

gigtoday

winter 2005 / 2006

Welcome to 2006 !

Welcome to the first edition of GIG Today for 2006. By the time this reaches you I expect that the Christmas break will be but a distant memory, our post-Christmas waist lines will have receded to their more usual dimensions and we will be stuck in to the issues the New Year will bring.

Sadly, just as we were going off for Christmas the scientific world was rocked by the revelation that the ground breaking work on stem cells and cloning which seemed to offer hope of significant progress in many currently intractable disorders had in fact been faked by the South Korean Professor who published it. While this is a set-back, the fact that it came to light is a reflection of the robustness of appraisal processes such as peer review. This gives confidence in the basic soundness of the system, and of its ability to detect "rotten apples". We should not allow this scandal to obscure the progress that has been made, and the important contribution that the study of embryonic stem cells has already made to our understanding of a large number of serious genetic diseases, nor our appreciation of the lead that the UK has taken in creating a regulatory framework that allows British based scientists to be at the forefront of developments in this important field.

A strong message about the importance of all forms of stem cell research, whether from embryonic, fetal or cord blood on adult sources and of the need to provide proper funding to allow it to proceed smoothly, was sent by a conference attended by over 400 + delegates (over half of whom were patient representatives) at the European Commission in Brussels to Europe's Science policy makers. Votes were taken using inter-active polling systems on issues raised throughout the conference, and each time the overwhelming majority was in favour of research and of pushing ahead to the development of therapies if and when this becomes possible.

Closer to home, the consultation by the Human Fertilisation and Embryology Authority about using PGD (pre-implantation genetic diagnosis) for late onset inherited cancers concluded on January 16th. GIGs response to this is on our website (www.gig.org.uk). In line with previous policy, and also with the expressed views of our members on PGD, we supported the extension of PGD to cover these later onset conditions for those who might wish to use them to avoid the risk of familial cancers in their sons and daughters (and, of course, in subsequent offspring).

New policies and programmes in the Department of Health mean that the five Genetic Knowledge Parks (GKPs) in England will need to look to new sources for continuation of their funding when their current grant runs out in 2007. GIG will be collaborating with colleagues in the GKPs to try and make sure that the programmes we are jointly engaged on continue. We are also starting a new project using funding from the Jeans for Genes appeal and Genzyme. More details on this will follow in the Spring newsletter.

Few things in life are certain apart from (so someone once said) death and taxes. Indeed some of the challenges that GIG faces arise from the fact that we work in an area that is constantly changing. Over the years I have been fortunate to work with an exceptionally talented group of colleagues, all of whom have contributed to GIG's development in their own way. None more so than John Gillott, our Policy Officer for the last 11 years. We have relied greatly on his insight, his analytical skills and the intellectual firepower he has been able to bring to bear on complex and sensitive issues. Now he is moving on to a new phase in his life, writing and contributing to public understanding of science. We will not lose touch with John, but we will miss him. We wish him well!

Alastair Kent.

Farewell but not Goodbye.

I remember, in those distant days when I had a full set of hair (well, nearly), turning up on my first day at GIG to be greeted by Alastair in a cosy two-room unit in Farringdon. GIG has grown and moved on since then—to an even more cosy two room setup in Islington / Canonbury / Hackney (depending on who's interested). Much has changed over the past eleven years, most notably perhaps GIG's higher profile with Government, quasi-governmental agencies and of course not forgetting European institutions. Much of that, in some cases pretty much all of that, is down to Alastair's hard work.

But in addition to office geography some other things have stayed the same. In my time at GIG I had the good fortune to work for and with Joanie, then Maggie, and throughout the whole period Alastair, with all of whom I was always able to have lively, informed and frank conversations, a crucial aspect of the kind of work I did. More broadly, GIG has always skilfully combined formal and informal mechanisms in its working, a practice that, as it grows, it should try to maintain in my opinion.

Reflecting on some of the issues I have worked on over the years, it is clear that there is also much continuity there as well. The search for effective therapies has been shown to be a long and hard one; whether it is gene therapy or more recently stem cell therapies. A group like GIG has to show

enthusiasm for the work without getting sucked into talking up or exaggerating new developments, something it is sometimes under pressure to do. In the absence of effective therapies, selection prior to birth has continued to be an important and slowly expanding service for GIG's members; and a staple of GIG press and policy work, especially recently, accentuated by the various reviews of the Human Fertilisation and Embryology Act. A third continuity is GIG's interest in research policy. There have been many components to this interest. A specific issue that sticks in the mind is the difficulty thrown up by the familial character of genetics given trends in regulatory policy. The most recent example of this is that under the terms of the Human Tissue Act an individual has an absolute veto on the analysis of tissue for the benefit of a relative in a clinical context. GIG has always highlighted the familial character of genetic information, and has put the effort in to working through and proposing solutions to the difficult cases this can give rise to.

It is with some anxiety that I am leaving GIG, but it feels like the right time for a change. It has been a pleasure to work with the range of informed and committed people that constitute GIG and its wider circle of friends and contacts. I hope to stay in touch and in discussion in the years to come.

John Gillott

 Can you help?

Would you like to

 be interviewed?

My name is Monica Bonaccorso. I am a social anthropologist working at the University of Cambridge, in the Department of Social Anthropology, on a Wellcome Trust funded project (2002-2006). The study explores the cultural idioms, images and representations used by media professionals, interest groups and science exhibitions and experts to articulate ideas and views in favour and/or against the most controversial genetic technologies. The project focuses more specifically on views of therapeutic/reproductive cloning, stem cell research and xenotransplantation.

I would be very interested in hearing from patient and families affected by genetic disorders for their thoughts

and opinions on these areas.

The interview would be very informal and last about an hour. I am happy to travel to where you live and meet at a convenient time, alternatively I can do interviews over the phone.

If you would like further details about the project or are interested in speaking to me then please do contact me at the details below.

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What would you like your health care professionals to know about genetics?

This was the question I posed at my presentation to the SADS UK Patient Support Seminar on Saturday 8th October 2005. I had travelled to Watford on a bright and sunny autumnal day for what promised to be an interesting and very worthwhile discussion with members of the charity, and I was richly rewarded with a warm welcome and much enthusiasm.

The Ashley Jolly Sad Trust, SADS UK, is a voluntary organisation that exists to help prevent the early loss of life by raising awareness about heart conditions that can cause a Sudden Adult/Arrhythmia Death (SAD) and to improve the lives of those adversely impacted by cardiac arrhythmia. The Trust promotes heart monitoring and donates heart-monitoring equipment to doctors surgeries for this purpose. SADS UK works to initiate Government legislation to safeguard those who may be at risk from a fatal cardiac arrhythmia and raises awareness about the possible hereditary nature of cardiac conditions.

Professor Richard Sutton gave the first presentation, which



included an overview of his work as an NHS Consultant Cardiologist and his involvement with the development of Pacemakers. He continues to research in the field of vasovagal syncope and now his interest has spread to cardiac resynchronization. Needless to say I felt his was a hard act to follow so I took the opportunity to ask everyone to stand up and have a stretch, smile at the person next to them and hopefully forget what a fantastic Speaker they had just had!

Laughing and relaxed the audience took their seats again and I introduced myself and my role as Development Officer for Patient and Public Involvement on behalf of the NHS National Genetics Education and Development Centre, and the West Midlands Regional Clinical Genetics Unit. I outlined the work of the new Centre and described how we want the involvement of patients to inform the development of education and training in genetics for NHS staff. In developing our learning support materials we aim to use first hand accounts of patients to demonstrate the clinical utility of genetics and to tailor them to the needs of the different health care groups.

The current experience of patients, and their views on the knowledge, skills and attitudes that health care professionals need for a positive patient experience will help focus genetics education appropriately. Therefore we need to gain an understanding of what patients

want their health professionals to know about genetics.

Activities where patients with genetic disorders, their families and carers, may wish to help include;

- Advising on what you expect health professionals to know about genetics,
- Recounting the usefulness (or otherwise!) of genetic information,
- Sharing experiences,
- Reviewing learning resources,
- Reviewing our website.

The SADS Patient Support Seminar also included discussions on aspects of Insurance and the new National Service Framework Chapter 8 Arrhythmia and Sudden Cardiac Death. It was clearly a very successful and enjoyable day, and I would like to thank Anne Jolly and SADS for inviting me.

If you would like further information on how your views, opinions and experiences will help the Centre to develop genetics education in the NHS please contact me, **Anna Lane**, on **0121 623 6905** or email **anna.lane@bwhct.nhs.uk**



Treacher Collins Syndrome and Support Group

Treacher Collins Syndrome is an autosomal dominant genetic condition that causes facial malformations and severe hearing loss.

Someone with Treacher Collins Syndrome may have malformed cheekbones, chin, nose, jaw and temples. Eyelids are often drooping, seeming not to support the eyes, and there may be a small nick in the lower lid. The ears may be malformed or completely absent causing a hearing loss. Hairline and palate may also be unusual.

The features of TCS are, however, variable so that whilst some people may be severely affected others can be so mildly affected that it is very difficult to say whether they have the condition or not. A child born with a severe form of the syndrome could also have choanal atresia and such a receded lower jaw that they require a tracheotomy. Jaw distraction surgery at a later date often improves this situation.

Although the gene that causes the syndrome was detected a few years ago it is still not easy to detect if you have the effected gene because of its complexity. The gene does not break in the same place for every family. It is easier if there are several effected family members to map the break, but if it is a fresh mutation then it can take a considerable time to detect.

The TC Family Support Group was set up in 1987 by a group of parents of TC children and TC adults.

The first priority was to find as much information as we could about the syndrome as there was very little written and very little awareness. In 2003 we were fortunate to be awarded a lottery grant to employ a project worker for the group and have been able to produce a pack of information, had awareness campaigns and increased our

membership. We also produce a regular newsletter and have at least one get together every year, usually a family weekend or a conference, to give opportunities for people and children with TC to meet each other.

Meeting someone else who has the same condition is so valuable. TC people are often very isolated due to the rarity of the condition, so to meet and chat with someone who goes through the same things as you do every day is invaluable support. There have been a few medical advancements which have benefited people with Treacher Collins Syndrome and our group has been involved in these developments, namely the Bone Anchored Hearing Aid and the research into locating the gene. Treacher Collins Syndrome presents as 1 in 50,000, however because you can have the syndrome very mildly it is often difficult to detect until you have a child born with the obvious characteristics.

Our group is fairly small. We have a mailing list of just over two hundred and so it is difficult to provide as much as we might like because our members are few and far between, however we are always at the end of the telephone to talk to newly diagnosed families and I know from personal experience and my work with the group that this support can be invaluable.

Since the development of the internet and the world wide web we have increasing contacts with TC people in other countries who we find are much

more isolated than us with no information or medical support.

**If you would like more information please contact Sue Moore, 01603 433736
sue@treachercollins.net
www.treachercollins.net**

What is Treacher Collins Syndrome?

The syndrome was named after an ophthalmologist called Edward Treacher Collins in 1900. It is a condition that causes facial malformations and severe hearing loss.

Treacher Collins Syndrome is inherited so for those that have the Syndrome there is a 50-50 chance of having children affected by it. The severity of a parent's condition is not an indication of the possible severity of the child. Each case will be different and must be assessed individually. The psychological effects of Treacher Collins Syndrome are significant due to facial malformations. It is important to have correct consultation on everything involved.

Over half of Treacher Collins Syndrome cases are a result of fresh genetic mutation, not direct inheritance from parents. These children in turn will have a 50-50 chance of passing it on.

The James Lind Alliance



The James Lind Alliance has been established to promote partnerships of patients and clinicians to identify and prioritize unanswered questions about the effects of treatments.

So, what's the big deal about that?

Four years ago I worked for a Bank, and had done so for 35 years. I knew a lot about Banking but, being busy all my life, had a much more vague appreciation of what went on in the wider world.

Then, our eldest son died of variant CJD (The human form of mad cow disease) and I became closely interested in Medical Research. Whilst vCJD may not be a Genetic illness (though a great deal is unknown) there are other forms – one of which is genetic – “familial” CJD. Once in a family this normally affects 50% of members – with devastating effects.

Because of the high media profile of CJD we were “fortunate” enough to benefit from detailed consultations with researchers and funders on our views and trials etc. As a group of Patients and Carers we were involved at every stage – and even now have a strong presence within the whole spectrum of research.

I thought this was the norm – but now I know differently.

I had wrongly assumed that patients were always working hand in glove with those in the medical world researching for new therapies.

Whilst this may occur in isolated areas it does not appear to be the established practice. The James Lind Alliance (JLA) has been formed to try and address part of that process – namely the agreement and prioritization of uncertainties about the effects of treatments.

The JLA was conceptualised during the summer of 2004. by its three founding conveners - The James Lind Library, the Royal Society of Medicine, and INVOLVE. The JLA Secretariat is supported by a grant from the Medical Research Council and the Department of Health.

Our aim is to promote a meeting of minds of

Patients and Clinicians about priorities for research. One of the ways in which we are trying to do this help is by ‘harvesting’ patients’ and clinicians’ unanswered questions about the effects of treatments, so The Database of Uncertainties about the Effects of Treatments (DUETs) has been established, and you can visit it at www.duets.nhs.uk.

We are currently piloting our first JLA Working Partnership of patients and clinicians – in Asthma, and we have been delighted by the enthusiasm and commitment of the partners. Needless to say, the ownership for the Partnership always remains with the Clinicians and Patients.

We realise, of course, that every health condition has its own idiosyncrasies and we are keen to work with different groups to develop our understanding of the support that we wish to offer.

To us, a successful outcome of this whole initiative would be a schedule of possible research areas that, because they had the explicit support of Patients and Clinicians, would be of greatest interest to research funders and researchers

Those research proposals should be easier to achieve funding because of the active involvement of the potential participants – and the sheer logic of putting first those issues agreed upon by the people affected most. Several funders have already expressed an encouraging interest in our early results

The normal route into the JLA is for organisations and individuals who identify with its objectives to become Affiliates of the Alliance (for which there is no charge). They can then consider whether they would like to help establish a JLA “Working Partnership”.

If you are interested in learning more of the JLA, or becoming affiliated to the Alliance, please visit our website – www.lindalliance.org Information on DUETs is at www.duets.nhs.uk. **Lester Firkins is Chair of the JLA Development Group, a member of INVOLVE and Co-Chair of the MRC Prion-1 Clinical Trial Steering Committee.**

Nail Patella Syndrome - Working with the Media.

When I heard about the opportunity of working with the BBC and doing a documentary on highlighting various genetic conditions, I was both very excited, but equally anxious that everything would be ok. I had worked with the BBC a few months before as they aired a programme on BBC Radio 4 which was hosted by Candida Harris, a journalist for the BBC and who also lives with a rare genetic condition. I live with a condition called Nail Patella Syndrome and share it with my son, Stephen.

I realised that Candida and I had so much in common with one another and talking to her was extremely therapeutic in many ways. I decided that the cameras should not be a problem and gave my consent to go ahead with the programme.

A few weeks before the filming, I met with the Producer/Director, Dickon Le Marchant, who was incredibly understanding and gentle. He was the type of person I knew instantly that I would not have a problem working with and the initial fears of the intrusiveness were slowly disappearing from my mind. He explained a little about the programme and that he would be coming with one other person, Lucy, who would help with the sound control amongst many other jobs. This was a great relief as I am sure you can imagine, I had visions of cameras all over the place and the house swamped with people for a few days. He explained that they would film over the period of 3 or 4 days and that we should start to think about what we would like to see on camera. I had some ideas immediately and we discussed these and agreed that it should be shown. Over the next few weeks, we planned out what was

needed very easily by email and telephone conversations, all the time building up a trusting relationship.

On the 1st November, 2005, Dickon, Candida and Lucy arrived at my house at 9.30 am. I have to say that I was a lot more excited, than nervous to see them as I hadn't seen Candida for a few months! It was very calm and we all settled in before the decisions were being made as to where the "Main interview" should take place. We decided at my dining room table, and I am glad that I gave it a good clean beforehand! The camera was set up, my mike was put in place and Candida and I chatted naturally, all the time with the camera focussing on our conversation. It wasn't difficult at all as it was completely natural to discuss the subject matter, but at one stage, I completely welled up and the tears began to flow! I had no idea that this would happen and I had no control over it, but Dickon, very compassionately, switched the camera off. I didn't feel uncomfortable as these people had become "friends" and knew how emotional this was to talk about. The interview lasted approximately 90 minutes.

After the interview, we planned on picking up my son, Stephen, from school. From school, we were to go to Yorkhill Children's Hospital where he had an appointment with the Renal specialist, Dr. Heather Maxwell, followed by an ultrasound scan of his kidneys.

We left the house at 12.20 pm and picked up Stephen from School. It was quite bizarre in many ways as I was still wearing the radio mike, but became very natural over the course of time. We drove from the school to McDonalds for a quick bite in motion as we continued on to the hospital. As we arrived at the

Hospital, we were met by the Press Officer for the Greater Glasgow Health Board. She is the one who clears all the presence of the cameras with the people concerned. We then met up with Dr. Maxwell, who has been caring for Stephen since he was a year old! We discussed what would be nice to be filmed and then we were asked to do a re-take on entering the clinic. They filmed it all and then we were asked to go to the ultrasound scan room where he would get his scan of his kidneys. It was all well organised with very little waiting time and no hitches at all. It wasn't too intrusive for Stephen and all he wanted to do was to get outside to play in the little park!

We returned home and Dickon filmed little scenes of general family life. Nicola, my daughter was home from High School by this time and was introduced to everyone. We basically carried on as normal, I made dinner, Nicola started her homework and Stephen went outside to play with his friends. Dickon and Lucy followed Stephen and filmed him playing football and then followed him to a little park close to our home. They came back to the house afterwards and filmed us moving around at home and then decided that the day should come to an end. It had been a very full day and we should start afresh in the morning. Unfortunately, Keith, my husband, was working late, so it meant that the interview with Candida would need to be postponed as she needed to go home to London to her family. It was emotional for me to say goodbye to this incredible woman who I will always maintain, is a true inspiration. I am sure that we will keep in touch regularly now.

The following day, 2nd November, started a little later,

approximately 12 noon. I was on the phone to Stephen's TaeKwon-Do instructor Mr. Brian Leckie, when they arrived. We were discussing what would happen at the class later in the day as this would show the inspirational side of Stephen's character. This was the bit I was looking forward to as the students were all well informed and I had worked on permission slips for them to sign and hand back to say that they were comfortable about being filmed.

Dickon explained to me that he would like to film me walking in a park somewhere locally. We drove to a little park and I brought some bread for the ducks and swans. We waited for the rain to stop and grabbed our opportunity. He told me to walk towards the camera and stop at a point where I was to feed the swans. I am sure that we provided some entertainment for an elderly couple who were parked next to us!!! Dickon was filming me leaning towards a beautiful swan who had come right out of the water for the bread when another came up behind him and hissed loudly at him and suddenly bit him in the nether regions!!! I have to say he was so professional about it and continued filming!

We then left the park and drove to Stephen's school where we planned on filming Stephen coming out of school. The school had all been informed and were a great support to us on this. He came out swamped between two of his classmates, who I think were happy to be Stephen's friend on that day! We then returned home and filmed a little more at home and then it was time to go to the Taekwon-Do class.

We left just after 5 pm and introductions were made by Mr. Leckie and the BBC crew. They spoke about what would happen and Mr Lecke was wonderful and so accommodating. He was asked if he would wear a radio mike for the filming and he agreed. As the students arrived, I

have to say I was becoming a little overwhelmed at the support shown. They were all immaculately groomed and were fine examples of the discipline taught to them throughout the years. The class numbers were immense and I had to gather myself on more than one occasion by leaving the room for some fresh air. It was just amazing to see. Stephen worked so hard and was very keen to show off his talents!

We returned home and I was totally elated at what had just happened. I will never forget that hour as long as I live and breathe. It was simply magical!

When we arrived home, Keith was home from work and he finally got the chance to meet with Dickon and Lucy. The difficult part was always going to be for Keith to talk on camera as he is such a private person and as Candida had to leave to be with her family, I would be the one who would have to "interview" him on camera! I was anxious about this as Keith is not a great conversationist, but he knew how important this was to all of us. We quickly grabbed the opportunity as Stephen and Nicola disappeared upstairs. Keith was miked up and initiated the conversation. At first it was a little uncomfortable, but as the time passed it became quite natural and the conversation flowed. It was an incredible few minutes as we were sitting there being filmed as we discussed something that was very emotional for us both. I realised during that time just how much Keith loves and accepts his family. He just accepts the fact that his wife and son both share a rare genetic disorder and views his life as normal. He knows the implications, but he has adjusted his life to suit.

After the filming, we decided that we probably would need no more. I was asked to find a few

photos and make a little picture story of our experiences together. I will send these to the BBC and Dickon will keep in touch regarding the screening of the programme. It is likely to be seen on BBC 4 first in January and should be then transferred to BBC2 at a later date.

It has been a wonderful opportunity to raise awareness for our rare genetic condition, Nail Patella Syndrome. I know that I could never refuse this chance as this platform does not come around every day. I hope that we are able to find many others in the UK with this condition and are able to help them. I hope that we are able to prevent many of them from losing their eyesight and encourage them to attend renal units to prevent kidney damage. I hope that we make many medical professionals sit up and listen to us and realise that even though statistically we are 1/50,000, we are ALL important and need medical care.

None of this would have been possible without the incredible support from the Genetic Interest Group. I am indebted to you now and will never forget the love and support you have shown us over the past couple of years. I absolutely encourage everyone to seriously think about grabbing the chance to do something like this too and I feel pretty sure that many people will be helped as a result of your work and commitment to your cause.

Carol Dobbins, NPS UK
<http://www.npsuk.org/>



Carol Dobbins with Candida Harris

Eurogentest Project - an update

Over the last year I have been busy gathering data and doing research for Unit 6 of the Eurogentest project. This is a European Network of Excellence which aims to improve all aspects of genetic testing across Europe. The unit that Alastair, Pritti and I are involved in is concerned with improving patient education about genetic testing. In order to do this we firstly decided to assess the quality of patient information from across 5 European countries, namely the UK, Holland, Belgium, Italy and Sweden. We did this by gathering examples of written information that related to genetic testing, in the form of personal letters, leaflets, brochures and web pages. These were gathered from numerous sources including geneticists, genetic counsellors, hospital websites, patient organisations and national health departments. The written information was then assessed to see if it contained a number of themes that related to genetic testing.

The findings were that much of the literature contained information about the condition (background and effects, heredity and risk, treatment and management), about half the information contained issues concerning the test itself (candidates for testing, purpose of the test, test procedure, test accuracy etc) and very little contained information about the social aspects of genetic testing (patient rights, discrimination, cost, shared decision making, psycho-social consequences etc). Another interesting finding was that although there was lots of information available concerning genetic testing for hereditary breast cancer, there was very little information available concerning genetic testing for the more rare conditions such as tuberous sclerosis and 22q11 deletion. In addition, information about hereditary breast cancer dealt with far more of the key issues to do with the social aspects of genetic testing than any of the other literature did.

Our questionnaire results also showed that many people do not receive written information before genetic testing. In fact, whilst 96% of respondents said they provide written information at the end of the consultation episode, only 53% said they provide written information before genetic testing. This means that information about genetic testing is only

being communicated orally, if at all.

The findings were recently written up in a draft report and presented at the national co-ordinators meeting in Leuven.

This was where representatives from patient organisations were invited to

discuss the findings of the unit so far, and to share ideas about how to progress in the future. This was a very fruitful meeting that gave participants the opportunity to meet members from similar organisations in Europe, and a number of interesting experiences and ideas were aired. It was agreed that one way to proceed with the patient education unit would be to refine the evaluation tool that had already been developed (to assess the written patient information), and to use it to form a 'patient centred checklist'. This will be a tool that will guide and inform patients and families when speaking to a health professional, so that they can make well informed decisions regarding genetic testing. This checklist will be in the form of a number of questions and discussion paragraphs, as well as providing local relevant information. It will be developed with the help of patient organisations and professional health educationists in order to ensure its accuracy and accessibility. It will be translated into a number of languages and disseminated across Europe through patient organisations, hospitals, genetic clinics and national health bodies.

With our findings showing that the quality of patient information about genetic testing across Europe is poor, it is obvious that there is a large amount of work to do in this area. Through the Eurogentest project, we aim to improve the standard of patient information across Europe so that patients are able to access good quality information about genetic testing, whichever country they are in.

Celine Lewis, Eurogentest Project Officer
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www.eurogentest.org



Celine Lewis

HITS (UK) Family Support Network

Supporting families affected by the Hypomelanosis of Ito Syndrome

It all started in the year 2000 when a small group of families got together in London and formed a small committee, armed with only a piece of paper with a dozen or so names on it, a lot of determination and willpower and just over £120 to start off the bank account.

Five years on, we are still a small group but we have come a long way during that time and are now supporting families in 21 countries - the UK & Ireland, Australia, Belgium, Brazil, Canada, Cyprus, France, Germany, Holland, India, Italy, Mexico, Malta, New Zealand, South Africa, Singapore, Spain, Sweden, Turkey & the USA.

Each year we have held our annual Family Day in London and families from as far away as Sweden, France and Northern Ireland have managed to come along to join in with the UK members. We have an



Family gathering in the UK

on-line chat group at Yahoo and send out 2 newsletters a year. In 2005 Dr Celia Moss and her team at the

Dr Celia Moss and Dr Saleem Taibjee from Birmingham Children's Hospital in conjunction with the Skin Laboratory at University Hospital Birmingham have recently



HITS UK family day

set up a research project looking into the genetics of hypomelanosis of Ito. They are trying to find out if the diagnosis of the condition can be improved by taking skin biopsies and looking at skin cells called keratinocytes. The hope is that they can help families find out what is the precise genetic abnormality (mosaic) in their child. This research may also help us to understand which pigment genes are disrupted, causing the characteristic lightening and darkening of the skin in this condition.

Membership is free, but donations are gratefully accepted. For more information please email : **Terri Grant at tgrant@uk.ey.com or phone 07940 114943**



US members of HITS at a family gathering

Birmingham's children's hospital embarked upon the following research project:

Moving GIG Policy Forward

As you will have read earlier in the GIG newsletter, John Gillott, GIGs Policy Officer has decided that it is time for him to move on after 11 years with us. GIGs Director and Assistant Director along with GIG trustees have been looking at ways in which GIGs policy work can be continued and we are therefore delighted to announce that Nick Meade, who has been working with GIG for three days a week as

Project Implementation Officer will be working for the other two days on policy issues.

GIG is also aiming to increase its profile and carry out more proactive policy work lobbying MPs, policy makers and holding events. Melissa Winter will be working with Nick to do this. Pritti Mehta will be working on issues of equity and access within the policy field as well. In the devolved governments Buddug

Williams and Gillian Scott will be playing a critical role in raising GIGs profile.

Alastair Kent will remain active in this area as well, working on UK and European policy issues and he will be leading the new GIG Policy Team as it develops over the coming months.



The Haemophilia Society: AGM and national conference feedback "Reaching out"

The Haemophilia Society's Annual Conference and AGM "Reaching Out" was held at Stansted Airport on 17-18 September 2005 with 189 adults and 59 children attending over the two days. Two popular motions of interest to all GIG Today readers were debates about access to treatment and the future of gene therapy.

'This house believes gene therapy is the way forward'

Haemophilia is an area of active research into gene therapy as a temporary or permanent cure for haemophilia A (factor VIII deficiency) and B (factor IX deficiency). Dr Simon Waddington, researcher at Imperial College London, spoke for the motion: "Gene therapy is the way forward for haemophilia" Dr Paul Giangrande, co-director of the Oxford Haemophilia Centre spoke against. The motion was defeated by 35 to 20 votes. For the motion, Dr Simon

Waddington Simon spoke with a five-part presentation to support the motion, covering the history of gene therapy, a broad description of three different models of it, the utility of gene therapy, alternatives to it, and current clinical practice and research. He shared people's concern about the safety of the interventions. In summary, the case for was:

- It is a mature area of clinical research
 - The technology covers a broad area and gene therapy research had contributed to many other therapeutic interventions and medical understanding
 - Gene therapy research is closely allied to the production of clotting factors by cloning genes and their expression (recombinant technology)
 - More specifically, research into clotting factors will benefit other disease areas
- Against the motion, Dr Paul Giangrande Dr Giangrande wanted to point out he was not against new technologies, having been pro-recombinant from its earliest days and beginning to

investigate gene therapy from 1993. He is conservative about gene therapy in haemophilia, agreeing it is not so much a question of if it will be introduced but when, but it is "twenty years off".

In summary, the case against was:

- Current treatment for haemophilia is safe and effective; gene therapy research should concentrate on genetic conditions where no such interventions exist
- We need to follow Hippocrates's advice: "First do no harm"; there are dangers in introducing new medical therapies which look promising but prove problematic once in routine use (eg the recent problems with COX-2 inhibitors and the death of a US patient on a gene therapy trial)
- People have unrealistic expectations of gene therapy believing, for example, that they will be able to play first class rugby or that the cost will be lower (not experienced with the switch from recombinant to plasma-derived clotting factor concentrates)

'This house believes I have the right to extra treatment so I can go skiing'

Treatment for haemophilia is generally effective although considerably expensive. This financial year has seen the final phase of the switch from plasma-derived factor VIII and IX to recombinant (genetically-engineered) production of these concentrates. The conference debated the motion 'I have the right to extra treatment so I can go skiing'. The issue at stake was that many with severe haemophilia, particularly children and young adults, are on prophylaxis therapy. This means they take treatment every two to three days to prevent painful bleeding into joints and muscles. Those who go on skiing holidays may need additional treatment on top of the usual regime. In a world where NHS resources are limited, is this right?

Speaking for the motion was a mother of a son with severe haemophilia. Speaking against the motion was Dr Brian Colvin, centre director at the Royal

London Haemophilia Centre. The vote was 69-55 in favour.

For the motion, Gill Jolley introduced herself as a mother of a well-travelled and enthusiastic sportsman. She detailed three key arguments concerning the nature of the activity and proper preparation, the right to treatment, and the enhancement of quality of life.

Against the motion, Dr Brian Colvin He felt that traditionally sports such as swimming, running and tennis have been encouraged, whilst boxing and rugby have been disallowed. Some degree of choice might be exercised for sports with risks in between these extremes; this is really the subject of this debate.

A report on the content of plenary and workshop sessions is available from the Haemophilia Society (www.haemophilia.org.uk).

John Morris, Services development manager,

Charting the patterns of genetic variation.

Alastair Kent, Director of the Genetic Interest Group is part of the International HapMap Consortium. This Consortium involves scientists from the United Kingdom, USA, Canada, Japan, China and Nigeria. The project began in October 2002 and set an ambitious goal of creating a human haplotype map within three years.

A haplotype map charts the patterns of genetic variation that are common in the world's population. The results from the first phase were published in a paper in *Nature* in October 2005 and announced the attainment of that goal as well as providing overwhelming evidence that variation in the human genome is organised into local neighbourhoods, called haplotypes, these are usually inherited as intact blocks of information.

Any two unrelated people are 99.9 percent identical at the genetic level. However, it is important to understand the 0.1 percent differences because it can help explain why one person is more susceptible to a disease or responds differently to a drug or an environmental factor than another person.

Scientists are calling this a milestone in medical research building on the foundations laid by the human genome sequence. The HapMap will provide a powerful new tool for exploring the root causes of common diseases such as asthma,

diabetes, cancer and heart disease.

"The HapMap is a phenomenal tool that is making possible research that was impractical, if not unimaginable, only a few years ago" said Yusuke Nakamura MD, PhD - Director of the University Of Tokyo's Human Genome Centre. The data that is produced from the HapMap project will help biomedical researchers simplify their searches for information and enable them to search much faster.

Many other discoveries lie on the horizon as the HapMap empowers studies of other common diseases, including, Alzheimer's disease, asthma, cancer, diabetes, hypertension, heart disease and schizophrenia. In addition to assisting in the identification of genetic factors involved in disease, the HapMap can help to pinpoint genetic variations that may affect the response of people to medications, toxic substances and the environmental factors. Such information will hopefully be used to help doctors prescribe the right drug in the right dose for each patient.

As expected, a vast majority of both rare and common patterns of genetic variation were found in all of the populations studied.

However, the consortium did find

evidence that a very small subset of human

genetic variation may be related to selection pressures related to geographic or environmental factors such as micro-organisms that cause infectious diseases. While more follow-up studies are needed to explore these differences, researchers say some of the most striking examples serve to confirm well-known genetic differences among populations, such as the Duffy blood group, which plays a role in response to malaria, and the lactase gene, which influences the ability to digest milk products.

"Like the Human Genome Project before it, the key to the International HapMap Project's success lies in the shared vision and hard work of hundreds of researchers from many different nations and many different disciplines" said NHGRI Director Francis S Collins, MD, PhD who served as the project manager for HapMap.

As was the case with the data generated by the Human Genome Project, HapMap data is being made swiftly and freely available in public databases.

To find out more about this project and the research that is taking place please look at www.hapmap.org



EVENTS and NEWS

The XP Support Group 5th Owl Patrol Night-Time Camp

The XP Support will hold it's 5th Owl patrol night-time camp for those with Xeroderma Pigmentosum and other light sensitivities from 10-13 February 2006.

The camp provides light sensitive patients with the opportunity to meet in a protected environment during the day and to enjoy outdoor activities at night. The camp also presents speakers on various topics related to light sensitivities and will hold a special clinic over the weekend.

For further information, please contact the XP Support Group. Tel:01494-890981 E-mail: info@xpsupportgroup.org.uk

Sharon Terry to Receive Honorary Doctorate for her Work on Community Engagement and the Haploype Map

Sharon Fontaine Terry, president and chief executive officer of Genetic Alliance, will be presented with an honorary doctorate at the Fall Honors Convocation, on Sunday, November 13 at 3:00 pm in Joyce Auditorium, Iona College, New Rochelle, NY. Terry is a co-inventor of the patent for the ABCC6 gene and provides leadership to the world's largest coalition of genetic advocacy organizations including more than 600 advocacy groups, community groups, health professionals, researchers, hospitals and clinics. Her address to the convocation is entitled "Envisioning the Future."

New Years Honours List

GIG was delighted to hear of two colleagues working in the genetics field who received New Years Honours and we would like to take this opportunity to pass on our congratulations to Susy Leather and Neva Haites.

O.B.E.

Professor Neva Elizabeth Haites, Vice-Principal and Head of the College of Life Sciences and Medicine, University of Aberdeen. For services to Medicine.

D.B.E.

Ms Susan Catherine Leather, M.B.E., Chair, Human Fertilisation and Embryology Authority. For services to the Regulation of Infertility Treatment and Embryo Research.

CMT UK launch a brand new website for 2006

CMT United Kingdom are pleased to announce the launch of their brand new - and greatly improved - website. It features all you ever wanted to know about Charcot-Marie-Tooth Disease (CMT), how to live with it, how to manage it, and general information with coping with disability in day to day life.

There is also an area to children with CMT, which will, no doubt, develop further over time - as will the professionals area, which we are hoping will become a repository for all the latest scientific news on CMT. The website, which has been created by ResonantMedia Ltd of London, can now accept online donations, membership and has a shop for Christmas cards and other CMT related goodies.

Visit CMT United Kingdom at www.cmt.org.uk

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