

GIG is one of four charities to benefit from fundraising by the Royal Logistics Corps Band



A group photo with the four charities who benefitted from the bands performances

The Genetic Interest Group were delighted to be nominated earlier this year to receive a donation from the Royal Logistics Corps Band, based at the Princess Royal Barracks, Deepdene in Surrey.

This donation was made through Musn Paul Starbuck ATCL who was one of four band members nominated to choose a charitable organisation for the proceeds raised over the festive period by the band's performances.

Alastair and I travelled to Surrey to make a small presentation and to collect the donation. Amongst the other charities to benefit were the Frimley Hospital Children's Ward and Riding for the Disabled, who were very kindly given a box of old army boots.

Although this may seem a little odd, it was the ideal

donation to such a charity, as without the correct footwear disabled children cannot have riding lessons..

We are extremely grateful for the donation made to GIG, which is helping us to continue working to benefit those who are affected by genetic conditions.



Musn Paul Starbuck ATCL, third from left and Alastair Kent.

GIG TEAM GET TOGETHER IN MANCHESTER



(Back row) Johanne Ayres, Anna Lane, Buddug Williams, Melissa Winter, Lucy Ullmann, (Front row) John Gillott, Tom Barclay, Pritti Mehta

The GIG extended family got together for a group meeting in February of this year.

As always, it was a good opportunity for us to catch up on what we had been doing and to share information and ideas.

This was also the first meeting that Lucy Ullmann and John Gillott were able to attend.

We had decided for this meeting to discuss fundraising issues, and how GIG could generate further income and we were very grateful to Anna Lane for giving us an excellent presentation drawing from her wealth of experience in fundraising.

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

Directors Report Alastair Kent



The ideal that underpinned the NHS from the moment of its founding was that treatment should be based on need and free at the point of delivery. Few would argue with this sentiment, but for many this ideal has been more rhetoric than reality. Just recently there seems to have been some

movement. Hopefully the postcode lottery that has characterised access to genetic testing is showing signs of crumbling, to be replaced by a more equitable system that allows access wherever you live. The UK Genetic Testing Network Steering Committee (a body that represents the scientists that do the tests, the doctors who request them, the commissioners who pay for them and the Department of Health) on which GIG has a place, has agreed a list of tests which should either be provided by all DNA laboratories or to which samples should be sent for analysis wherever they come from.

The details can be found on the Department of Health website www.dh.gov.uk

This is an important step forward. Just as important, the list is not a closed one, so there is a mechanism for new tests to be added.

Another encouraging development is the news that we have been awarded a project grant by the Department of Health to roll out the work that Pritti Mehta has been doing to check that people from minority ethnic groups are actually getting to genetic centres for advice and support. Over the next three years GIG will be working with the regional genetics centres to provide training and support so that the professionals working in those centres can be confident that everyone who needs access to the services they offer is actually able to benefit - even those who are in traditionally "hard to reach" groups.

GIG's website www.gig.org.uk continues to go from strength to strength. Every month recently we have been visited by over 10,000 people - and this looks likely to rise when we put our teaching pack "Genes and You" on the site later in the

summer. This will be fully downloadable, and so will be available to teachers and lecturers wanting to explore genetics with their students. The Genes and You folder will continue to be available from the GIG offices, so long as stocks last, if you would like a copy just email Melissa or Lucy at (melissa@gig.org.uk or lucy@gig.org.uk) and they will put one in the post for you.

We have recently published a set of leaflets emphasising the important role of diagnosis as a route to help and support. Sadly there are still people "out there" who think that, because many genetic disorders are incurable, the need for a diagnosis is reduced. The stories in this new set of leaflets should soon correct this notion. These are available in paper format or on the website in a downloadable format.

We were very disappointed that our application for a Development Officer in Scotland has not been successful. Despite this setback we remain committed to establishing such a post and are approaching new sources of funding.

Elsewhere we continue to develop and expand our input through the Genetic Knowledge Parks and the Genetic Education Centre in Birmingham.

We have a new member of staff; Max Ullmann has joined us (part-time) to help run the office, boosting the support we have at the centre for our work - which, given everything that's going on, is a welcome boost.

Looking ahead, the summer will see the Human Tissue Bill complete its route through Parliament. GIG has been lobbying hard to try and ensure that the interests of families with genetic disorders are properly represented and taken account of. Some of the developments outlined in last year's white paper on genetics are now taking shape and we continue to be involved with many of these. The number of orphan designations continues to rise steadily at the European Medicines Agency and new treatments for rare disorders are being granted licences quite regularly. Making sure that they are available for all who need them will be a growing challenge as Primary Care Trusts struggle to fund new developments, meet government targets and do everything that is expected of them. This is an issue that we will be keeping a close eye on.

Finally, I would like to take this opportunity to thank all our members for their support and encouragement. We value it



VCFS Family Support Group Saturday March 27th

Report by Anna Lane, Development Officer, GIG and West Midlands Clinical Genetics Unit

Leaving behind grey menacing skies, I approached Gloucester to a wash of pale blue sky with not a hint of rain. I was looking forward to spending some time with families belonging to the VCFS Family Support Group for those with Velo-Cardio-Facial (Shprintzen) Syndrome and Di-George Syndrome and 22q11.2 deletion.

Velo-Cardio-Facial Syndrome is a genetic condition for which it was only possible to test in 1992, but which is believed to affect 1 in every 2-3,000 live births (the second most common cause of congenital heart defects and learning disorders after Downs Syndrome).

Families brought food to share and the day was packed with fun for the children, including a seasonal Easter

Egg Hunt! Susie Mitchell, a parent herself and co-organiser of the group, told me that they have a hectic calendar of events and always try to do something that children with a range of abilities can enjoy. The recent Pony Riding activity was hugely successful and is now being offered on a regular 6 monthly basis. Future plans include activities for teenage members in addition to those

European Health Day 2004 Tom Barclay, Assistant Director



Over the past few months, I have attended several meetings in Brussels of the European Health Forum involved with the organisation of European Health Day. The objective of European Health Day is to provide a platform for an exchange of ideas and views of different stakeholders of the European health community. The aim is to

particularly involve those who are not normally part of the 'EU circuit'. The event will offer the participants an opportunity to hear the views and visions of the European policy-makers and other actors in the health field, and to network with other organisations.

In the longer term, European Health Day will highlight particularly important health-related issues, and be open to all interested parties. It could be linked to the date of World Health Organisation's World Health Day, which is usually scheduled to take place in April.

However, the format of the first European Health Day is unlikely to be the final style of the event in future years. Rather, this first event will be a "transitional event", and therefore its scope and duration and the number of participants will, in this instance, be limited in order to be able to produce a good quality event within the very limited resources and time available. The very modest budget that was finally made available for the event also places considerable restraints on what is possible, and I feel sure that future European Health Days are likely to increasingly become the exciting, interesting as well as influential events that they deserve to be.

At the time of writing, the event is planned to take place on Monday, 17th. May 2004 at the Charlemagne building, Brussels. However, this date should be regarded at the moment as somewhat provisional.

In order to achieve a well focused, one-day event, one main theme has been selected, under which various related items will be discussed. Given the planned timing of the event within a few days of the forthcoming enlargement of the EU, "Health in an enlarged Europe" has been selected as the overarching theme as a welcoming gesture that will engage and enthuse not just participants from existing member

for younger children, for example, an evening of bowling.

I had been invited to talk to the group and agreed to present information on the subject of 'Participation and Partnership - Patient-Centred Services'. This led to some lively debate and discussion and most people there had a positive view toward ensuring a step in this direction. Of course, the key is to get involved. The ideal for the future is that patients and the

public will 'share a seat at the decision making table'.

By the end of the afternoon I found myself to have been a guest at a very special gathering of like-minded people who share a keen sense of fun and commitment to empowering their children to reach their full potential. I felt privileged to have been invited to this quiet enclave where so much mutual support is being given so generously.

countries but also those from the new member states. Furthermore, as 2004 will be an important transitional year for EU institutions and, potentially, for European health policy, this theme will provide various speakers with an opportunity to discuss the EU's role in health and cast a look at experiences of the past and perspectives for the future.

Given the size of the plenary room available, the overall number of invitees cannot exceed 400 people. The invitation of participants will be done in parallel both by personalised invitations and by placing an open invitation on the Public Health web-site. The latter group will be allowed to participate on a first-come, first-served basis. GIG has been able to nominate a very small number of suitable groups with pan-Europe interests who may receive personalised invitations.

An outline programme for the event has been agreed:

Opening of the conference and keynote speech:

09.30 - 10.00 Opening of the conference and keynote speech:

"Experiences of the past, perspectives for the future"
Commissioner Byrne

10.00 - 12.30 Plenary session: panel of ca. 8 speakers, chaired by Mr Byrne

"Role of the EU in an enlarged Europe of health"
"Developing the health community in enlarged Europe"
(10 min/speaker + 55 min for questions and discussion)

12.30 - 14.00 Lunch

14.00 - 17.00 Parallel sessions (3-4 speakers/session, 20 min/speaker, time for discussion)
Topics will include the following:

1. Healthy lifestyles in an enlarged Europe: Role of the voluntary section, health promotion, role of health professionals/NGOs...
2. Health and internal market in an enlarged Europe: Health professionals, pharmaceuticals, health services...

Final Plenary Session/End of conference

After the conference, it is proposed to post the conference papers on the EU's public health web-site http://europa.eu.int/comm/health_en.htm. At the time of writing, there was also the possibility that the event would be broadcast in real-time by video-stream on the same website. These will include speeches and presentations given by the various speakers, photographs of the event and minutes/conclusions of the parallel sessions, drafted by the rapporteurs. A list of participants may also be published.

I hope to be able to report upon the event itself in a future edition of 'GIG Today'.

Tuberous Sclerosis Association

I was very pleased to meet Janet Medcalf and Anne Carter of the Tuberous Sclerosis Association in February of this year. They very kindly gave me a brief insight into the association and the work that it carries out.

Melissa

What is Tuberous Sclerosis?

Tuberous Sclerosis is a genetic disorder that affects 1 in 6000 of the population and is associated with tumours in any organ on the body, but mainly the brain, kidneys, skin, heart and lungs. The brain tumours cause the epilepsy, learning disabilities, autism and attention deficits that create so many of the problems for those with Tuberous Sclerosis and their families.

This condition was first described by Bourneville in 1880 but until the 1980s, there had only been sporadic research into the condition.

Tuberous sclerosis (TS) is also known as tuberous sclerosis complex (TSC) due to the complexity of the condition. It is difficult to explain because it affects

individuals in many different ways and with varying degrees of severity. Some children and adults with TS remain largely unaffected and go through life free from symptoms, whilst others who are less fortunate experience one or more of a range of symptoms.

- 85% of sufferers carry one of the two identified genes, TSC 1 on chromosome 9, discovered in 1997, and TSC 2 located on chromosome 16, discovered in 1993.
- There is an 80% chance that people who are affected by this condition will have had a tumour on their kidneys by the age of 10.

The Tuberous Sclerosis Association (TSA) was launched in 1977, by Ann Hunt (who is also now a trustee of GIG) who became the secretary and Esther Galbraith who was the founding Chair. Since its beginnings, the TSA has grown into an established organisation which supports sufferers, promotes awareness, and seeks the causes and best possible management of Tuberous Sclerosis (TS). There are now over 1700 families who belong to the association.

To date the TSA has raised over £7million for research into the causes and the symptoms of tuberous sclerosis and for family care and support. This challenge has been met by a team of dedicated workers who began, as many charities do, by volunteering their time and skills.

The TSA now has eight employed members of staff. Janet Medcalf is Head of Support Services. Janet is the first

point of contact for new and existing families, she is able to offer support, advice to new and existing member families and offer help to those who need it, she also co-ordinates a team of Specialist Advisors. Anne Carter works as Head of Appeals and Publicity and both have been involved with the TSA virtually from the beginning, first on a voluntary basis and then as paid members of staff since the early 1990's. Both Janet and Anne have daughters affected by this condition and Ann Hunt is now Head of Research.

Supporting Members

A key area that the TSA have been integral in, has been the establishment of medical clinics around the UK for people with Tuberous Sclerosis. There are now six centres offering services to children and adults who are affected. Recently the TSA secured funding for a third Specialist Advisor to help share the work of Harriet Spencer and Janet Bower. This will help enormously as each



Janet Medcalf, Head of Support Services



Anne Carter, Head of Appeals



Anne Hunt, Head of Research

advisor will be covering a smaller region and will therefore be able to meet and help more people within their own areas. At present the distances that are a necessity can often mean that staff are unable to meet in person as many people as they would like to. The Specialist Advisors have been enormously successful and both Janet and Anne feel they were one of the best investments that the organisation had made. The TSA have three established local support groups and are hoping to expand these to other areas in the near future.

Events

Professional and parent study days have been going since the late 1980's. They are aimed at doctors and other medical and health professionals, as well as teachers and social workers. The TSA usually arrange accreditation for such events where possible. Most of the Professional Study Days have been multi-disciplinary –covering a wide range

of issues such as epilepsy, kidneys, skin, genetics, behaviour, development. More recently, they concentrated on a single aspect; neurology, which enabled the day to go into greater depth. The next professional day they hope to organise will be one on behaviour.

The TSA try to organise at least one family day a year, following a similar format to the professional days but, as with many charities, this can be dependant on the funding that they receive, but they feel very strongly that it is important to have such days as it really gives people a chance to air their concerns and also to learn from others. Many of the events are over a weekend and are in places such as Center Parcs, where people can relax and enjoy themselves in the company of friends. There are now established groups such as the "outlook group", which is specifically for over 18 year olds who are only affected mildly with TS. With such a diverse group of people, it has been very beneficial to do this. The "outlook" members have also agreed to help chaperone other members in a weekend organised for those with moderate disabilities.

The other part of their events calendar is of course the fundraising events. The TSA organise golfing days and awareness days. They are also very keen to get their members fundraising too and have set up local fundraising groups so that members living near each other can campaign within their local community.

Research

Much of the research that has been carried out into TSC is genetic. The TSA funded research into the cloning of the two genes responsible for the condition and helped in getting a DNA test for those with TSC. Another important area for the TSA is the research that they have funded into defining the very serious cognitive, behavioural and emotional problems that many of their members suffer from. A TSC renal registry has been established which is collecting data on the renal problems

that people suffer from. One of the most recent studies is a longitudinal study looking at the clinical problems of a group of children with TSC, who were diagnosed in the UK from 2001 onwards. This is the first study of its type that has been carried out for TS and the association are, naturally, keen to gather the findings that may well have an impact on the lives of its members now and in the future.

Linking with Others

As the symptoms of this condition are so varied it has been an enormous help to the TSA to link up with other charities who represent conditions such as Epilepsy, Autism and kidney

Key developments for the TSA

- Finding the two TSC genes
- Involving siblings within the group
- DNA Testing
- CVS now being available.
- Recognising Autism as part of TS
- A raised level of understanding and acceptance of TS
- More people are being diagnosed and the symptoms are being picked up faster and more often.
- Improvements in behavioural care

problems. It has been an extremely positive experience to share information and ideas with other groups and to also work together in research projects where both parties benefit.

The TSA are very pleased to have a long standing relationship with GIG. There are many issues, especially the more over arching genetic issues that they, as a group, need to respond to but often feel they simply don't have the time to. So by extending their networks to organisations such as the Genetic Interest Group they are able to work in the most efficient and cost effective manner.

Haemophilia Society News



The Haemophilia Society has recently made five small grants covering a range of issues from pain management in haemophilia and the Factor VIII gene, to testing of young carriers and information provision for siblings of children with chronic health conditions. Launched in 2002 in adherence with AMRC guidelines, the Society's new research fund was made possible thanks to the Society's acceptance as a guest charity of Jeans for Genes.

Jeans for Genes, a national appeal organised by four main national charities and partnered by four guest charities, raises funds for research into serious genetic disorders affecting thousands of children. Donations also help to fund valuable advice and support services for families. The idea is simple: wear your jeans and pay £1 for the privilege.

Members of the Haemophilia Society were keen to get involved and have helped raise awareness of bleeding disorders, as well as other genetic conditions, by acting as case studies for education packs, talking to their local media, and even presenting directly to schoolchildren about their experience of living with a long-term medical condition. Many came up with some innovative and exciting ways of raising extra funds on the day too!

The deadline for the Society's next round of small grants for medical, scientific and psycho-social research studies in the UK is 29th February 2004.

For more information visit www.haemophilia.org.uk or www.jeansforgenes.com.

What's happening in Wales?

geneteg feddygol
GWASANAETH | GYMRU



medical genetics
SERVICE FOR WALES



More than half of the £1.5 million cash injection from the Welsh Assembly Government (WAG) to the Wales Medical Genetics Service has been allocated. Neurogenetics will receive funds to secure a consultant post following Professor Peter Harper's retirement later this year. Genetic counsellor posts around Wales and support staff will also be funded, strengthening the future of Neurogenetics in Wales. The Cancer Genetics Service will also receive money to support its work. Another consultant post, genetic counsellors and support staff posts will be funded and will enable the Service to deal with increasing numbers of referrals.

Alongside the clinical work is the laboratory

service. It also received funding to offer testing for certain types of colorectal cancers. Other awards made to the laboratory section of the Service include new tests which have a quicker turnaround time for results, which may help alleviate anxiety levels for families; laboratory staff and new equipment to carry out chromosomal investigation to help make diagnoses in children with certain learning difficulties.

The remaining portion of the WAG money will be awarded to other projects within the Service later this spring. GIG is involved in this process and will be working to make sure the money will provide the best patient and family care.



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In March, the **Wales Gene Park** held a successful two-day meeting on ethical issues that occur during social research on family issues. Those participating were from Cardiff University's Genetics and Society group and the Centre for Family Research, Cambridge University.

The aim of the meeting was to discuss particular issues and areas of possible concern that researchers may come across when interviewing and talking to participants. Talks given included different perspectives within the research process.

Maggie Ponder (GIG) and Claudia Downing (Cambridge) talked about the perspective of the interviewee, what people may think of research and their possible reasons for

participating. The discussion also focussed on the importance of good practice of research and the important considerations to remember when asking people about personal issues in their lives.

Liz France (Wales Medical Genetics Service) presented the group with her experiences of working as both a professional within the healthcare service (genetic counsellor) and her own active involvement in research, in particular interviewing people with cancer.

Smaller groups convened to discuss particular topics in further detail. Some issues raised were potential closer links between social scientists and research ethics committees to understand each other's work. Another point of discussion was

about training for less experienced researchers - should there be practical resources available?

All the groups who attended were keen to continue the good work and interesting discussion that came out of the meeting. Over the next few months Flo Ticehurst, Wales Gene Park's Genetics and Society Research Officer will be co-ordinating a report on the event and continuing the work from the two days. If you would like some more information, please contact Flo at the Gene Park address below.



small group discussions at the Wales gene park research seminar

Neurological Services Review

A review of healthcare services for all neurological conditions in Wales is currently underway. The review, which is underway until June this year, is commissioned by the Welsh Assembly Government. The aim is to get an accurate picture on the current situation for affected individuals and families. By identifying shortfalls in the service, opportunities to improve this may, hopefully, be recognised. The Wales Neurological Alliance is working hard to promote the needs of individuals, families and carers during this consultation period. The WNA is a group of charities (including GIG) working together to promote the needs of all

people affected by neurological conditions in Wales.

If you or a member of your family is affected by a neurological condition, or if you are a carer and living in Wales and you would be interested to have an opportunity to have your say, please get in touch with me in the near future, I would be very pleased to talk to you.

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Progress in Gene Therapy for Cystic Fibrosis

2004 is the 40th anniversary of the Cystic Fibrosis Trust. We look to the future with hope, whilst never forgetting those whose lives have been lost to CF. 40 years ago, the prognosis for those with Cystic Fibrosis was bleak. Most died in infancy and few made it to school.

Research and an improved understanding of how to treat the symptoms of Cystic Fibrosis have changed the outlook for those with this condition markedly. The average life expectancy is currently just over 30 years and children with CF do well at school and indeed in higher education. However, this achievement has not been painless for families and individuals. All we are able to do at present is to hold back the progression of the symptoms of CF for a while. These symptoms all stem from the inherent genetic flaw which causes CF, that of a fault in the mechanism for producing mucus. We all produce thin, lubricating and anti-bacterial mucus to protect and support our vital organs. In those with CF, this mucus is thick, congealing and attracts infection. Eventually, key organs become clogged, heavily infected and non-functional. The gut and the lungs are the

main organs to be affected. 90% of those who eventually succumb to CF die of lung disease.

To control the volume of this unhealthy mucus and to limit its potential for damage has become a life-long and daily battle for those with CF. Hours of daily treatment, including physiotherapy, a huge drug intake, oral, nebulised and intravenous and dietary adjustments have to be accommodated in everyday routines, as well as regular hospital and in-patient stays. Eventually, this treatment begins to be less effective and ultimately fails, at which point the only option is a lung transplant.

Gene Therapy offers hope, not only of a normal length and quality of life, but of a life free of this gruelling and burdensome regime of treatment, which has to be done relentlessly. Not all families can cope with this and many individuals rebel or just ignore their CF, especially during adolescence.

The Cystic Fibrosis Trust has had to give very serious thought as to its priorities, especially given the fact that we could effectively spend every pound we raise many times over. We have had to consider other research possibilities and

what they might offer, our contribution to improved NHS clinical care, which has amounted to over £10 million since 1997, and our direct support to families and individuals coping with CF.

We have decided to prioritise gene therapy research whilst protecting family support. This is because no other research offers better significant improvements to the length and quality of life for those with CF than gene therapy in the foreseeable future. Whilst there are, of course, no guarantees about gene therapy, we have strong and reasonable expectations that it will be effective. We have had to recognise, though, that success in this area is only possible as a result of a detailed and focussed programme of research, systematically tackling each of the obstacles currently facing us.

To do this, the Cystic Fibrosis Trust has facilitated an arrangement whereby the three world class research teams in the UK, who were working as competitors, will work as a consortium. Although geographically spread out, being based in Edinburgh, Oxford and London, they act as one, having formed a virtual laboratory and acting as a problem solving team to achieve very clearly

defined objectives, those of

- Identifying a clinically relevant gene therapy formulation within three years
- Carrying out phase I proof-of-principle and phase II dosing human CF studies within the following two years
- At the end of the five year period, to be negotiating with an industrial partner for a phase III study to assess clinical benefit in CF subjects.

To do this, they are addressing key problem areas, including the best vector, the most appropriate delivery route and viable ways of measuring gene delivery and its effectiveness. This, of course, all has to be set against a background of ensuring safety.

Results so far are encouraging, although the obstacles still to be surmounted cannot be underestimated.

We hope to be able to announce

significant progress within five years. It is possible that effective gene therapy treatments will be identified even sooner, whilst we also have to accept the possibility that some obstacles will hold back progress for longer.

We are sure about two things. The first is that it is only a matter of time before gene therapy for Cystic Fibrosis becomes a reality. The second is that without a focussed and detailed research programme, it will take much longer.

To find the initial £15 million for this research, over and above our other commitments, is an enormous challenge for the Cystic Fibrosis Trust. We are not finding it easy. The prize, though, is priceless. A life free of the life threatening lung disease that plagues those with CF.

Reality Not Hype: the new genetics in primary care

A joint Knowledge Park event held in London 30th January 2004

This event was a joint venture organised by the six knowledge parks, London, Cambridge, Manchester, Newcastle, Wales and Oxford.

We began the day in a filled-to-capacity lecture theatre at the Royal College of Physicians where we were introduced to the clinical context of genetics in primary care. This really gave the delegates, who were mainly practicing GP's, a quick introduction to genetics, the prevalence of genetic disease, a swift overview of genetics in relation to insurance and pharmacogenetics. Ian Hopkinson noted in his talk, about prevalence of genetic disease, that one of the difficulties he faced was that GP's were always telling him "All this genetics was all very good but it's always to do with those rare disorders, we never see anything like that in our own practice" However, researchers working with Ian Hopkinson gathered data on genetic prevalence of genetic disease in North

London across several general practices. They reviewed 1823 sets of notes out of which 189 contained genetic diagnosis. (Using the criteria of a) single gene disorders and b) Multifactorial conditions where a person had 2 or more family members who had had the same condition). If these results are generalised, then roughly 10% of all patients seen have a genetic component.

We then broke for coffee, giving everyone time to digest all the information, catch up with colleagues and replenish their brain cells for the second part of the day. This second part was rather smoothly presented by members of the Gene Park team from Wales, who spoke about the implications for Clinical Practice and how they communicate risk to their patients, what the definitions of risk are and in reality how patients interpret the risk they are told they have. People were surprised to hear that, research of people who had been placed,

following genetic counselling, in a low risk category, showed they still believed they were, in fact, at a high risk. Many wanted to be at a higher risk so they could be monitored more closely. They gained a sense of security from this.

In the afternoon the delegates were split into workshop groups. There were four workshops altogether and everyone had the opportunity to attend at least one. The issues covered were: Cardiovascular Genetics, Cancer Genetics, Ethics of predictive testing and Anti Natal Screening.

Full reports on each of these breakout sessions are available on the London IDEAS Genetic Knowledge Park website www.londonideas.org

I attended the Ethics of Predictive testing workshop followed by the Ante-Natal Screening workshop. Pritti Mehta was asked to minute the latter session on Anti Natal Screening, so I will let you read her account of both sessions on the

London IDEAS website at <http://www.londonideas.org/internet/>

Dr Ainsley Newson chaired the workshop on Ethical issues in predictive genetic testing. She gave the group a whirlwind tour of ethics and the areas we should be focusing on for the debate. The group was then presented with a scenario and, in smaller groups, discussed the options that we felt were open to us and what we should, ethically, do.

In our two groups we were to answer one of the following questions.

1. Should genetic tests such as that for BRCA1 and BRCA2, which have a low clinical validity and no effective intervention, be available? For anyone who wants it? Who should provide the testing?

2. Imagine that Gloria carries a BRCA1 mutation. Rachel (the GP) also treats Gloria's 19 year old daughter Amy. Amy is worried about her family history of breast cancer and is interested in finding out her risk; however she is not aware that her mother has been tested.

How do you approach this problem?

We then came back together for one

final debate. Although we could have spent much longer in discussion this was a great opportunity for the group, who were mainly primary care workers, to look at an issue from a different angle. It was also an insight into looking at the issues from an ethical point of view and seeing what the outcome of that would be.

I was in the group answering question one. We decided that these tests should be made available, because if they were not made available through the NHS here, then people would simply buy them over the internet and they may not have the necessary support systems in place to help them deal with the consequences of such knowledge. Also we decided that you could not deny people a test if one was available. This test, we felt, should not be made available to everyone, at present, as there was not enough evidence that it would help. Health professionals should provide this test as they have access to support networks for the patients; if people buy tests over the internet they may not know where to turn to for information and advice following the results.

Scenario

Breast Cancer

Rachel is a GP in South London. She has been treating 45 year old Gloria (and her family) for 15 years.

Gloria reports that several members of her family have developed breast cancer in recent years and that a mutation had been identified in her sister Claire.

Gloria expresses interest in being tested. Rachel refers Gloria to a clinical genetics service for counselling.

Facts

5-10% of all breast cancer/ovarian cancers are caused by a genetic mutation

A woman with a mutation in BRCA1 or BRCA2 is at 50-80% lifetime risk of developing breast cancer.

If a mutation is detected in a healthy person:

Effectiveness of surveillance/intervention is still under debate

Increased screening?

Chemo-prevention?

Prophylactic mastectomy?

GENES AND LIFE - NEW INTERACTIVE VIDEO CD LAUNCHED with a 20% discount offered by Primed Communications to GIG members

Genes and Life is a new, concise, public-education, interactive video-programme on CD, produced in partnership with the University of Central Lancashire and Roche Genetics, a unit of F.Hoffman - La Roche Ltd. It has been produced to offer a practical perspective on the significance of recent progress in the field of genetics and medical science.

This is a timely programme, given the increased public interest in genetics and in particular the role our genes may play in our health and future medicines. It is designed to offer guidance and support to healthcare professionals as well as the general public in order to give a better understanding of how genes can impact on health.

The program is divided into three main sections, Genes and Health, Genetic

Tests and Genes and The Future of Healthcare and offers answers to questions such as:

- Why should we know more about our genes?
- How might my genes impact on my health?
- What is a genetic disease?
- How can I find out if I am at risk of developing a genetic disease?
- Should I test for a genetic disease?
- How might genes improve the safety and effectiveness of medicine?
- How might genes influence the development of more targeted medicines?

The programme is designed to provide short sequences of information in such a way that common questions are addressed in a simple and appropriate

manner for people aged from 15/16 upwards, with little knowledge of genetics. This video CD, which runs on a computer, has been reviewed by healthcare professionals and members of the general public throughout the development stage with very encouraging feedback.

Should you wish to order a copy we are pleased to say that the company behind the production of the CD has offered to give 25% of each sale through this newsletter to GIG. Please use the coded flyer inserted in this newsletter.

For further information on the CD please contact info@primed.co.uk or call 01483 861300.

Having A Rare Genetic Condition: The Baffling World Of Setting Up A Support Group

My personal story by Emily Sole

"Multiple Endocrine Neoplasia Type 1! What's that?" I remember saying. I was not ready for what I was about to be told. In fact I wasn't really sure if I wanted to hear about it either as it didn't sound particularly pleasant and I was the one who had this strange sounding condition. "It's a rare, hereditary genetic condition," the doctor explained. The DNA test had come back positive: a fault on one of my chromosomes meant that this was my result. I'd done science at school but had never been very good at biology and now had to try and remember what chromosomes and genes were. My head was spinning.

I was lucky the condition was found really as it was just a routine blood test that had picked up raised calcium and prolactin levels. After I was referred to an Endocrinologist, he had put two and two together and realised that something was not quite right with my endocrine system. He had suggested I go for this genetic test. "Your mother or father will have the condition," the doctor stated. Now this was a blow. So it wasn't just me. "We'll have to test your family."

THE FAMILY IMPACT

It turned out that I had inherited this condition through my Mum. At first she felt guilty but it wasn't her fault, she had lived most of her life without realising this condition was there. Then came the triple whammy: both my brother and sister had had inherited it too. It had been a 50% chance of us getting the condition but unfortunately all 3 of us had inherited it. It was all in our 'genes'.

A genetic counsellor came to talk to us, to give us support and reassurance. Looking at her notes she said "Multiple Endocrine Neoplasia Type 1! What's that?". So we explained to her what we had found out already. It's easy to understand once it's broken down into chunks: multiple (lots of), neoplasia (new growths or tumours), endocrine (in your

endocrine system), type 1 (one of the types, type 2 being quite different).

Our Endocrinologist was really nice and helpful. He had explained that you could, and probably would, get benign tumours in your endocrine system and that these would mainly be in your pancreas, pituitary and parathyroids, and sometimes had the potential to become cancerous. He had also explained that you would not necessarily get tumours in all three or in any particular order, except that your parathyroids would probably be first. We had also started to look on the internet although this had been quite scary as it talked with more certainty about cancer and the possible risks/symptoms. All the tests, scans, treatment and later surgery, soon proved that we needed to increase our knowledge about MEN.

THE BEGINNINGS OF A SUPPORT GROUP

All the time we were thinking: where are the other people with this condition that we can talk to? Confidentiality meant that our Doctors couldn't give out the names of other MEN sufferers so it was difficult to find others to talk to. It was said that about 1 in 100,000 had the condition in the UK so there had to be more out there, it was just a question of locating them.

It was while we were trawling the internet for information that Mum came across a Canadian MEN Society and contacted them through numerous emails, letters and phone calls. We could finally talk to others with similar problems: what a relief! Why wasn't there such a support group in the UK, we wondered? These people were so chatty and friendly and made us feel so much better and confident about the future. We no longer felt alone. That was when we realised that a UK support group was needed. The more knowledge we could get about the genetic conditions of MEN 1 and 2 to

help ourselves, and others, the better.

We were invited over to Canada to stay with the family who set up the Canadian MEN Society and a Great Aunt helped to finance the trip covering the much needed cash for the travel costs. What a fabulous learning experience it turned out to be. The society had a whole list of people with MEN from throughout Canada, the USA and even New Zealand, far more than we had ever anticipated.

It was great to learn from their experiences: how they had set up their society; the difficulties they had faced and how they worked through them. Ultimately we worked out how we could make a way forward together. We forged an alliance with them and from then on saw the creation of the UK MEN Society (later to be called AMEND - Association of Multiple Endocrine Neoplasia Disorders) and a joint website.

THE WORK BEGAN

Once back home we had a lot of work to do. It was hard at first to know where to start. Initially, we checked with our Endocrinologist and other key MEN figures who gave us their backing and provided us with the encouragement we needed to get it up and running. The help of a local retired businessman further assisted us. He set us targets to meet and created the website. My Mum produced a leaflet about the conditions and the aims of the support group, and a friend designed a logo for us to use. We attended conferences, wrote letters, liaised with Medical Professionals, joined other helpful support groups and started to gather lots of information about MEN.

However, it was not until we placed an article in the Pituitary Foundation Newsletter that we began to get people contacting us. It had taken a long time, a lot of hard work and effort, and a trip to Canada, to get to that point but now we could finally contact others in the UK and start to offer help and support for those

with MEN.

AMEND is currently growing all the time. We gained charity status in October 2003 and our Trustees include the leading UK MEN Researcher, an Endocrine Consultant and Surgeon. AMEND has attended a number of Endocrine Consultant and Surgeons Conferences and hopes to attend the International Conference MEN2004 in Bethesda, USA in June of this year. We provide regular Newsletters for our members, are in the process of producing detailed patient information booklets, and are set to hold our 2nd Annual Assembly/AGM in July.

We could not have got to this stage

without all the help given by volunteers, consultants, surgeons, nurses, pharmaceutical companies, the Canadian MEN Society, other support groups and all the kind people who have provided assistance along the way. We still have a lot of work to do: spread the word so that others with the conditions gain support and information; inform GP's and specialists about the support group and MEN; keep up-to-date with recent medical/surgical advances; and ultimately increase the awareness and understanding of all aspects of the conditions. Our main goal however, remains the same: to provide support and understanding to those with the rare

genetic conditions of MEN types 1 and 2, their families and carers.

For further information about MEN 1 and 2 please contact

Liz Dent

Association of Multiple Endocrine Neoplasia Disorders (AMEND)

Lyndene

The Green

Bewerley

Harrogate

HG3 5HS

Tel: 01423 712131

liz@mensociety.com

<http://www.amend.org.uk>

NEWS AND EVENTS.

We would like to invite all GIG members, friends and family to this, GIGs first major fundraising event.



CASINO NIGHT

9th July 2004

7pm till 11pm

The King Alfred School, London NW11

**£12.50 entrance in advance/ £15 on the night
including a buffet dinner and your first set of chips**

Events on the night will include, blackjack, roulette, live music, bar and food.

If you would like more information or to purchase tickets please send an email to casino@gig.org.uk or contact Lucy or Melissa on 020 7704 3141



A-T Society Family Day Saturday 8th May 2004

Ataxia-Telangiectasia (A-T) is a rare, neurodegenerative, inherited disease which affects many parts of the body and causes severe disability. Ataxia means poor co-ordination and the Telangiectasia are extra blood vessels which can be seen, especially on the whites of the eyes.

A-T is progressive and affects the cerebellum (the body's motor control centre) and in about 70% of cases, weakens the immune system as well,

leading to respiratory disorders.

A-T first shows itself in early childhood, i.e. the toddler stage. The symptoms are lack of balance, slurred speech and perhaps a more than normal number of infections. All children at this age take a little while to develop good walking skills, coherent speech and an effective immune system so it may be some years before A-T is properly diagnosed.

Because A-T is such a rare disease, families who are affected often feel isolated. Our family meetings provide an important opportunity for families to meet together for mutual support and to gather information both from each other and from our speakers. We provide speakers in the fields of A-T research, clinical management, disability management and disability rights.

At our meeting on 8th May 2004, speakers will include:

- Professor Malcolm Taylor, University of Birmingham,
- Pauline Pope, Consultant Physiotherapist,
- Louise Hickson from the Willow Foundation will talk about special days out for young adults with A-T.
- David Owens, Trustee,
- Derek Sinclair from "Contact a Family" will talk about benefit issues.

For further details of this year's Family Day, please contact Kay or Nick:

Tel: 01582 760733.

E-mail: atcharity@aol.com

NEWS AND EVENTS.

ASSERT

Angelman Syndrome Support
Education and Research
Trust

Bi Annual Family Conference
3-5 September 2004
Loughborough University

Topics: Education/ Epilepsy/ Genetics/
Communication/ Transition/ Wills and
Trusts/ Alternative Therapies

For more information please contact:
ASSERT, PO Box 13694, Musselburgh,
EH21 6XZ
01268 415940
email: assert@angelmanuk.org
www.angelmanuk.org

The Fragile X Society

Fragile X Society
Family Conference
Saturday 12 June 2004
in Birmingham
Topic: Fragile X and Communication
and workshops

Fragile X Society
Family Conference
Saturday 2nd October 2004
in Belfast
Topic: Genetics of Fragile X

For further information please contact the
Fragile X Society, Rood End House, 6
Stortford Road, Greatr Dunmow, Essex
CM6 1DA Tel: 01371 875100

Prada Willi Syndrome Association

Multidisciplinary Training Day on
Prader-Willi Syndrome
Monday 5th July, 2004
Hayes Conference Centre,
Sawnnick, Derbyshire.

Talks and discussion groups aimed
at professionals in the fields of
health, social services, residential
care and education, who would like
a broader knowledge of PWS.
Parents/Carers also welcome

Further details from:
PWSA (UK)
125a London Road
Derby
DE1 2QQ
Tel: 01332 365676
Email: [admin@pwsa-
uk.demon.co.uk](mailto:admin@pwsa-uk.demon.co.uk)

Ataxia - Telangiectasia Society International Conference

University of Birmingham
14 - 15th October 2004

Continued Professional
Development approval from the
Royal College of Physicians worth
13 CPD points are available for this
conference. .Please contact:
Maureen Poupard
Tel: 01582 7607
Email: atcharity@aol.com

Gorlin Syndrome Group

new contact details
gorlin.group@btconnect.com or
info@gorlingroup.co.uk -
www.gorlingroup.co.uk

Do you need help with your accounts?

Tony Matassa a registered chartered
accountant with extensive experience in
the voluntary sector would like to offer
his skills and experience in this area to
our members. Auditors can be an
expensive resource for charities and you
may be able to benefit from using this
freelance accountancy service. The
following are areas of work that can be
typical to many smaller charities and in
which Mr Matassa is experienced in
professionally: -

- The recording and reporting of
restricted funding and fund analysis
- Gift Aid tax reclaims
- Providing advice and support on
regulatory obligations in compliance
with SORP and The Charities Act
- Setting up accounting software such
as Sage Line50, and payroll systems
- Preparations of accounts for audit
- Budgeting and cashflow forecasting
- VAT related issues where charities
have subsidiary trading companies to
generate income.

Having worked for the Colon Cancer
Concern and the Haemophilia Society,
Tony Matassa is well versed in the
requirements that charities have and is
able to offer competitive rates on a
freelance basis. For further information
please contact Tony Matassa, 1 Pant
Mawr, Broadlands, Bridgend, CF31 5BB
Tel: 01656 647 520 or email
antonio@matassa.fsbusiness.co.uk

Association for Glycogen Storage Disease(UK)

Family conference and AGM is being
held on Saturday 9th October 2004 at
the Novotel in downtown Birmingham.
For more details contact Ann Phillips at
0161-980-7303 or keep an eye on our
website at www.agsd.org.uk