

gig today

winter 2007/08



Rare Disease Day

Rare Diseases a public health priority in Europe

The 29th February 2008 will mark the first ever Rare Disease Day in

Europe. This is an EU wide collaboration between patient organisations and is being coordinated by the patient group Eurordis the European Organisation for Rare Disorders. The aim of the one day activity is to raise awareness of rare disorders in each country to politicians, policy makers, health care providers and professionals as well as to patients and the public.

By working at a European level it is clear that we can highlight that collectively rare diseases are not rare! There are 25 million people living with rare disorders throughout Europe and 75% of those are children. There is certainly lots that can be done to improve the awareness of the issues patients with rare conditions face in accessing services, information and treatment.

Strike while the iron's hot!

The European Commission's nearest equivalent to England's Department of Health (DH), the Health and Consumer Protection Directorate (DG Sanco) has recently published a Communication called Rare Diseases; Europe's Challenges, so this is an ideal time to raise awareness of rare diseases as discussion is taking place right now about how to improve services for patients. A communication issued by the European Commission is not a legally binding document in member states, however it is an excellent document to help in publicising the need for improved communication and networking amongst health professionals throughout Europe for patients living with genetic diseases. This is the first document of its kind to be produced on rare diseases by the Commission and it is currently in a consultation phase.

Taking Part in the Day

GIG will be taking part in this first ever Rare Disease Day to highlight the work being carried out to improve the lives of those living with genetic disorders. With many advances taking place in genetic research it is really important to highlight

the need for these advances to be transferred into practice for patients.

Other events will be happening around Europe too, including a conference and rare diseases march to Copenhagen Town Hall in Denmark, to schools projects and posters being developed in Germany.

GIG is delighted to have secured funding to help us to organise two events here in the UK.

The Events are open to GIG members and interested parties

To mark Rare Disorders Day GIG will be holding two parliamentary receptions. The first will be hosted by Evan Harris MP at the House of Commons, Westminster on 26th February from 4 - 6pm. This event will highlight work carried out by GIG helping patients to access services and information, with particular focus on the Family Route Map Project, followed by drinks and discussion.

We will also be holding a lobby day and reception in the Welsh Assembly in early March in Cardiff hosted by Jane Hutt AM. This event will incorporate work that is being carried out with GIG and the Wales Gene Park to help families affected by inherited heart disease. The event will be followed by drinks and discussion.

If you, one of your trustees, advisors or members would be interested in attending the free Westminster Event for Rare Diseases please contact

Melissa Hillier 020 7704 3141 or
melissa@gig.org.uk

If you, one of your trustees, advisors or members would be interested in attending the free Welsh Assembly Event for Rare Diseases please contact Buddug Williams 029 20 68 21 40 or
williamsbg@cf.ac.uk

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EuroGenGuide Project Annual Meeting

The annual meeting for the first year of the EuroGenGuide project was held in Milan in late October. The culmination of ten months hard work by the team, the meeting was a great success. As well as being a good way for everyone to discuss the work with each other in person after so much email and telephone contact over the year, the two discussion sessions helped enormously in pointing out the final amendments necessary to the work so far produced, in order to meet the project targets for 2007.

The focus of the work in Milan was to assess the drafts of five 'protocols' which will form the central information core of the EuroGenGuide when it is complete. These protocols will provide information and practice guidelines to patients and health professionals about relevant aspects of genetic testing and research, such as informed consent, biobanking procedures, benefit sharing, and the various concerns that both able-bodied and disabled men and women from different ethnicities and backgrounds might have when considering taking a genetic test or becoming involved in research.

I have been producing and re-working drafts of the protocols over the course of 2007 in collaboration with the EuroGenGuide team, and the main focus of the meeting in Milan was to work out and agree the final changes that need to be made to them such that they are ready for the next stage of the work which begins in 2008.

Some of the protocols have been more challenging than others, for a range of reasons including the availability of relevant information from which to carry out research, and the need to find a way of explaining complex information in a way that is both understandable to all users and also sensitive to every reader, irrespective of the nature of their particular situation. In view of these challenges, the

two sessions of the Milan meeting were divided as follows:

Session one was a general discussion and feedback of all the five protocols, with time dedicated to each individually. The feedback from the less problematic protocols was taken back to the UK to be fed straight into new versions. However the feedback from the two more challenging protocols, i.e. the ones concentrating on gender issues, and on disability and vulnerability, was taken and used as the basis for session two.

Session two was a more in-depth discussion session, where the group was split into two and the key points from session one were worked on intensively to provide three or four clear and central strategies that needed to be employed in developing the final drafts of the protocols. Both sessions were really useful and I came away with invaluable feedback and advice that I have been using since the meeting to finish a new set of drafts in time for the end of the year, in readiness for the next stage of work that will begin in January.

Once the final drafts of the protocols are agreed they will be assessed by focus groups comprised of a range of health professionals, patients and their relatives. The feedback from these focus groups will be used to produce versions that are completely ready to be included in the first full draft of the EuroGenGuide, which will be ready in May 2008. At this point the EuroGenGuide will 'go public' online and in print, which will be a very exciting time for the project. So you can see that the first annual meeting in Milan was a crucial milestone in the project, as the work that it has set in train leads towards the first publicly accessible and useable draft of the EuroGenGuide, which will appear in six months time.

Alex McKeown

Could Government stand in the way of research developments for patients with genetic disorders

I have written on the subject of the Human Tissue & Embryology Bill a number of times since it was first drafted in late 2006. Since then, it has been renamed as the Human Fertilisation & Embryology Bill, as plans to amalgamate the Human Fertilisation and Embryology Authority with the Human Tissue Authority have been shelved.

The Bill is now at the Parliamentary stage which has begun with a reading in the House of Lords. GIG is involved with lobbying to change the Bill on a number of issues. We have plans to collaborate with groups such as: Muscular Dystrophy Campaign, Motor Neurone Disease Association, Huntington's Disease Association, and the Parkinson's Disease Society. These groups share many of GIG's objectives, especially those that look toward the creation of an environment that encourages research towards the goal of cures and treatments.

There are two issues in the Draft Bill which concern all of our groups; we plan to write to the Health Department Minister that is responsible for the Bill to raise our concerns.

Licences for therapy

Therapies derived from Embryonic Stem Cell technology are still a light at the end of the research tunnel. Many avenues of research concerned with conditions supported by GIG members hold significant potential and are the subject of much hope for patients with these conditions. For some of these conditions, there is no cure or palliative care currently available to patients. It is vital that this research continue at the exciting pace that it currently shows.

Licenses for this kind of therapy are not currently necessary, but therapies that will require such licenses are just around the corner. An estimate of five years for some treatments is not overly optimistic. The Bill, as it stands, has no provision for the granting of licenses for treatments derived from embryonic stem cells. Without an alteration to this Bill, a full new Parliamentary Bill would be required before treatments such as these become eligible for licenses.

It is the view of GIG and other patient groups that this lack of an outlet for products of these research avenues will impede investment and slow the pace of research. The worst case scenario, of course, is the development of a safe, viable treatment that is then forced to languish unused for years whilst the Government empowers the HFEA to grant licenses for its use.

We would like the Bill to reflect these views, and provide for the exploitation of research towards treatments making use of Embryonic Stem Cell technology, by facilitating the licensing of such treatments.

Consent for Cell Nuclear Reprogramming /Replacement

As the Bill currently stands, scientists wishing to create an embryo from a line of somatic cells will need to obtain consent from the donor of the cell line. This new requirement is likely to severely diminish the amount of cell lines available for use by researchers in the UK.

The creation of embryos from somatic cell lines is a valuable research tool. Embryos can be created to have a specific condition, and the embryo's stem cells studied, to gain greater knowledge of the condition. Currently, before the new Bill comes into effect, scientists in the UK have at their disposal a large resource of anonymously donated somatic cell lines; a second resource is the hundreds of commercially available somatic cell lines. Both of these comprehensive resources will become unavailable as soon as any requirement for donor consent comes into effect. This is not because the consent would not be forthcoming; it is because all of these cell resources are already anonymised. Those cells donated for the purpose of research were donated on the condition that the patient consents for their usage in research; donors would not be able to find out any more specific information on their usage.

A specific example is that of Spinal Muscular Atrophy (SMA). There are one hundred somatic cell lines that have been donated by patients with SMA available commercially. As the Bill stands it would be illegal to use these due to lack of consent. An optimistic estimate as to how long it would take to build up such a valuable resource by collecting cells under consent is five years.

Any interruption, let alone an interruption of five years or more in research and development of treatments derived from stem cells would be contrary to overall research policy and would appear to deliver no particular benefit. All of the groups mentioned above strongly feel that the Bill should be amended to avoid such an undesirable impact.

I hope that we should be able to get some movement from the Government on this, as the coalition of groups making these points is an important representation of patients' wishes.

Nick Meade

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Animal Research in Cambridgeshire



Jumping in a taxi at the train station we announced where we were going and I watched anxiously as

A few months ago on a crisp sunny morning I made my way to Cambridgeshire to meet with Andy Gay at Huntingdon Life Sciences (HLS). I was going on an organised visit along with Sense About Science (<http://www.senseaboutscience.org.uk/>), Pro-TEST (<http://www.pro-test.org.uk/>) and organised and attended by the Research Defence Society (<http://www.rds-online.org.uk/>). None of us had been before and we were all keen to see what the facility was really like and to get behind the hype and media coverage that I am sure everyone will be familiar with.

GIG is a pro-research organisation and this includes the use of animals in research. We are vocal about the regulations and safety precautions that need to be in place in order for this work to be carried out and are aware and pleased that the UK has some of the most stringent safety laws in protecting animals used in medical research in the world. However, I was still quite anxious about the visit, and could not help but think of the media coverage I had seen of Huntingdon Life Sciences. I wasn't really sure what to expect from such a research facility and how I would feel on a personal level about being there.

We attended on a Friday as this is the usual day for protests outside the facility and it would give us a sense of what it is like to work there and how this affects staff and the centre in general.

the taxi driver took this on board. I thought that he might call us names or say how evil we were and I sat nervously looking out of the window. Funnily enough though he discussed with us how much the local community of Huntingdon support the research facility. He said that everyone knew that the work carried out there was done well and that the animals used were kept in excellent conditions. He was very animated at the disruption that the animal rights protesters had caused and how utterly unreasonable he thought their actions were to the staff who worked there. He had lived locally for most of his life and he did say that things had become much better recently with much less trouble and intimidation, which everyone was very pleased about. It was not the sort of endorsement I had expected and it reassured me about our visit.

Arriving at the facility security was understandably extremely tight, with ID required to enter the building and an escort to where we were going. The first part of our day would be a presentation on HLS and the current work and history of the company. After lunch we would be taken round some of the animal laboratories. We chose on the day which facilities we would like to visit as we needed to be shown around by the staff who cared for the animals in each building and this was arranged whilst we were given a presentation in the morning.

WHAT DOES HLS LOOK FOR IN THE WORK THAT IT CARRIES OUT?

- Toxicity (poisoning effect)
- Allergic reaction
- Residue (accumulation over time of a particular product)
- Inter-reaction (combinations of drugs or products)
- Efficacy (does it work)

Huntingdon Life Sciences

Huntingdon Life Sciences has over 50 years experience of working in the field of research. It has three centres, two in the UK and one in the United States. It employs over 1400 people and 50% of its staff are scientifically qualified.



HLS helps its customers to develop and bring to market safe and effective new compounds, 99% of the work carried out by HLS is due to the regulatory requirements of governments around the world. Companies often contract this type of research to companies such as HLS as it has the experience and facilities to carry the work out.

There are various pieces of legislation that require the use of animals in research within the UK

- Medicines Act 1968
- Health and Safety at Work Act 1974
- Food and Environmental Protection Act 1985
- Consumer Protection Act 1987
- Food Safety Act 1990

97% of animals are consumed for food - 12 animals per person in the UK per year

HLS assesses the risk of new products before they are exposed to humans, animals and the environment. A small amount of work is also carried out on herbicides and pesticides looking at their environmental impact. Any new compound must satisfy regulatory bodies before it can be brought onto the market and it is a legal requirement that it must be safe and effective. The production of new products is carefully controlled by government regulations all around the world, although the details of such regulation differ from country to country. Consumers also rightly demand that the products they use are safe and effective and that they "do what it says on the tin".

Some of the main therapeutic areas that HLS work on are cancer, heart disease, diabetes, Asthma, Parkinson's, Alzheimers, pain control, epilepsy, AIDS, a number of vaccines and many more.

As a company HLS has interactions with three main government departments in the UK, Department of Health - Medicines Control Agency, Department of the Environment, Food and Rural Affairs and the Home Office - Animal Welfare.

following the mornings presentation we were shown to the two laboratories that we had decided to visit earlier that



morning. We were going to visit the mini-pigs and the primates. Each animal house is a separate building and we needed to ensure that we were covered up appropriately for each area. Whilst waiting to see the mini-pigs we were also able to visit an environmental research facility where we saw quail and earth worms. Both were housed in very clean and light environments.

We then moved to the mini-pig enclosure. The pigs are breed specifically to be smaller than normal pigs because otherwise they become so large they can be unmanageable. The mini pigs we saw were housed in large pens and the room reminded me very much of a large market area with

THE SORTS OF QUESTIONS THAT MIGHT BE ASKED OF A NEW PRODUCT.

- Is the drug absorbed ?
- How long does it last in the body ?
- How is it excreted ?
- Are there metabolites ?
- ... are they toxic ?
- What is the best route of administration?
- What dosage, how frequently ?
- Is the drug safe ? What are the major side effects ?
- It the drug effective ?

lots of straw and lots of noise! The pigs we saw were being used in a dermatological test for a potential new drug to treat psoriasis. This meant they had to be housed singularly in pens to ensure that they didn't try to lick and swallow the drug being tested, or the bandage that covered the treated area. Normally the pigs would be housed in two's as they enjoy company. However they could all see each other through their pens. The product was tested in three different strengths on the animals and the toxicity tests would be lasting for three months. I asked what would happen if one

of the pigs had a bad reaction to the drug or product being tested, and was told that it would be stopped immediately and the dose would be revised or stopped completely depending on the type of reaction. The aim of the tests being carried out was to find out what the effects were of a range of doses of the drug without causing any obvious side effects. The aim, I was told, was to keep the animals healthy.

Much of the information gained from these tests would be found out in post-mortem, looking at how the product had affected internal organs (22 organ systems are examined), right down to the cellular level. All the animals in

pharmaceutical development research would be kept for this purpose and would be used in the findings.

Our next port of call was to visit the primate house, however as we needed to wait a while we had a chance to look at some farm animals being used in the development of new

veterinary drugs - horses and sheep. These species are not generally used in animal research and are only test subjects when they are to be the final beneficiaries of the potential new medicine.

The primate house was in another building and in order for us to go inside we had to put on full scrubs, including glasses and protective face-wear so that we would not carry any germs in with us.

The cages where the monkeys were held where in brightly lit corridor rooms, and at each end

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of the corridor were large clear glass doors which mean that the monkeys could look to see what was happening outside. This they did, a lot. There was also a lot of food all over the floors! After a few moments though it was clear where this was all coming from, the monkeys were having a great time eating and throwing things down, they were peering eagerly out of the cages to catch a glimpse of us all. The handler said that they had newly designed cages which had been developed by the handlers themselves and they had little areas that looked rather like bay windows where the monkeys could look out and see what was going on. The cages were also very high, from floor to ceiling. This helps because if monkeys are feeling nervous they like to climb higher and look down on whatever they see as a threat. It was explained to us that all the animals that lived there had been bred in captivity and that HLS never bought animals from the wild. It was much better for the animals to have been born into captivity and to be very used to people, as it would otherwise be a very stressful transition. This is not the case in other countries around the world where monkeys used in research can be taken from the wild

My nerves at entering the primate house had subsided considerably, all the animals we saw looked healthy, well, curious and at ease with humans. We were allowed to enter the room where the monkeys were kept and were told not to poke our fingers in the cages. This was not because they may be bitten off, but that our protective blue gloves may be snatched away! And of course this nearly happened to two of the party, the monkeys were strong and probably associated the bright blue gloves to their handlers and their food. On average the monkeys would be kept for one year or more and following this they (as with the mini pigs we saw earlier) would be put to sleep.

Monkey's are only used in medical research when no other species is appropriate, and only account for 0.15% of all the animals used in the UK (83% are rodents, mostly mice). Monkeys are sometimes used in the development of new vaccinations, in this instance they may receive the new vaccine only once or twice and then be kept and monitored for another year of observations.



Wherever possible smaller animals are used in research and the staff that we

chatted to were well aware of the sensitive nature of their work.

On leaving the primate enclosure I felt very happy with what I had seen.

Contrary to what I had heard the animals were well looked after by people who cared deeply for their well being. The animal handlers that I had spoken to had an immense amount of pride in the work they

The use of animals by society (in 1 year)

1000 million - used in food
14 million - domestic pets
8 million - destroyed as pests
3 million - medical research
¾ million - abandoned pets
1000 cats and 1000 dogs

did and held all the animals with respect and care. Due to the sensitive nature of the work that is carried out at HLS there are various procedures in place to ensure animal welfare standards remain high. For example if someone witnessed something they didn't feel comfortable with, they were encouraged to report it immediately. The best thing about this policy is that people don't have to speak

to their line manager, who they may feel uncomfortable dealing with, or even the vets and others within the company, but they could directly phone the Home Office Inspectors (who regulate UK animal research) should they feel they need to. The centre is visited a couple of times a month by these inspectors and also has daily visits from the companies who have research projects at HLS that carry out their own independent audits of the facilities.

Visiting Huntingdon Life Sciences has reassured me in many ways; the facilities that house the animals are clean, light, spacious and not at all gloomy. The people who work with the animals that we went to see were interested in there welfare and made every effort to ensure that whilst they were at HLS they were well looked after. Learning about the work that is undertaken at HLS dispelled many



misconceptions that I had about the facility and I also learned a lot about the types of animal research that is undertaken and why this is necessary.

Melissa Hillier

Genetic Testing Information for Patients - Where next?

This year's annual EuroGentest Unit 6 meeting took place in Milan at the end of October, and was attended by genetic professionals and patient group representatives from across Europe. This year we were pleased to have a number of new participants who had come from Eastern Europe including participants from the Romanian Prader Willi and Williams Syndrome Society and participants from the Bulgarian Gaucher Society and Bulgarian Information Centre for Rare Diseases. Altogether there were participants from 15 different European countries including Serbia, Belgium, Italy, Turkey and the Netherlands, which in itself was a great achievement. The main aim of the meeting was to update the group about the work carried out over the past 12 months, namely the development and translation of the patient information leaflets and the draft framework of core competences in genetics for health professionals, and to decide upon the agenda for the year ahead.

As many of you will know the 11 patient information leaflets developed over the past 12 months at GIG have been a great success. The leaflets cover issues relating to genetic testing including Inheritance Patterns and Chromosome Problems, Information about Genetic Testing and the Genetic Appointment, a Frequently Asked Questions about Genetic Testing and a Genetic Glossary. Much of the content of these leaflets was decided at last years annual meeting. Since then the leaflets have gone live on both the EuroGentest and Genetic Interest Group websites, and are currently available in English, Romanian, Bulgarian, Polish, Turkish and Estonian. Translation is also underway on 15 more European languages.

www.eurogentest.org/web/info/public/unit6/patients.xhtml

Following on from this work, it was decided at the Milan meeting to continue developing information for patient and families, translating and disseminating it across genetic clinics and patient groups throughout Europe. This time it was agreed that we would focus more specifically on the

practical implications as well as the psychological effects of genetic testing. This is because EuroGentest research has shown that there currently exists very little pre-written information in this area for patients and families at genetic clinics.

At the beginning of the session the patient group was asked to discuss ideas for the themes of the booklets. Due to the varied countries and conditions that were represented at the meeting, we were able to take advantage of views from a variety of different conditions and healthcare systems, and a number of interesting ideas were put forward. These included: Information for people considering carrier testing; Implications of having a child with a genetic condition - implications for parents, siblings and grandparents; Information for families when no diagnosis can be found or results are inconclusive; Information for children whose parents have been diagnosed with a late onset disorder and Practical legal information relating to patient rights and genetic testing. Participants worked together in small groups to discuss the possible content of these leaflets and when ideas



Participants at the EuroGenTest Meeting in Milan

had been exhausted the groups came together to discuss their thoughts and findings. The discussion was very lively and some useful ideas and experiences emerged during the afternoon. Over the coming 24 months we will be utilising and building on this work by developing practical and helpful guides for patients and families which will be translated and made freely available.

The meeting was a great success and feedback has indicated that the work begun is important and timely. However this meeting was only the tip of the iceberg. In the coming year we will be continuing the development of these information resources and speaking directly with patient and families who have personal experience of the above mentioned topics. We hope that GIG members will be willing to share their thoughts and experiences with us.

Celine Lewis

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The Family Route Map Project will conclude with formal Launch

We are delighted to announce that the Launch of the six condition-specific Family Route Maps together with the generic template and Final Report will be held at The Wellcome Trust Conference Centre in London on March 5th. We very much hope that the Support Group representatives and individuals and families who helped with the project will be able to attend as we aim to invite clinicians with an interest, the Department of Health and NHS decision-makers to join us in this launch and workshop.

Developing Family Route Maps as a Tool to help families with genetic conditions to access appropriate information and services in the UK was the primary objective for this groundbreaking and unique project. It was agreed that listening to the concerns of patients, families and carers was essential. Workshops were held for each condition with the aim to explore information and services currently available to these families as the first stage in the development of the Route Maps and also to ask them what they would like to see included. Additionally, an on-line questionnaire was made available through the GIG website with links to the websites of the six Patient Support organisations, and interviews with clinicians and other healthcare professionals with expertise and experience in these conditions were undertaken. In primary care in the UK we know very little, from the point of view of the patient, about caring for patients with rare genetic disorders and therefore primary care services were talked about in all of the patient focus groups. A focus group with General Practitioners (GPs) discussed the concerns and issues raised in the patient focus groups through a series of vignettes presented for discussion. From this collective information a template for a Family Route Map that could be used generically by other Patient Support Organisations was also created.

Seven important over-arching themes (below) were identified from the qualitative data with a number of sub-categories and these are discussed in the report in relation to current care and future possible developments:

- Information
- Communication
- Diagnosis
- Treatment and Surveillance
- Education of Healthcare Professionals
- Ethical, Legal and Social Issues; and
- Empowerment of patients, parents and carers.

Overcoming barriers

The patients, parents and carers involved in this project were by and large well informed as many had already been in touch with Patient Support organisations and some were themselves involved in providing support to others within one of the six charities representing the chosen conditions. However, it should be noted that during the course of the focus group discussions, people were actively exchanging relevant information and peer-learning took place. Newly diagnosed patients, the less articulate, less forthright and less resilient patients have an incredibly difficult time trying to get what they need or in some cases remain ignorant of the care they should be receiving because the current system of health and care services doesn't proactively seek to provide information in a timely and comprehensible fashion. Instead many people need to ask the right questions or for some finding relevant information is left to chance.

"It was only by chance that we came across the [Support] Group...it's really a case of who you know." (Gorlin Syndrome focus group)

What we found out from the project

Findings from this project suggest that patients with rare genetic disorders are not given sufficient information about their condition, services are considered 'patchy' and some families are still not aware of, or accessing, NHS Clinical Genetic Services. Many patients report: delays in being diagnosed; difficulties accessing the treatment and surveillance they need due to a lack of knowledge about rare genetic conditions in the medical profession and to an absence of coordination and continuity of care; and little psychosocial support leaving patients and families frustrated and 'stuck' in the system.

"It felt like you were on your own!" (Myotonic Dystrophy focus group)

Participants in the GP focus group were well informed of the risk factors and protocols for referral to a Clinical Genetics Unit for more common genetic cancers but for rare genetic disorders it appears less clear. Discussions between healthcare professionals, and individuals and their families about genetics-related information crucial for informed decision-making are often ad-hoc or absent, as those affected by genetic conditions move through their life-stages. Information through Patient Support Groups and the media (including

the internet) fulfilled some of their needs and helped them ask questions of professionals in order to make informed choice which is core to service delivery in the NHS.

"I didn't realise my daughter could be a carrier".
(Barth Syndrome focus group)

Many people expressed dissatisfaction with the lack of co-ordination and continuity in their care, and were frustrated by having to constantly explain the condition and fight for what they need. They describe themselves as 'slipping through the net' with no-one taking responsibility for them, as everyone thinks that someone else is caring for them.

"It makes me feel a bit like I've got to fight for everything I need" (SWAN focus group)

It is apparent that not all patients with the same condition are receiving the same surveillance and/or treatment. This may be appropriate due to different manifestations of the same condition, but patients feel unsure about whether they are receiving the most appropriate care, as many healthcare professionals do not have knowledge and experience of these rare genetic conditions.

"...in terms of eyes, kidneys [surveillance]...I have nothing whatsoever." (Nail Patella Syndrome focus group)

Importantly, even when lack of knowledge of these rare conditions by healthcare professionals was openly expressed to patients in a supportive way, participants felt there was an opportunity to learn together, valuing each others experience.

Those affected by the six conditions appreciate that it is not possible for healthcare professionals to know about every condition, but feel they should have the option to be treated by an expert, even if this involves travelling outside of their Primary Care Trust (PCT). People said that they would be prepared to travel in order to see an expert in the condition or to be cared for by a Centre of Excellence.

"Patients should be under the care of a specialist centre." (Multiple Endocrine Neoplasia Disorders focus group)

Conclusion

Although this study focuses on six specific conditions, it is an important source of information about the patient experience of clinical management and commonalities such as the need for clinical guidelines are likely to apply to the majority of

genetic conditions. This project has found that patients' needs are not being met and has generated suggestions for possible strategies that could be used to provide better care. A possibility favoured by patients is the option to attend a specialist centre, as this would ensure they are receiving the appropriate treatment and surveillance and allow for coordination and continuity of care. Patients currently find it difficult to identify experts within the field, so require more signposting, and experience difficulties with getting referral to these experts. Some people suggested that a further role for clinical genetics is to be part of the multidisciplinary care for patients with rare genetic conditions, providing information, coordinating care, and acting as a point of contact for any queries or concerns.

Another suggestion from some participants in one of the patient focus groups is that practice nurses in primary care could carry out the role of 'champion', coordinating care to ensure appropriate surveillance and treatment, and providing support to patients. It was felt that they did not need to have extensive knowledge of the condition, provided they knew what surveillance should be taking place, as they would be able to follow guidelines that had been put in place by experts. They are viewed as friendly and supportive, and it was felt they were ideally placed to be an advocate for patients who feel that their voices are often not heard.

"I think we all need an advocate." (Myotonic Dystrophy focus group)

The development of Family Route Maps will give patients access to information that they may otherwise have had to wait months or even years for, due to the rarity of the conditions and the inconsistent information that is available. The Route Maps will also help the health professionals who work with these families to explain all the treatment options, where to go for further information and which other disciplines the condition requires contact with. We anticipate that the Family Route Maps will also help to educate patients and professionals and will raise awareness of the complexity of these conditions and what patients may need to know.

If you would be interested in attending the launch of this project or receiving further information on the work please contact Melissa Hillier on melissa@gig.org.uk or 020 7704 3141

Anna Allford, Melissa Hillier

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New developments in PGD helping ALD Patients

Following the ALD Life conference on living with inherited disease in November last year, ALD Life had some extremely welcome news for female carriers wishing to start a family. (Adrenoleukodystrophy (ALD) is a rare, inherited metabolic disorder, which in most forms is an X-linked condition).

Previously, the only options to patients affected by ALD were to go ahead with normal pregnancy and have the foetus tested for ALD at the earliest opportunity if desired, or to embark on a process called pre-implantation genetic diagnosis (PGD) (see the box). This process involves IVF and then testing viable embryos to see if they are male or female and only replacing female embryos. Understandably a lot of people were worried about having a female child with the gene that they could pass on to future generations and the potential problems that they themselves may face, and so many have been waiting for a definitive test for an ALD gene.

At the ALD Conference Alison Lashwood, a consultant nurse in genetics for Guys and St Thomas' Hospital, London was pleased to be able to announce a new test that will be available at the Guys Assisted Conception Unit in the latter half of 2008. This new technology is called pre-implantation genetic haplotyping (PGH) (see the box). The process involves a course of IVF treatment,

What is PGD?

The PGD process takes a single cell from each embryo (following IVF treatment) and analyses them to distinguish between the embryos which carry a genetic disorder and those which don't. One or two unaffected embryos are then transferred to the woman's womb in the hope that this will result in a pregnancy unaffected by the specific genetic condition.

Due to the complexity of diagnosing genetic disorders in embryos up until now clinicians have only been able to diagnose relatively common conditions and more importantly those where the gene change (mutation) is the same for all those affected.

What is PGH?

PGH involves clinicians firstly amplifying the DNA from the single cell they have extracted from the embryo to replicate the cell's genetic material by a million times.

Then a technique called DNA finger printing is used on the cells. This finger printing enables clinicians to distinguish between the chromosomes carrying the affected gene and those that do not. This means that they can track the affected gene without having to look at the actual mutation.

For couples at risk from an X linked disorder, such as ALD it will also increase the number of embryos that are able to be implanted. As using PGD can only sex embryos for X linked disorders making sure only unaffected females are used. It is not possible to diagnose affected from unaffected male embryos, however this new technology will allow that.

following which a single cell is taken from each embryo. Clinicians firstly amplify the DNA to replicate the cell's genetic material by a million times. DNA finger printing is then used to distinguish between chromosomes carrying the affected gene and those that do not, meaning they can track the affected gene without having to look at the actual mutation. This means that PGH is looking at a lot more information and has the potential to be used for a wide variety of conditions such as ALD. PGH offers an opportunity to remove the ALD gene from a family tree permanently as parents will be able to use embryos which are free from ALD. In practical terms, this treatment will only be available at Guys & St Thomas' Hospital in South London. However, this does not mean that only those local to the area can apply for the treatment. Anyone in the UK can ask their local genetics centre to refer them. Timings for the treatment are usually eight months from initial consultation, so those interested can in theory start the process now.

Alison Lashwood said that "The development of PGH takes PGD to a whole new level allowing us to help even more couples have unaffected babies." Alison and her colleague Sarah Ross are readily available to discuss this and can be reached at the Assisted Conception Unit at Guys Hospital on 020 7188 1364.

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

Batten Disease Family Association celebrates its 10th Anniversary year

The BDFA, set up in 1998, is a supportive, informative, national networking organisation for the families, carers and professionals giving care to children and adults with Batten Disease and for promoting awareness of, and research into, the disease. Here are some events we are organizing in 2008 as we celebrate our 10th Anniversary by working towards our new Vision: To bring light to Batten Disease by being the central point of excellence in the UK for supporting affected families and to facilitate research into the disease.

28th March 08	Scientists and clinicians invited to a 'Bringing Therapy to Batten Disease' Workshop at University College London
17th April 08	Learn more about Batten disease and support resources for carers and professionals at the Batten Disease Training Day at Guy's Hospital, London
10th May 08	Join in our BDFA 3rd Sponsored Walk Challenge on The Ridgeway, Oxfordshire
5th June 08	Network and share expertise with fellow professionals at the Batten Disease Interest Group meeting, Evelina Children's Hospital, London
17th June 08	Keep up with the latest in developments in Batten Disease at our 'Taking Steps Towards Therapies for Batten Disease' conference and PSDL open day, King's College London

Please see the BDFA website at www.bdfa-uk.org.uk for further details or contact the BDFA Office on 0115 965 4815/email bdfa.info@btinternet.com

BDFA is a Registered Charity No 1084908

Ectodermal Dysplasia Society Launches New Website

Just before Christmas the Ectodermal Dysplasia Society announced the launch of their impressive new website which they hope will increase the profile of this rare group of genetic conditions.

Ectodermal Dysplasia (ED) is a group of closely related conditions of which more than 150 syndromes have been identified. ED causes abnormalities in two or more ectodermal structures such as hair, teeth, sweat glands, nails, cranial-facial structure and other parts of the body.

www.ectodermaldysplasia.org has been created as a valuable resource for its members and to raise awareness of ED among health professionals and other

organisations. Visitors to the site will be able to view general information about ED or find answers to more complex issues. The website will be regularly updated with the latest information and news.

The Ectodermal Dysplasia Society gained charitable status in 2001 and has contact with over 400 sufferers and their families in the UK and around the world. The ED Society has a Medical Advisory Board, the members of which are medical professionals with experience of ED, some of whom are involved in research into the condition. The Society has established relationships with similar organisations in other countries.

President of the Society, Diana Perry, said 'We are delighted with the new site. It will raise awareness of ED with the public and professionals. We are very pleased with the new Members Section which will greatly increase the amount of information we can offer.

The site is clear and very easy to use with lots of up to date and useful information. It's very easy to navigate and to learn more about the group of conditions that ED covers. Patients and health professionals can also locate other ED support groups throughout the world.

EVENTS and NEWS

The Ataxia - Telangiectasia Society Organise an "X scape" Weekend for young people with A-T.

An "Xscape" Weekend in Milton Keynes is being organised for young people with A-T from 8th to 11th March where they will be able to try skiing and simulated sky-diving at "Airkix" as well as enjoy the shopping and all the other facilities Milton Keynes is able to offer to wheelchair users because it is so wheelchair friendly.

International Research Workshop in Japan on AT between 22 -26th April 2008.

10th May the AT Society will be holding their AGM and annual Family Day at Nottingham.

For more information about any of the above please contact Maureen Poupard, Hon Secretary on atsociety@btconnect.com

New Years Honours List

We are delighted that the following people have been given awards in the New Years Honours List.

Knight

Professor Bruce Anthony John Ponder, Head, Department of Oncology, University of Cambridge and Li Ka Shing Professor and Director, Cancer Research UK Cambridge Research Institute. For services to Medicine.

Dame

Professor Kay Elizabeth Davies, C.B.E., Dr. Lee's Professor of Anatomy, University of Oxford and Director, Functional Genetics Unit, Medical Research

Professor John Irving Bell, Regius Professor of Medicine, University of Oxford and President, Academy of Medical Sciences. For services to Medicine.

Angioma Alliance (UK) hold their Forum, Saturday June 7, 2008. Admission free. Orion Suite, Grange Holborn Hotel, Southampton Row, London. The morning, (registration from 0830,) will consist of speakers; the afternoon, member workshops. Angioma Alliance (UK) have negotiated a special accommodation rate at the GrangePlease call Ian Stuart for further details (01305) 213876. The event is sponsored by the Batty Charitable Trust.



The UK Thalassaemia Society National Doctors' Conference in association with the Royal Society of Medicine 10th June 2008

"Thalassaemia - a childhood condition comes of age" will be held in The Lecture Theatre, The Royal Society of Medicine 1 Wimpole Street London W1G 0AE.

Contact Elaine Miller at the UKTS office for further details on 020 8882 0011 or office@ukts.org

www.ukts.org

The Marfan Association has a new website www.marfan-association.org.uk please update your address books as the old site (marfan.org.uk) will no longer work.

Annual Marfan Association UK Information Day Saturday, 8th March, 2008 at St.George's Hospital in London-

For more details please contact the Marfan Association at Rochester House, 5 Aldershot Road, Fleet, Hants. GU51 3NG. marfan@tinyonline.co.uk or 01252 810472

Behaviour and Mental Health in Prader-Willi Syndrome Reading University - 27th March 2008

A conference to explore the complex issues which surround behaviour in PWS, and the mental health problems which sometimes accompany the syndrome.

This will be of interest to healthcare, education, social services and residential professionals who are helping to manage the behaviour of both children and adults with PWS. For more details, contact PWSA (UK), 125a London Road, Derby DE1 2QQ, tel 01332 365676, or visit our website at www.pwsa.co.uk