary Burton
Professor Shirley Hodgson
Dr Tessa Homfay

Advisors

Professor John Burn, MD, FRCP, FRCPCH, FRCOG
Professor Michael Connor, MD, DSc, FRCP
Professor Timothy Cox, MD, FRCP (Glas), FMedSci, FRSE
Professor Kay Davies, CBE, MRCP (Hon), FMedSci, FRS

Dr Roz A Eeles, MA, PhD, FRCP
Professor Alan Emery, MD, PhD, DSc, FRCP, FLS, FRSE (Retired June 2009)
Professor Peter Farndon, BSc, MD, FRCP, DCH
Professor Nick Hastie, FRSE, FMedSci, FRS
Professor Shirley Hodgson, DM, FRCP
Professor Peter Lachmann, PMedSci, FRS
Professor Norman Nevin, MD, FRCPE, FRCPath, FFCM (Belf)
Professor Martin Richards, MA, PhD, ScD
Professor Mike Ratton, MA, MB, MSc, FRCP, FRCPCH
Professor Marcus Pembrey, MD, FMedSci, FRCP, FRCPCH, FRCOG

Members

Action for Sick Children (Scotland)
Action on Gilbert's Syndrome
Adrenal Hyperplasia Network
Albinsim Fellowship
ALD Life (support group for those affected by adrenoleukodystrophy and adrenomyeloneuropathy both personally or medically)
Alkaptonia Society
Alpha 1 Support UK
Alstrom Syndrome UK
Alzheimer's Society
AMEND (Association of Multiple Endocrine Neoplasia Disorders)
Androgen Insensitivity Syndrome Support Group
Anirida Network UK
Anorchidism Support Group (ASG)
Ante Natal Results and Choices (ARC)
The Arrhythmia Alliance
The Arthrogyrosis Group (TAG)
Assert (the Angelman Syndrome Support Education and Research Trust)
Association for Glycogen Storage Diseases (UK)
Ataxia - Telangiectasia Society
Ataxia UK
Barth Syndrome Trust
Batten Disease Family Association
Beckwith-Wiedemann Support Group
British Heart Foundation
The British Porphyria Association
Cancer Research Genetics UK
Cancer Research UK
Cardiomyopathy Association
The Cavan Tommy Hoey Trust
Cavernoma Alliance UK
CCHS Support Group (congenital central hypoventilation syndrome)
CDLS Foundation (Cornelia De Lange Syndrome)
CGD Research Trust (Chronic Granulomatous Disorder)
Childhood Eye Cancer Trust (CHECT)
Children's Mitochondrial Disease Network
CLIMB (Children Living with Inherited Metabolic Diseases)
CMT UK (Charcot-Marie-Tooth Disease)
The Cogent Trust
Cohen Syndrome Support Group
Congenital Adrenal Hyperplasia Support Group

Costello Support Group
Cri Du Chat Syndrome Support Group
CRY (Cardiac Risk in the Young)
The Cystic Fibrosis Trust
DEBRA (National Charity for Epidermolysis Bullosa EB)
Down's Heart Group
Duchenne Family Support Group
Dyskeratosis Congenita Society
East London Branch Sickle Cell Society
Ectodermal Dysplasia Society
Ehlers-Danlos Support Group
The Eyeless Trust
Fanconi Hope Charitable Trust
FAP UK (Familial Adenomatous Polyposis & Gardners Syndrome)
The Fragile X Society
Friends of Kabuki Syndrome
FSH Muscular Dystrophy Support Group
FuchsFriends UK
Galactosaemia Support Group
Gauchers Association
GeneAid
Gorlin Syndrome Group
Gwent Huntington's Disease Association Support
Haemachromatosis Society
Haemophilia Society
Headlines (The Craniofacial Support Group)
HITS (UK) Family Support Network (supporting families with neurocutaneous disorder also known as Hypomelanosis of Ito)
HME Support Group (Hereditary Multiple Exostoses Support Group)
Huntington's Disease Association
Huntington's Disease Association (Colchester Branch)
The Hypermobility Syndrome Association
Hypermobility East Anglia Group
Hypoparathyroidism
Information Point for Centronuclear & Myotubular Myopathy
The International Autistic Research Organisation
The Jennifer Trust for Spinal Muscular Atrophy
Keratoconus (KC) Global
Klinefelter Organisation (UK)
Laurence Moon Bardet Biedl Society
The Lowe Trust
Making it Better - The Daniel Courtney Trust (Microvillious Atrophy Support Group)
Manchester Sickle Cell and Thalassaemia Centre
Marfan Association UK
The MAX Appeal (Di George Syndrome(VCFs & 22q 11.2 deletion)
The Medical Advisory Service
Mosaic Down Syndrome UK
Motor Neurone Disease Association
Mowat-Wilson Syndrome Support Group
Muscular Dystrophy Campaign
Muscular Dystrophy Support Group
Myrovlytis Trust
Narcolepsy Association - UKAN
NPS UK (Nail Patella Syndrome)
NASPCS (charity for incontinent and stoma children)

National Sickle Cell Programme
Nemaline Myopathy
The Neurofibromatosis Association
Niemann-Pick Disease Group (UK)
NSPKU (charity for those with Phenylketonuria PKU)
Nystagmus Network
Ohdo Syndrome Family Network
OSCAR (Organisation for Sickle Cell Anaemia - Reading)
Osteopetrosis Support Group
Patients Know Best
Pemphigus Vulgaris Network
Peutz Jeghers Syndrome Support Group
Pick's Disease Support Group
Primary Immunodeficiency Association
PKD Charity (Polycystic Kidney Disease)
Plagiocephaly Care UK
PPUK (Parent Project UK fighting for a cure for Duchenne Muscular Dystrophy)
Prader Willi Syndrome Association UK
Primary Ciliary Dyskinesia Family Support Group
PXE Support Group
Restricted Growth Association
Rett Syndrome Association UK
Rubinstein Taybi Support Group
SADS UK (The Sudden Adult Death Syndrome Trust)
Scottish Huntington's Association
Schwachman Diamond Support Group
Sickle Cell and Young Stroke Survivors
Society for Mucopolysaccharide Disease
SOFT UK (Support organisations for Trisomy 13/118 and related disorders)
STEPS (Association for People with Lower Limb Abnormalities)
Stickler Syndrome Support Group
Syndromes without a name (SWAN)
Tar Support Group
Talangiectasia Self Help Group
TOFS (Tracheo-Oesophageal Fistula Support)
Treacher Collins Family Support Group
Triple X Support Group
Tuberous Sclerosis Association
Turner Syndrome Support Society (UK)
UK Thalassaemia Society
Unique - The Rare Chromosome Disorder Support Group
Usher Service - Sense
Williams Syndrome Foundation Limited
Worster Drought Syndrome Support Group
XLP Research Trust
XP Support Group (Xeroderma Pigmentosum)

Support

The Genetic Interest Group has been supported by a wide range of organisations over the last year.

With many thanks to:

The following organisations for supporting our work through grants and donations:

The Association of the British Pharmaceutical Industry (ABPI)

The Big Lottery Fund

Bourne End and Cockham Rotary Club

BUPA

The Department of Health (Section 64 Programme of Grants)

The European Commission (FP6)

The European Commission (FP7)

GE Healthcare

Genzyme

GlaxoSmithKline

GlaxoSmithKline IMPACT Award

The Hospital Saturday Fund Charitable Trust

The Medical Research Council (MRC)

Merck, Sharp &Dohme Ltd

National Institute for Health Research (NIHR)

National Services Division, Scotland

Reneuron Ltd

Roche

SAFE

Schering Plough

The Scottish Executive

The Wellcome Trust

The Wales Gene Park for supporting the work of our Development Officer in Wales

The following organisations for providing pro-bono support for our work:

Bird & Bird

Clifford Chance

TudorReilly

The following organisations for supporting the work of Rare Disease UK:

The Association of the British Pharmaceutical Industry (ABPI)

Genzyme

Information about GIG

Registered Charity Number: 1114195

A Company Limited by Guarantee: 05772999

Principal address: Unit 4D, Leroy House, London, N1 3QP

Auditors Nyman Libson Paul, Regina House, 124 Finchley Road, London, NW3 5JS

Bankers: CAF Bank Ltd, 25 Kings Hill Avenue, Kings Hill, West Malling, Kent, ME19 4JQ