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We have now been working in partnership with the shopping club KidStart for over a year and our income from this source is steadily growing. Many, many thanks to all of you who have supported GIG by shopping through KidStart over the last year. Please carry on supporting our work in this way and encourage your friends, contacts and members of your group to do the same. If we can continue to increase the number of people saving for GIG through KidStart, it could prove to be an effective and sustainable way to support our ongoing work in the long-term.

For those of you who have not yet used KidStart, it is a shopping club that works with 350 well known online retailers such as M&S, Waterstone's, John Lewis, Argos and a number of High Street partners such as Goldsmiths, House of Fraser and Jo Jo Maman Bebe. Anyone can join KidStart for FREE and every time they make a purchase via KidStart online or in the high street having registered their credit and debit cards, they earn a percentage of every £1 they spend which can be saved for GIG.

How to save for GIG through KidStart:

Log on to www.kidstart.co.uk/GIG.

If you are not a KidStart member, you will need to register. You will be asked to accept the invitation to save for GIG. If you are already a member of KidStart, just log in and accept the invitation to save for GIG.

Go to MY PROFILE and you will see GIG as one of the beneficiaries to receive savings as you shop. You can also add a child or school that you wish to save for and you can share your savings amongst your beneficiaries.

In order to save for GIG you MUST state what % allocation

you wish us to receive. This is done in your profile once you have accepted the invitation to save for GIG. Of course we would like 100% of your savings to go to GIG, but we will leave that to you....

You can start to shop immediately and collect savings which you can view in your "Kiddybank".

Every time you shop with KidStart, a percentage of what you spend is automatically saved. It is transferred to GIG on a six monthly basis.

We have produced a leaflet that tells you all about KidStart. If you would like copies of the leaflet to distribute to your members, friends and contacts or if you have any further questions about KidStart please contact helen@gig.org.uk. Thanks again for your support.

Helen Parr, Fundraiser

Albinism Conference

We were delighted to be asked to attend the Albinism bi-annual conference in November last year and were very pleased to be able to bring our GIG stand and to speak to delegates about the work of GIG and also the work of Rare Disease UK



Stephen and Ariadne at the Albinism Conference.

Winter 2010

The Genetic Interest Group AGM 2009

At this year's AGM in November we were very pleased to have two excellent speakers who made presentations about patient involvement in research, but from very different perspectives. Dr Sue Pavitt is Director of the Comprehensive Health Research Division, Clinical Trials Research Unit at the University of Leeds and she spoke about her experience of patient and public involvement in research and particularly in clinical trials, whilst Neil Townsend from the theatre company Y Touring spoke about how they work to engage young people in decisions regarding their healthcare and the healthcare of others.

Involving Patients and the Public in Clinical Trials

Dr Sue Pavitt, at the University of Leeds gave a presentation on involving patients and the public in clinical trials. Despite the differences between the groups represented at GIG, this was clearly an area of shared interest and common goals.

Clinical trials are not new, and Dr Pavitt gave several interesting examples. There is archaeological and historical evidence to suggest that the ancient Egyptians attempted this kind of research. In 1537 Ambroise Pare carried out a clinical trial of a mixture of turpentine, egg yolk and rose oil to promote wound healing. Perhaps the best known is the work of one Dr Lind in 1747 who undertook trials using lemons and limes to prevent scurvy.

Although recent in comparison, the University of Leeds has had a Clinical Trials Research Unit for over 20 years, and plays a crucial role in strengthening the links between clinical research, the pharmaceutical and related industries, clinicians, academics and patients. It also helps to ensure that the findings coming out of clinical research are translated into practical medicine and of tangible benefit to service users.

Although rare diseases vary enormously in their presentation and implications for the people who live with them, there are trials which are relevant to many of them, regardless of their intrinsic differences. For example, trials about retro-viral delivery systems will have applications for future gene therapy trials for a wide variety of different genetic disorders.

Research should be carried out 'with and by' patients, not

merely 'about' them. Involving patients and public (who may be carers, family members or potential patients) is useful during every part of the Clinical Trial cycle. (See diagram).

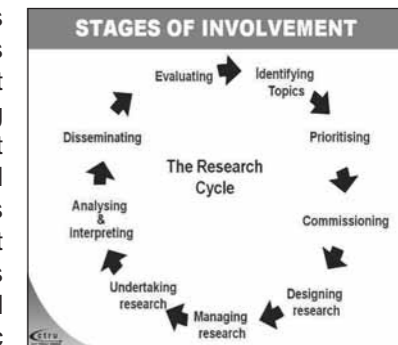
Dr Pavitt gave examples of practical pointers patients had given about the practicalities of running trials.

Patient representatives on a trial involving Senior Citizens highlighted that consideration of the times of when bus passes could be used for public transport and running

clinical appointments to accommodate this constraint led to a logistically designed trial that would improve trial participation. Involving patients also highlights the fact that sometimes their expectations and aspirations may differ significantly from those of the researchers. This was illustrated in an example in oral cancer where researchers and clinicians talked about a five year survival rate, whereas this was of secondary importance for patients who felt that high level functioning (being able to eat and talk) and appearance were more important, even if this meant a shorter survival time.

When patients' views are valued and respected, and they are able to contribute at every stage, there are benefits to both sides. Scientists and clinicians working together with patients and their carers and representatives are able to construct research trials that patients and public are willing to partake in, where the programme design is practical, carrying out the research is achievable, and delivers meaningful results. In Dr Pavitt's Unit, patients are given support and information about what is expected of them and how they can contribute. They are also provided with practical assistance where necessary. (Trials involving young parents for instance would factor in the provision of a crèche.)

Everyone at the meeting represented a user group whose individual members have the potential to be involved in clinical trials. We all came away with a much clearer idea of what is involved in the setting up of this kind of research. Most of all, the presentation emphasised how important it is to combine the knowledge, experience, insight, and practicality of patients and public with the specific knowledge, expertise and interest of scientists, industrialists and other stakeholders, to achieve the best possible outcomes.



PatientPartner – A GIG Project



The PatientPartner Project is one of the 7th Framework programs funded by the European Commission. The project's focus is based on patients partnering in clinical trials and fits in nicely with the talk that Dr Pavitt gave at the GIG AGM. One of the project's aims is to develop a stronger relationship and improved communication between patients/patient organisations and the other stakeholders involved in the clinical trial process. As Dr. Sue Pavitt mentioned in her presentation, clinical trials have been around for quite some time but only recently have patients been working alongside researchers to help drive research and input into the design of clinical trials. In the past there has been a lack of direct involvement as partners in clinical trials by patients, patients were often seen as contributing a sample but little else. This is now slowly changing and the work of PatientPartner will have far reaching consequences as it will set out what patients, researchers and industry all want from patient involvement in clinical trials and will make recommendations to governments on how this can be achieved.

There are some cases across Europe, in which Pharmaceutical companies, Academia, and Patient Organisations have developed good practice in terms of communication, with which all parties are satisfied; however, this is currently not something that happens as a routine part of the clinical trials process. As Dr Pavitt highlighted in her talk, a patient's input and involvement can significantly help the clinical trial progress. In the examples that PatientPartner has found it is often with practical suggestions that patients can help adjustments which, in the long term, will benefit both the participants and researchers in clinical trials.

PatientPartner is conducting three regional workshops across Europe to try and identify whether there are any regional patterns that differ as to patients' current participation in the clinical trial process. The North-Western (London) and Central-Eastern (Budapest) European workshops have recently taken place, with the Southern European (country to be confirmed) workshop remaining.

If you would like to find out more about these workshops please visit the PatientPartner website: www.patientpartner-europe.eu or contact me at GIG Ariadne Stamatopoulou, ariadne@gig.org.uk

The second speaker at the GIG Annual Conference was Nigel Townsend from Y Touring. Y Touring are an award winning theatre company, established in 1989. They focus very much on developing dramas that tackle current and often difficult issues. They perform to young people and adults across the UK to help stimulate debate and raise awareness of often complex issues.

Nigel spoke to us particularly as they have recently developed a play called Starfish which explores the need for clinical trials and the ethical issues that surround them. It also aims to raise awareness of the use of electronic patient data for research and its impact on health. Usually the play will take place in schools and colleges where students will have a full afternoon to listen to the play and to then debate some of the topics. Nigel kindly gave our AGM audience a small taster. We were asked to vote "yes", "no" or "don't know" to a series of questions. Nigel explained how this would usually happen before and after the play. The questions are quite provocative to stimulate debate, such as "I'm happy for anyone to know all the details of my medical history" or "I'm happy to reveal details of my medical record if it will help someone get better treatment" or "I would be happy to reveal details of my medical record to allow the development of a new abortion pill". It was really interesting to see how our audience voted, and Nigel then showed us the preliminary results from the voting at their live shows. Ariadne and I were very pleased to be invited to a special showing of Starfish, just before Christmas at the Y Touring offices. The play was not only written, directed and acted superbly but it also managed to cleverly draw in many issues about clinical trials. The play was also very dramatic and I have to confess that both Ariadne and I shed a few tears towards the end. We were both quite surprised at how engrossing the play was and how much it stimulated discussion, even being familiar with the issues audience. In schools and colleges it is easy to understand how the play and discussions take up a full afternoon.

Y Touring have also produced dramas on animal research, plus a series of five plays inspired by the ethical issues raised by current biomedical research.

www.theatreofdebate.com
www.ytouring.co.uk
www.geneticfutures.com

The section of this article referring to Dr Pavitt's talk has been reproduced with kind permission from GIG member The Jennifer Trust for Spinal Muscular Atrophy.

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Policy Update

GIG's policy team has been active on many fronts over the past few months. These are some of the issues we have been dealing with recently. For more information, you can always check our website or get in contact directly.

Preimplantation Genetic Diagnosis (PGD)

Since the recent Human Fertilisation & Embryology Act changed the law governing PGD, the Human Fertilisation & Embryology Authority (HFEA) has to be satisfied of the "seriousness" of the condition being licensed for PGD. GIG has recently been giving evidence to the HFEA's Licensing Committee on specific conditions under consideration. Where applicable, we have also been approaching our members and assisting them in with the preparation of evidence.

We have also been involved in a consultation to change the way PGD for late onset conditions and preimplantation tissue typing (saviour siblings) are licensed. Currently, due to greater ethical concern surrounding these two methods, they are licensed on a case-by-case basis, rather than the condition-by-condition basis that other PGD is licensed. We would like to see all PGD licensed in the same way to reduce stress on families and time delays, especially in these uses of PGD where time is of greater value.

Changes to Critical Illness Insurance

The Association of British Insurers (ABI) launched a review of critical illness cover, focusing on the especially difficult area of total permanent disability (TPD), a cause of a large number of refused claims. GIG worked together with member group Ataxia UK, to produce a strong response resisting moves to ban the product, as this catch-all definition is often the only way in which members can claim on critical illness cover.

Following a workshop at the ABI, at which GIG was one of only two patient groups represented, the ABI has abandoned plans to drop TPD, and has instead focused on better communication.

Animal Research Legislation in Europe

The European Commission and the Council of Member States has finally, in mid-December, reached a compromise agreement with the European Parliament. GIG has been working on this issue for many years now, and most recently coordinated a patient lobby by five major pan-European patient organisations calling for good quality ethical review of research in Europe, rather than bans which could force studies out into countries with poorer regulation.

National Commissioning

The House of Commons Health Committee launched an inquiry into healthcare commissioning in the UK to which GIG submitted evidence. GIG praised the response to the 2006 Carter Review, whilst calling for a strengthening of the powers and budget of the National Commissioning Group.

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org
.uk



We highlighted the differences between commissioning arrangements in different home nations and called for better commissioning of services for those in need of care from multiple areas of the health service. We look forward to seeing the recommendations of this inquiry in 2010.

The Department of Health have launched a consultation entitled "Strengthening National Commissioning", which will close in February. GIG is concerned as to the extent to which proposals in this document do improve the commissioning process for our members and will make a response in the New Year.

As always, we are delighted when our members come to us for advice, assistance, or to collaborate, on any policy issue that interests you. GIG's policy team has been active on many fronts over the past few months. These are some of the issues we have been dealing with recently. For more information, you can always check out website or get in contact directly.

Nick Meade, Policy Analyst
nick@gig.org.uk

EuroGenGuide - Project draws to a close



At the time of writing, the three year EC-funded EuroGenGuide is about to end, following the success of its official launch in Warsaw on November 27th, 2009. The finished physical copies of EuroGenGuide are now being distributed across Europe to national genetics societies, patient organisations, doctors, researchers, and scientists. This hard copy, both printed and on CD contains all the material developed by the team, which is also currently available on the project website at <http://www.eurogenguide.eu>. After the project ends, the website will be reduced in size, with the home page and all the key information being incorporated into GIG's website. The EuroGenGuide URL will remain active, and will re-direct you automatically. The material will be downloadable from the website as a PDF, and if you or any of your members would like to have a copy of EuroGenGuide sent to you on CD, please don't hesitate to contact the GIG office.

EuroGenGuide has been a highly rewarding experience, and not without its challenges. One of the biggest of these was the task of co-ordinating a large group of individuals from a wide range of health organisations from right across Europe. GIG represents a diverse group of over 130 UK patient organisations, and what the UK has in common with Europe as a continent is the range and diversity of its people, their backgrounds and cultures. Mediating between these differences, and attempting to find ways to improve access to the benefits offered by genomic research is a huge challenge but one that represents an opportunity to improve the lives of millions of people.

Warsaw was chosen for the launch of EuroGenGuide for several reasons, an important one of which was what the location symbolised for the project. Warsaw, as the capital of a rapidly developing country in the centre of Europe, represents a boundary between the western and eastern regions of the continent. This geographical boundary has for many decades also been an economic one, east of which access to genomic healthcare has been more limited than to the west of it. One of EuroGenGuide's aims is contributing to the elimination of this disparity by improving access to the areas where it is needed the most. Poland is a big, prosperous, rapidly developing country in a region which can benefit from the work of EuroGenGuide. Attended by and with speeches from representatives of the Polish government, professionals from the health, education and science communities, as well as patients and patient representatives, the event was a fitting way to launch the project. Widely covered by Polish media, we hope this high profile launch will give the project the platform it needs to be a useful resource where it is needed the most.

This article for the newsletter will sadly be my last, as I will be leaving in the new year. This marks the end of three great years, during which I have had a hugely rewarding and enjoyable time working for GIG. You will know how great the GIG team is, and it has been a pleasure to be a part of it. I have learnt a huge amount—the variety of GIG's work and the close knit nature of the staff means I have gained a real insight into genetic diseases and the issues affecting those who live with them, as well as an understanding of the connections between patients and patient groups, government and policy making, and the clinical, research and pharmaceutical communities. This is not something that can be taught, and it has been an experience that will be invaluable to me in the future. I would like to thank everyone at GIG, and those of you who have helped me with EuroGenGuide, I wish you all the best for 2010 and beyond.

Alex Mckeown, alex@gig.org.uk



The Experience of Uncertainty in Parents of Children With an Undiagnosed Medical Condition

The National Human Genome Research Institute at the National Institutes of Health (NIH) in the USA is sponsoring a study that seeks to learn more about how parents of children with an undiagnosed medical condition think and feel about their child's condition. We hope that this knowledge will improve the health care and counselling for these parents. Men and women who are 18 years or older and have at least one child with a medical condition that has remained undiagnosed for more than 2 years are invited to take part in this study. Participation involves one survey that takes about 45 minutes to finish. The survey can be taken online or a paper copy can be mailed to you.

For this study we are keen to gain input from patients and

families in the UK as well as in the USA and I would therefore be very grateful if you could let people know about this research.

For additional information about this study, you may review the Notice to Participants <http://www.surveymonkey.com/StudyNoticeUncertainty>.

If you have questions or would like a paper version of the survey, please contact:

Anne C. Madeo, MS, Principal Investigator Genetic Counselor National Human Research Institute National Institutes of Health Bldg. 31, Room B1B36, 31 Center Drive, MSC 2073, Bethesda, MD 20892-2073
Phone: 301-443-2635 Email: anne.madeo@nih.gov

GIG congratulates Michael Griffith of Fighting Blindness Ireland

Fighting Blindness is delighted to announce that their Chairman, Mr Michael Griffith, has been selected by the Senate of Trinity College Dublin to receive the honorary degree of Doctor in Science (honoris causa) on December 11 2009.

'Chief Executive of Fighting Blindness (1996-2008) and founding chairman of this charity in 1983. For more than 20 years, he has played a crucial role in the development of biomedical research in Ireland in general and of Genetics at Trinity in particular. His support and fundraising efforts since the 1980s led to the mapping of the human genes responsible for Retinitis Pigmentosa, a form of blindness. He has been the driving force behind Fighting Blindness, which today supports more than 14 research projects on eye diseases. He has been a member of the Health Research Board between 2002 and 2005 and again since 2007 and of many international health bodies' –Trinity College Dublin

Michael Griffith also served as a member of The Retina International Management Committee for many years and has been instrumental in the promotion of research into degenerative and inherited blindness not only in Ireland, but also in Europe. He is committed to the cause of rare diseases and has worked with policy makers at home and abroad to maintain and develop research into these conditions.

Michael has also worked to ensure that access to therapies was fair and equitable. He played an important role in the creation of a number of organisations such as the European Platform for Patients' Organisations, Science and Industry (EPPOSI) in the 1990s and founded the only national equivalent; the Irish Platform for Patients'

Organisations, Science and Industry (IPPOSI) in 2002. He also worked with colleagues in other Irish patient led charities to establish the Medical Research Charities Group (MRCG) and has contributed to the formation of the Genetic and Rare Diseases Organisation (GRDO).

His work is marked by his great generosity of spirit and genuine interest in the people and families affected by all forms of blindness and other rare disorders, including the rare genetic skin condition Epidermolysis Bullosa (EB). For a number of years, Michael held the position of Chairman for the patient group Debra Ireland, which supports those affected by EB. He remains active on the Debra Ireland board.

I am sure you will join us at Fighting Blindness Ireland in congratulating Michael on this prestigious and well-deserved honour. This is a wonderful recognition of Michael's work in the fight against degenerative and inherited blindness over the years. It is also a validation of the importance of patient involvement in the development of medical research.

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.ie

FEATURE ON THE MYROVLYTIS TRUST

Progress Report for the MYROVLYTIS TRUST

London, 27th November 2009

Introduction

Since our last article in the GIG newsletter, July 2008, the Myrovlytis Trust (www.MyrovlytisTrust.org) has continued to promote research into Birt Hogg Dubé Syndrome (www.BHDSyndrome.org). BHD Syndrome is a monogenic, autosomal dominant condition, caused by mutation of the gene folliculin (FLCN), and characterised by renal cell carcinoma, pneumothorax and skin fibrofolliculomas. Our activities over the last eighteen months include:

Funding Research

Awarding several basic research grants to university research groups in the UK, Europe, North America and elsewhere. Examples include:

- Grant to Professor Maria Czyzyk-Krzeska, University of Cincinnati, to fund a post-doc to look at the regulation of FLCN expression by VHL in renal cancer.
- Grant to Dr Vera Krymskaya, Associate Professor, University of Pennsylvania, to examine the role of FLCN in cell proliferation, cell-cell contact formation and cell-cell interactions.
- A small clinical trial will look at the effect of rapamycin on skin fibrofolliculomas. Ethical approval has been obtained, patients have been recruited, and the trial is due to start in January 2010.
- In collaboration with Partnership for Cures [www.4Cures.org],

awarding three drug-repurposing pilot grants. The projects aim to take drugs currently being used for one condition and 're-purpose' them to provide a 'new' treatment for BHD syndrome.

- Awarding three research grants in the exciting field of renal gene therapy:

Dr Yosef Haviv (Hadassah-Hebrew University Medical Centre, Israel) will take an SV40-based kidney gene therapy approach;

Dr Richard Harbottle (Imperial College London, UK) will use non-viral S-MAR plasmid vectors to develop prophylactic gene therapy for BHD

A collaboration between Professor Andy Baker and Dr Laura Denby (Glasgow, UK) and Professor David Curiel and Dr Justin Roth (University of Alabama, Birmingham, USA) will develop virus- and cell-based targeting platforms for renal gene therapy.

The European BHD Consortium [www.EuropeanBHDConsortium.eu] is supported by the Myrovlytis Trust. Its guidelines on the diagnosis and management of BHD syndrome will be published in Lancet Oncology. Members of the Consortium are diagnosing increasing numbers of BHD patients across Europe

Sequence Variation Database

A new, free, online database [www.lovd.nl/flcn] of all known FLCN mutations has been created by Dr. Derek Lim (Clinical Research Fellow, University of Birmingham Medical School, UK), in collaboration with the European BHD Consortium. The database: has been recognized by the Human Genome Variation

Society; is hosted by Leiden Open Variation Database; consolidates all known FLCN mutations, and includes several novel unpublished mutations as well as all published ones. A description of the database has been recently been published - Lim D et al. 2009.

Conclusion

We hope that the momentum that is continuing to build within the BHD research community will bring us closer to therapy for BHD syndrome.

Duncan Azzopardi and John Solly
<http://www.myrovlytistrust.org/travel-grants/>

BHDSyndrome.org

www.BHDSyndrome.org has been developed over the last few months and now includes:

BHD Forum This 'user-led' area of the site enables people to introduce themselves, discuss any aspect of living with BHD syndrome, or address a particular issue that may be of concern.

BHD researchers section This is continually updated and includes:

- **BHD-specific literature library** (www.bhdsyndrome.org/research/resources/bhd-article-library); downloadable database (www.bhdsyndrome.org/research/resources/bhd-literature-database); details of all, potentially useful published and unpublished lab resources (www.bhdsyndrome.org/research/resources/laboratory-essentials) including antibodies, animal models, cell lines.
 - **Detailed information about:** the diagnosis process; symptom information; treatment availability.
- BHD research blog (www.bhdsyndrome.org/topics/bhd-research-blog), which analyses scientific literature and discusses research. Initially aimed at researchers in the field, BHD families might also find it useful.

BHD Pamphlets

The Myrovlytis Trust is producing a series of pamphlets about BHD syndrome. Aimed at families as well as GPs, the pamphlets are intended to raise awareness and increase knowledge of BHD. Since different people have different interests in BHD, and are affected differently by the condition, different pamphlets will discuss specific aspects: diagnosis; specific symptoms; current treatments etc.. We would be very interested to receive advice from organisations that have already gone through this process.

BHD Symposium

The Second BHD Symposium will be held on 22nd April 2010 at the Washington Marriott at Metro Center Hotel, Washington DC, USA. The American Association of Cancer Research's Annual Meeting will take place Washington immediately before the BHD Symposium. More details will be available at (www.bhdsyndrome.org/the-second-bhd-symposium).

Birt-Hogg-Dubé Syndrome Research and Support

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December 8th 2009 Researcher Resources Updated
Both the BHD Literature Database and the BHD Article Library...
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December 1st 2009 BHD Diagnosis and Management Guidelines
The European BHD Consortium has just published guidelines...
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Introducing Hypoparathyroidism UK (HPTH UK)

What is Hypoparathyroidism

Mention 'hypoparathyroidism' and people will probably say 'hypo-splutter-WHAT?' Most people probably don't know where the parathyroid glands are, or what they do.

That is what Liz Glenister discovered, when she was diagnosed with hypoparathyroidism in 1992 after surgery to remove her thyroid gland.

Hypoparathyroidism (or HPTH) is a rare condition, in which there are lower than normal levels of calcium in the blood due to insufficient levels of parathyroid hormone (PTH).

Sometimes HPTH is acquired as a result of neck surgery or from autoimmune destruction of the parathyroid glands, but it can also be inherited and transmitted through autosomal recessive genes or associated with other disorders.

What all forms of hypoparathyroidism have in common is that the body cannot manage its own calcium levels, and it can present challenges in getting the right diagnosis. Information is needed for patients and doctors alike to help them manage the condition and avert crises. Too little calcium can result in tetany; muscle twitching, tingling, or numbness of lips, tongue, fingers and feet. Symptoms may include tremors, dizziness, brain fog, mood changes, anxiety, weakness, chills, and headache. Severe cases can progress to painful muscle cramps and seizures and sometimes people have ended up in A&E. Treatment is usually life-long with calcium and vitamin D analogues such as Alfacalcidol®.

Setting up UK Support Group

After a long search for information and support, Liz found the website of the Hypoparathyroidism Association run by James Sanders in the United States. She suggested starting up a UK branch to address the specific needs of UK patients, and in 2005 was given a small set-up grant to build and independently run the first UK website and online forum.

As 'HPTH UK' the organisation has grown rapidly and we now have over 500 members - patients, carers, and supporters. We plan to apply for recognition as a charitable Trust in 2010.

The organisation also provides support to people with related disorders. We have members with pseudo-hypoparathyroidism (PHP) and pseudopseudohypoparathyroidism (PPHP) - sometimes collectively referred to as Albright's Hereditary Osteodystrophy (AHO) - and DiGeorge Syndrome; and many who have been treated for primary hyperparathyroidism.

HPTH UK is run by a small group of volunteers with the help and advice of its medical advisors and other interested professionals. We have been supported by a grant from the Society for Endocrinology and we are also recognised by the British Thyroid Association, two British medical professional

societies representing researchers and clinicians. We work closely with other HPTH groups around the world.

Liz Glenister herself is a former teacher who works tirelessly on a volunteer basis and is a huge inspiration to her team in spite of having to battle with her personal HPTH issues. She says: 'Often, finding that you are not alone and finding people who understand what you are going through is a huge relief, and is the first step to understanding and taking control of your condition.'

As Hypoparathyroidism is not well-known or well-understood we have spent much of our first four years raising awareness and providing information for patients and doctors alike. Here are some of our present and future activities.

Raising awareness

We are launching our new website on 5 January 2010 to coincide with the worldwide Hypoparathyroidism Awareness Day. We also publish a newsletter, and we are on Facebook and Twitter.

Peer support for HPTH patients

We run a 24/7 online forum as a place of contact for patients and we have also set up two telephone help lines: 01342 316315 (South) and 01623 750330 (North).

Guidelines for treatment

No official medical advice on managing HPTH exists despite the fact that medication must be tailored to individual needs and crisis management is often necessary. HPTH UK is involved in the first stage of development of guidelines. We are working with the Society for Endocrinology's Bone & Mineral Special Interest Group to produce a patient information leaflet. Next, we plan to collaborate on an information leaflet for doctors and hope eventually to see NICE clinical guidelines developed.

Better treatment

A step towards better treatment was realised recently when a trial in the use of parathyroid hormone (PTH-184) (Pretact) in the treatment of hypoparathyroidism began in the UK as part of a global REPLACE study sponsored by NPS Pharmaceuticals, Inc. HPTH UK has been active in raising awareness for this trial and collecting information from people who wish to participate.

For more information about HPTH UK please contact Judith Taylor (Public Affairs) at Judith@hpth.org.uk.

If you would like to offer help or to make a donation, please contact **Liz Glenister (Director, HPTH UK)** at liz@hpth.org.uk.

Alström Syndrome UK Secures £344,000 Big Lottery Research Funding



LOTTERY FUNDED

Mike Hales (ASUK Grant Adviser and Programme Manager), tells how it was achieved.

Alström Syndrome UK (ASUK) is a small patient support group that has now secured Big lottery funding for a 3 year medical research project into this very rare single gene condition which leads to blindness, hearing loss, diabetes, obesity and heart, kidney and liver disease among many other complications. ASUK have three partners who will deliver the research – Torbay General Hospital, Birmingham Children's Hospital and Cambridge University.

The programme encompasses clinical research at the two hospitals, genetic research at Birmingham and stem cell work investigating the fundamental cell biology at Cambridge University's Metabolic Research Laboratories. This will be the world's first structured research programme using Alström patients during which we will set up the first-ever AS clinical database; establish the first-ever AS tissue bank and initiate the first stem cell investigations into this syndrome. If all goes to plan at the end of three years we expect to:

- discover more people who have never before been correctly diagnosed with AS and therefore not been given the best treatments
- be able to improve management of this difficult-to-treat condition
- be able to better predict the course of the disease
- take the first steps to genetically-engineered treatments
- potentially, provide insights into type 2 diabetes, cardiomyopathy and obesity in the general population

This is heady stuff for a small patient support group with 50 members that is still run out of the front room of a house in Paignton! It might therefore be worth recalling how we got here.

In the charity's early days at the end of the last century, ASUK started by pulling together an annual weekend family conference to which local doctors who had become familiar with Alström Syndrome through treating the founder, Kay Parkinson's two Alström children, were also invited. They gave their time and skills freely and began to hold a clinic for children who came to the conference.

Over the years this grew until a major breakthrough happened in 2006 when the NHS National Commissioning Group agreed to adopt and fund regular Alström clinics at Torbay and Birmingham. Although not the prime purpose, this established a platform for research by presenting doctors with a group of patients that could be seen regularly and tracked over a period of years. Basically, it put in place a foundation for potential research which provided a unique selling point when it came to arguing our case for funds. However, we had no real experience of constructing a complex medical bid such as this or of initiating or leading research. Furthermore, funding of this type goes generally to the specialist laboratory or university department – not small inexperienced charities.

Fortunately, the Big Lottery takes a different view. They will only fund charities and one of their key objectives is to foster charities to initiate and manage research. When the first bidding round was open in 2008 we applied for and succeeded in getting a small development grant to pay for the work involved in constructing the main bid. This turned out to be a wise move because completing the main application was a daunting process which occupied most of my spare time for 5 months. We were fortunate in having support from the two clinic hospitals and found further enthusiastic support in Stephen O'Rahilly and Robert Semple at Cambridge. We marshalled out backers, stakeholders and arguments, analysed our budgets and eventually became one of the 57 charities funded in this round.

I would encourage any charity - however small - to believe that such things are possible. Looking at the awards made this year, conventional laboratory-based research which we are undertaking seems to be the exception. Many are more focused on social science projects. I am sure the criteria for success are no less stringent but it does mean you do not have to have started free clinics over ten years ago to stand a chance!

For more information about the project contact mike.hales@alstrom.org.uk

For more information about Alström Syndrome contact info@alstrom.org.uk



John Parkinson (ASUK Family Liaison); Kay Parkinson (ASUK CEO); Cathy Carey (Torbay Hospital Consultant Cardiologist); Richard Paisey (Torbay Hospital Consultant Endocrinologist); Mike Hales (ASUK Grant Adviser and Programme Manager)

New Study on the Genetic Basis of Brain Abnormalities and Learning Disability

The Oxford Brain Abnormalities research group, a diverse group of scientists and medical professionals, have recently received funding from the NHS for a new study. This group is studying microcephaly (small brains), lissencephaly (smooth brains), polymicrogyria (brains with too many folds), and hydrocephalus (excessive water on the brain). While some genes, such as DCX, have been identified that cause brain abnormalities, many remain to be identified. The goal of their research is to identify new genes that cause structural brain abnormalities and to develop novel diagnostic tools to aid genetic diagnosis. The group is using state of the art technology to identify small deletions and duplications in genes, as well as mutations. They are seeking participants for their study. Participation requires donation of a DNA sample, which in most instances already exists in your local genetics unit, and clinical information which they will obtain from your doctor. While the group cannot guarantee they will be able to identify the genetic cause in all cases, they have been able to do so in some instances. It is also hoped that by understanding the biology that underlies these diseases, better treatments will be available to future generations. If you are interested in participating please visit our web site <http://www.brainabnormalities.org.uk>.

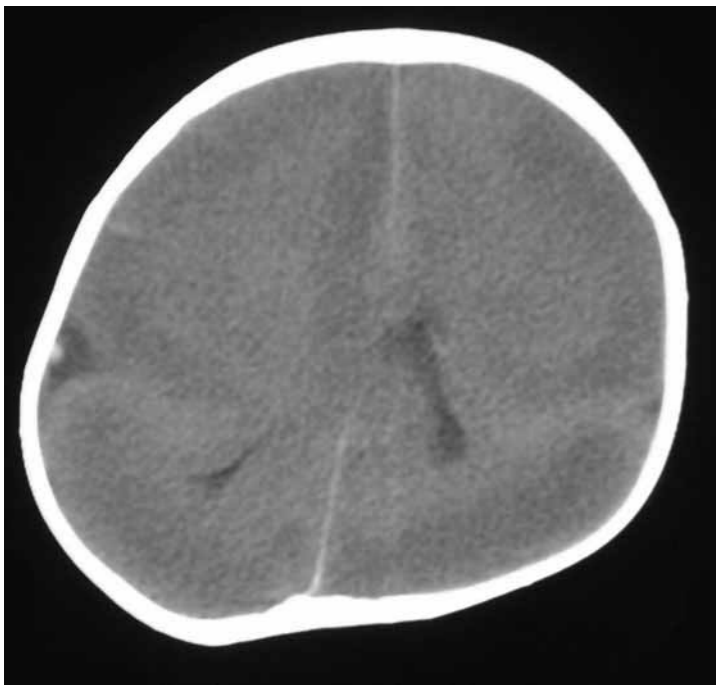


Figure 1: An MRI scan of an individual with lissencephaly, which literally means smooth brain. Patients with this disease, lack the characteristic folds in the cortex, and often exhibit learning disabilities with epilepsy.

Rare Disease Day 2010



Rare Disease Day is marked internationally on the 28th February and Rare Disease UK is responsible for co-ordinating activities in the UK. The purpose of Rare Disease Day is to raise awareness, both among decision makers and the general public, to reinforce rare diseases as a public health priority and to provide a voice for people with rare diseases and their families.

The theme for Rare Disease Day this year is "Patients and Researchers: Partners for Life" as a result we will be seeking to highlight that a lack of research is being conducted into rare diseases. Research into rare diseases is crucial, both in order to gain a better understanding of the condition and the numbers of people affected, but also in order to translate basic research into meaningful outcomes for people with rare diseases and their families. Research into rare diseases can also lead to a better understanding and the development of therapies for more common disorders, for example Gleevec was originally approved for a rare blood cancer but is now used in the treatment of six other life-threatening diseases. The European Commission's Communication on Rare Diseases highlighted that the development of therapies faces three hurdles: the lack of understanding of underlying pathophysiological mechanisms, the lack of support for early phases of clinical development and the lack of opportunity/cost perception from the pharmaceutical industry. As a result Rare Disease Day is an opportunity to call for innovative, multidisciplinary approaches to rare disease research and an investment in sustainable research programmes.

Rare Disease UK will be holding three parliamentary receptions at the Welsh Assembly, the Scottish Parliament and the Northern Ireland Assembly to mark Rare Disease Day 2010. We will be asking our members to contact their MP/MSPs/AMs/MLAs to inform them about Rare Disease Day. We have also been collecting case studies from our members to try gain media coverage of real life stories to raise awareness of rare diseases.

If you are interested in getting involved in our Rare Disease Day activities, please contact us. Also, we encourage our members to mark the day - if your organisation has plans for Rare Disease Day, please do share them with us and we are happy to publicise it on the Rare Disease Day website and in our newsletter.

For more information on Rare Disease Day 2010 please contact Stephen - stephen@raredisease.org.uk or Melissa - melissa@raredisease.org.uk 02077043141

More information is also available on the Rare Disease Day website: www.rarediseaseday.org

PATIENT EMPOWERMENT

Rare Disease Patients can voice their opinion and propose policy scenarios with new Play Decide Games



Summary

As part of its mission to empower rare disease patients in Europe, EURORDIS is proposing a new consultation method that will give rare disease patients the opportunity to voice their opinion about issues that affect them. Read on to find out more and get involved!

Rare disease patients want to be involved in the definition of strategies and plans for rare diseases that are currently being developed at the national and European level. Policy makers understand the need to involve patients so that decisions on science and technology reflect public needs and concerns. How can patients be more effectively involved in decision-making? This is the premise behind PLAY DECIDE.

"The PLAY DECIDE games are designed to empower patients and their representatives to become advocates for their cause, explains Anna Kole, Public Health Project Coordinator at EURORDIS. "The idea of the PLAY DECIDE exercise is two-fold: provide a structure that allows patients to feel safe while learning and discussing a topic that they may know little about, and also equip patients with the tools they need to advocate – facts, examples, and well defined arguments. Although the tools take the format of a game, they are in fact interactive exercises intended for a very serious audience with very serious needs."

In order to achieve this goal, EURORDIS is mobilising its extensive network of patient representatives to organise as many PLAY DECIDE Sessions alongside their membership events in as many countries as possible. The target is to facilitate between 600 and 1000 discussions across 27 countries, in 23 languages!

PLAY DECIDE is part of the POLKA project which seeks to facilitate the consultation of the European rare disease community at large, with the aim of building consensus on preferred public health policy proposals for rare diseases. In the scope of the POLKA project, several PLAY DECIDE Games about topics concerning rare diseases will be made available to EURORDIS members and other rare disease patient groups across Europe until the end of the project. The PLAY DECIDE kits on these subjects will be adapted to most EU languages so that more people can participate.

The following games are available for download in 23 European languages on www.playdecide.org:

- Stem Cell research
- Pre-implantation Diagnosis
- Neonatal Screening
- Cross-border health care
- Is there any upper limit for spending on a single patient: the case of Orphan Drugs
- Diagnosis, Information to the patient and Genetic counselling

Polka

Patients' Consensus on Preferred Policy Scenarii for Rare Diseases

EURORDIS and its partners have selected these subjects because they deal with access to treatment, orphan drugs and with genetic issues, both areas of particular importance for rare diseases. "These issues have ethical and financial implications. They stir great emotional debate in today's society," explains Anna Kole. "It is therefore important that rare disease patients and their representatives are well acquainted with them when they carry out their advocacy activities."

The PLAY DECIDE initiative kicked off in May 2009 at EURORDIS Annual membership meeting in Athens. Since then several national alliances have already promoted the games amongst their membership and games have been organised in Denmark, Italy, Germany, Finland, Hungary and Spain. Many more sessions are planned in the weeks leading up to Rare Disease Day, next month, and two parallel sessions of the European Conference Rare Diseases in May. A prize of 40.000 euros is being reserved for those patient organisations that organise the highest proportion of sessions!

So, do you want to become involved in decisions on critical issues that affect you and other rare disease patients and representatives? Is providing your views in surveys just not enough? Practice voicing your opinion and learn from those who may have other arguments. Perfect your advocacy skills. Go to www.playdecide.org and start playing now! See the game action!

To download the PLAY DECIDE Games and instructions go to: www.playdecide.org

If you have questions about how to organise sessions in your country, contact Anna Kole at anna.kole@eurordis.org.

