

# gigtoday

autumn 2007

## GIG attends the British Society for Human Genetics Conference 2007

The British Society for Human Genetics (BSHG) hold an annual conference for clinicians, scientists, patient organisations and industry to meet together, sharing and debating current and breakthrough information for genetic conditions and the implications for clinical practice. This year GIG had a strong presence throughout the conference held between 17th and 19th September at the University of York. In addition to the GIG information stand in the main exhibition hall four of us attended as delegates as we'd had the following titles accepted for poster presentations:

- Boy Genius: Exploring Genetics through Interactive Theatre for Young People.
- Development of a culturally sensitive translation protocol.
- Development of a Set of Patient Leaflets about Genetics and Genetic Testing Testing for Use in Genetic Clinics Across Europe.
- Finding a route through the maze of current information and services for individuals and families affected by or at risk of six rare genetic conditions: the patients' perspective.
- South Wales FH Forum - Improving information & policy input for families.

With a full programme, and international speakers, reflecting themes from cutting-edge research and studies of specific conditions to discussions of ethical decisions around genetic testing and funding for NHS Genetic Services, we found it difficult to choose which sessions to attend. There were also satellite workshops, lunchtime seminars, evening debates and if you could cram in sports or social activities these were catered for too!

Our posters were on display throughout the conference for those who wished to browse. Additionally, specific times for authors to stand by their posters were scheduled into the busy Monday and Tuesday and this gave us chance to talk to delegates who had a particular interest in the subject of our individual posters. Disseminating information and findings from the patients' perspective in this way is both important and essential to fulfil GIG's role to inform and influence decision-makers on behalf of our membership. Raising awareness amongst this group of

professionals helps ensure that they make use not only of the resources GIG has produced, but also the experience of GIG's staff members.

Highlights this year included:

- Prof Hal Dietz (Baltimore, USA) in describing his work with Marfan Syndrome and related disorders told us of a promising breakthrough in drug therapy for heart complications.
- Prof Francesco Muntoni (London) outlined future planned trials of treatment for boys with Duchenne Muscular Dystrophy and in particular one form of gene therapy that can be delivered by intravenous injection that potentially will affect virtually all muscles. Initially however, the trial would be designed to look at what this therapy achieves using one muscle in the foot.
- Dr Lynn Chitty (London) reported on the first year of a systematic review of use of free fetal DNA testing. This is a method of prenatal diagnosis which does not require an invasive procedure (such as amniocentesis, which carries a risk of miscarriage) in order to collect the sample for testing. Instead, it has been found that DNA from the fetus is present in the woman's blood stream, so that a simple blood sample is taken from the woman in order to analyse fetal DNA. So far, free fetal DNA testing is only being used for finding out the sex of a fetus (where X-linked conditions are in the family). The review has established that the technique is accurate for this purpose from about 7 weeks (much earlier than ultrasound scanning).

An added bonus for us as GIG staff was the time spent together at the conference, discussing our own work for GIG and getting to know each other better as colleagues and friends. GIG is grateful to all at BSHG for kindly donating space in the exhibition hall for GIG to be represented at the conference and for the warm welcome we all received.

**Anna Allford, Amy Hunter, Celine Lewis, Buddug Williams.**

Autumn 07

## Dealing with controversial policy issues.

It was the introduction of the Human Fertilisation and Embryology Bill to Parliament in 1989 that led to the formation of the Genetic Interest Group as a co-ordinated campaign to preserve the opportunity to carry out embryo research under properly controlled circumstances and to address the problems rising from serious genetic disease.

Today, nearly twenty years later, embryo research is still a hot topic, but the advance in our knowledge has made possible, at least in part by GIG's pioneering advocacy on behalf of patients and families keen to promote the opportunity to undertake high quality, ethical research on embryos. This means that issues that would have been seen as "science fiction" are now providing real opportunities to advance our knowledge and understanding.

One such issue has been the creation of what have been called "hybrid embryos", which we have mentioned in previous newsletters. A hybrid embryo is created by taking an animal egg, removing the nucleus and inserting instead the nucleus of a human cell. The resulting fusion is stimulated to divide thus creating an embryo from which stem cells can be obtained for research purposes. This technique allows researchers to overcome the shortage of human eggs available for research (they are nearly all needed for use in in-vitro fertilisation). It also makes it possible to study the early stages of a particular disease, as cells from a person with the condition can provide the necessary DNA, thus faster progress will be possible.

GIG, with the support of many of its members, was an active and vocal participant in the campaign to persuade the Human Fertilisation and Embryology Authority (HFEA) to license research using these "hybrid embryos". This is something that the Authority has now agreed to do in principle, and it will soon make a decision on specific project applications later this year.

The issue of embryo research is a controversial and challenging one. There are other issues that GIG has taken up, many of which we are working on today. We can be sure that there will be more in the future, as scientific advances open up novel possibilities and create new ethical and political challenges. It is the support from our members that

gives us the confidence to address these issues head on, and to put forward views and opinions derived from our members and the individuals and families they represent, to those in positions of power and influence.

It is not just on embryos that GIG is in the forefront of patient and family advocacy for research and its translation into improvements in therapy and support. Animals in biomedical research, gene and tissue therapy, rationing and the postcode lottery, patenting and intellectual property as well as genetics and insurance are all issues where GIG has been and continues to be a strong voice for patients and families here in the UK and in Europe.

Most recently, GIG has responded to the Department for Communities and Local Governments consultation on *Discrimination Law Review - A Framework for Fairness: Proposals for a Single Equality Bill for Great Britain*. Many of GIG's members and those they support kindly returned a questionnaire sent out by GIG helping us to highlight real life experiences in our response and to highlight the need for people to be protected from unfair discrimination due to their current genetic status, or their potential risk to a genetic condition. This information also helped the Human Genetics Commission (HGC) to formulate their response and highlighted to them the real need to understand and respond to people's perception of genetic discrimination.

With the support of our members, and the engagement of our allies in science, medicine, industry and politics we will continue to try and meet this responsibility for as long as the need remains.

**Alastair Kent**

### Further reading

GIG website  
<http://www.gig.org.uk/consultation.htm>  
 (Discrimination Law Review, Sept 2007 - GIG response)

Dept of Communities and Local Government.  
 (the full consultation document page)  
[www.communities.gov.uk/publications/communities/frameworkforfairnessconsultation](http://www.communities.gov.uk/publications/communities/frameworkforfairnessconsultation)

## The HFEA's "cybrid embryo" decision is good news for patients.

Following its public consultation, the Human Fertilisation and Embryology Authority (HFEA) has announced its decision that there is 'no fundamental reason to prevent cytoplasmic hybrid research' and that 'individual research teams should be able to undertake research projects involving the creation of cytoplasmic hybrid ('cybrid') embryos if they can demonstrate, to the satisfaction of an HFEA licence committee, that their planned research project is both necessary and desirable'. UK scientists want to use enucleated animal eggs - those from which the nucleus, containing the vast majority of an egg's genetic material, has been removed. Genetic material from human patients could then be added to these empty eggs, and the resulting cybrid embryos used to create embryonic stem cells that are virtually human.

This is good news. Although it will be some time before tangible benefits to patients emerge, the decision is endorsed and welcomed by the patients and families who make up the Genetic Interest Group (GIG) membership because of the potential such research holds for investigating the fundamental biology of many disabling conditions; conditions which can be serious, incurable or untreatable. Patients with these conditions depend on an ambitious and innovative biomedical research programme to deliver future treatments and/or cures. The HFEA decision will at last speed up progress in this area.

It has taken almost a year for the HFEA to be ready to assess the two specific applications for research using cytoplasmic hybrid embryos that were submitted to them (from Newcastle University and King's College London). The lack of progress in this area while the HFEA process has unfolded is regrettable as the uncertainty will have been a disincentive to researchers in the UK. It is to be hoped that the response to the consultation, with almost two thirds of respondents expressing support for cytoplasmic embryo research, will increase the HFEA's confidence in the powers given to them and in their decision-making process. Even when there

is doubt that their stance is aligned with public opinion, they have a mandate in law to regulate without recourse to public consultation. Having said that, this latest consultation result should reassure the HFEA as it regulates in future, because it adds to the trend of public support for high quality, properly regulated research aimed at decreasing human suffering, even when that research presents moral challenges to some sections of society. Notable earlier examples include embryo research in general, preimplantation genetic diagnosis (PGD) and saviour siblings.

A second benefit of the HFEA consultation process is that while responses to the consultation inevitably came thickest and fastest from religious and patient groups, the frequent press coverage created a valuable opportunity for increasing awareness among the general public of what the proposed research actually entails. The 'Franken-bunny' stories have just about disappeared and the more nuanced reporting now seen across the media is very welcome.

As legislation in the shape of the draft Human Tissue and Embryos Bill is scrutinised, it is to be hoped that the Government hears the message that the public and patients value medical research and have confidence in existing regulatory procedures. Any change in the regulatory process that makes it more difficult to perform medical research would be a retrograde step. GIG urges the Government to take this into account in its overhaul of the HFEA and the Human Tissue Authority (HTA), and to ensure that the proposed new authority (Regulatory Authority for Tissues and Embryos, RATE) is able to respond quickly and effectively to new regulatory challenges created by scientific progress. Progress that can and will throw up as yet unknowable prospects.

### **Dr Amy Hunter**

*This article first appeared in BioNews 424:10/9/07 ([www.bionews.org.uk](http://www.bionews.org.uk)), and is reproduced with kind permission of the Progress Educational Trust.*

Autumn 07

## Ethnic Monitoring Project draws to a close.

It hardly feels like I've been working at the Genetic Interest Group for three years, but when I look back, it seems amazing that we've managed to do so much in that time. I first started with GIG to work on a three year project on ethnic monitoring in clinical genetics which has now ended. The project followed on from work done by our then-project manager, Dr Pritti Mehta, who now enjoys a role at the RNIB as Senior Research Officer. She piloted ethnic monitoring in clinical genetics centres, demonstrating both a need for ethnic monitoring, and the efficacy of the practice. The success of her project led to GIG securing funding for a three year part-time project, implementing ethnic monitoring in clinical genetics centres in England.

The project has had its ups and downs, but once centres have been convinced of the reasons why they must perform a permanent ethnic monitoring programme, they have been universally successful. The primary reasons are two-fold. One, to be able to demonstrate that the centre is aware of the ethnic profile of its patients; and two, to provide evidence to direct any strategy necessary to deal with any disparity in healthcare between patients of different ethnic origins. The first stage, before any strategies can be implemented, is to implement the ethnic monitoring system.

We have modelled more than one ethnic monitoring strategy to take account of the different ways in which various centres in England provide their care. Genetics care in the NHS is unusual. It is delivered from a relatively small number of regional centres that care for large areas of the country. To provide for such large areas the centres provide care in

clinics distributed through their catchment areas; this allows them to reach patients who would have to travel large distances to reach the main centre. This model is a problem for a watertight ethnic monitoring system. A usual NHS monitoring programme would make use of the administrative staff at a hospital or clinic, they act as gatekeepers to the healthcare, and provide a perfect opportunity to record the ethnicity of patients. Genetic Clinicians do not have this luxury, they are often the only member of their centre at any outreach clinic, and will not have any administrative staff to help.

Our collection models provide centres with a choice on how to perform their ethnic monitoring: either at the point of healthcare delivery, in which case the clinician must perform the monitoring task; or at the point of appointment provision, this allows the administrative staff at the centre to keep control of the monitoring programme. We have created educational packages that will allow centres to train new staff, and refresh the knowledge of their current staff; there is also a resource that answers most questions that staff may have about the reasons for, and the uses of ethnic monitoring.

Once the project is all tied up, I look forward to moving full-time to my role as Policy Officer, supporting our Director Alastair Kent, and our new Senior Policy and Research Manager Dr Amy Hunter in the policy team

**Nick Meade**

## Family Route Map Workshop at the NPS(UK) Conference, Chester.

**A**rriving in Chester to glorious sunshine I entered the lovely Conference hotel and found a throng of chattering adults and children already exchanging information and shared experiences. I had been invited to present a workshop to develop the Nail Patella Syndrome (NPS) Family Route Map and was expecting to have a small gathering in a room off the main conference hall. Instead, I was ushered into a hall full of people seated behind tables and was told there was about 70 people here as not everyone could make it! Slightly anxious I took my seat and listened to experts from the USA who were providing new and breakthrough information about ADHD (Attention Deficit and Hyperactivity Disorder) associated with NPS, and education and care for affected children and adults.

The enthusiasm, interest and genuine friendliness of everyone there was the spur for me to stand up and deliver a workshop originally intended only for about 10 participants! I had prepared a set of questions around both the NPS related issues and the previous outcomes of

the combined focus groups we'd held across all six conditions that we are currently working with to develop condition-specific Family Route Maps for: Barth Syndrome; Gorlin Syndrome; Multiple Endocrine Neoplasia (MEN; Myotonic Dystrophy; Nail Patella Syndrome; and Syndromes without a name (SWAN). A generic template for other support groups to develop their own Route Map will also be freely made available through GIG at the end of the project.

The positive response from those present was overwhelming and by the end of the day the A3 sheets for each individual question had all been filled up with ideas, information and contacts. This



**Carol and the webmaster for NPS UK.**

provided me with sufficient information to develop the first draft of the NPS Route Map and also gave Carol Dobbins, Chair of NPS(UK) and her Committee plenty to think about in terms of what their members wanted and importantly, information about healthcare services

accessed by members which had been helpful.

After a great lunch (and more new friendships being forged) the main Conference

continued with further clinical information about the kidney and eye structural abnormalities associated with NPS and how surveillance is required to detect any complications in these systems. Additionally, many people present benefited from the detailed facts relating to the joint involvement and best practice when surgery is required presented by a Surgeon who also has a son affected by NPS. The Speaker discussing the molecular genetics of NPS put into context how the altered gene influenced the development of the affected systems and led to the way the condition presented in individuals and families.



**Carol - Chair, Kath - Project Manager, Shirley - Treasurer and her daughter Rosie singing "I am what I am"**

Other events at the Conference included a children's activity group and the opportunity to have a professional manicure for free! Now

many of you may wonder why on earth this luxurious pampering session was included as part of the Conference? Well, in addition to those who took advantage of it feeling totally relaxed and beautified, the name of the condition, Nail Patella Syndrome, indicates that the nails are involved and are often absent or misshapen, especially the thumb nail, exposing the nail beds or leading to pitting or discolouration of the nails. Many people, especially females, feel their self-confidence to be affected by this feature and had never considered having a manicure. Lisa, the Manicurist showed each person individually how to make the best of their nails giving them a huge boost in their confidence. Saturday evening saw the social events begin with Karaoke and a disco as I slipped away to spend the night in a spooky house (but that's another story).

My thanks go to all those who participated in the workshop and helped in the development of the NPS Family Route Map.

**Anna Lane**



**Lisa the manicurist at work**

Autumn 07

## TREAT-NMD

### Addressing Rare Inherited Neuromuscular Diseases: From Molecular Basis to Cutting Edge Therapies



**T**he TREAT-NMD (Translational Research in Europe for the Assessment and Treatment of Neuromuscular Disease) network currently brings together 21 partner organisations across 11 European countries. Some of Europe's leading clinicians and scientists have formed this 'network of excellence' - the first of its kind - which has been funded by the European Union (FP6). The network also includes patient organisations and industry partners to help establish a common roadmap for the progression of cutting edge therapies from the lab to the clinic. The overall aim of the network is to integrate European researchers and clinicians in the field of neuromuscular diseases, and to create an infrastructure that lasts beyond the current funding period. This European network is leading the world in terms of an integrated approach for neuromuscular diseases.

Neuromuscular disorders affect over 300,000 people in Europe. The term refers to a large group of conditions that affect either the muscles themselves, or the nerves controlling the muscles. Most conditions result in chronic long term disability and early death may eventually result from respiratory or cardiac failure. TREAT-NMD will encourage experts in this field to work together to share good practice and to improve global standards of care. The network will produce treatment guidelines that describe the basic standards of care that all European patients with neuromuscular disorders can expect. At the moment, standards of care vary between different member states, thus making it difficult to conduct multi-centre clinical trials because baseline data is not consistent among these centres.

Scientists will work closely with doctors to test and apply new research into these inherited disorders, in order to develop new ways of caring for patients with conditions like Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA). Close links with industry partners will also be further developed in order to advance technological and methodological tools with a view to promote further clinical trials for potential new treatments and cures for rare neuromuscular diseases. Patients and patient charities will be involved at all levels of the project, which will also include a programme of training and education.

#### Impact of the Network

- Across Europe there is a lack of common standards for diagnosis and care of neuromuscular disorders- TREAT-NMD will propose and disseminate common standards to solve this issue
- Pre-clinical evaluation of new therapies is vital and the TREAT-NMD network will ensure standardised procedures to validate relevant models and subsequent testing and evaluation protocols
- Production, toxicology and safety of potential therapeutic agents is a vital step in the clinical trials process-TREAT-NMD will ensure common standards for regulating these processes, in close collaboration with the CLINIGENE network of excellence. These standards will also be extended to ensure



better targeting of systemic delivered therapeutic agents

- TREAT-NMD has established a Clinical Trials Coordination Centre that will support the translation of research into new drugs for clinical practice
- TREAT-NMD will complement other 'networks of excellence', such as MYORES, CLINIGENE, and EUROAGENTEST to optimise collaborations and partnerships and to ensure that duplication of efforts is avoided
- TREAT-NMD will offer all the services needed to stimulate industry to develop new and effective treatments for neuromuscular diseases, such as pre-clinical testing and clinical trials

Other areas in which TREAT-NMD will make an impact include:

- Creation of harmonised, durable, and relevant biobanks and databases
- Optimising efficacy measurements of therapeutic treatments
- Dissemination of results to the scientific community
- Develop training and education programmes
- Promote knowledge transfer to industry partners
- Ensure gender equality in the

scientific community

- Extension of the network to Eastern European countries

Other parties are welcome to take part in the network via a broad international club of interest, so that TREAT-NMD will be a resource for the whole community. This group currently contains around 100 researchers, including colleagues from outside Europe, who do not receive funding directly from the EU but who are likely to be part of the network in the future. This will help ensure cooperation between Europe and the rest of the world. TREAT-NMD is currently implementing an international patient registry for the more common neuromuscular diseases that will contain information on both genotype and phenotype, so clinical trials and subsequent treatments can be offered to those patients who are most likely to benefit.

For more information about the network please contact the TREAT-NMD Coordinating Office.

Contact Information

TREAT-NMD Coordination Office  
Institute of Human Genetics  
Newcastle University, UK

info@treat-nmd.eu  
+44 (0)191 241 8605

Autumn 07

## Policy and Research at GIG



A few short weeks ago I joined GIG as Senior Policy and Research Manager, coming from my previous post as manager at the London IDEAS Genetics Knowledge Park. You will already know about the sterling work being carried out by Alex and Celine on the European projects

EuroGenGuide (developing information resources for professionals and the public about issues around informed consent and accessibility to genetic services and research) and EuroGenTest (producing and evaluating information leaflets for patients who use clinical genetics services across Europe). With Alex and Celine's help I am gradually being inducted into the labyrinthine workings of pan-European initiatives and part of my role is to support this work and ensure GIG's contributions and those of our partner organisations are properly integrated and delivered on time.

In parallel with this, Nick is actively engaged with policy developments in Europe and the UK, and I will be overseeing and developing this area of GIG's work with Nick, Alastair and Melissa. Nick and I are already working together, and with Rebecca in

Scotland, on responses to consultations from NHS Scotland and the Medicines and Healthcare Products Regulatory Agency (MHRA), and on the move by a group of MEPs to call for an end to medical research on non-human primates.

Excitingly, GIG has just been awarded funding by the Scottish Government for a new regional patient advocacy post which I will be supporting to ensure our work there is relevant to patient needs and emerging regional policy issues. The new person will be based in an office with Gillian when she returns from maternity leave in November. More news on this development will appear in later issues of *gigtoday*.

Looking further to the future, I will be focussing on growing our policy and research activities, making sure that the research we carry out feeds into development of our policy positions and lobbying work. I hope that my role will contribute to improving GIG's already impressive place within national life (and further afield) so that the interests of GIG's members continue to be taken into account at the earliest stages of policy development.

**Amy Hunter**

### New European Network of Centres of Expertise for Dysmorphology Launched

Over 2,500 rare and difficult to diagnose conditions have been identified. However, the experience of patients affected by a rare disease is that even in EU-designated Centres of Expertise, clinicians' experience of some of the rarer diseases may be limited and a diagnosis might be delayed or not made at all.

In December 2006, the European Task Force on Rare Diseases looked in detail at this problem and published a report recommending that centres of expertise for rare diseases be encouraged to form EU-wide networks. This recommendation then became a funding priority in the 2006 annual Work Programme for grants in the framework of the Programme of Community Action. A group from the University of Manchester, headed by Professor Jill Clayton-Smith submitted a bid and were successful in obtaining funding to develop a network for dysmorphology<sup>1</sup>. The project will run for three years and comprises six designated Centres of

Expertise in this area (UK, Belgium, France, Italy, The Netherlands and Poland). The University of Manchester, will be the coordinating and managing centre for the Network.

The Dyscerne project will be formally launched in Manchester at the Nowgen Centre on October 5th 2007. The main aims of the project are to raise current standards for the diagnosis and management of rare dysmorphic syndromes, and to improve dissemination of information on these conditions. The project will identify existing centres of expertise for dysmorphology, and create a formal network of centres

A web-based electronic dysmorphology diagnostic system will be established, enabling clinicians to submit difficult to diagnose cases electronically to an expert diagnostic panel. Recommendations and opinions from the panel will be collated and sent back to the referring clinician. Clinicians using the system will be guided on the description of clinical features and an on-line training tool will be developed to facilitate optimum use of the system. *continued opposite*

## Receiving genetic information from healthcare professionals: people's experiences and preferences

Last year, a range of people with or at risk of genetic conditions and parents of children affected by a genetic condition shared their experiences and views with us. This was part of a project carried out by the NHS National Genetics Education and Development Centre to explore people's experiences of receiving genetic information from healthcare professionals and their views on how genetic information should ideally be provided. We are grateful to all those who took part for providing such valuable information, which is now being used by the Centre to inform its work in supporting genetics education for healthcare professionals. We would also like to thank GIG, the North West Genetics Knowledge Park and the Patient Consultative Panel of the Human Genetics Commission for their support.

Between September and December 2006 my colleague Julie Bedward and I talked to 27 people over the telephone. One of the main issues raised by the people we spoke to was the need for greater awareness of genetic aspects of conditions amongst healthcare professionals. The people interviewed did not expect all healthcare professionals to have detailed knowledge of all genetic conditions, but did feel that healthcare professionals should be more willing to consider the possibility of a genetic condition, to refer patients for investigations and to take their concerns seriously.

The interviewees gave useful suggestions for healthcare professionals who provide genetic information. They felt genetic information should be up-to-date, given without bias or judgement, and that healthcare professionals should be mindful of

their use of technical terms. They suggested that, because some people want a lot of information at diagnosis and others don't want this information until later, it would be helpful if healthcare professionals tailor the amount of information given and tell people where they can access more information later on. They also felt healthcare professionals should be aware that genetic information can have an emotional impact for individuals and the wider family.

People also talked to us about the roles of different healthcare professionals. GP's were viewed as the best group to provide ongoing support and co-ordination of information, roles that were considered very important. Some people said they would prefer to receive detailed genetic information from the specialty consultant with whom they had regular contact and with whom they had established a rapport. Others felt that such information was more likely to be available through referral to specialist genetics services.

A full report of the project and its findings will be available soon on the Centre's website at: [www.geneticseducation.nhs.uk](http://www.geneticseducation.nhs.uk). If you have any particular queries about the project please contact me,

**Sarah Burke**, at [S.E.Burke@bham.ac.uk](mailto:S.E.Burke@bham.ac.uk).

National Genetics Education and Development Centre  
c/o Birmingham Women's Hospital  
Edgbaston  
Birmingham B15 2TG

*continued from the previous page*

Four conditions will be identified where guidelines for clinical management are needed. Expert review groups for each condition will be created which will produce draft management guidelines. These will include criteria for diagnosis, protocols for review and screening, and information on management at different life-

stages. The guidelines will be piloted in participating centres and their impact and their usage audited and evaluated.

Dyscerne will also serve as a model for future EU Networks of Centres of Expertise.

1Dysmorphology is the study of birth defects or malformations which form recognisable patterns

of growth, development, behaviour and physical features.

For further information, please contact Pam Griffiths, Project Manager:  
[pam.griffiths@cmmc.nhs.uk](mailto:pam.griffiths@cmmc.nhs.uk)/0161 276 3209

Autumn 07

## RDS Resource Centre

The RDS Resource Centre was set up in July 2005 to help research institutions deal with Animal Rights Extremism. During its first year it produced a comprehensive set of guides offering advice and information on what to do about animal rights extremism:

1. **General information** about animal rights extremists including the main groups active in the UK and description of their tactics
2. **Preparedness measures** including contingency planning and raising awareness amongst staff
3. **Security**, control of access to sensitive buildings, control of information and CCTV monitoring
4. **Legal and law enforcement** resources
5. **Miscellaneous**, covering issues such as new building projects and monitoring of trends in extremism
6. **Crisis Communications** planning for an ARE emergency

After a successful first year the Resource Centre expanded its services to include advice and support on communications and developed two communications handbooks (for Universities and Charities) to help research institutions feel more confident about their communications on the use of animals in scientific and medical research.

There has never been a better time for openness and transparency: the Government has been more openly supportive of the use of animals in research, and is taking measures to deal with extremism; while the public is becoming less tolerant of animal rights extremists and their tactics.

In addition, opinion polls show that the vast majority of the public accept the need for animal research provided that there is no unnecessary suffering, the research is for serious or life-saving purposes and there is no alternative.

Until recently the scientific community's communications policies on the use of animals have largely amounted to ducking 'below the parapet'. Experience shows, however, that this does not prevent animal rights extremists from finding out what they want to know. If an activist or extremist group decides to pay attention to an institution, it will not be because of official public communications from the institution but the result of careful research which may include published papers, database

searches (eg Lexis-Nexis, PubMed, Medline), or other publicly accessible material.

In fact, by not proactively communicating, institutions are neglecting to inform those members of the public who support research using animals because of its potential benefits.

We understand that history or even internal pressures may keep many from 'putting their heads above the parapet' and that change will not happen overnight. That is why the communications handbook offers a series of recommendations - all of which have been 'tried and tested' by a number of institutions without any adverse results.

More specifically, it outlines measures that research institutions can take to improve communications on the use of animals without attracting unwanted attention. In the handbook we:

- Demonstrate through examples that research institutions can be proactively explain their use of animals in medical research without attracting attention from animal rights extremists or antivivisectionists
- Give general advice and guidelines on how to prepare and implement a communications strategy
- Give general information and materials that can be used for Q&As or briefings

Communications about the use of animals will help garner support and understanding while non-communication will only prolong opposition and mistrust. Research institutions can play a significant role and help illuminate the complex social issues of animal research, its welfare costs, and its benefits to human and animal health.

**During the past year the Resource Centre has organised two regional events (in Scotland and the Midlands) to address openness and animal research. The next event is taking place in Cambridge in 13 November, while two more are in the pipeline. In addition, the Resource Centre team has visited a number of research institutions across the UK. If you think that your organization would benefit from a meeting or presentation, or if you would like to attend our next event contact:**

**Corina Hadjiodyseos, Communications Officer; tel: 020 7478 4333; email: corinah@rds-net.org.uk**  
**Kirk Leech, Project Officer; tel: 207 478 4332; email: kleech@rds-net.org.uk**



## The Rare disease patient solidarity project (RAPSODY)

The Genetic Interest Group was asked earlier this year to become involved in the RAPSODY project. This is a two year project being carried out by Eurordis, the European Organisation for Rare Diseases, ([www.eurordis.org](http://www.eurordis.org)) along with nine partner organisations. The overall objectives are to improve the quality of care, information and social services for people living with a rare disease. In order to help do this there are a number of different work strands or "units" which are being carried out with the help of partner organisations.

**Help lines for rare diseases** - offering quality information throughout Europe and helping those who feel very isolated to make contact with others affected by the same or similar disease.

**Therapeutic Recreation Programmes** - These are for children and young adults living with a rare disease. Standardising the quality of therapeutic recreation programmes for children and young adults, helping centres to learn from each other and create 'best practice' and standards.

**Respite Care Services** - This will identify existing respite care facilities and create a network to provide patients and families with improved respite care. The project will provide facilities for dialogue between respite centres to help improve services and development.

**Patient's Survey** - Measuring patients' expectations on the administration of medical, paramedical and social care, through a pan European survey, tailored to the needs of thirteen specific rare disease networks.

**Centres of Expertise** - A European Network. This work will enable patients to engage in the European parliament's discussions on Networks of Excellence. The project will organise 10 workshops around the EU to gain people's views and opinions, these will be fed back to the EU Parliament.

### UK Workshop

GIG was contacted and asked to be part of the last strand of work mentioned, the Centres of Expertise

and we agreed to organise a workshop which would bring patients, health professionals, health economists and researchers together to discuss the issue of centres of expertise, and how these may help patients to access care and services. Each workshop that was hosted, as part of this project, had the same agenda and the same format, this was to help Eurordis in their analysis of the data they received back.

The workshop was split into three sections and with over 20 delegates participating we had three smaller groups so that people were able to talk and share ideas more openly. Each group had a facilitator and a scribe who then gave all the information back at the end of the day to me so that I could write the UK report.

The three topic areas we were given to discuss were

1. The needs and expectations for centres of expertise for rare diseases
2. Proposals for the evaluation of national centres of reference in your country
3. Cooperation with other countries and recommendation for European Reference networks.

Within each topic area we had a list of questions and suggestions to help the facilitator move the discussion through the issues, and there was so much to talk about, we ran out of time in each session!

However, we had many positive outcomes from the break out sessions, in that within each group many of the ideas, suggestions and wishes for improving health services were similar and on most areas people agreed - clinicians and patients.

The outcome of this extremely important workshop was fed back in a full report to Eurordis, who in turn looked at all 10 reports from each European State to make a full digest and has since fed back to the European Commission's department responsible for Health which is called DG Sanco.

*continued on the next page*

## Autumn 07

*continued from the previous page*

### European Workshop

In June this year I attended the RAPSODY project's Centres of expertise Workshop in Prague, along with many other patients and patient representatives who had helped in, not only this strand of the RAPSODY project, but also in the Patients Survey strand. I was accompanied by some GIG members who had attended the UK workshop - Lynne Zwink, The Fragile X Society; Michaela Damin, The Barth Syndrome Trust; and Jane Cox, Tuberous Sclerosis Association. We had talks and discussions on the findings from the workshops which had been held and also how we felt these findings could be used and how services could be improved in our own countries. We also discussed the importance of collaborating and how it could help even more through networks by raising awareness and knowledge of rare conditions and sharing information with a larger audience.

The next step following this workshop is the Rare Disease Conference which is taking place in November this year in Lisbon. At this conference further findings from the Rapsody project will be announced and discussed.

By looking at the health systems across Europe, which vary considerably, it enables both patients, health professionals, politicians and commissioners to gain an insight into new and innovative ways of getting health interventions, treatments and surveillance to patients in ways that may not have been recognised or considered within their own Member State.

The findings from this project will be reported to DG Sanco and I will certainly keep you up to date with any news and feedback that I receive.

**Melissa Hillier**

### Patient UK website

Patient UK was first launched in 1997 by PiP (Patient Information Publications). It started as a directory of UK websites which provided information of health, disease and related issues.

Since the internet the database has added "online" in 2002 and is now run by PiP and EMIS (Egton Medical Information Systems). It contains considerably more content and information on health issues. The database contains patient leaflets as well as an appendix of patient support groups.

The aim of this website is to

provide non-medical people in the UK with good quality information about health and disease. The authors do this by writing evidence based information leaflets on a wide range of medical and health topics. They also review health and illness related websites and link to many of these from the web directory included on this website.

The database is used by over 50% of GP practices throughout the UK, and the website receives over 2 million hits per month, not including the in-house doctor surgery use. Many of the people

that access the information are the public and patients.

Patient UK enables GP's to print out further, reliable information during a consultation whilst discussing issues with the patient directly. It was started over 10 years ago as a partnership between two GP's living and working in the Tyne and Wear area, they both have a particular interest in writing and providing evidence based patient information. They are editors and lead authors for the content on Patient UK.

If you would like your patient organisation to be included on Patient UK please do contact Tim Kenny one of the co-founders on tkenny101@btinternet.com  
www.patient.co.uk

**Genetic Interest Group**  
Unit 4D, Leroy House  
436 Essex Road  
London  
N1 3QP

**T: 020 7704 3141**  
**F: 020 7359 1447**  
**M: mail@gig.org.uk**  
**W: www.gig.org.uk**

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

Patient  UK