

## Celebrating 50 years of DNA



When Watson and Crick wrote their now famous letter to Nature half a century ago they knew they were on to something big! Just how big that something would turn out to be could not begin to have been imagined.

Today, fifty years on from the publication of the structure of DNA we can begin to appreciate the potential of their discovery. Throughout this year activities are taking place here in the UK, in America and across the world to celebrate the golden jubilee of this discovery.

Today, as a result of research into the genetics of health and disease we have, for the first time in human history, the tools to explore how our biology works at the most basic and fundamental level.

Knowledge of the structure of DNA and the techniques developed to enable us to read our own genetic code have enabled us to understand what goes wrong at the molecular level when mutations result in serious genetic diseases. This is an incredibly powerful tool, and already we have seen considerable benefits from the application of genetics to the resolution of previously intractable health problems. Recombinant technology (“genetic engineering” to most of us) has produced human insulin for the treatment of diabetics, clotting factors for people with haemophilia, enzyme replacement therapy for those with some rare metabolic disorders, to name but a few of the advances in use today. Of course, there is a huge way to go, and we do not know how long or how difficult the journey will be, but at least we are on the road and moving forward.

To commemorate the golden jubilee, GIG is producing three new publications this year. The first of these is out now, called “Genes and Risk”. It explores the extent to which

genetics enables us to predict future health events. Whilst some genetic changes result in a high degree of certainty in predicting future outcomes, others are much less reliable as guide to the future, as a genetic and environmental or other factors all work together to affect the outcome. All our members should by now have received their copy of this booklet but if you would like any further copies then please do let us know. It is also available to download from the website [www.gig.org.uk](http://www.gig.org.uk)

The second publication called “DNA Delivers” will be out very shortly. This looks at the impact that our knowledge of DNA has had on families and on the doctors who work with them. Although many of the families featured are affected by conditions that remain incurable, nevertheless the knowledge we have now has made advances in treatment possible and given people some options where previously they may have had none.

The third publication to be published in October focuses on the importance of

diagnosis as a means of giving choices and empowering families. Whilst a diagnosis may be difficult for families to come to terms with, being in ignorance is usually more so. The issues and the experiences of individuals and families with a range of different types of genetic conditions will be featured in this booklet, to be published later this year.

We are very grateful to all those who gave their time and shared their experiences with us, and to the sponsors who made these publications possible.

**Alastair Kent**

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

**Tel: 020 7704 3141  
Fax: 020 7359 1447**

**Email: [mail@gig.org.uk](mailto:mail@gig.org.uk)  
Website: [www.gig.org.uk](http://www.gig.org.uk)  
A registered Charity (Number 803424)**

**Please note that the views and opinions expressed in this newsletter are not necessarily those of the Genetic Interest Group**

# “Our inheritance, our future – realising the potential of genetics in the NHS”

At last, the government’s white paper on genetics “Our inheritance, our future – realising the potential of genetics in the NHS” has been published. After a prolonged gestation, the government finally announced its intentions for the future of genetics in the NHS and the money it will put up to make its plans a reality!

In all, about £50million of spending commitments were contained in the white paper. Anyone wanting its full details can read the whole document on the Department of Health’s website at <http://www.doh.gov.uk/genetics/whitepaper.htm> but a number of important commitments stand out.

First amongst these is the funding for new scientist and genetics counsellors, together with money to buy new equipment for testing laboratories. This will allow the introductions of targets for the delivery of list results. This will be a very important step forward – especially for those families who now find themselves waiting for months and months and who have to live with the anxiety that this causes.

Another important commitment is the promise of funding for gene therapy research. £3million has been earmarked for rare disorders with a further £2.5million for cystic fibrosis gene therapy. Congratulations are due to Ann Hunt, who served on the Gene Therapy Advisory Committee for her tenacious

advocacy, keeping gene therapy for rare disorders on the Department’s radar, and to Rosie Barnes, CEO of the Cystic Fibrosis Trust for her tireless campaigning to get money into CF research. The signs are promising – more so now than they have ever been. Let us hope that this cash injection, together with money from other sources (such as Genes for Jeans day to be held on 3rd October this year) will let us see real progress towards prevention and cure for some at least, of the rare genetic disorders.

Other initiatives in the white paper include funding for education and training for health professionals, and it’s announcement of GP Specialists – although only 10 are envisaged in the short term, so they will not have an immediate impact when you realise that there are over 30,000 GP’s in the NHS. But it is a start; and as the Chinese proverb says it “a journey of a thousand miles starts with a single step”

Of course, without public support and encouragement for genetic research and the application of its results making progress will be much more difficult. The white paper emphasises the need for public communication, and the importance of ensuring the ethical application of new knowledge, and the prevention of abuse. In particular it raises the question of consent for paternity testing, and promises legislation to prevent a free for all in paternity testing. So trawling through the bins of celebrities for hair samples ought to become a thing of the past and a good job too!

Delivering the new genetics across the board will be a long and expensive process. The £50million earmarked in the white paper is only a drop in the ocean. A very welcome “drop” to be sure, but more importantly the money is the commitment, the recognition that, in future genetics and medicine will be intertwined, and if that intertwining is to be supportive, rather than a stranglehold, then we have to start building the framework now. This white paper is an important foundation stone on which to build for a secure future for genetics in the NHS.

**Alastair Kent**

---

## A New Face at GIG

With the expansion of GIG’s work in the Genetic Knowledge Parks, we need to support staff working away from the London office. To do this we have appointed Tom Barclay as GIG’s new Assistant Director. Tom writes to introduce himself in this edition, but some of you may have spoken to him on the phone or met him at one of the meetings he has attended since he took up his post a few weeks ago. He is keen to be involved, so please do make contact and get to know him as he settles in to his new role.

## Gig’s New Staff Member in Wales

I am delighted to introduce myself as the development officer for GIG in Wales, based within the Wales Gene Park. I will be working with GIG staff in London and with clinicians across Wales to promote awareness of families with genetic disorders by generating and communicating their viewpoints on the developments of genetics in healthcare in Wales.

Over the next few months I will be establishing a network of individuals and groups to inform the political and

professional discussion about the services and resources needed to continue providing high quality genetic services for the people of Wales. I hope to continue the strong links that GIG has with various groups. I am also very eager to meet with new groups and families.

If you would like to know more about GIG in Wales, or if you have any matters you would like to discuss, please contact me and I will be happy to talk with you.

**Buddug Williams**, GIG Cymru  
Development Officer, Wales Gene Park,  
Medicentre Heath Park, Cardiff, CF14  
4UJ, T: 02920 682140, F: 02920 682141,  
E: [williamsbg@cf.ac.uk](mailto:williamsbg@cf.ac.uk)

# GIG

## NEWS & EVENTS

### New members of GIG

I am very pleased to say that since I last wrote to you all we have eight new member groups and one individual member joining the Genetic Interest Group.

**Mrs Lillian Ramsay**, The Eyeless Trust  
Quemerford Cottage, 50 Malthouse Sq  
Lakes Lane, Beaconsfield  
Bucks, HP9 2LE  
[www.eyeless.org.uk](http://www.eyeless.org.uk)

**Hannah James**, Aniridia Network UK  
PO. Box 6444, Colchester  
Essex, CO4 3XU  
[http://www.geocities.com/aniridia\\_uk](http://www.geocities.com/aniridia_uk)

**Andrew Wells**  
Peutz Jeghers Syndrome  
Support Group  
17 Barton Road, Harlington  
Dunstable, Bedfordshire, LU5 6LG

**Liz Dent**  
Association of Multiple Endocrine  
Neoplasia Disorders (AMEND)  
Lyndene, The Green,  
Bewerley, Harrogate, HG3 5HS  
<http://www.amend.org.uk>

**Martin Armstrong-Fox**  
Fuchsfriends UK,  
21 Broomhill Road  
Woodford Green,  
Essex, IG8 9HA

**Peter Cardy**  
MacMillan Cancer Relief  
89, Albert Embankment  
London, SE1 7UQ

**Gill Bromilow**  
Dept of Clinical Genetics  
R D and E (Wonford) Hospital  
Barrack Road  
Exeter, EX2 5DW

**Jennifer Bowden**  
LOOK, Queen Alexandra College  
49 Court Oak Road, Harborne,  
Birmingham ,B17 9TG  
<http://www.look-uk.org>

**Eileen Gascoigne**  
The Dystonia Society  
45 - 47 Britton Street  
London, EC1M 5UJ  
<http://www.dystonia.org.uk>

I am delighted that we are continuing to grow, and hope that this will continue. I have also asked our new members to write a small piece about the conditions that they represent, which I hope you will all find helpful. They will appear in this issue and the following one. **Melissa Winter**

### The Aniridia Network UK

The Aniridia Network UK represents people with aniridia and their families. Aniridia is a congenital genetic eye condition, in which the gene for eye development (PAX6 at 11p13) does not function correctly and this causes the eyes to be underdeveloped. Aniridia is almost always present in both eyes and the most obvious feature of aniridia is the absence of the iris, the coloured ring around the pupil. Most people with aniridia appear to have very dark eyes. People with aniridia also frequently develop cataracts, glaucoma, cornea scarring, low vision and nystagmus (involuntary movement of the eyes). Aniridia is also associated two other syndromes, WAGR (Wilms tumour, Aniridia, Genital abnormalities and mental Retardation), which is caused by a larger deletion, and Gillespie syndrome, this involves aniridia and cerebella ataxia. The genetics of this condition are unclear.

### The Dystonia Society

Dystonia is a neurological movement disorder, characterised by uncontrollable, involuntary muscle spasm. The spasm can affect almost any muscle group in the body, and may lead to permanent deformity, disfigurement and disability.

Recent research has identified up to 17 different possible genetic factors, which relate to different forms of the condition. The most severe type of dystonia is generalised dystonia, and this usually first appears in childhood or adolescence. A majority of people with this form of dystonia have been found to carry a defective gene - DYT1.

The Dystonia Society exists to support people who have any form of dystonia, and their families, through the promotion of awareness, research and welfare. There are 20 local branches around the UK, and a central information and support line.

### Peutz-Jeghers Syndrome (PJS)

is a rare disorder that is either inherited from a parent or occurs spontaneously. Patients with PJS have hamartomas predominately in the small intestine, as well as fewer polyps in the large intestine and stomach. Purple / black pigment spots often dot the lips and other areas of the face and body. Most of the time, the spots disappear sometime in the teen years. Age at diagnosis depends on the severity of symptoms. Research has shown that there is an increased risk of cancer over the general population. The gene predominantly responsible for the syndrome has been discovered, but genetic testing can still be inconclusive.

The PJS support group operates through a listserv operated by ACOR (the Association of Cancer Online Resources). The group is meant to end the isolation of those affected by PJS, their family members, friends and medical professionals

### Gender Trouble Film

Gender Trouble's film (see [www.wonder-dog.co.uk/gender.html](http://www.wonder-dog.co.uk/gender.html)) is funded by the UK's Wellcome Trust and in which four UK group members (3 AIS, 1 CAH) talk to the camera about their experiences.

A number of medical departments/clinicians have already purchased copies of the film and the producer Roz Mortimer and AISSG are keen to make it available to as many as possible. If you need further information, do please contact Roz ([roz@wonder-dog.co.uk](mailto:roz@wonder-dog.co.uk)).

Androgen Insensitivity Syndrome  
Support Group (AISSG)  
[www.medhelp.org/www/ais](http://www.medhelp.org/www/ais)



# Institute of Ideas – Genes and Society Festival

26th – 27th April 2003, Battersea Arts Centre  
Lavender Hill, London

This weekend organised by the Institute of Ideas was of course linked to the 50th Anniversary Celebrations of the discovery of DNA. The organisers brought together a host of scientists, writers, social commentators, regulators, philosophers, artists and campaigners to discuss the many implications of genetic discoveries and advances.

There were many sessions throughout the day and on filling out the form to attend you needed to specify which talks you would like to go to. I have to say that it was rather a hard decision as they all looked so fascinating! Alastair was speaking in two of the sessions and John Gillott in another, so that made those choices easy for me. The other session was up to me to decide on. ....

Here is a brief run down of the sessions which I attended, if you would like any further details then please do not hesitate to contact me and I will do my best to fill you in.

## Plenary Session “Genetics: Hype and Reality”

Alastair Kent was on the panel for this plenary session. The session reflected on what has been achieved to date in genetics, what is on the horizon and also the wider social implications of this technology. The three other panel members were Dr. Michael Fitzpatrick, GP and columnist for the Lancet, David Goldstein, Wolfson Professor of Genetics at University College London and Geoff Watts, presenter of BBC Radio 4's Leading Edge and a member of the HGC.

David Goldstein was the first to speak and he posed two questions at the beginning of his talk. The first was What has genetics delivered? And the second was What are the practical implications in medicine?



Genetics has delivered the human genome project, and it was delivered early! We now know how information is transferred within our bodies, how to write it, and how to understand quite a bit of it, and all this in just 50 years. If you had to print out your own human genome to read out loud it would take 40 days and nights non-stop. He then went on to discuss how in the last 50 years we have managed to discover so much about our ancestors by using DNA technology. Mr. Goldstein worked on the BBC's Blood of the Vikings programme and used the example of Viking genes, stating that we can now find out whether the UK's gene pool has been affected by old invasions, such as the Vikings and we can now find out if we have Viking genes.

In regards to his second question, he argued that one of the implications for medicine now and in the future is that pharmacogenetics will become more widely used. We will try to make the drugs we take work for individuals, taking into account what their genome tells us. This could have amazing potential for many, ranging from headache pills to patients with serious medical conditions such as epilepsy who at present are drug resistant due to their genetic make up. If we could isolate why, pharmacogenetics could provide the drugs that work.

Dr Michael Fitzpatrick was the second person to speak and he spoke about how the new genetic technology we have attracts and repels people. He argued that DNA offers tremendous promise, it is the key to the secret of life but that because of this it preys on people's fears that others may not interpret or manipulate the information for the benefits of everyone.

He argued that genetics has become politicised. There is much more knowledge at the molecular level but the treatment of patients had not really improved. “We now have a biological revolution all the time, this has brought fear into society, people feel out of control of their own lives. Genetics can be seen as a threat”, Dr Fitzpatrick felt this was a rather pessimistic view but that it was one he saw from all walks of life in his surgery. It will take time for therapeutic treatments to come about after the biology has been made. It took 400 years from knowing how the human heart works to the first heart transplant, 80 years from understanding how germs spread to getting antibiotics.

Dr. Fitzpatrick felt that although DNA hadn't contributed in the day to day treatment of patients that it was still a great discovery, even if it did not bring about any clinical benefits in our lifetime. It had already delivered more than that.

**(l-r) Alastair Kent, Dr. Michael Fitzpatrick, Tony Gilland, David Goldstein, Geoff Watts**

As you can imagine a heated debate began after these two speakers. Alastair quite rightly pointed out all the benefits to patients over the last 20 years. He didn't think that DNA was over hyped at all as the progress we have made has been astounding. As he said "We just don't know how long the walk is, and how far we have come. We have made huge progress and we should not lose sight of this."

## Genes and Longevity – How long should we want to live?

The subheading to this was really "Do we want to live forever?!" I found this to be a fascinating debate. It looked at the implications for individuals and for society as a whole with an aging population. How has science input into this? And how will it change our identity? We had four brilliant speakers, Richard Ashcroft-Head of Unit and Leverhulme Senior Lecturer in Medical Ethics Imperial College, John Hands – novelist, Phil Mullan - author, David Wynford-Thomas – Professor of Pathology and Director of Cancer Research UK at University of Wales College of Medicine.

Immortality has been part of human life since time began, Egyptians, vampires, Christians, Muslims. There has been a continued obsession with living forever, through our souls.

The facts today are, that we are living longer. However we are living longer – older but not living longer – younger. 40% of the NHS budget is spent on caring for the over 65's. We also discussed the philosophical argument, which was how long do we exactly want to live for? Would life not become monotonous? Would the fact that we live forever mean that we never really bother to do anything? Is the fact that life is something which finishes which gives us the momentum to do great things? And who benefits from humans living forever? The rich?

The question about pensions arose, along with the retirement age. Who would get the jobs if people lived for an extended period of time, would there simply not be enough jobs to go round? Certainly for me, more questions came out of the discussion than answers and we, as an audience, did not come to any joint conclusions. Although it appears that scientists have isolated a certain gene that is related to the aging process, and some animals do not carry this gene and still age, so there will be many factors to look at when attempting to make people live for a significantly longer amount of time.

## Genes, Privacy and a Genetic Underclass?

Medical researchers are interested in collecting DNA in their quest for understanding diseases, and employers and insurance companies are also interested in our DNA with regards to future health problems. So the question raised was should we worry about our privacy? John Gillott, Policy Officer at GIG was speaking at this meeting along with Mark Littlewood, Director of Liberty, Lee M Silver Professor of Molecular Biology and Public Affairs, Princeton University, and Robert Terry Senior Policy Advisor, The Wellcome Trust.

Professor Lee M Silver spoke about the biology (a little!) He explained that almost 99.9% of each person's DNA is identical and so the notion of a genetic underclass is too simplistic. There is just so much information that you would not be able to pigeon-hole a person into one simple category.

Robert Terry, spoke about the Biobank Project, which as I think most of you are all quite familiar with, I will only recap briefly. Biobank is the worlds largest study of the role of nature and nurture in health and disease. Half a million participants aged between 45 – 49 will participate by giving a sample of their DNA, lifestyle details and their medical histories. Their health will then be monitored and it is hoped that scientists will uncover genetic and environmental factors that lead to common disorders including cancer, heart disease, and Alzheimers.



**John Gillott**

John Gillott spoke about the concerns that the general public were "supposed" to have with regards to genetic information. He felt that there was a tendency to over exaggerate the tests available.

The focus of people's concerns is on predictive information on healthy people and doesn't take into account the people who have genetic conditions and who are already being discriminated against in regards to insurance.

Mark Littlewood, of Liberty had a certain scepticism about research and genetics. He thought that there is a need for a "regime" to be put in place for our genetic information. He was concerned about other people's access to personal information,

and of the storage of such information, as he felt this could be a potential problem in the future. The reason he was so concerned about the genetic information was that a) it is medical information b) it can be a very detailed profile of who you are c) It's likely to be very accurate 4) Information is valuable to certain parties, such as researchers and insurance companies. The difficulty with this information is that it can be highly sensitive and at the moment the advances are taking place so quickly.

## Stem Cells prospects and barriers

This was another very stimulating discussion and I am pleased to say that even though it was late in the day, everyone was certainly actively participating. It was good to hear contrasting views and opinions, too. The focus of the discussion was on embryonic stem cell research, which has the potential to lead to new treatments for patients with degenerative diseases of the heart, liver, kidneys and cerebral tissue. After much discussion in the UK it is now legal to clone human embryonic stem cells for research but in the US it has been denied any public funds, and moves are being made to make it completely illegal. What is the future for this research? As you can well imagine, this was a passionate debate and we had four brilliant speakers. Simon Best, BioEthics spokesperson for the US and UK Biotechnology Industries, Robin Lovell-Badge Head of Division, Developmental Genetics, MRC National Institute for Medical Research, Alastair Kent, Director, Genetic Interest Group, Dr. Stephen Minger Centre for Neuroscience Research, GKT School of Biomedical Sciences, Kings College London.

The first speaker, Robin Lovell-Badge explained in excellent lay terms what embryonic stem cells are – which was extremely useful for someone like myself! As I am sure many of you know already, stem cells are found in many tissues of the human body and their primary function is to replenish cells. Stem cells are used in the foetus to generate other tissues on our body. They generate all the various cell types that make up the body. In adults, stem cells focus on certain areas, replacing the skin for example. However, in the beginning, the embryonic stem cells can give rise to any cells in the body. The reason there is so much media hype surrounding them is that these are cells that have the potential to become anything within your body. However, by studying these cells we can learn how they will work later in life, and this could have clinical implications for helping people who suffer from chronic conditions.

Alastair Kent, spoke about the benefits of using embryonic stem cells to treat patients with genetic conditions. He also spoke of the need for appropriate use, and the sources of the embryonic cells.



**Alastair Kent**

As you can well imagine, this was a very heated discussion, with lots of conflicting views coming from the audience, which made it all the more interesting. The whole event was superbly organised, and certainly the talks I

attended were informative and thought provoking. I will be keeping up to date with the events that the Institute of Ideas organise and you can do so also by looking at their website at [www.instituteofideas.com](http://www.instituteofideas.com)

---

## Education for Health Professionals

### The Patient Perspective, workshop held on 28th April

Report by **Melissa Winter**

This second of two workshops was held in conjunction with the Public Health Genetics Unit (PGHU) in Cambridge, who have been commissioned by the Wellcome Trust to identify the needs for education in genetics among non-genetic healthcare professionals and to develop a national strategy to meet these needs. The PGHU have held seven workshops covering the following areas: - patients, Users and Carers – held by GIG, Postgraduate medicine, Nurses, Midwives and Health visitors, Dietitians, Pharmacists, Primary Care and Health Service Managers. In each workshop there was a representative from the patient workshop, to do a small presentation voicing patient experiences. It was extremely useful and was certainly a real eye opener for some!

The second patient workshop was held at the National Council for Voluntary Organisations, at Euston. (A very nice, and quite reasonably priced venue for charities, should any of you wish to use them, I would certainly recommend it) and followed on from the one first held on 17th February where we had members from Ataxia UK, PIA, Huntington's Disease Association and Ataxia - Telangiectasia Society.

The day worked very well and was chaired by Albert Njindou, a trustee of GIG who works at the Huntington's Disease Association as a Regional Care Worker. The purpose of the workshop was to invite patients, carers and their families to

contribute their experiences with health professionals and to explain how their interaction with these health professionals had helped or hindered their overall experience. It was a very informative and interesting day and the outcome will be ploughed into the report that the Public Health Genetics Unit in Cambridge are putting together. I will keep you abreast of the outcomes of this report in forthcoming newsletters. If you would like to read the full report of the patient workshop that was held on 17th February then please look at [www.medschl.cam.ac.uk/phgu/about\\_phgu/patients-wshop.asp](http://www.medschl.cam.ac.uk/phgu/about_phgu/patients-wshop.asp)

I would like to take this opportunity to thank you for your wonderful response to both workshops and in encouraging your families to attend and discuss issues that are very personal to them. At the workshop held on 28th March we had GIG members representing The Eyeless Trust, Diabetes UK, DEBRA, Unique, Cystic Fibrosis Trust, HITS UK, Downs Heart Group the XLH Network and The Association for Glycogen Storage Diseases.

On 19th – 20th May I attended the strategy building meeting held by the PHGU to bring together members from each of the workshops, to gather and discuss the points raised by all the stakeholders that took part, in order to form an overall group strategy. The PHGU will now report their findings, which will contain recommendations for a UK programme for genetic education of health professionals to the Department of Health and the Wellcome Trust. The final document will also go out for consultation and will be made available on the PHGU website. [www.medschl.cam.ac.uk/phgu/](http://www.medschl.cam.ac.uk/phgu/)

---

## What's New in Fragile X Research?

**The Fragile X Society, National Family Conference, Saturday 28th June 2003, at the Holiday Inn Birmingham City Centre**

**Report by Anna Lane, Development Officer-Patient & Public Participation, Genetic Interest Group.**

Experiences were shared and friendships forged when families and professionals met on a hot and sunny Saturday in Birmingham's bustling city centre.

Research Scientist, Dr. Mark Hirst, gave a summary of the history of the research to find a diagnostic test for Fragile X, through to his (and others') work today searching for routes which might provide opportunities for a possible therapy. Lively

questions from families in the audience helped clarify the interpretation of results concerning the number of CGG repeats reported as part of the test results, with 200 repeats being the critical number for a positive result. However, it became apparent that smaller numbers than this had significance to those family members given 'carrier status' and also to people who displayed characteristics that are part of the syndrome. Importantly, Dr Hirst placed into context how the work being carried out in laboratories behind the scenes is relevant to the clinicians helping families.

Barbara Carmichael, Genetic Nurse Specialist, explained the role of genetic counselling and emphasized that because Fragile X is such a complex genetic disorder it is important for continued contact and dialogue with families. She suggested that for some people who had been tested many years ago it may now be appropriate to request further testing. This is because those early tests only looked at the chromosome itself and now DNA tests are conducted that can provide more information for individual families about the risks of inheriting this condition.

During lunch there was time to chat about some of the points raised and soak up the atmosphere created by the positive attitudes of the children and their parents enjoying this opportunity to meet old friends and make new ones.

Dr Jeremy Turk, Child & Adolescent Psychiatrist, provided an update on his long-term study of a group of young boys from the early '90s which looked at aspects of their developmental behaviour. His ongoing relationship with the Fragile X Society is recognised as being extremely valuable and he was greeted as an old friend. He reported that the study showed features such as inattentiveness, restlessness and fidgetiness persisted in boys with Fragile X, unlike boys who only have learning difficulties. The subject of autism caused much interest and Dr Turk said a substantial minority of people with Fragile X have autism (29%). Further studies from Dr Turk and his colleagues include Boys with Fragile X Pre-mutations and Females with Fragile X Syndrome, both of which help to shed light on the affects of this inherited disorder. Finishing his talk with a little about 'Aggression', Dr Turk gave some insights into how incidents can sometimes occur and some practical ways of dealing with this.

I felt inspired, informed and grateful for this unique experience of attending an event at which parents, professionals and those with this condition were equal partners, in helping to find out as much as possible about this baffling genetic disorder. We all left feeling optimistic that in the future there will be a therapy to treat its effects.



# Rett Syndrome Association UK

A while back I was kindly invited to visit one of GIG's members the Rett Syndrome Association UK and on arrival I was shown around by Talya Hilburn one of the Associations Family Care Workers. At the RSAUK there are two Family Workers, and their main role is to give support and advice to families living with this condition. **Melissa Winter**

Rett Syndrome was given its name in 1966 and as with many disorders it is named after the Doctor who first discovered it, an Austrian, Professor Andreas Rett. Please look in the box to find a full description of Rett Syndrome.

The Association for the condition was set up in 1985 and began as a support group for parents and families. There were no offices or staff at that time. It has grown considerably since then and in 1996 a permanent office was established in London and there are now 6 members of staff.

When Rett Syndrome UK (RSAUK) was established there were no centres for the treatment of Rett Syndrome at all. Dr Alison Kerr, who is the leading expert in this condition in the UK, started travelling clinics around the country. They would operate for 2 – 3 days in a particular hospital and then move on to another area. This meant that the expert professionals were able to see more children and make further diagnoses, which in turn helped families in finding out the correct treatment for their family member. Now 23 years on, there are six permanent clinics throughout the United Kingdom, each with full time staff.

Patients with this condition have direct and indirect problems. Not only do they suffer from chronic disability, they also have indirect difficulties such as breathing abnormalities. Patients hold their breath at irregular intervals, they can also hyperventilate and have problems with air -swallowing. There is currently a lot of research taking place to try and establish the reasons behind these breathing complications and also the physical complications. RSAUK is very proactive in helping parents and patients get the treatment that they need as many of the indirect problems can be alleviated with the help of regular therapy which

includes:

- Physiotherapy and hydrotherapy – to help with the muscle stiffness that is characteristic of this condition
- music and language therapy – A person with Rett Syndrome can learn and improve their skills, this is an extremely slow process but it can be done through using these types of therapy.

Rett Syndrome is not a degenerative disorder and so many patients do have a normal life span. Although as patients get older they do have other complications which can lead to an early death, for example in older people their muscles become increasingly rigid, joint deformities and muscle wasting occurs.

Epilepsy can also be a difficulty, over 50% of people suffering from Rett Syndrome also have some kind of epileptic seizure at some time during their life. The RSAUK has however identified moments where families can go into what they call “crisis mode”. This is normally around the time a child needs to be statemented, or when they begin schooling and also when they move into adulthood. At all these key points the Family Workers are there to offer their support, coupled with information that the RSAUK have produced about these particularly difficult times.

RSAUK has also set up a networking group through parents. If a parent has a difficulty, they can speak to a designated person that has volunteered to the RSAUK. This parent will have already been through a similar situation and is able to offer advice. This is invaluable information for a parent, as I am sure you are aware, and it can make a huge difference. Along with this, RSAUK have also set up local support groups which are run and are co-ordinated by the parents themselves.



## What is Rett Syndrome?

Rett Syndrome is a neurodevelopment disorder mainly seen in females. Children with Rett are born and develop normally. Usually between 6-18 months they enter a period of regression, specifically in language and purposeful hand skills, this occurs between 9 months and 30 months. At this point a development of repetitive hand movements are developed and the head circumference decreases, gait is also impaired and becomes stiff or clumsy.

This condition is caused in 80% of cases by a gene on the X chromosome called MECP2 which is faulty. This gene was identified in 1999. It is suspected that another gene might be involved in causing the disorder and researchers are currently working on finding it. The diagnosis of Rett Syndrome however is based on clinical criteria. If the child meets the criteria, regardless of a negative test she/he has Rett Syndrome. Almost 99% of Rett Syndrome cases are sporadic and caused by a new mutation in the gene. The incident rate is roughly 1: 10,000 of female births. Patients of Rett Syndrome are generally very happy and they are often called “Angels” as they have extremely expressive eyes and people say it looks as if they are looking into your soul.

# Congenital Adrenal Hyperplasia Support Group (CLIMB)



Congenital Adrenal Hyperplasia (CAH) is a metabolic condition, which affects the production of steroids from the adrenal glands. The three main types of steroids are, cortisol (which is the stress hormone), aldosterone (salt retaining steroid) and androgens (male hormones). Both boys and girls with the condition are exposed to an excess of androgens whilst in the womb and although this has no visual effect on male babies, the females are usually, to some extent, virilised and should therefore be identified at birth. These girls obviously have female chromosomes (XX) and have a normal uterus, vagina and ovaries internally. However, they often require surgery to correct the appearance of their external genitalia.

Boys are more difficult to identify. In CAH there are salt losers and non-salt losers. Salt losers generally become ill within the first 2 weeks of life (they suffer what is known as an adrenal crisis). Hopefully, they then receive the appropriate tests and are put on the replacement steroids required. If not, unfortunately, this is a potentially fatal condition. Non-salt losing boys are usually identified by tall stature and signs of precocious puberty between the ages of 2-6 years. These tall boys will have an advanced bone age which will have affected their final height potential and they will therefore be quite short adults.

All parents expect their children to be healthy and most are devastated to learn that their child has a chronic life

threatening disorder, which will require life-long treatment. The CAH Support Group was set up to help families with CAH sufferers and was formed approximately 11 years ago. It is a sub group of CLIMB (Children Living with Inherited MetaBolic diseases), which is a registered charity.

Both the CLIMB and the CAH Support Group exist to: -

- a) Give support to families and sufferers
- b) To increase awareness of the condition(s) to the public and to the medical profession
- c) To raise funds to support research.

The support group committee is made up entirely of volunteers and our current membership stands at approximately 350 families and 150 professionals. We hold conferences at regular intervals and a newsletter is sent out 3 times a year. Our key to success is getting information to where it is needed as soon as possible after the need is recognised; i.e. at diagnosis. Also we believe support at this time is crucial. We believe that the existence of support groups is very reassuring and the work they do is vital in helping to cope and come to terms with the future.

Fortunately, the treatment for Congenital Adrenal Hyperplasia has improved dramatically over the years and the outlook for those affected is extremely encouraging.

The current fee for annual membership is £17.50. This entitles members to a free

information pack, newsletters, reduced admittance fees to conferences (the next event will be on 18th May 2003, in London), support and advice and of course a vote at the AGM in selecting committee members and a say in how the Group is run.

## The Eyeless Trust

The Eyeless Trust was formed by Lillian Ramsay to help children born with ANOPHTHALMIA, MICROPTHALMIA and COLOBOMA.

Anophthalmia is the absence of one or both eyes from birth and microphthalmia indicates the eye or eyes so small and poorly developed that they can barely be seen. In anophthalmia there is total blindness but with microphthalmia the child sometimes has small amounts of vision. These conditions are not new but both are very rare having an occurrence of one in one hundred thousand births for anophthalmia and one in ten thousand births for microphthalmia.

Coloboma describes a situation where a child has a portion of the structure of the eye missing. This may occur in a range of areas and be large or small.

All three conditions can be in one eye or bilaterally. About 2/3rds of the children known to the Trust have one or more additional disabilities and therefore need a lot of support. The number of children with these conditions continues to grow and as the Trust becomes better known more families are referred to us.

The Eyeless Trust has a team of qualified Social Workers who cover England, Scotland, Wales and Northern Ireland and are managed by a Social Work Administrator. Well over 330 families are now registered with the Trust and the majority receive regular home visits.

**For more up to date information, please contact our website: [www.cah.org.uk](http://www.cah.org.uk)**

**Mrs Sue Elford** (Chairman), 2 Windrush Close, Flitwick, Beds, MK45 1PX

Tel: 01525 717536 E.mail: [Sue@cah.org.uk](mailto:Sue@cah.org.uk)

**Miss Melissa Cull** (Secretary), 17 Newton Road, Lichfield Staffs, WS13 7EF

Tel: 01543 252961 E.mail: [Melissa@cah.org.uk](mailto:Melissa@cah.org.uk)

# The Primary Immunodeficiency Association (PiA)



The Primary Immunodeficiency Association (PiA) is a national charity which provides information and support to people with primary immunodeficiencies and their families. Our aims are to promote awareness and early diagnosis of the various primary immunodeficiencies, to ensure that all those affected have access to the best possible treatment, and to encourage and support original research.

Primary immunodeficiencies are disorders of the immune system. The World Health Organization recognizes approximately 70 primary immunodeficiencies including X-Linked Agammaglobulinemia (Bruton's Disease), Common Variable Immunodeficiency and Severe Combined Immune Deficiency (boy-in-the-bubble disease).

People with primary immunodeficiencies have little or no natural defence against infections. Mostly genetically based diseases they are treatable, but as yet, often not curable. The year 2002, however, saw some pioneering advances in gene therapy, particularly in the treatment of X-linked Severe Combined Immunodeficiency (one of the most life threatening and severe forms of primary immunodeficiency and also known as 'baby in the bubble syndrome'). Little Rhys Evans from South Wales received extensive national media coverage last year as he was heralded as the first 'baby in the bubble' to be successfully treated with gene therapy. Most patients however, can only be treated with antibody replacement therapy, aggressive antibiotic therapies, and in a small minority of cases, bone marrow transplantation.



As many of our members depend on lifelong infusions of immunoglobulin, great importance is attached to surveillance of the international blood plasma industry to ensure the safety and availability of supplies of immunoglobulin. The PiA is a founder member of IPOPI (international Patient Organisation for people with Primary Immunodeficiencies) and a member of EPPIC (European Patient's Primary Immunodeficiency Collaboration)

The team of 5 staff at the PiA are led by the indefatigable chief executive David Watters, who is also Chair of the Jeans for Genes Campaign. Jeans for Genes provides much of the revenue pumped into gene therapy and other research projects which the PiA fund every year. The PiA has a medical advisory panel of clinical immunologists and a research Grants Committee which awards grants to doctors and other researchers looking for improved treatments and a cure for primary immunodeficiencies. The PiA offer an extensive range of services to their members

which include a helpline, a network of specific condition contacts which puts members in similar circumstances in touch with each other, a full-time benefits worker, regional days, workshops and holidays for our younger members.

A recent addition to our member services has been the recruitment and training of volunteers to visit clinics around the country where patients with primary immunodeficiencies receive their treatment. The aim of these visits is ultimately to raise awareness of the PiA as well as providing any additional help or support needed (publications, benefits advice, etc). The project, spearheaded by Jenny Jackson, is now underway and developing in great strides. Jenny has now made contact with nearly all the immunology centres in the UK and provided much needed 'field' support to members and gained valuable feedback from patients and

medical staff alike. The measure of her success has been seen in a substantial increase in membership over the past year.

Our membership base consists mainly of people who have a diagnosis of primary immunodeficiencies (currently around 1600 both in the UK and overseas), their family and friends, people in the medical professions, the media and other organisations.

As with many a small charity, member fundraising plays a vital role and supporting our hard-working members is high on the agenda. We plan to incorporate a fundraising section within our website this year which will contain new downloadable materials (including a new fundraising pack). It is certainly true that PiA members never seem to tire of walking, running, climbing and organising a multitude of events around the year to raise both money and awareness so we can continue our work. We are certainly proud of them!

**Our publications too have received some attention in the past year with improvements made to our quarterly newsletter inSIGHT and the forthcoming revised editions of our 'Understanding' series of booklets (due later this year). Many of our publications are readily available on the website.**

**If you would like more information about any aspect of our work please visit our website [www.pia.org.uk](http://www.pia.org.uk) or email [info@pia.org.uk](mailto:info@pia.org.uk)**

# A note from the editor...

A big thank you to all our members to attended The Royal Society meeting "The National Forum for Science 2003, the Peoples Science Summit" on 4th March 2003, which focused around the issue of "genetic testing". We had, I believe, over 30 GIG members from far and wide across the UK attending. It was an extremely busy and sometimes quite an intense day, but it was such a pleasure to meet a few more members of GIG, and also do a bit of networking and in doing so gain another member to GIG – The Anirida Network. This was a public forum and it was a good opportunity to meet lay members of the public, doctors, and researchers who had all come to participate in the debate about genetic testing.

We were all divided into working parties, to discuss a hypothetical questions which was posed to us "In 2023, every newborn is issued with a genetic birth certificate. What are the implications of this information, and what are the kinds of issues that we are or should be concerned about, given that this technology may be available by this time?" The day was structured extremely tightly and we did sometimes find that we simply didn't have enough time to go into an area to the extent we would have liked. However, it was extremely important that so many GIG members attended, as we were really able to stand up and push forward the issues that

affect those who suffer from genetic disorders, which for some people was a new area they had not come across or been aware of prior to this meeting.

After talking with some of GIG's members after the forum, I found that many had felt a little frustrated on the day, but did feel that it had been a worthwhile exercise, in raising awareness and bringing our message to others.

A report from this meeting is now available to read on the Royal Society's website, and all those who attended should also have received a hard copy. [www.royalsoc.ac.uk/scienceinsociety](http://www.royalsoc.ac.uk/scienceinsociety). It is a little lengthy, but if you make it to the back in the appendix you will see that the group I was in, Group 6, Ethics of Genetic Testing with Larry Winger from the XLH Network and Hannah James from Aniridia Network, has our recommendation at the top of the list...

## XLH Network – Internet forum

After meeting Larry Winger at the Royal Society he kindly asked Hannah James and me from the Aniridia Network to participate as guest speakers in an email forum for the week of 24th March. We were linked into the XLH Network email mailing list which has over 280 participants worldwide. Larry then kindly

introduced us and his members were able to ask Hannah and I questions regarding our organisations. It was certainly a very interesting week and I learned a lot from the members who were emailing back and forth, not only to Hannah and me but also to tell other members of the problems they had come across or any new developments with their treatments etc. I was also very pleased to receive an email from a UK member of the XLH Network who then kindly gave up some of her free time to attend the workshop we held on 28th April with the Cambridge Knowledge Park called "Public Consultation on Genetic Services" - which I have already spoken about in this newsletter.

## Another New Staff Member

I hope you will all join me in taking this opportunity to welcome Lucy Ullmann to the GIG team, Lucy replaces Claire Foster as our Administrator here and is coping well in her first weeks, coming to grips with the accounting and filing systems! Lucy is at present hiding from my camera, but eventually I will snap a shot of her and she will appear on the GIG website! If you would like to contact her at present she is still using Claire's email address which is [claire@gig.org.uk](mailto:claire@gig.org.uk) or you can reach her through the [mail@gig.org.uk](mailto:mail@gig.org.uk) **Melissa Winter**



## Congratulations

Pritti Mehta, GIG's expert in ethnic minority issues is marrying Jonathon Sanderson in July. The ceremony will be a fusion of Indian and European traditions (although apparently the groom has vetoed the

suggestion that he should arrive on a white horse.) I am sure you would all join me in wishing them both a long and happy life together and embracing our congratulations to the couple.

Instead of wedding presents, Pritti and Jonny are looking for sponsorship for a 470 kilometre cycle ride from coast to coast through Sri Lanka in which they are participating, in aid of the International Childcare Trust, who work in the UK and in developing countries, in direct partnership with local groups, to implement innovative and cost-effective development and relief initiatives, for the poorest members of the community. They are currently working in India, Sri Lanka and Kenya. Anyone wishing to sponsor them should send cheques to "Dr. P Mehta Cycle Challenge". This is a very good cause, so please be generous if you can.

**Alastair Kent**

# An Introduction

A few weeks ago, very soon after I started here at GIG, Melissa asked me to contribute an article to 'GIG Today' to introduce myself as the new Assistant Director, and to say something about my rôle. This task is pleasant enough and sounds relatively easy, but does require a degree of inspiration to strike at just the right moment. I don't think that I could get away with using the excuse of 'writer's block' for not being able to produce the goods – I am not JK Rowling and this is not 'Harry Potter and the Geneticist's Stone'!

It has not been as easy for me as I thought it was going to be to put pen to paper (or should we nowadays say 'fingers to keyboard?') to great effect to produce this article. Part of the problem of writer's block is not always about not knowing what to say or not having enough to say, but quite the opposite – having so much to say that, literally, one doesn't know where to begin! For me, this was very much the case when it came to writing about GIG and its activities (as well as my life at GIG since starting here!). It is also true to say that, although I had a very clear idea of what I wanted to say, how to make a beginning when writing it was a major problem.

I notice that it is now quite common on television for 'bloopers' and 'out-takes' of mistakes, fluffed lines and missed cues (sometimes probably contrived) that would previously have found their final resting place on the editor's cutting-room floor, to be used as amusing pieces in the final version of the programme that is broadcast. I suppose I could say that what is good enough for Delia Smith is good enough for me! Although I have had a fairly clear idea of all that I wanted to say here (while not always being clear about how I wanted to say it!), at one stage while grappling with how to begin this article, my mind actually toyed with ideas along the lines of: 'Fifty years ago, Watson and Crick elucidated the structure of DNA. At around the same time, and in a maternity ward not five hundred miles away, I was born. Little did I then realise....'!

Fortunately for us all, something resembling inspiration finally hit me, allowing my thoughts to be galvanised, arranged and organised to be able to tell you how enormously impressed with not only all that is currently happening in the world of genetics and the frenetic speed at which everything is happening, but also by the extent to which GIG is directly involved in so much that is happening. It is to GIG's great credit that much of what it does

and is involved in is directed to helping to ensure that the frenetic activity that I mentioned is not simply being allowed to be confined to abstract, exotic academic issues, but for it to also directly relate to, involve and engage real people and their lives, and across the community, and that the voices of real people, especially those who have encountered, or who live with, genetic disorders, have opportunities to be heard and have influence, whether it be on clinicians, scientists or politicians.

In many ways, my rôle at GIG is still evolving and developing, and it is not easy to be specific and exact. However, much of my time will be (and already is) spent liaising and working with GIG's colleagues in the Genetic Knowledge Parks, especially with regard to the various projects and initiatives that GIG is directly involved in. Particularly, I will be working with and providing management and support to Pritti Mehta in London, Anna Lane in Birmingham, Buddig Williams in Cardiff and to two colleagues yet to be appointed (at the time of writing) in Manchester. Much of the financial and budgeting dimensions of GIG's projects and activities seem to be becoming a significant part of my remit, and I will also be actively involved in providing management and support to colleagues within GIG's offices here at Leroy House, as well as deputising for Alastair at various times and in various ways.



I very much look forward to meeting more and more of you in the future, as and when our paths inevitably cross. If any of you feel that you wish to short-circuit this process and arrange for us to meet, and/or to discuss anything directly with me, I can be contacted at GIG's offices or by e-mail at [tom@gig.org.uk](mailto:tom@gig.org.uk).

**Thomas Barclay**  
Assistant Director

Back in March, GIG went on an outing to the London Eye. A good evening was had by everyone, and the views were splendid across London, I would certainly recommend it to you all.

(l-r) Pritti Mehta, Melissa Winter, Kavita Sangha, Maggie Ponder, Alastair Kent, Claire Foster, Parul Vansadia, Janet and John Gluckstein.

