



## GIG Welcomes Two New Members of Staff



**Lauren Limb**  
- Research Assistant

I am very pleased to have joined GIG as Research Assistant. My role here will be to coordinate the Rare Disease UK Working Groups, and to research into their ideas to produce a draft National Strategy for Rare Disease. This will bring together all aspects of rare disease - prevention and diagnosis, best practice in treatment, and the dissemination of information on rare disease to the public, patients and professionals. I am looking forward to progressing this project and working to make a difference to the many people affected by rare conditions.

lauren@gig.org.uk and lauren@raredisease.org.uk



**Anna Allford, Research Assistant/Project Officer**  
- Familial Cancer Risk in Ethnic Minorities

I am delighted to be back at the Genetic Interest Group working on such an important project. GIG has produced groundbreaking reports previously around access to genetics services for minority ethnic communities (Darr, 1999) and ethnic monitoring (Mehta, 2003) and now in partnership with the

Division of Primary Care at the University of Nottingham this project will identify:

- Why there is a significant under-representation of cancer referrals to clinical genetics services amongst people from minority ethnic groups; and
- What strategies and approaches might address this.

Funded by a grant from the Big Lottery Fund Research Programme, the project will firstly identify why people from minority ethnic groups with a significant family history of cancer are less likely to access clinical genetics services than the mainstream UK population and, secondly will inform the development of interventions in order to improve access to genetics services for people from minority ethnic groups.

The project will focus on bowel, breast and prostate cancer. Based on the NICE Amsterdam Criteria, these are

recognised as the commonest cancers where preventative measures exist if familial risk is identified. We will use the experiences of three established minority ethnic communities in England (South Asian, Black Caribbean and White Irish) as exemplars to improve understanding of;

- Patient-related contexts and perceptions, and
- Service-related facilitating or hindering factors shaping access to genetic service care for common cancers in ethnically diverse contexts.

Two centres will collaborate with the research; the Medical Genetics Unit of St. George's University of London, and a Regional Genetics Service in the Midlands. This qualitative study will seek to discuss the research questions with clinicians and key service informants in addition to patients and their families who have accessed cancer genetics services. Crucially, we aim to also invite people from the community who have a history of one of these cancers in their family but have not been seen by genetic related healthcare professionals, to take part in the study.

This research has received full ethics approval and we are preparing to commence at the first centre where we have gained their Research and Development Committee (R & D) approval.



LOTTERY FUNDED

## New post of Child Development Officer for Alström Syndrome UK

This vital new post has been funded by BBC Children in Need and will support children who have a very rare genetic condition called Alström Syndrome. The condition is devastating leading to progressive blindness and deafness, heart and kidney failure, diabetes, liver dysfunction and associated problems.

The position is being shared on a job share basis; the North of England is covered by Kerry Leeson-Beevers and Catherine Lewis will be covering the South of England. They are both looking forward to this challenging and rewarding position.

If you need any additional information please contact Catherine Lewis on 01803 613117 or 07970071675 catherine.lewis@alstrom.org.uk



anna@gig.org.uk

## Spring Newsletter 2010



GIG and Rare Disease UK (RDUK) marked Rare Disease Day 2010 with receptions at the Scottish Parliament, the Welsh Assembly and, for the first time, the Northern Ireland Assembly. A wide range of stakeholders got together to share experiences, raise awareness and demonstrate the need for better services for rare diseases. Many attendees had participated in our contact campaign and took the opportunity to bring some of the issues affecting people with rare



Wilmut who summarised developments in research. Susan Green also gave a moving yet fascinating talk on her life as a parent of two children with Niemann-Pick Disease and their involvement in research. A Motion tabled by Dr Richard Simpson welcoming Rare Disease Day was signed by 36 MSPs.

Our event in Belfast on the 4th March was hosted by Lord Browne and Jim Wells MLA, the Chair of the Health



diseases to the attention of their elected representatives.

We kicked off at the Welsh Assembly on the 24th February. Around 80 attendees and 19 AMs (almost a third!) attended the event, hosted by the Plaid spokesperson for Health and Social Services, Helen Mary Jones AM. Speaking at the event was Alastair Kent, Chair of RDUK, and Chris Humphries who talked about her experiences as both a mum to a young man with a rare condition and as national coordinator for the Laurence Moon Bardet-Biedl Society. Professor Julian Sampson spoke about some exciting developments in research for families affected by rare diseases including his research into Tuberous Sclerosis. We were pleased to welcome Dr Tony Jewell, the Welsh Assembly Government's Chief Medical Officer, who described how NHS Wales had to work smarter with its resources and work alongside the whole of the UK so that families in Wales could access the right care and treatment.

On the 2nd March we held another successful reception at the Scottish Parliament which was attended by over 120 people and many MSPs. The event was hosted by Christine Grahame MSP, the convenor of the Health and Sport Committee, who remarked on the large turnout and support and her surprise on discovering how common rare diseases actually are. Alastair spoke again along with Professor Sir Ian



## The Focus of Rare Disease Day 2010 "Patients and Researchers - Partners for Life"

- The focus of Rare Disease Day 2010 was on the importance of research into rare diseases.
- There is no cure or treatment available for the vast majority of people with rare diseases. Without research this will remain the case. As a result, research is fundamental to improving the lives of people with rare diseases and their families.

The tagline "Patients and Researchers - Partners for Life" reflects that patient organisations can be valuable partners in research projects by raising funds, as an important source of practical knowledge and in recruiting the cohorts needed for clinical trials. Patient organisations can act as catalysts for collaboration between researchers, clinicians, industry and patients.

- Research and development for rare diseases is restricted by a number of factors including:
- A lack of mechanisms e.g. registries for recording patient information (and most patients with rare diseases are not recorded by the NHS)
- Small numbers and fragmented patients
- Difficulties in recruiting patients for clinical trials
- A lack of opportunity/cost perception by the pharmaceutical industry
- Difficulties in finding sustainable sources of funding

As a result, research into rare diseases requires collaboration including supranational collaboration, long-lasting infrastructure and sustainable sources of funding.



Committee. There was a great deal of interest in the event with around 120 people in attendance and many MLAs. Due to the importance of the event, the Health Committee postponed their meeting so that members could attend and meet their constituents, which as Mr Wells remarked, doesn't happen very often! Speaking alongside Alastair was Debbie Hoey, mother to Cavan, who has three rare conditions. Consultant in Medical Genetics, Dr Fiona

Stewart, spoke about the importance of research and examples of good practice in Northern Ireland and finally Eibhlin Mulroe, the Chief Executive of the Irish Platform for Patient Organisations, Science and Industry (IPPOPI) spoke about collaboration between the Republic and Northern Ireland and on a European level. Although the timing of the general election meant that we didn't hold a reception at Westminster this



year Dr Evan Harris MP kindly tabled an Early Day Motion welcoming Rare Disease Day. Many MPs have signed up (and still are at the time of writing) prompted by letters from GIG and RDUK members. We will be holding a reception at Westminster in the autumn to bring rare diseases to the attention of new MPs; more information to follow in due course and we hope many of you can attend.

We would like to thank everyone who helped make Rare Disease Day such a success: the hosts of the events, the speakers, all attendees, those who sent media case studies and all who participated in our contact campaigns - the turnout of elected representatives showed that your communications really do make a difference.

**For full event reports and to see the photos from all the events, please visit the Rare Disease UK website:**

[www.raredisease.org.uk](http://www.raredisease.org.uk)

#### About Rare Disease Day

Rare Disease Day takes place annually on the 29th February (i.e. a "rare day") When it is not a leap year it is marked on the 28th February.

This year was the 3rd Rare Disease Day. What began as a European initiative has now spread to become an international event.

#### Aims of Rare Disease Day

To raise awareness of rare diseases – rare diseases have generally not been considered a public health priority. This is partly due to the mistaken belief that rare diseases affect a small number of people; that there is little that can be done to help people with rare diseases or that what can be done would be unfeasibly expensive. Rare Disease Day is an opportunity dispel some of these misconceptions and highlight that there are over 6000 rare diseases affecting approximately 3.5 million people across the UK (1 in 17 people). Collectively rare diseases are not rare!

- To emphasise rare diseases as a public health priority
- To ensure that by acting simultaneously and collaboratively nationally and internationally the voice of rare disease patients is heard by more people
- To highlight the issues affecting people with rare diseases
- To bring all of the stakeholders involved in rare diseases together. This includes patients, families, carers, policy makers, healthcare providers, clinicians, researchers, health workers, industry and patient organisations.
- To coordinate policy actions for rare diseases at each level – in England, Scotland, Wales and Northern Ireland; across the UK; at a European level; and internationally.

## Research Study in VCFS in children VOLUNTEERS NEEDED FOR CHILDREN'S STUDY

VCFS is short for Velo-Cardio-Facial Syndrome. VCFS is a medical condition which is caused by the deletion of a small part of one of a person's two chromosomes 22. The deletion occurs on the longer arm (or Q arm) of the two arms of this chromosome 22. Another name for VCFS is therefore 22Q11 Deletion Syndrome (or 22Q11DS). VCFS is sometimes also referred to as DiGeorge syndrome, particularly when children have heart problems.

Children with VCFS can experience learning difficulties. They may also experience behavioural problems at home and in school and may find it more difficult to make friends. Sometimes, their thinking and speech may be difficult to follow or may seem unusual to others. It is not clear at the moment to what extent children with VCFS are at risk for these problems.

Researchers also don't know to what extent the genetic deletion plays a role in the behavioural and thought problems some children with VCFS experience. It is possible that the learning difficulties that children with VCFS can have contribute to their risk of behavioural and thought problems. However, if and to what extent they do is not understood.

Understanding more about the relations between learning, behavioural and thought problems in children with VCFS may lead to more effective methods of treatment and prevention, thus improving the lives of children and families affected by this disorder.

Our study aim is to better understand the developmental pathways of VCFS. We therefore are interested in assessing the behaviour, and the psychological and intellectual functioning of children with VCFS.

The study has ethical approval and is conducted by University Hospital Cardiff. Dr. Marianne van den Bree is the principal investigator.

#### Participation involves:

- Assessment of the child's symptoms via clinical interview with the parent.
- Interview with the child about how they experience themselves and others.
- Assessment of the child's cognitive ability. This involves completing a number of puzzles.
- Blood or saliva sample from the parents and the child for DNA extraction

Participation in this project is entirely voluntary. All participating families will be reimbursed for their time. All information gained from the study will be confidential. You are free to withdraw from the study at any stage.

If participating families like to, we can provide their clinician with the information gathered in the study and this may be helpful for treatment. We hope that the project will provide greater insights into the development of 22Q11DS and this may in turn contribute to the development of new treatment strategies in the future.

#### Your family may be eligible for the study if:

- You have a child who is 7-11 years who has been diagnosed with VCFS.
- If your child has a sibling who does not have VCFS we would also ask them to take part. However, families of children with VCFS who do not have a sibling can still take part.

#### For more information please contact:

Dr Marianne van den Bree, Tel: 029 2074 4002

E-mail: [echo@cardiff.ac.uk](mailto:echo@cardiff.ac.uk)

Cardiff University Hospital, Department of Psychological Medicine, School of Medicine, Cardiff University, Heath Park, Cardiff, CF14 4XN.

The National Human Genome Research Institute at the National Institutes of Health (NIH) is sponsoring a study that seeks to learn more about how parents of children with an undiagnosed medical condition think and feel about their child's condition. We hope that this knowledge will improve the health care and counseling for these parents. Men and women who are

18 years or older and have at least one child with a medical condition that has remained undiagnosed for more than 2 years are needed to take part in this study. Participation involves one survey that takes about 45 minutes to finish. The survey can be taken online or a paper copy can be mailed to you. For additional information about this study, you may review the Notice to Participants disclosure. [www.surveymonkey.com/StudyNoticeUncertainty](http://www.surveymonkey.com/StudyNoticeUncertainty)

#### If you have questions, please contact:

Anne C. Madeo, MS, Principal Investigator Genetic Counselor National Human Research Institute National Institutes of Health Bldg. 31, Room B1B36 | 31 Center Drive, MSC 2073 | Bethesda, MD 20892-2073

Phone: 301-443-2635 | Email: [anne.madeo@nih.gov](mailto:anne.madeo@nih.gov)

## Healthcare delivery: Commissioning or Planning?

Much of GIG's policy work is aimed at bringing treatments to patients. This is why we put a lot of effort into supporting research and ensuring it can go ahead in a permissive and well-regulated environment; our view is that research will be the source of solutions for most currently unmet health needs. GIG looks at the whole road, from research to treatment development, to clinical trials and regulation, to healthcare technology assessment, and to the delivery of treatments for use in the NHS.

Commissioning is the process by which treatments, medicines and services are purchased for provision in the NHS England (the Purchaser-Provider split). In Wales, Scotland and Northern Ireland, the term is not used. In this brief article, I wish to cover some of the current issues occurring in the commissioning and planning of healthcare delivery in England and Wales now; in a future article I will discuss the planning of healthcare delivery in Northern Ireland and Scotland.

GIG's member groups as a whole have a strong interest in the existence of a commissioning framework which is capable of planning and funding many differing types of care such as: complex therapies for which there will be very few patients; multi-disciplinary care from various specialities for patients with multifactorial conditions; and good quality local and convenient care provision for chronic conditions. Good quality healthcare requires good quality commissioning and GIG looks to inform the commissioning process, so it is best place to commission appropriate services for the patients our member groups represent.

### Strengthening National Commissioning

National specialised commissioning is the method by which just over fifty specialised services for rare disease are paid for and provided by the NHS in England. The annual spend is around £0.5 billion. The logic behind providing specialised services in this way are a) that the best quality of care can be provided by experts in their field working in specially equipped centres, b) that this model is most cost effective, and c) that Primary Care Trusts' budgets aren't affected in an unpredictable manner by peculiarities in incidence of rare disease in their catchment area, and that this doesn't lead to post-code prescribing.

The Department of Health recently consulted upon proposals to clarify the structure of and relationship between the two bodies responsible for commissioning services on a national level, which were, and as we go to press still are, rather confusingly called the National Specialised Commissioning Group (NSCG) and the National Commissioning Group (NCG). The proposed changes were to make NCG the sole body responsible for advising ministers on commissioning services to ensure that there is one consistent message to ministers, and to change its composition

to represent all stakeholders.

Regional specialised commissioning is the process in which groups of PCTs gather to form a Specialised Commissioning Group (SCG), (these cover the same area as Strategic Healthcare Authorities) to commission services that, due to the small populations that require them, are best commissioned for populations of more than one million people. SCGs are supposed to commission specialised services based on the Specialised Services National Definition Set (published by NSCG). Commissioning of these specialised services is very patchy.

It is GIG's view that at the time of the consultation, arrangements for national commissioning were functioning much better than the arrangements for regional specialised commissioning.

Both the consultation document and GIG's response to the consultation can be found on our website in the current campaigns section of the policy area. GIG's response praised the intent of these proposals, but at the same time we took the opportunity to call for more far-reaching changes that would address more of the problems surrounding both national commissioning and regional specialised commissioning.

#### We called for:

- A budget for national commissioning which increases at the rate of NHS growth.
- An empowered NSCG, able to hold SCGs to account on their responsibilities to commission services from the Specialised Services National Definition Set; and to ensure coordination between SCGs.
- NSCG to be responsible for ensuring there is coordination between providers of specialised care and primary care.
- NSCG and/or NCG to be proactive in seeking services that would be best commissioned on a national or regional basis, and to assist the providers of such services to achieve such a status.

The Department of Health's response to the consultation responses is on our website also. We were broadly satisfied with the response, although it focused narrowly on changes to NCG and did not address any of our concerns regarding NSCG or regional commissioning.

### House of Commons Health Committee Report on Commissioning

As the deadline for this article looms, the House of Commons Health Committee has published a report (here: <http://bit.ly/dqTyD3>) following its inquiry into commissioning. I have been unable to examine it fully for this article, but a number of interesting points are apparent from a first look.

- The Committee estimates that as much as 14% of NHS spend goes on the process of commissioning, a tool originally intended to improve efficiency.
- Many PCTs are poorly equipped to fully understand the health care needs of their catchment area and properly commission local services for them.
- The Committee recognises that there is a lot of regional variation in regional specialised commissioning, and that PCTs are "disengaged" from specialised commissioning.
- The Committee calls for commissioners to be properly trained and properly equipped to analyse data and better commission services.
- The Committee stated that "If reliable figures for the costs of commissioning prove that it is uneconomic and if it does not begin to improve soon, after 20 years of costly failure, the purchaser/provider split may need to be abolished."

As GIG stated in our response to the inquiry (available on our website), the purchaser/provider split already has been replaced in both Scotland and in Wales, and GIG believes England should learn from the results of these changes. GIG will post our response to this report on our website in due course.

### Wales Health Specialised Services Committee

The purchaser/provider split in Wales no longer exists. In Wales, healthcare is planned and delivered by seven Local Health Boards (LHBs), statutory bodies responsible for the planning and delivery of healthcare to their resident population. This new process was launched on 1st October 2009.

From the 1st April 2010, specialised services in Wales will be planned by the Wales Health Specialised Services Committee (WHSSC pronounced "whisk"), on which the seven chief executives of the LHBs sit. This committee is responsible for deciding which services are planned and paid for on a national level, and which services must be planned and paid for individually by LHBs.

As I write this, WHSSC has not had its first meeting. However GIG does have some concerns regarding this structure. One of the weaknesses of the regional commissioning structure in England has been the tendency of PCTs to try to keep as much money for themselves, and not to recognise the benefits of sharing the costs of best serving their patients. GIG hopes that LHBs and WHSSC will not fall into this trap. Buddug Williams, our Wales Development Officer looks forward to working with LHBs and

www.gig  
.org.uk

#### WHSSC

to ensure this doesn't happen.

GIG will continue to monitor arrangements for the delivery of healthcare services to patients across the UK, and to call for the changes that we believe are necessary for better services for the patients our members represent. We would be very happy to hear from you if you have any comments or contributions to make. Please contact [nick@gig.org.uk](mailto:nick@gig.org.uk)

GIG will continue to monitor arrangements for the delivery of healthcare services to patients across the UK, and to call for the changes that we believe are necessary for better services for the patients our members represent. We would be very happy to hear from you if you have any comments or contributions to make. Please contact [nick@gig.org.uk](mailto:nick@gig.org.uk)

By Nick Meade, Policy Analyst, [nick@gig.org.uk](mailto:nick@gig.org.uk)

## From Chrysalis to Committee - the story of a Self Help Support Group

Many support groups for health conditions are formed by individuals who are affected by the condition because they or a family member have the condition themselves. The FSH-MD-UK Support Group is such a group, formed twenty five years ago by a couple whose teenage daughter was newly diagnosed with FSH, a form of muscular dystrophy. For many years this couple took on almost all the responsibility for this which included:-

- Talking to new members over the phone
- Organising regular events for members
- Developing leaflets and later a website with information for people who were newly diagnosed
- Maintaining a membership database
- Producing a newsletter

From an early stage the group had a chairperson who had FSH but otherwise most of the work of running the group rested on two people, with no formal structures or written constitution. While it was a successful group it was not sustainable and there were times when it was very difficult for them to have the time to keep the group running.

Over the last three years the group has changed to formal structure, involving more than twenty members and spreading the responsibility and workload much more widely. It has done this by

- Developing a Memorandum of Understanding with the Muscular Dystrophy Campaign (MDC) who now provide support to the group
- Identifying the tasks that need to be done, breaking them down to almost twenty different roles
- Creating a Committee to oversee the running of the group
- Developing and agreeing Aims and Objectives and Terms of Reference

And what happened to the couple who founded the group twenty five years ago? We invited them to become Honorary Life Presidents and are delighted that they have accepted so we'll have a formal lunch for them this year, which can become another social event to support people with FSH.

### What made it all work? For any group on this journey there are a number of things to bear in mind:

- People are more likely to volunteer for tasks and roles when they are made clear - it's much easier to respond to a request to edit the newsletter than a general request for help.
- Be prepared to split or rearrange roles and responsibilities if that helps people to volunteer, rather than having a rigid structure. For example, the FSH Support Group split the secretary's role into a correspondence secretary and minutes secretary to suit the skills and commitments of the individuals concerned.
- People who have been involved in other organisations bring useful knowledge and understanding of how groups work. When the group first forms - having individuals who are used to chairing meetings, taking minutes, reporting on

finances and resolving conflict really helps the group in starting up.

- Focus on people's experience apart from their condition. Self-help groups support people in their role as a person 'with a condition', but most people have lots of other roles and talents which they bring to the table.
- Understand that some people they don't want to be too involved with the group because to do so would mean that being a 'patient or service user' would be a greater part of their life than they'd like.
- People with specific health conditions are in this for the long haul but may choose to move in and out of the group according to changing personal circumstances. Enable them to take on a role for a while and then drop out - that way they may come back later. If it's too difficult to step down some people will never offer to step up.
- Change within the group is healthy as it brings new ideas, skills and experiences. Many groups limit the amount of time an individual can spend in any one role to help create that turnover.
- Accept there will be some conflict - groups go through processes as they develop and 'storming' is part of this. Disagreements show that people care about what is being discussed. Differences are good and can usually eventually be resolved - so long as they don't get personal.
- Spread the work around. If there are fifteen roles, fifteen people each carrying out one role will be much more sustainable than one or two people trying to do everything.
- Accept that other people will do things differently to the way you would- and most of the time it doesn't matter
- A larger committee is harder to manage but a more diverse committee will mean that you will develop more ideas and creative ways of working- which will broaden your appeal to a wider membership.
- Always acknowledge what people have done - they will continue to contribute if they feel valued.
- Be careful not to misrepresent individuals or what they have said or done - it often antagonises them.
- When a new role or task appears, think 'who would like to be asked to take on this role?' rather than 'how can I fit this in along with everything else?'

**Sheila Hawkins**

**Former chair of FSH Support Group UK**  
<http://www.fsh-group.org/>

## Announcement of New Support Group

David and Samantha McDowell lost their daughter Jessica in August 2009 (aged 11) to Fanconi Anaemia (FA) and they also have a 2-year-old son Alexander with the condition. As a result of a lack of Family Support in the UK, they decided to set up "Fanconi Anaemia Family Support" (FAFS) so other Families would not suffer alone, as they had. This group actually started in January 2010 and on 18 March 2010, it received its official registered Charity status (No. 1135020); it is the only FA Family Support group in the UK or Ireland. The group is BY parents, WITH parents, FOR parents and its main focus is Family Support but if fundraising is successful and the funds exist, they plan to drive relevant research. They have close connections and great relationships with a number of International FA organisations; primarily FARF in the USA and the FA Support Groups in Germany, France and Spain, with contacts now being made with other FA groups around the world.

Although FA is a rare genetic condition, it has serious implications for the wider population. The Fanconi Pathway is the means by which the human body repairs and maintains healthy DNA, and it is also responsible for resistance to Chemotherapy. The ability to understand and manipulate the Fanconi Pathway will not only improve Cancer treatments but also the ability to make early detections and improve prognoses, ultimately saving lives. With research that has already taken place and recent developments, FA researchers are pioneering new methods of detection and treatment for a number of different Cancers. Research into this "orphan" disease is now beneficial to all of us.

On Saturday 20th March 2010, at the Holiday Inn in York, FAFS held its first Family Support Meeting. A total of 14 Families attended - the make up of these Families was - 22 "parents", 4 young adults, 9 teens and 19 children under the age of 11. Of the people actually at the meeting, there were a total of 9 people with FA (aged 1, 2, 4, 8, 13, 13, 16, 20 and 25). It was a lovely opportunity for Families to meet each other and chat, discussing a range of issues relating to FA and providing mutual support. Even though there had been

an opportunity in the morning for Families to introduce themselves and talk freely, it was lovely to see Families and professionals mingling and chatting over lunch. Plans are underway for support networks for Families and future meetings.

The organisation is planning a number of fundraising events over the coming months and these will be added to the website. The Trustees welcome help and support from any quarter and are happy to discuss joint initiatives / corroborative working.

Contact details are available on the website at

<http://www.fanconisupport.info>  
or by calling the Chairman (David McDowell) on 07939 593993. If any FA-affected Families have not already been in contact, they are encouraged to do so. Donations can either be made online or buy sending a cheque.



Registered Charity No. 1135020

[www.fanconisupport.info](http://www.fanconisupport.info)



## Pronuclear transfer may give families affected by mitochondrial disease a choice

By Dr Kristina Mills and Dr Marita Pohlschmidt  
Research Communications Officer  
and Director of Research, Muscular Dystrophy  
Campaign, UK

Research at Newcastle University funded by the Muscular Dystrophy Campaign has shown that it might be possible to prevent mitochondrial diseases being passed from mother to child. These often devastating conditions, which affect 3, 500 people in the UK, can affect almost every part of the body, with symptoms including muscle weakness and pain, loss of sight and hearing, learning difficulties, seizures, diabetes, heart, liver and kidney disease and breathing difficulties. In the most severe cases it leads to death in early childhood.

Mitochondrial diseases are caused by damaged DNA in the mitochondria which are the 'batteries' of the cell and supply it with energy. Every cell contains thousands of mitochondria and each carries a small piece of DNA which contains 13 protein coding genes. This is compared to 23, 000 genes in the chromosomal DNA located in the nucleus of the cell. Mitochondria are passed down the maternal line, from a mother's egg to her child.

The research led by Prof. Doug

Turnbull (in collaboration with Dr Mary Herbert and Prof. Alison Murdoch) showed for the first time that it is possible to transfer the nuclei containing the chromosomal DNA of a human embryo, into a donor egg. The donor egg previously had its own nuclear DNA removed. The human embryos manipulated in this way had minimal amounts of mitochondria transferred to the donor egg and encouragingly a proportion of them successfully developed in the laboratory for 6-8 days. The success rate of embryo development to the blastocyst stage was rather low at 8.3%, but the embryos used were those donated to research because they had fertilised abnormally and could not be used for IVF. The researchers expect higher success rates with normal embryos, but this requires further research.

There is currently no effective treatment available for mitochondrial diseases and due to the way that these conditions are inherited and their unpredictable nature, genetic counselling and prenatal testing are extremely difficult. If this technique, known as 'pronuclear transfer', were to come into clinical use it would give families the option of having healthy children. The abnormal mitochondria would be exchanged for the healthy mitochondria of a donated egg. Less than 0.1% of the resulting baby's DNA would come from the donated egg, and these genes would only be involved in the production of energy via the mitochondria, and not any other characteristics. This is rather like replacing the batteries in a laptop; the laptop itself is unchanged and all of the information on the hard drive remains the same but the new batteries allow it to work properly.

Recent research in the US has also given strength to this approach- it was shown that a similar technique tested on monkeys gave rise to apparently healthy offspring. The current Human

Fertility and Embryology (HFE) Act does not allow embryos modified in this way to be implanted into the mother. If this legislation were to be changed, the leading scientists are confident that it could be offered to families with mitochondrial disease within the next three years. Before this can happen, further studies to demonstrate that this technique is safe and efficient are required and Prof. Doug Turnbull and Prof. Alison Murdoch are currently liaising with the HFE Authority to discuss the details of the next steps. The case can then be made to the Secretary of State who, since the amendment to the HFE Act in 2009, has the power to make provisions for this to technique to be introduced into clinical practice.

Pronuclear transfer has been developed specifically for mitochondrial diseases and is not applicable to any other types of genetic conditions. It is an option that the families affected by these potentially devastating conditions have been so desperately hoping for, because it means that it would break the chain of inheritance within a family line. The technique can not stray into the realm of designer babies, as some may fear, because it can not be used to alter the genes that are located in the chromosomes. Instead it gives some parents the choice to have healthy children and also improves the quality and length of life of future generations.

The Muscular Dystrophy Campaign urges the government to carefully consider this new technology and take a positive view as it will address the needs and rights of families affected by mitochondrial disease to have healthy children.

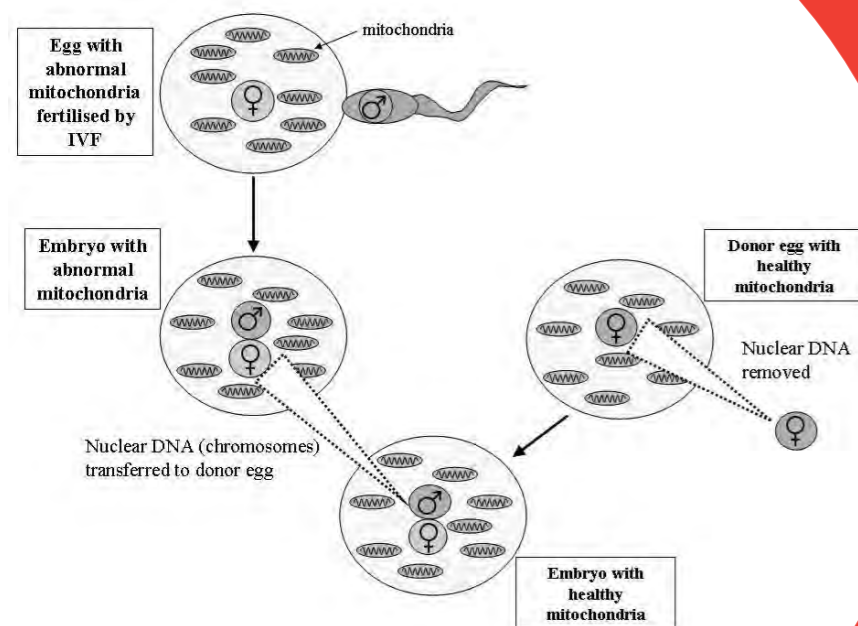
### Reference

Craven L, Tuppen HA, Greggains GD, Harbottle SJ, Murphy JL, Cree LM, Murdoch AP, Chinnery PF, Taylor RW, Lightowlers RN, Herbert M, Turnbull DM. Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. *Nature*. 2010 Apr 14. <http://www.nature.com/nature/journal/vaop/ncurrent/full/nature08958.html>

### Further information

<http://www.muscular-dystrophy.org/research/news/2194>

This article has been reproduced with kind permission of Bionews [www.bionews.org.uk](http://www.bionews.org.uk)



## Genetics Gets Creative - CGD Research Trust Get Together

Six months ago at the Chronic Granulomatous Disorder Research Trust (CGDRT) Get-Together weekend in Derby, the main conference focussed on the medical and practical aspects of living with CGD. There were presentations on the CGDRT paediatric nurse service, diagnostics, lung problems in CGD and establishing a good relationship with GP surgeries, as well as the latest news in CGD research, the work of the charity and fundraising. Other parallel sessions included a guide to effective communication with medical professionals by the CGDRT clinical psychology support team and individual consultation sessions. Combined with the opportunity to relax and socialise with a visit to Alton Towers and over meals, the programme offered much in depth information but genetics was about to get creative! Previously the CGDRT offered the children and young adults entertainment. In response to requests, the charity asked Julia Thomas to devise a programme that was enjoyable but more productive. The charity and the participants were delighted with the outcome. Whilst the adults were discussing CGD in the main conference room, the children and young adults were having some fun of their own.

Saturday began with visual art using lots of hands-on practical activities which centred around aspects of genetics. I wanted to incorporate both art and genetics, aiming to deliver a creative and fun session that also educated. (Julia Thomas, visual artist)

Some of the activities concentrated on physical attributes and the fact that 'Everyone's Different', exploring the idea of heterogeneity. Other activities focussed on 'Taking a Closer Look' within our bodies using photo-mosaics as well as marble painting in order to make their own coloured chromosomes.

Sunday's 'Movement & Balance' workshop, led by dancer Anushiye Yarnell, aimed to build up confidence about the body. By controlling and choreographing their movements, the participants developed an appreciation of their own strengths and limitations through exploring movement and

balance within their routine. This workshop was a chance for the participants to engage in a non-competitive physical activity celebrating their individuality.

When I first spoke to Rosemarie about CGD itself and how it affected people, it struck me that balance was a key feature, wanting to lead as full a life as possible whilst weighing up the health risks of taking part in certain activities. I'd worked with Anushiye on a previous project and was impressed at how she incorporates themes into the routines within her workshops. I felt that complementing the genetics education in my visual art workshop with a physical activity, based around movement and balance, would encourage participants to interact with and support each other as well as to help build their self-confidence. (Julia Thomas, visual artist)

After the Movement & Balance workshop had finished and the adults were coming out of the conference room, we displayed the artwork the children had been doing and the boys then gave a demonstration of the routine they had been working on. It was amazing to see they had become confident enough to perform in front of an audience and obviously proud to show off what they had created over the weekend.

The CGD Research Trust is grateful to the Jeffrey Modell Foundation for sponsoring the Creative Workshops.

[Julia Thomas is an artist with a bioscience background, having studied and worked in genetics & statistics. She is currently working on projects with the Wales Gene Park and CESAGEN (Centre for Economic and Social Aspects of Genomics) as well as studying part-time on the MA in Arts & Health at Glamorgan University.]

### Contact Details

**Julia Thomas | Email: [julia.m.thomas@talktalk.net](mailto:julia.m.thomas@talktalk.net)**

**Mob: 07711 269101**

**Rosemarie Rymer | Email: [rosemarie@cgdrtr.co.uk](mailto:rosemarie@cgdrtr.co.uk)**

**Tel: 01725 517977**



### Report 10th Partnering for Rare Diseases Therapy Development Workshop

It is my pleasure to inform you that the report of our 10th Partnering for Rare Diseases Therapy Development workshop which took place in Brussels in 2009 is now available on our website. For a direct access please click on the following link: [http://eposi.org/web/publications/\\_docs/publications/Documents.aspx](http://eposi.org/web/publications/_docs/publications/Documents.aspx)

Of course you will receive a printed copy in the coming days but do not hesitate to ask me additional copies for dissemination!

For more information please contact [jonathan.ventura@eposi.org](mailto:jonathan.ventura@eposi.org) or visit the website on [www.eposi.org](http://www.eposi.org)

## The 7th international 22Q11.2 deletion meeting and the 10th annual max appeal conference

Ricoh Arena, Coventry 29th - 31st July 2010

You are cordially invited to this landmark event, taking place over three days as "back to back" meetings for parents and professionals. The scientific programme will run on the 29th and 30th July, with the family programme being run on the 30th and 31st July.

A common social function on the second evening will bring all together for a terrific time. The venue is the spectacular Ricoh Arena, home of the Coventry City Football Club, close to the historic Warwick Castle and the Royal Shakespeare Company where some of the children's activities are planned to take place.

### "One Step Beyond"

Families are warmly invited to attend this meeting, we will be able to make good use of the national and international experts to bring you new and exciting information. In addition there will be opportunities to explore common issues in smaller group sessions with professionals and other parents who 'have the t-shirt'. We know that Max Appeal families have become accustomed to a high standard at our annual event but this year plan to exceed your expectations!

The children's and young people's programme is exceptional! Visit Warwick Castle and the Royal Shakespeare Theatre, enjoy outdoor activities such as high wires and climbing wall in an outward bound trip. Create video clips and music CD to take away with you.

### "Treatment As We Move Into A New Decade"

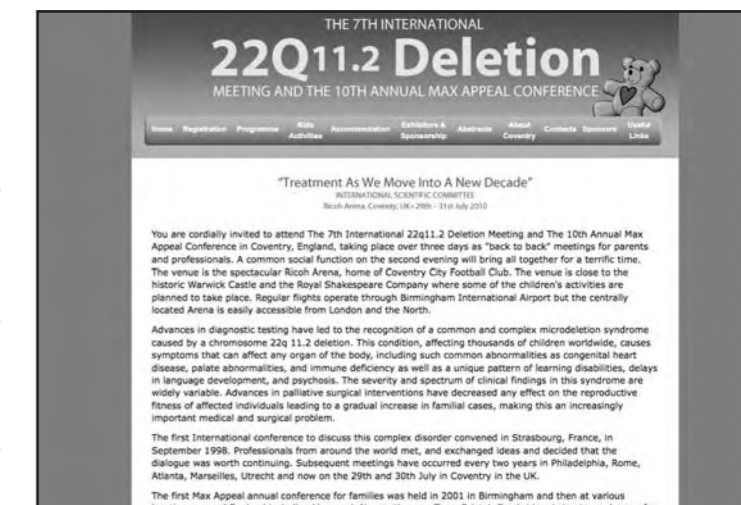
Professionals interested in presenting at this meeting being held on the 9th and 30th July 2010 are invited to submit an abstract at [www.22qdeletion.com](http://www.22qdeletion.com) by the 22nd February 2010. All those seeking information about the 22q11.2 deletion are encouraged to attend!!

The first International conference to discuss this complex disorder convened in Strasbourg, France, in September 1998. Professionals from around the world met, and exchanged ideas and decided that the dialogue was worth continuing. Subsequent meetings have occurred every two years in Philadelphia, Rome, Atlanta, Marseilles, Utrecht and now Coventry.

This conference is targeted to all health care professionals and basic science researchers, including physicians (both general and specialists), scientists, genetic counsellors, psychologists, speech therapists, audiologists, educators, nurses, and other health care professionals.

The scientific committee: Donna McDonald-McGinn, MS, CGC, Peter Scambler, MD, PhD, Anne Bassett, MD, Bernice Morrow, PhD, Bruno Marino, MD, Solveig Oskarsdottir, MD, PhD, Nicole Philip, MD, Kathleen Sullivan, MD, PhD, Ann Swillen, PhD, Jacob Vorstman, MD, PhD

To attend this conference please click onto this website [www.22qdeletion.com](http://www.22qdeletion.com) or alternatively contact the secretariat is Tarquin Scadding-Hunt of Maximize Group, Virginia House, High Street, Partridge Green, Nr Horsham, West Sussex, RH13 8HX Main: +44(0) 1403 711301 Direct: +44(0)1403 711366 Mobile: +44(0)7866 319222 Fax: +44(0) 1403 710058 Email: [tarquin.scadding-hunt@maximizegroup.com](mailto:tarquin.scadding-hunt@maximizegroup.com)



Get your local support groups and services listed on NHS choices

**NHS**  
**choices**  
[www.nhs.uk](http://www.nhs.uk)

NHS choices is expanding its popular directory of services to include local support groups and services for people living with genetic disorders.

Some 2 million people use the directory each month, so including details of your local support groups will significantly increase public awareness of your organisation and services.

To find out more, please email **Tamsin Ford, Directories Development Manager at NHS Choices** for call 020 7 972 6109 [tamsin.ford@dh.gsi.gov.uk](mailto:tamsin.ford@dh.gsi.gov.uk)