



## Money back for your kids when you shop through KidStart

1. Shop online - choose from over 300 retailers



2. You earn money back from your shopping



3. We give it to your children for their future



# KidStart

The smart way to save for children

Please support GIG's work - shop through the KidStart Website!

If you ever shop on-line, PLEASE consider supporting GIG's work by using the KidStart website. All the money we make through KidStart will go towards funding the work we do representing and supporting our member groups.

KidStart is a shopping club that works with almost 300 on-line retailers and a growing number of high street shops and service companies, including Marks and Spencer, Waterstones, Expedia, Argos and PC World.

Anyone can join KidStart, free of charge. Every time you shop on-line through www.kidstart.co.uk you earn savings, generally between 5-10% of your spend (the percentage is set and paid by the retailer). Your savings are transferred to your "Kiddybank" and you can then allocate them to GIG, to a school or to a child's trust fund or savings account. The leaflet enclosed with this newsletter tells you more about GIG's partnership with KidStart.

I joined KidStart a year ago and I have saved nearly £175 for GIG, simply by doing my regular shopping through the site.

PLEASE consider supporting our work by joining KidStart and saving for GIG.

PLEASE also consider promoting KidStart amongst your members and asking them to save for us too.

### How to save for GIG through KidStart:

- Log on to www.kidstart.co.uk/GIG.
- If you are not a KidStart member, you will need to register. You will be asked to accept the invitation to save for GIG. If you are already a member of KidStart, just log in and accept the invitation to save for GIG.
- Go to MY PROFILE and you will see GIG as one of the

### Five Good Reasons to save for GIG with KidStart

1. GIG is the ONLY charity working for people with genetic disorders that you can save for through the KidStart website, so supporting GIG in this way WILL NOT divert money away from your charity,
2. Saving for GIG via the KidStart website won't cost you anything. Savings come from the retailers.
3. You can also save money for your children or for a school through the KidStart website.
4. If enough people save for GIG through KidStart it will provide a sustainable source of funding for our work. This will enable us to continue to represent and support charities for people with all types of genetic disorders in the long-term.
5. Saving money for GIG through the KidStart website is very easy and very satisfying.

beneficiaries to receive savings as you shop. You can also add a child or school that you wish to save for, and you can share your savings amongst your beneficiaries.

- You can start to shop immediately and collect savings which you can view in your "Kiddybank".
- Every time you shop with KidStart, a percentage of what you spend is automatically saved. It is transferred to GIG on a six monthly basis.

Please contact me at Helen@gig.org.uk if you have any questions about KidStart or if you would like copies of the enclosed leaflet to distribute to your members.

With many thanks for your support.

Happy shopping!



summer 2009

## Innovative Therapies and Rare Diseases. Are we there yet?

GIG's annual conference this year was held at the offices of the law firm Clifford Chance in London. We are extremely grateful for the pro bono support we received from Clifford Chance for this event.

The morning of the conference looked at the therapeutic possibilities arising from cutting edge research, Professor Peter Hillman explained how he became involved in the development of a treatment called eculizumab that has now become used in an ultra rare condition called Paroxysmal nocturnal haemoglobinuria (PNH) a rare blood disorder (although the treatment was not originally developed for this purpose). There was over 10 years of research prior to any discussions of the uses of this technology in patients and Professor Hillman explained clearly the long process and how as other developments for rare conditions such as Gauchers were marketed that it became clear there could be a viable way to provide this treatment to PNH patients. The first trials were carried out in 2002 and in 2007 eculizumab was licensed to treat PNH patients.

Dr Peter Hollands then spoke to us about the developments in stem cell therapies focusing particularly on the current and potential uses of cord blood stem cells. Cord blood stem cells are taken from newborn children and can be stored to be used at a later date, for example in blood conditions. The collection of the cord blood is a very simple procedure, which takes just a couple of minutes after the birth of a baby. To store the cord blood, stem cells are isolated from the other blood cells and a protective substance is added to allow freezing. Cells are packaged and frozen in computer controlled freezers and stored at -196 degrees Celsius. There have been a few cases of successful transplantation of cord blood cells particularly in the blood disorders including Fanconi's anaemia, sickle cell and Thalassemia. There have to date been over 10,000 successful transplants worldwide, and Dr Hollands was very positive about the potential for this treatment to be used in other conditions in the future.

Following on from this Dr Adrian Thrasher and Dr Robin Ali spoke about gene therapy and how this relatively new

technology has been developed. Moving on from the "boy in the bubble" syndrome (SCID) to other conditions such as Cystic Fibrosis and Epidermolysis Bullosa. They explained the processes that have to be undertaken by patients for this treatment and also some of the amazing outcomes that have taken place to date particularly in SCID and also in X-CGD. Dr Robin Ali made a powerful presentation about the use of gene therapy in a particular form of Retinitis Pigmentosa called Lebers Congenital Amaurosis (LCA) which is early onset and can lead to blindness in patients. The outcomes of his research demonstrated the improved vision that patients gained having had gene therapy. This research is still in the clinical trial stage and is not a treatment as yet but Dr Ali has found that this treatment is safe, there have been no adverse effects or immune responses. Dr Ali gave a similar talk at the RP Fighting Blindness Conference in 2008 and you can see the full talk on YouTube...

<http://www.youtube.com/watch?v=mpO9qYDPCvY> this includes a talk from Steven Howarth who was one of the first patients to benefit from this new type of gene therapy.

Our final speaker for the morning was Melissa Smith, a patient and Trustee of Debra, the charity working on behalf of people with Epidermolysis Bullosa (EB). Melissa was able to put the patient perspective onto all the research we had heard about. Melissa is taking part in a clinical trial using cell therapy for EB.



GIG team members: Helen Parr, Melissa Hillier, Ben Francis and Krystle Kontoh at the GIG Annual Conference.

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## Who is at GIG?

### Head Office

- Alastair Kent - Director
- Melissa Hillier - Assistant Director
- Amy Hunter - Senior Research and Policy Manager\*
- Nick Meade - Policy Officer
- Helen Parr - Fundraising Officer\*
- Celine Lewis - Project Officer (Eurogentest)
- Alex Mckeown - Project Officer (EurogenGuide)
- Ariadne Stamatopoulou - Project Officer (Patient Partner)
- Stephen Nutt - Public Affairs Assistant
- Heather Law - Administrator
- Krystle Kontoh - Facilitating Networks Project Officer
- Benjamin Francis - Insurance Template Project Officer
- Rebecca Pitfield - Finance Officer\*

### Regional Staff

#### Cardiff

- Buddug Williams - Wales / Cymru Development Officer

#### Scotland

- Gillian Scott - Scottish Development Officer\*
- Claire Cotterill - Patient Engagement Project Officer

\*part time

One of the reasons that patients with EB have such difficulties with the blistering of their skin is that their bodies do not produce enough (or any) fibroblasts (fibroblasts are cells of connective tissue that produce and secrete fibers) that produce Collagen VII. This means that the skin is less able to recover and regenerate, which can lead to chronic non-healing. Melissa has taken part in two clinical trials looking at injecting collagen VII into a patient to assess whether or not this will help to make the skin more resilient and able to heal better.

This new cell therapy has shown very positive signs of working and after a trial in a very small area, researchers are now testing a larger skin area on Melissa. In October 2008 Melissa had 106 injections into healed skin on her back as well as 4 injections into a wound on her shoulder. The results were amazing!

The healing time was reduced considerably and the skin was more stable afterwards as well, meaning less blistering. This treatment is more effective than dressing and topical treatment which is the only source of treatment currently available to patients with EB. Melissa has found that by taking part in this clinical trial she now feels like she has some control over her condition. Melissa has since had other injections and although they can be painful, Melissa felt that the long term gain was definitely worth it. Other patients have also now had the treatment with success.

Although a cure is the ultimate goal to end the blistering completely it was felt that the use of fibroblast injections are a guiding light on the journey, a ray of hope that will help patients to beat EB and it encourages others to become involved in the fight.

Following workshops (findings are mentioned on P8) we had a lively debate, which posed the question "Innovative therapies for rare diseases offer value for money". Our two debaters were Steve Bates from Genzyme Therapeutics and Ed Jessop from the National Commissioning Group. Both gave powerful arguments Steve started the debate with a short summary of Genzyme and the products they produce. Steve felt that innovative therapies offer value for money as they have dramatic effects on patients quality of life, and offer treatment where there was none before. Ed Jessop was keen to look at the improvements treatments made and felt that in a case where the improvement to a patient was dramatic and life changing then as long as the NHS was paying a "fair price" the treatment should be provided. Where the decision making process was less clear cut was in regards to treatments that could extend life by a few weeks or months, or where the cost to society was felt to be too high. We had a lively debate and concluded that innovative therapies needed to be judged by a fair and rigorous system to ensure that the society was gaining value for money but also to ensure that patients were able to access treatments that could have life enhancing benefits.

## NEW STAFF MEMBERS AT GIG

GIG is delighted to welcome two new members to our team at GIGs Head Office in London. Ariadne has recently graduated from Brunel University where she was studying Psychology .... and joins GIG on a new project called Patient Partner. Steve, comes to GIG from The National Autistic Society Cymru, in Wales and will be joining our ever busy Policy Team. Steve will also be working as the Secretariat for the Rare Disease UK initiative that GIG has recently been instrumental in setting up.

### Ariadne Stamatopoulou

Project Officer - Patient Partner



I am a new project officer working on the Patient Partner project at GIG. The Patient Partner project is an EU funded project that focuses on the development of patients', organisations', sponsors', doctors', and pharmaceutical companies' awareness of clinical trials, what they entail, and the relationship between the patients and doctors concerning clinical trials in

Europe. The main focal points of the project will be the types of relationships between the patients and doctors, and suggestions as to how one would be able to further those relationships, resulting in better and more clinical trials. These topics will be addressed with the help of Guides which will be produced for both patients, and the organisations/sponsors, as well as a list of recommendations, and a thematic website which will be accessible to all.

[ariadne@gig.org.uk](mailto:ariadne@gig.org.uk)

#### What are clinical Trials?

They are based on research studies that involve people who have a certain medical condition or make use of health services, which compare a new or different type of treatment with the best treatment currently available.

### Stephen Nutt

Public Affairs Assistant



My name is Steve and I've just started working as a Public Affairs Officer for GIG.

I'm from South Wales originally; I studied Law at the University of Manchester and then I went on to study for a masters in International Politics. During this time I decided that I wanted a career involving politics but I also wanted to work for an organisation that I felt was

pursuing a worthwhile cause and whose aim is to help people – this led me to public affairs.

Following my MA I moved back to Cardiff to complete a Public Affairs and Campaigns internship with the National Autistic Society Cymru(Wales) and an internship with the Liberal Democrats. I was then offered a job with the NAS Cymru as an interim Policy and Public Affairs Officer, communicating the needs of people with autistic spectrum disorders to the Welsh Assembly Government. I also provided the secretariat for the Cross Party Autism Group in the National Assembly for Wales.

At GIG I will be supporting the public affairs, policy and communication activities in raising the profile of GIG and our campaigns in Westminster and Brussels. I will be working with Melissa and Nick as well as supporting public affairs activity in the devolved nations when appropriate. A large amount of my work will be on Rare Disease UK; I will be providing the day-to-day secretariat and co-ordinating and publicising activities.

I'm very excited about getting stuck in at GIG and Rare Disease UK and to highlight the needs of people with rare and genetic disorders and their families to government and politicians.

[stephen@gig.org.uk](mailto:stephen@gig.org.uk)

## RAPID kicks off

Many women at risk of having a baby affected by a serious genetic condition opt to take a diagnostic test while pregnant. Such prenatal genetic diagnosis currently requires invasive testing involving the insertion of a needle directly into the fluid surrounding the foetus, or into the placenta. This is carried out from 11 weeks of pregnancy and on average 1 in every 100 women tested will miscarry due to the test itself. Because of this, healthy foetuses are lost each year after invasive testing.

Recently laboratory techniques have been developed that have allowed DNA from the foetus to be obtained quite safely from a normal maternal blood sample, carrying no miscarriage risk. This also means earlier diagnosis is possible and could lead to health service savings with fewer invasive tests and complications.

Some NHS laboratories already offer limited non-invasive diagnosis and there is an urgent need to evaluate it thoroughly from a technical and clinical point of view, and to develop laboratory and clinical standards before it 'seeps' further into practice.

'RAPID' stands for 'Reliable Accurate Prenatal non-Invasive Diagnosis' and is a major new project that aims to address this situation.

GIG is a co-applicant in the project which is being led by Dr Lyn Chitty of Great Ormond Street Hospital NHS Trust / Institute of Child Health. The project is funded by the National Institute for Health Research.

### Work to be done

GIG's involvement will ensure that couples' views and information needs will be taken into account from the outset. The decision to take a prenatal diagnostic test is an important one for any woman or couple and should be taken with access to accurate information and with the support of appropriately trained health professionals. This should not change just because the test itself becomes more straightforward to carry out and may be seen as more 'routine' than the current invasive test procedure.

We will use one-to-one interviews, questionnaires and focus groups with those at risk of the disorders being tested to determine couples' preferences as to what and how information should be presented, and which health professionals they would prefer to guide them through their decision about testing. The results of this work will be used in

'RAPID'  
stands for  
'Reliable  
Accurate  
Prenatal  
non-Invasive  
Diagnosis'

the creation of good patient information and the education of doctors and midwives offering patients the tests.

The project will focus on three distinct uses of the technology:

- Sex determination of the foetus, where it is medically significant, such as for X-linked conditions
- Diagnosis of single gene disorders, such as cystic fibrosis
- Diagnosis of Down's syndrome.

### Costs

Before the NHS can adopt significant changes to prenatal services, their impact on budget must be understood – many questions must be addressed, such as what will the tests cost compared to current invasive testing, will more people take the non-invasive test, what will be the impact on training costs for medical staff? The project will therefore include an economic analysis of non-invasive testing, for the three uses described above.

### Updates

A dedicated website for the project is being created, and the first information day (to which GIG members were invited) was held on 7 July. The website address and regular updates on progress will be published here as they become available.

The official start date for the project was 1 June 2009, and it will run for 5 years. GIG is recruiting a Project Officer to carry out our part of the project which will be overseen by Senior Research Manager Amy Hunter. For more information email amy@gig.org.uk

## Launch of a nurse-led, single gene, community partnership project

A new and innovative project for people living in the community with complex needs as a consequence of single gene conditions has been launched in Scotland. (There are several thousand single gene conditions; examples include Friedreich's Ataxia, Myotonic Dystrophy, Neurofibromatosis, Huntington's Disease).

Over at least the last decade our understanding of genetic conditions, genetic science, diagnosis, prognosis, and how to effectively manage care has transformed considerably. This increased awareness and interest in genetic issues naturally increased speculation about the potential application of this knowledge across all areas of health and social care provision. Dr Mary Porteous (consultant geneticist) acknowledged that "it was vitally important that advances in genetic technology are matched with improvements in the holistic care of individuals affected by complex genetic disease".

Against this backdrop, a review of genetics in relation to Health in Scotland was commissioned. The review group's remit was to assess what developments and resources would be required to enable the NHS in Scotland to be in a position to harness developments in genetics and to maximise the advantage for people in relation to health.

The Single Gene Complex Needs (SGCN) project is an outcome of that review.

This SGCN project marks a significant expansion of resources provided by our genetic centres in Scotland. Marie McGill, the project lead explains, "the commitment to effectively manage a person's care and monitor activity and outcomes at every stage of a person's journey has been recognised by genetic services as an essential pre-requisite to maximise people's outcomes and improve the quality of service provision. The SGCN project will work in collaboration with our third sector, public and private sector partners towards this goal".

The SGCN project plan has considered very carefully the significant changes to the way health and social care is delivered in Scotland (within a rapidly changing social environment) but more importantly puts the interest and concerns of patients at the centre of all that the project does to

build the capacity of local service providers and so improve quality of life.

It is critical to the success of the project that we understand and implement strategically a coherent plan of care and support. People living with complex needs as a consequence of genetic change are faced with sizeable challenges. Many people living with complex needs who want input from relevant disciplines tell us they face challenges with access to, and co-ordination of, services. Many professionals have little or no experience of working with families with complex needs as a consequence of genetic change. All require support to optimise care pathways and peoples' outcomes.

The SGCN project team is grateful to our Scottish Government, Sir Kenneth Calman, the Review Group, the Clinical Genetic Centres, the Scottish Huntington's Association and all other individuals and organisations who are already working incredibly hard to improve outcomes for people in Scotland living with the extraordinary challenge of genetic conditions. Reminding us all of how important this new service is to people living in the community, Amanda McNab, a service user described her initial response to the project,

"I was absolutely ecstatic that our voices had been heard, our perseverance, our determination has paid off. This is so important. I phoned everyone that evening then sat down and had a glass of wine to celebrate". As a consequence of the commitment of all the aforementioned we now have this invaluable opportunity to put your vision into action.

### Project contact details

**Marie F McGill, Nurse Consultant and National Lead**  
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**Jacqueline Ellis, National Project Administrator**  
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**Single Gene Complex Needs (SGCN) Project**  
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Western General Hospital, Crewe Road South, Edinburgh EH4 2XU  
Telephone: 0131 537 1062 Mobile (Marie): 07932 628 604

## Facilitating Networks Project

The Facilitating Networks project will enter the second year of its three year life cycle in August. Since the project's initiation it has gathered momentum and seen a few changes made to the groups it had originally planned to work with.

In an effort to reflect a truer picture of Tuberous Sclerosis Complex we moved this group from the dermatological category and reclassified the condition within network group for rare neurological conditions. This reshuffling was coupled with the XP support group being unable to take part in the project and left a vacuum within this group. Having carefully considered which of our members would benefit from involvement and fit within the set, we will now be working alongside The Ectodermal Dysplasia Society and HITS (the Support group for Hypomelanosis of Ito). These two support groups have great potential for collaborative work and we look forward to establishing what areas of work their network will focus on.

For further information on the project please go to the GIG website's Facilitating Networks page.

[http://www.gig.org.uk/projects/facilitating\\_networks.htm](http://www.gig.org.uk/projects/facilitating_networks.htm)

## Patient Leaflets Promoted in Europe



As the Eurogentest Project (www.eurogentest.org) enters its last phase, we have been attending a number of international genetics conferences, promoting the work of EuroGentest and GIG. In particular promoting the patient leaflets that have been written which discuss how genes are passed on through families and provide information about the various genetic tests that currently exist. This information has been written with the help of our members and other European patient groups to ensure that it provides the scientific as well as the more personal, human aspects that patients undergoing genetic testing experience. In March GIG was invited as a speaker at the Czech Working Day for Human Genetics in Prague. This was a conference attended by geneticists from across the Czech Republic and Celine Lewis, EuroGenTest Project Officer presented a talk titled 'Developing and translating genetic information for patients; experience of the EuroGentest project'. One of the aims of this presentation was to inform clinicians about the availability of the leaflets, explain the processes that took place in developing and translating them, and to highlight that they are free to download and print out. Feedback was positive and we are hoping that we will now see the numbers of clinicians using these materials in the Czech Republic rise.

EuroGenTest was represented at the 8th Balkans Meeting on Human Genetics in Cavtat, Croatia. Again, presenting our work developing patient information, and we also had a stand in the conference area where there were leaflets in numerous languages on display. We made a number of good contacts at this conference who have offered to help with future translation work. By the time you read this article we will have also attended the Eastern European Rare Diseases Conference in Plovdiv, Bulgaria. So all in all it has been a very busy few months with lots of travelling.

We have had a great response to the leaflets and HTML pages on which the patient information is also available. In May the resources were viewed over 8000 times and in 13 different languages. We are hoping as new translations are completed, and as promotional activity increases, these numbers will rise. Feedback so far has been very encouraging. We have received a number of personal e mails from clinicians from across Europe praising the initiative, and saying that they have found the leaflets to be a useful resource in their clinics. A recent article in the European Journal of Human Genetics also spawned more interest in the leaflets with a number of clinicians from inside as well as outside of Europe contacting GIG for further details.

If you would like to view the information on line, or if you would like to add a direct link from your website to any of the leaflets, they can be found at the following address:

[www.gig.org.uk/publications.htm](http://www.gig.org.uk/publications.htm)

## EuroGenGuide

As the EuroGenGuide moves into its final six months, there is no better opportunity to give you an update as to what has happened so far this year, and what the future holds for the project beyond its completion and its launch. This piece will be the final article about EuroGenGuide for the foreseeable future, and so if you are interested in using the material that we have developed, or have any comments or advice to offer on the work that the EuroGenGuide team has been doing, then please visit the project website at <http://www.eurogenguide.eu>.

At the time of writing, from the website it might appear, if you have been following what has been happening with EuroGenGuide, that not much has changed since the last time you looked. However, this is because the focus of the past six months has been on updating and refining the material 'behind the scenes'. The fruits of these labours will be visible on the site over the Summer and towards Autumn. So, in the meantime let me explain what it is we have been doing.

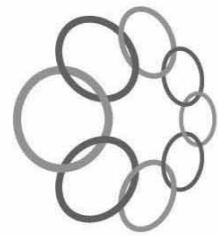
The main focus of the work for the period immediately preceding this, during the spring and up to the present, has been a wide-ranging user survey. This was carried out in order to get an impression of how the guide has improved from Version One – which went online last July - and the more recent Version Two which has been the version visible online since January of 2009.

The survey was only possible thanks to one of the improvements to the site which was added to become a part of Version Two, and this was the online survey system. This

enables the website administrator to devise a questionnaire or survey, contained within the website and with questions about how well the material is working. The link is then emailed to patient groups and networks of contacts and the data is automatically directed back to the website to be collected by the project team.

We were pleased to find that the results of the survey were resoundingly positive. 74 different patient groups from 17 different countries across Europe replied, rating EuroGenGuide according to criteria such as relevance, intelligibility, usefulness, useability and appearance, and offering comments as well. Nonetheless, some very valuable suggestions for improvement were made, and it turning these suggestions into reality that is the aim of the EuroGenGuide team for the remainder of the project as we look towards its launch in Warsaw in late November. Keep checking and over the coming months you will see significant changes and improvements all over the project website.

The past two and a half years have been fascinating and rewarding, and developing EuroGenGuide has been a hugely satisfying experience for the whole of the team. We all hope that by the time of its launch, EuroGenGuide will be a useful resource for all of those across Europe who want to find out about their options in respect of genetic testing, research and counselling. EuroGenGuide is what is known as a 'Specific Support Action', for the larger EuroGenTest network. It is GIG's and the EuroGenGuide team's aim that the work that we have carried out over the course of this project will help to ensure that all those who need support, advice and information about their choices are able to access it in the best interests of their own health and the health of others affected by genetic diseases.



# RARE DISEASE | UK

## Patients highlight the lack of Communication and Coordination as having significant impact on the healthcare they receive.

In the afternoon at this year's Annual Conference we had an update from Rare Disease UK on the work they have been undertaking, followed by workshops to gain patient experience of current NHS services and where we would like to be in the future.

Rare Disease UK is a joint initiative of the Genetic Interest Group, and others in response to unmet health care needs of families who currently struggle to get access to integrated care and support from the NHS.

We posed two questions in the breakout sessions, "What are your current experiences of NHS Services?" and also "What would a better health service look like?" The results have been summarized below.

### What are your current experiences of NHS Services?

Three main themes came out across all four workshops when addressing the current issues surrounding patient and families experience of the NHS. These three themes were unanimous across the workshops.

#### Diagnosis

The first of these can be gathered under the banner of "Diagnosis". There are various issues within this including the delay in diagnosis and also the misdiagnosis of conditions. *"A child was diagnosed with the incorrect form of our condition, the prognosis for these two conditions is very different and it was a very upsetting and unnecessary experience for the family".* Another delegate commented that in their condition *"over 90% of patients were misdiagnosed".*

The

phrase "postcode lottery" came up when discussing diagnosis, as it was felt that it depended where you live and who you saw as to whether your diagnosis would be timely or not. It often seemed down to "luck" more than anything else when an accurate diagnosis is made. Delegates commented that they felt it was like a "fishing expedition" rather than a linear process of gaining an accurate diagnosis.

There was perceived to be a severe lack of knowledge surrounding rare conditions. It was felt by delegates that GPs are ill equipped to deal with, or to recognise the often disparate symptoms of rare conditions. Information is lacking – a suggestion was made later about having an 'NHS Direct for Doctors' or an accessible resource so they had a central hub they could contact if they were unsure or needed further information. Some delegates felt that GPs (and other healthcare professionals) have a tendency to look at individual symptoms and never to 'join up the dots' and link the symptoms to a condition. There was also felt to be a reluctance by some clinicians to admit the limit of their knowledge.

Although some comments were critical of GPs, it was clear that delegates were not expecting them to be knowledgeable about the several thousand rare conditions in existence, but rather that they did expect the GP to be the gatekeeper to services: and that they be able to manage the patients care and to help them seek a diagnosis and ongoing management and further treatment where necessary.

#### Communication

The second major theme that arose, again with consistency across all four groups was the lack of Communication. This theme encompassed various issues and at different levels, including the lack of, or inappropriate communication to patients from healthcare professionals, the lack of communication between healthcare professionals, between hospitals and between departments in hospitals.

Due to the nature of rare conditions it was seen as vital that clinicians and patients work together as partners. This would enable the families to share their knowledge with clinicians and enable information flow to improve. It was felt that this currently happened far too infrequently.

## The National Alliance for people with rare diseases & all who support them

### Lack of Coordination for Service Provision

The final theme encompassed an even larger topic area. However it did move the workshops on very nicely to the second question. There was also general agreement across groups that the way services are commissioned means that there is a real lack of standardisation in the treatment of rare conditions. With the lack of formal registries and coding for rare conditions it often means that it is hard to gather the solid data required to move forward with rare conditions and to identify what is best practice. Delegates whose condition had a "Network" or "Centre" always, without exception, said they had dramatically improved the care families received. It was felt that more care pathways were needed, and again for them to be standardised throughout the UK. This would also help clinicians to know where to signpost families to and could provide a mechanism to ensure that patients weren't "kept" because they were seen as "interesting" by clinicians, who although meaning well, may not be providing them with the optimum service due to their lack of knowledge and communication with others.

### What would a better health service look like?

#### Structure

Improved structure of NHS services for those affected by rare conditions was felt to be the key issue and with a structure in place many of the other issues raised could well be resolved. As part of the structure there is a greater need for the systematic gathering of data on rare conditions, eg registries. Using the ICD 11 Codes when they are issued in 2014 will be extremely beneficial to rare conditions. There also needs to be thought put into the development of disease prevention and screening initiatives. Often rare conditions rely heavily on one clinician who is a champion in the field. There needs to be more consideration for succession planning to ensure long-term service provision and sustainability. A whole health service across a region can disappear if a clinician retires. With more planning, improved structures, recognition of specialised services and funding for rare diseases, patients can expect more coordinated services.

#### Funding

Funding was also raised in various groups particularly the need for a national funding programme for all rare conditions. This would again feed into a more systematic and structured service provision package.

### Centres of Expertise/Excellence

These were seen as best practice for rare conditions. It was felt important that if such centres existed for all rare conditions that it was vital that families were sign posted to them.

Currently many delegates who were either involved directly in a "managed clinical network" or where a "care pathway" or "centre" had been developed felt that this had happened due to many years of campaigning by patient organisations and clinicians with a special interest in a rare condition. The services have grown up organically, and are often funded in an ad hoc way. For example, Thalassaemia centres have been running for some time now but will only be funded formally in the coming year. Prior to this centres have evolved and been funded by patient groups and in kind by time donated by clinicians and hospitals. Centres currently also tend to be in areas where there are lots of patients with the condition who have got together to develop such a centre. This needs to be more structured and coordinated so that all rare conditions can be included and guidelines can be developed.

Care pathways can be a cost effective way of using limited resources. Currently due to the delay in diagnosis and the sometimes incorrect treatments that patients undergo, resources are being wasted. It is critical that care pathways are seen as a benefit and not a huge extra financial burden on already stretched NHS resources.

Raising awareness of patient support groups would also be helpful, as they have a wealth of knowledge about the condition and can often help newly diagnosed families find an expert.

## Genetic testing and implementing the new HFE Act 2008

By Danny Edwards, Policy Manager at the Human Fertilisation and Embryology Authority (HFEA)

Over the last 12-months we have been working closely with our stakeholders in preparing for the 1st of October, when the amendments contained in the Human Fertilisation and Embryology (HFE) Act 2008 come into effect. This included holding in January a consultation event with patients and clinicians on new requirements for genetic testing in fertility treatment as well as inviting written comments and suggestions from interested parties. It was important for us to understand fully people's views on how best we can apply the new legislation and we are grateful to the thoughtful and valuable contributions we received.

### New PGD Licensing Arrangements

The Human Fertilisation and Embryology Act 2008 has brought the regulation of preimplantation genetic diagnosis (PGD), HLA tissue typing and PGS (known collectively as embryo testing) onto a statutory footing for the first time.

Part of this change in the law is a new statutory requirement that the HFEA must be satisfied that PGD is carried out only either where there is a significant risk that a child born will have or develop a serious condition. The HFEA has taken this chance to look again at the way the Authority licenses PGD.

In March of this year, the Authority took a decision to remove the requirement that an application from a clinic to test for a condition (that they have not tested for before) be based on the facts of a particular family. From October 1, conditions will be licensed by the Authority 'in principle' meaning that any clinic already licensed by the HFEA to carry out PGD will be able to test for any

condition which the Authority has agreed meets the statutory requirement for seriousness. Clinics will be expected to continue to decide on the appropriateness of PGD in individual cases, using the guidance in the HFEA Code of Practice. We expect this change will help those people needing PGD to access their treatment faster by removing the need for the licensing of each clinic for each condition.

The licensing of genetic testing involving tissue-typing (what is often described in the media as the creation of 'saviour-siblings') and testing for conditions which are late onset or low penetrance, will continue to be made on a case-by-case basis. These categories of testing will be the subject of a policy review by the HFEA later this year.

### The 8th Edition of the Code of Practice

Over the last year, following extensive consultation with the fertility sector, we have completely revamped the HFEA Code of Practice. It is now much easier to navigate, read and understand, and presents guidance more clearly and accessibly. For example, all the guidance the HFEA provides on preimplantation genetic screening (PGS) and on embryo testing is contained in two specific guidance notes. There have been several additions and changes to guidance in the HFEA Code in the area of genetic testing, both as a result of the HFE Act 2008, and other policy decisions.

### Using donors or embryos in treatment known to have a serious genetic condition

The HFE Act 2008 includes a prohibition of deliberately preferring donors or embryos known to carry a risk of that donor or embryo having a serious genetic condition. However, the law does not completely rule out the use of donors or embryos such as these. The 8th edition of the Code gives guidance to clinics about action they would be expected to take in these rare circumstances, including considering the welfare of any child who would be born, and making use of a clinical ethics committee.

### Preimplantation genetic screening

The 8th edition of the HFEA Code contains our updated requirements for centres to follow when using PGS. This includes providing information to patients about the risks of PGS and ensuring that the patients understand that it is an unproven treatment. In addition, clinics will be expected to monitor the latest literature and professional guidance on the use of PGS, and adjust their practice accordingly.

### The use of a senior clinical geneticist

We provide additional guidance in the 8th Code that a senior clinical geneticist should be involved in the decision-making process when deciding whether a patient should receive treatment involving genetic testing.

### Notifying donors about genetic conditions they are found to have

We have included additional guidance in the 8th edition of the Code which recommends that a centre seek a donor's consent prior to screening as to whether or not that donor would want to be informed of a genetic disease they are found to have as a result of that screening. Also, we recommend that the donor state as part of this consent whether or not they want their GP to be informed of a genetic disease they are found to have.

If you have any questions about the new licensing process for PGD, or about Code of Practice guidance around genetic testing, please get in touch. In addition, if you would like to be kept informed about the coming HFEA policy review of the licensing of tissue typing and low penetrance/late onset conditions, please email Danny Edwards at [danny.edwards@hfea.gov.uk](mailto:danny.edwards@hfea.gov.uk).



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