

You're Unique! An event celebrated by over 2000 people in Birmingham



Sunday 5th October at Thinktank, Birmingham Report by Anna Lane Development Officer – GIG & West Midlands Regional Clinical Genetics Unit

Within a short time of commencing my new post as Development Officer for Public & Patient Participation based at the West Midlands Clinical Genetics Unit I had an opportunity to visit Thinktank, the Birmingham museum of science and discovery. Greeted by whirring, flashing, noisy gadgetry I knew this was the venue for the 'Open Day' I had been planning – now if only I could persuade Thinktank to open for free to the public! Soon after The Wellcome Trust launched the PEOPLES AWARD, a “Fast,

responsive mechanism for funding public engagement activities” and the rest is history...

Genes Day was the culmination of a collaboration between Thinktank, the West Midlands Regional Genetics Service and the charity, Genetic Interest Group. Designed to bring together clinical staff, scientists and other healthcare professionals, plus families and individuals belonging to the GIG member charities Tuberous Sclerosis Association and Neurofibromatosis Association, to communicate, celebrate and debate with the public the impact on society of the 50th Anniversary of unlocking the key to life—DNA.

Special activities for the day included:

‘Build a Beastie’ Workshop – fun for all the family!

This activity was especially suitable for children between the ages of 5 and 11 and they proudly displayed their hand-crafted ‘Beastie’ which they got to keep.

‘Past the Post’ DNA Quiz Show

A specially commissioned performance piece that was both funny and informative for the audiences. Cheesy Game Show Host, Charlie, kept the two teams of, ‘James Watson’ and ‘Francis Crick’ versus ‘Maurice Wilkins’ and ‘Rosalind Franklin’ on their toes as they answered questions about DNA.

Meanwhile the Show’s Floor Manager flashed cue cards at



‘Past the Post’ DNA Quiz Show

the audience to clap, cheer or laugh!

Interactive Debate on Genetics with Professor Peter Farndon, Consultant Geneticist

This took place in the Theatre where a computerized voting system gave people the chance to register their opinion. Professor Farndon also ensured that the audience could fully participate by throwing open the discussion, which emphasised that there is often no ‘right’ or ‘wrong’ answer to the question “Genetic Testing – What would you do?”

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

You're Amazing!

All the family got a chance to find out just how unique each of them are in this 'genetic



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passport' trail. Can you smell freesias or roll your tongue? These were some of the questions asked at the 5 Stations where Thintank staff offered activities and explanations as to the reasons why these are just some of our genetic characteristics that we inherit from our parents.

Behind the Scenes – Genetic Testing – How is it Done?

The public got to meet some of the scientists and clinical staff who work in genetics and found out what happens when a blood test is sent to the laboratory together with how useful this information is to the doctors. Some people later told me how helpful they had found this, allowing them to openly discuss their concerns in a friendly, informal way.



Tuberous Sclerosis Association Family day held at the think tank event



West Midlands Neurofibromatosis Support Group held a family day at the think tank

This was all in addition to the ten galleries of activities and entertainment on four floors that Thinktank offers!

The day was a very real success both in terms of numbers, 2,101 visitors, and also the positive feedback and comments we received. All of our aims were met and we know that people from the harder to reach communities also attended. There will be a

post-evaluation meeting in November and by then we should have the results of the independent evaluators report that we commissioned. We very much hope to build on the success of the event and look at further ways of collaborating in projects involving the arts and media, as a route for engaging the public in science and genetics. So here's to organising the next!

We gratefully acknowledge the support of The Wellcome Trust.



Professor Peter Farndon speaking at the interactive debate

Ethnic Monitoring in Clinical Genetics – Pritti Mehta

Pritti Mehta has recently completed this two year project which was funded by the Department of Health. On completion she submitted an abstract for a poster presentation to the British Society for Human Genetics (BSHG) conference in York. We were all delighted to hear that she was asked to do a 15 minute presentation on the project instead which she did at the BSHG conference in September. I was able to sneak in to hear this presentation as I was in York for the day.

THE PROJECT

The projects' main aims were to examine the issues around ethnic monitoring within clinical genetic departments and to develop an ethnic category framework that is relevant to Clinical Genetics.

GIG felt that there were a number of reasons for carrying out this project as only five out of twenty five regional genetic centres monitor ethnicity and GIG wanted to carry out a project that addressed this shortfall. Without monitoring this data it is impossible to identify any gaps in the service provision being offered by the NHS. Pritti carried out a multi centre pilot study with three

Regional Genetic Centres (RGC's) North West Thames, Leicestershire and South West Thames, to assess the feasibility of collecting ethnic data within the departments. These centres were chosen because of the prevalence of minority ethnic communities within the areas that they serve. (NW Thames 24%, Leicestershire 15%, SW Thames 9%) The national prevalence of the minority ethnic population is 9%.

The project used the Department of Health ethnic categories from the last census and adapted them slightly to include five additional categories, which Pritti felt were deemed to be needed for this clinical study, Arab, Iranian, North African, Jewish Ashkenazi, and Jewish Sephardic.

The main details recorded in the pilot study by the three centres were

1. Ethnic Origin
2. Parental ethnic origin
3. Diagnosis/ reason for referral
4. Preferred Language.

These details were collected over a period of one month in each centre. The results were collated together, anonymised and analysed by Pritti. The report showed

that 23% of all groups other than White British indicated that their preferred language was not English. There was also considerable under representation in cancer referrals from minority ethnic patients. This could be for a number of reasons, patients not being referred, or there being a lower incidence of cancer amongst minority ethnic groups, we don't know the full reasons at present but by recording ethnicity in the future it will provide opportunities for the variations to be studied in more depth.

THE CONCLUSIONS

General conclusions to this study were that the majority of staff felt it was a useful and worthwhile experience to collect ethnic data (although the most appropriate time to collect it remained undecided). The study has also shown the feasibility for collecting such data and that there is a need for wider and more details ethnic data collection across all clinical genetics departments. A template framework has been created along with guidance and recommendations to RGCs around the collection of ethnicity data. Pritta Mehta's report is available on the GIG website www.gig.org.uk

Melissa Winter

Work Placement at GIG.

I hope you will join me in welcoming Meredith Carter, who is at GIG for three months on her internship from George Mason University, USA where she is in her final year studying Psychology. Meredith is working with Pritti on the London IDEAS Genetic Knowledge Park project for translating genetic information into minority ethnic languages.

I know that many of you have spoken to Meredith already through her questionnaire which you all have received, but if you would like to email her then please do on mcarter4@gmu.edu or call her here at the GIG offices.



What's happening in Wales?



Since the last edition of GIG Today, I have been 'out and about' across Wales, meeting people and groups associated with various genetic disorders. Over the next few months I will be continuing this and getting to know existing members of GIG and encouraging others to join us. Looking at my calendar, I will be at a few gatherings for different associations and groups so if you are there, please come and say hello – the sooner we meet, the sooner we get to work together!

One of my main tasks is to raise awareness that GIG is more active than ever in Wales and I'll be writing to the Local Health Boards, NHS Trusts and the Welsh Assembly Government (WAG) to introduce myself and GIG's work and aims for the future.

Based in Cardiff at the Wales Gene Park, I am in the middle of an exciting and ambitious venture. There are four main areas of activity for the Gene Park: medical genetics research; developing new diagnostic tests and clinical services for the NHS; investigating the relationship between genetics and society which include educational programmes for the general public and for NHS staff; and ensuring patient needs are met to make sure we continue to provide and develop the best possible medical genetics service for the people of Wales (my role)! More information about the Gene Park is available on the website, you'll find the address on this page with my contact details. Since devolution in Wales in 1999, the provision of NHS medical genetics rests with the Welsh Assembly Government (WAG). This allows us to

work with the WAG to ensure that medical genetics services are developed to provide the best possible care for the people of Wales. Earlier this year, the WAG announced it would provide an additional £1.5 million investment each year for the future development of medical genetics services in Wales. Part of my role for the next few months is to make sure that we plan for the future. To do this, I am planning to bring together patient groups, individuals, families and support groups as well as people working to help those with genetic disorders and asking for their opinions and needs. By working with people who use these services, we will be making sure we tailor them to the best possible way. If you would be interested in sharing your views and thoughts and want to be involved in the consultation process about the future of medical genetics services in Wales, please contact me.

I'm hoping (with the ed's approval) to have a regular spot in GIG Today. That way I can keep you posted with what's been happening and what will be going on over next few months. In the mean time, if you would like to talk to me, please find my contact details below.

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Sads UK & Oxford Genetics Knowledge Park

Wellcome Trust Centre for Human Genetics

Cardiac Arrhythmias, Research and Therapy; A holistic approach

June 26th – 27th, 2004

Hanover International Hotel,
Daventry, Northamptonshire

At the 1st International SADS Conference held in London last year it became clear that the regular opportunity to gain up to date information and knowledge and to share experiences was crucial to people living with heart conditions that cause cardiac arrhythmias and those whose loved ones have sadly died through a suspected fatal cardiac arrhythmia. To follow up on the success of the conference last year we are running another event this year. In addition to providing opportunities to share experiences, the conference this year will include presentations on the latest clinical and research developments. We hope that you will be able to come along.

Research update

The Department of Cardiovascular Medicine at Oxford University is a leading cardiovascular clinical and research centre. Researchers there are currently developing routine clinical genetic testing for inherited cardiomyopathies and the Long QT Syndrome, conditions that can cause dangerous cardiac arrhythmias if not treated. Professor Hugh Watkins and Dr Edward Blair from Oxford will present an overview of current research into the genetics of these conditions and how these findings influence clinical practice.

Cardiac arrhythmia therapy

Professor John Camm, Professor of

Clinical Cardiology, St George's Hospital Tooting will provide information and debate about ICD's, drug therapy and drug induced arrhythmia.

Ethical issues in genetic conditions

Genetic testing and clinical diagnosis of inherited cardiovascular conditions raises a number of important ethical questions both for clinicians and for family members. These include questions such as: what are my responsibilities to other members of my family? What are the ethical implications of testing children and adolescents and discussing the test results with them? What are the psychological and social implications of diagnosis of a potentially fatal heart condition or sudden death of a young person? What are the emotional and practical implications of living with cardiac arrhythmias? At the conference, Dr Michael Parker who works as a medical ethicist in the clinical genetics unit at Oxford and also runs a national ethics discussion forum for members of clinical genetics teams (including nurses, counsellors and geneticists) will present some of the main ethical and social issues.

The importance of accurate diagnosis

Recent research funded by the British Heart Foundation suggests that there may be far more people dying suddenly and unexpectedly than is currently acknowledged. The lead researcher believes these unexplained deaths should be identified in order that they can be studied systematically, and proposes labelling these deaths as Sudden Adult Death Syndrome, SADS. The importance of accurate diagnosis at autopsy and the

significance for pathologists and family members of the deceased will be presented.

Who should attend?

The conference will be particularly valuable to people living with potentially fatal cardiac conditions, people who have suffered bereavement through suspected fatal cardiac arrhythmia and professionals and agencies who have knowledge and interest in the welfare and support of these people.

The format of the conference will take place through presentations and panel/audience debate, followed by workshops.

Key Speakers:

Professor Hugh Watkins MD PhD FRCP FmedSci, Professor of Cardiovascular Medicine

Professor A. John Camm QHP, FRCP, FESC, FACC, Professor of Clinical Cardiology

Dr Andrew A. Grace MB BS PhD FRCP FACC, Consultant Cardiologist

Dr Edward Blair BMsc (Hons), MBChB, MRCP (UK), Consultant Clinical Geneticist,

Dr Michael Parker BEd, PhD, MA, Reader in Medical Ethics, Ethox Centre

Dr Mary Sheppard BSc. M.D., MRCPATH, Senior Lecturer/Honorary Consultant in Histopathology

Further Information:

Anne Jolly, Sads UK

Tel 01277 230642

Email info@sadsuk.org

Do you live in a catchment area of one of the proposed Foundation Hospitals?

If you do, there is an opportunity for patients and staff to become a member of the Foundation Trust (within their catchment area). Once you become a member of the Foundation Trust you are then eligible to stand for governor of the trust. If you live in one of the catchment areas below, you can now contact your proposed Foundation Hospital and register your interest in becoming a member. These hospitals are in the process of bidding for Foundation status at the moment.

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- **Addenbrooke's NHS Trust** www.addenbrookes.org.uk/
 - **Basildon and Thurrock University Hospitals NHS Trust** www.basildonandthurrock.nhs.uk/
 - **Bradford Hospitals NHS Trust** www.bradfordhospitals.nhs.uk/
 - **Calderdale and Huddersfield NHS Trust** www.huddweb.demon.co.uk/
 - **City Hospitals Sunderland NHS Trust** www.sunderland.nhs.uk/chs/
 - **Countess of Chester NHS Trust** www.coch.org/
 - **Doncaster and Bassetlaw Hospitals NHS Trust** www.dbh.nhs.uk/
 - **Gloucestershire Hospitals NHS Trust** www.gloshospitals.org.uk/
 - **Guy's and St Thomas' Hospital NHS Trust** www.hospital.org.uk/
 - **Homerton University Hospital NHS Trust** www.the-homerton.demon.co.uk/index.htm
 - **King's College Hospital NHS Trust** www.kingsch.nhs.uk/
 - **Moorfields Eye Hospital NHS Trust** www.moorfields.org.uk/Home
 - **North Tees and Hartlepool NHS Trust** www.northteesandhartlepool.nhs.uk/
 - **Nuffield Orthopaedic Centre NHS Trust** www.noc.nhs.uk/
 - **Papworth Hospital NHS Trust** www.papworth-hospital.org.uk/
 - **Peterborough Hospitals NHS Trust** www.peterboroughhospitals.co.uk/
 - **Rotherham General Hospital NHS Trust** www.rotherhamhospital.trent.nhs.uk/
 - **Royal Devon and Exeter Healthcare NHS Trust** www.exeterhospitals.co.uk/
 - **Sheffield Teaching Hospitals NHS Trust** www.sth.nhs.uk/
 - **Southern Derbyshire Acute Hospitals NHS Trust** www.sdah-tr.trent.nhs.uk/
 - **Stockport NHS Trust** www.stockporthealth.nwest.nhs.uk/
 - **The Royal Marsden NHS Trust** www.royalmarsden.org/home.asp
 - **The Queen Victoria Hospital NHS Trust** www.queenvic.demon.co.uk/
 - **University Hospital Birmingham NHS Trust** www.uhb.nhs.uk/
 - **University College London Hospitals NHS Trust** www.uclh.org/

Continued Growth in DEBRA Research

New grants totalling well over £800,000 were agreed by DEBRA UK in August for research projects in the UK, France, Germany and Finland. The work focuses on our twin priorities of genetic therapies and combating the scourge of skin cancer in EB.

The genetic therapy work focuses on the preclinical research needed as an essential next step before trials on human patients. In particular, DEBRA researchers are working on the development of safe delivery systems, known as vectors, without the risk of malignant complications that may arise from the use of retroviral vectors. Alongside this, more basic research, but still with a strong focus on the development of treatments, will increase our understanding of the mechanisms behind the cell fragility and impaired healing underlying EB and allied conditions.

People with the type of EB known as recessive dystrophic EB are at very severe risk of developing a particular form of skin cancer, squamous cell carcinoma, leading to early death in most cases. DEBRA's cancer research programme is aimed at understanding why this happens and to identify promising leads within mainstream cancer research. A particular problem for researchers is obtaining sufficient tissue from these cancers, which are usually disposed of after operations, and a tissue bank will be established in Germany and the UK to collect this material.

Work that will have immediate benefit has not been forgotten and the new DEBRA Fellow, based at St John's Institute of Dermatology, will work on extending a technique called preimplantation

diagnosis. In essence, eggs and sperm from a couple at risk are fertilised to make an embryo. This is then grown in a test tube for a few hours to form a small ball of cells and one of these cells can be sampled to determine whether the embryo does or does not have EB, allowing only non-affected embryos to be implanted in the uterus. Alongside pre-natal diagnosis, which is already established for detecting different forms of EB, this work will increase the options open to parents.

DEBRA's total forward commitment to approved research projects now stands at £1.75 million (with £1.25 million already spent for current projects) and is set to continue to grow quickly over the coming years. The Chairman of the charity's international Medical and Scientific Advisory Panel, Prof. Robin Eady, said "DEBRA's research funds, whilst limited, are very carefully directed and the charity has had a major role, well in excess of its size, in setting the research agenda, not just in the UK but worldwide. As a scientist, I am always cautious about raising hopes prematurely but I am confident that the very great increase in understanding of EB achieved so far will continue and result in effective treatments."

Wherever possible, the effectiveness of DEBRA funding is increased by seeking access to finance from more wealthy sources after the initial work has been done. For example, two years' ago, DEBRA UK funded a collaborative project between Prof. Dennis Roop in Houston (USA) and Prof. Irwin McLean in Dundee (UK) to undertake the first stages of proof of principle research into gene therapy for EB Simplex. Prof. Roop was also encouraged to seek funding in

the USA and the message printed below, which we received in August, is self-explanatory.

"If you recall, one stipulation for funding our joint McLean/Roop grant by DEBRA.UK was that we would agree to attempt to find future support from the National Institutes of Health (NIH). I am pleased to inform you that a grant that I submitted last fall to the NIH to follow up testing gene therapy on our original EBS mouse model received the highest score of all grants reviewed by the study section and was funded for 5 years beginning July 1. The total amount of the award (direct plus indirect costs) was \$1,768,37.

I just wanted to thank you and DEBRA.UK for supporting our application and allowing us to obtain preliminary data that was vital in helping obtain this award.

This is certainly a success story in using seed funding from an organization such as DEBRA.UK to obtain additional support."

Sincerely,

Dennis R. Roop, Ph.D.

Director

*Center for Cutaneous Molecular Biology
Baylor College of Medicine*

DEBRA has two research grant rounds each year, with deadlines for the receipt of applications of 1 April and 1 October. Information on the scheme is available from the DEBRA Office (Tel 01344 771961).

The Retinoblastoma Society –

Society Annual Day
20th September 2003
by Melissa Winter

On a bright, sunny Autumn Saturday, Anna Lane and I attended The Retinoblastoma Society Annual Family Day at Birmingham Children's Hospital. I was especially pleased to have made it as others travelling from London, including some speakers, had terrible problems on the trains (a common problem, I realise)! However, with a few quick re-arrangements of the talks we were well on our way to what was an insightful and informative day.

I certainly gained more information about Retinoblastoma, and I think that the families attending also gained a bit more knowledge about the genetics of Retinoblastoma, as we had two excellent talks after lunch by Esther Jacobson a Clinical Nurse Specialist and Harry Wilshaw a Consultant Ophthalmologist from Birmingham. A well organised creche meant that mums and dads could relax a bit and take part fully in the discussions and workshops throughout the day.

The morning began with Kathleen Hassler, Schools Worker, from Changing Faces, a national organisation that helps people cope with the social and emotional consequences of living with facial or other disfigurements. Kathleen presented an insight into the practical solutions regarding the difficulties other family members experience, especially siblings, when a child is diagnosed with a genetic condition. Kathleen stressed that siblings often need something to say about what is taking place with their brother or sister as they are the ones who may have to field awkward questions from people outside the family eg friends in the playground, neighbours, or teachers. Parents were advised to tell their children that it is OK to say "I don't want to talk about it right now" to those people who may be wanting to find out information. There are also times when brothers and sisters need to just be kids, silly time for all the children, times when they don't have to worry and can let their hair down! Kathleen reported that the families that cope the best in these types of



The
Retinoblastoma
Society

Fighting Eye Cancer in Children

situations are the ones that have built up the most resilience to the circumstances they find themselves in. Families need to have time to reflect on their experiences together, which helps to build up this resilience.

Helen Costello a play specialist at the Birmingham Children's Hospital also discussed the benefits of informing children of their own condition and treatment procedures through play. Helens advice was

that during a play activity children often raised their concerns naturally, providing an opportunity to give them appropriate information in a way which suits their age and understanding.

Scheduled workshops followed and during the workshop I attended before lunch we discussed questions and issues surrounding artificial eyes. This complimented the talks we had just been given very well. Parents in this workshop said they found the artificial eye much easier to deal with if their children could talk about it and that in turn helped them, in some cases, to overcome their own uneasiness. Another very useful way of helping children to overcome their anxieties is to get them to speak with an older child who has an artificial eye that they are managing themselves. This practice happens in some clinics where younger children learn how to clean and remove their 'pretend' eye by speaking to and working with an older child.

After a lively lunch where children were able to play and left off steam in a safe garden area we returned to the Conference Auditorium to learn more about the genetic form of Retinoblastoma and how it is inherited.

Esther Jacobson informed us that Genetic Counselling for this condition is important in giving an indication of the risk to family members.

- The Rb (Retinoblastoma) gene is located on chromosome 13

- The gene is made up of different chemicals, referred to as 'letters' and there is a series of about 72,000 letters in the Rb gene.
- The condition is inherited in a dominant way which means that someone who has the faulty gene has a 50:50 or 1 in 2 chance of passing it onto their children.
- Almost everyone with the Rb mutation develops the condition.
- Testing in pregnancy is possible when the gene fault has been identified in that family.

There are two main types: -

- Unilateral – usually one tumour affecting one eye. 90% of this type of Rb is NOT genetic and 10% of case are.
- Bilateral – multiple tumours affecting both eyes, 99% of these cases are genetic in nature.

Finally, we heard from Harry Wilshaw about the treatments that are now being used for Retinoblastoma and what is on the horizon for the condition. By identifying the mutations in certain families doctors have been able to exclude many family members, where no mutation is found. Doctors are also now using a combination of treatments as it has been found that by giving a few cycles of chemotherapy (drugs) to help shrink the tumour(s) it has often allowed laser treatment to be more precisely targetted at the tumour causing far less damage to the surrounding area and preserving more of the remaining vision. This is a new treatment but it is fast becoming the preferred one for newly diagnosed children.

And for the future?

- Improved conventional treatment,
- Altered chemotherapy or radiotherapy,
- gene therapy
- molecular therapy.

Finally, Esther and Harry were joined by consultants who are part of the specialist Retinoblastoma team working out of the Birmingham Childrens Hospital. Families then

had an opportunity to ask the expert panel questions regarding the pros and cons of certain types of treatments and genetic testing. An important point was raised about children who had previously had treatment but were not tested for the gene, the experts agreed that it was still important to identify through genetic testing those individuals who were at risk of developing tumours in other organs such as the skin or bone.

All in all a thoroughly interesting day. Thanks for inviting us!



L-R Sonia Home, Chief Executive, The Retinoblastoma Society and Esther Jacobson, Clinical Nurse Specialist

Department of Health / Macmillan Cancer Relief Partnership Project

Development of Services for cancer genetics risk assessment and counselling in England

Genetic science is progressing rapidly. Over coming years our expanding knowledge of cancer genetics will have a major impact on our ability to predict an individual's level of risk of developing cancer, our ability to detect and diagnose cancer early and our ability to select treatments which are most likely to be effective.

A small proportion of breast, ovarian and bowel cancers are associated with particular inherited genes. Testing for the genes can identify whether someone with a strong family history of the disease is likely to develop it. In the future, more of these familial subsets of cancer may be discovered, for example in skin cancer. It may also become possible to test for genes or combinations of genes that predispose to cancer in a less clear-cut way, for example by increasing susceptibility to harmful environmental stimuli such as cigarette smoke.

This area of clinical and laboratory work is expanding as awareness of inherited cancers and public demand for appropriate services grow.

Some cancer specialists are working closely with geneticists and GPs to decide on the most effective way to identify and manage people at different risk of developing familial cancers but at present services are patchy. Primary care teams do not always have ready access to the information they require to assess whether a patient is at low, moderate or high risk and in the hospital sector the NHS has few expert geneticists working in the field of cancer. The National Institute for Clinical Excellence is preparing clinical guidelines on the identification and management of women who have inherited genes that predispose them to breast cancer. Publication is expected in February 2004.

The Government's White Paper 'Our Inheritance, Our future - Realising the potential of genetics in the NHS' was published in June 2003. Macmillan Cancer Relief, working closely with the Department of Health, service users and leading

experts in cancer and genetics, has developed a model approach to services for people at risk of, or concerned about, familial cancer. The model describes a continuum of advice and care involving

primary care, local cancer services and specialised genetic and cancer services. It includes:

- the provision of consistent, correct and appropriate information for service users
- risk assessment according to an agreed national framework
- streamlined referral in accordance with agreed pathways
- consistent management of individuals in the appropriate setting according to their level of risk

Macmillan Cancer Relief and the Department of Health, working in partnership, are now ready to pilot the 'Kenilworth' model and an invitation was issued to cancer networks, PCTs and regional genetics services at the beginning of September, for up to three year funding to test and evaluate all aspects of the model approach.

Glyn Purland

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10 Years of Co-operating with Industry

Threat or Opportunity?

On a day which saw patients, Patient Group representatives, clinicians and the pharmaceutical industry all sharing perspectives on 'Research and Genetic Disorders', Christine spoke of Partnership between The Society for Mucopolysaccharide Diseases (MPS Society) doctors and scientists, and the pharmaceutical industry. This partnership had developed and been cultivated from early beginnings into a flourishing mutually beneficial partnership that has allowed for the translation of scientific discoveries into treatments. Christine emphasised how whilst maintaining its independence the charity had been able to influence those with the funding and those with the knowledge to harness the opportunities for people affected by some of the lysosomal storage diseases.

- Lysosomal storage disorders, of which Mucopolysaccharide and Related Diseases are one group, affect about 1:5,000 live births in the United Kingdom
- Many children affected will have progressive physical and neurological disease that results in death in childhood

First Opportunity

Christine spoke of the first opportunities that the MPS Society had for working with industry which began back in 1993 when a company called CSL were interested in developing Enzyme Replacement Therapy (ERT) for MPS VI. Unfortunately in 1995 CSL changed their strategy and became a public company, and they had to withdraw their ERT development plan.

Hope on the Horizon 1997 – 2003

Hope was just around the corner though, and during the following seven years ERT treatments have been developed for lysosomal disorders by Biomarin, TKT Inc, TKT5s and Genzyme.

European approval was given for ERT simultaneously to two products developed by

TKT5s and Genzyme, Replagal™ and Fabrazyme™ in August 2001. In June 2003 Genzyme/Biomarin received European Approval for ERT treatment Aldurazyme™ for MPS I And only very recently TKT Inc launched a ERT pivotal worldwide clinical trial in humans for MPS II (October 2003). As these developments have been made the MPS Society has had a pivotal role in informing patients of the developments taking place and where treatment is available helping them to gain access to it.

As you can see progress is being made and at a rapid pace and this, as Christine said, needs careful planning and cooperation between both patient organisations, doctors, specialists and industry. In order to work effectively with the industry for the benefit of patients and their families it is important to be aware of the following areas:

- Never lose the focus of the goals of your patient association. – It is very important that the patient association remains impartial at all times.
- Be forever mindful of the goals of industry
- Have well thought out and tangible strategies for working with the industry
- Unfailingly act professionally and impartially

Christine discussed the threats and opportunities that could come out of such an alliance with industry and highlighted the areas of concern that should be taken into consideration when working in this type of collaboration, in order that patient groups avoid unnecessary pitfalls.

Opportunities

TREATMENT FOR PATIENTS

- Education for patients
- Education for patient organisations
- To educate the industry – Informing them of the condition.
- To educate professionals on the work of the

Presentation by Christine Lavery, The Society for Mucopolysaccharide Diseases at the Genetic Interest Group AGM and Conference.

continues overleaf

THE FRAGILE X SOCIETY

FAMILY CONFERENCE:
Saturday 8th November 2003

PROGRAMME

10am Registration and Coffee
Crèche opens

10.30am Reporting the Results of
Fragile X Research Adult
Male Carrier Study

Speaker: Dr Kim Cornish
McGill University,
Montreal, Canada And
Nottingham University

Development of Attention in
Young Children with Fragile X

Speaker: Gaia Scerif
Institute of Child Health,
London

12.30pm Lunch
Crèche closes

1.15pm Crèche reopens

2.00pm The Genetics of Fragile X
Explained and: The
Importance of Genetic
Counselling for Families

Implications for Carriers and
their reproductive options

Speaker: Barbara Carmichael
Clinical Nurse Specialist in
Genetics

4.00pm Close of conference with tea
Crèche closes

Places: Free to Fragile X families
£25 for non-members

- MPS Society
- Networking beyond the UK
- Sharing data from the MPS registry – MPS has been able to supply anonymised data of the weight and height of children with these conditions to the companies making the ERT in order that the correct prescribed dosage is given.
- Funding

Threats

LOSS OF INDEPENDENCE

This can be seen as one of the major threats of a charity working with industry but there are other areas to be aware of, and take into consideration, such as: -

- Being associated with a particular member of the industry – The MPS Society has worked with a number of pharmaceutical companies, and has always promoted its independence.
- Losing direction and becoming less transparent – It is important to inform people of the work that is taking place
- Losing credibility resulting in ineffective advocacy
- Becoming dependent on industry funding

MPS Society Conclusion

For the MPS Society co-operating with the Industry for the common goal of TREATMENT has been, and continues to be, a very positive experience because the threats were identified, managed and all opportunities taken in the best interests of the patients.

After outlining these key issues and developments Christine encouraged patient groups to look at ways of optimising their special relationships with their own members and those clinicians and scientists who have a keen interest in their disorder, thereby acting as a catalyst for promising future therapies and treatments. These

may include ensuring sufficient patients are available for research studies, keeping an accurate database of patients, contacting International societies with shared aims and values, inviting doctors and scientists to speak at meetings and being 'adult' when sought out by commercial companies who may be looking for a collaboration.

Christine added that the charity had taken some 'grown-up' decisions that were in the best interests of members of their society, for example, they made a policy decision that donations from pharmaceutical companies in the field of interest could not exceed 10% of their charitable income. This has enabled them to remain open and transparent at all times.

The enthusiasm and experience that Christine offered spurred a number of questions from the audience and clearly it was acknowledged as a successful and thought provoking presentation.

THE UKTS NATIONAL CONFERENCE

Sunday 16th November 2003
THALASSAEMIA IN
THE 21ST CENTURY

The United Kingdom
Thalassaemia Society
19 The Broadway, Southgate,
London, N14 6PH
Tel: 020 8882 0011
Fax: 020 8882 8618
www.ukts.org

To be held at the Royal Moat
House Hotel in Nottingham.

For further details and a registration form, please contact the UKTS offices on the above numbers, or download a form from their website.