



Association for Multiple Endocrine Neoplasia Disorders (AMEND) - Registered Charity 1099796 Trustee Vacancies

(Unpaid, out of pocket expenses reimbursed)

AMEND is a patient support group, providing a wide range of services for individuals and families affected by multiple endocrine neoplasia and associated endocrine tumours. We operate mainly in the UK but also internationally. Our annual turnover is approximately £24,000; we have no premises or paid staff.

We have vacancies for 3 new members to join our Board of Trustees.

We are looking for people who can help steer AMEND towards achieving its mission and ensure the organisation is effectively managed. You would need to be able to make a commitment to AMEND and devote the necessary time; have strategic vision, independent judgment, an ability to think creatively and a willingness to challenge constructively when appropriate. You will be used to operating at a strategic

countywide or regional level and will have extensive experience in one of the following areas:

- Health and social care
- The law
- Accountancy
- Education, learning and development

We are particularly keen to achieve a more diverse Board of Trustees and we welcome applications from all sections of the community.

We will pay Trustees' travel expenses and provide an induction into AMEND's work.

Please email for an application pack. For an informal discussion about the role of Trustee, contact the Chair of Trustees, Jo Grey on 01892 525308.

CaverHubs 2009

www.cavernoma.org.uk

Cavernoma Alliance UK is a charity created by people affected by cavernoma otherwise known as cerebral cavernous malformations or cavernous angioma.

Liverpool - Tuesday 17 November 2009

Venue: The Neurosupport Centre, Norton Street, Liverpool, L3 8LR

Time: 6.00pm - 8.00pm

Speaker: Mr Mohsem Javadpour

LHC: Ian Stuart

London - Saturday 21 November 2009

Venue: The Old Boardroom, National Hospital of Neurology and Neurosurgery, London, WC1N 3BG

Time: 10am - 1.00pm

Speaker: Mr. R. R. Vindlacheruvu

LHC: Ian Stuart

Unconfirmed

Oxford - Saturday 5 December 2009

Venue: TBC

Time: TBC

Speaker: Mr Richard Kerr/ Mr Richard Stacey, Consultant Neurosurgeons
John Radcliffe Hospital, Headley Way, Headington, Oxford, OX3 9DU

LHC: Ian Stuart

Southampton - Saturday 12 December 2009

Venue: Southampton General Hospital
Tremona Road, Southampton, SO16 6YD

Time: TBC

Speaker: TBC

LHC: Eliza Ellerby

Autumn 2009

GIG's Election Campaign Begins

Recent events in Parliament and the Press seem to indicate that the General Election Campaign has started even though it will probably not happen before May 2010. Health and the future of the NHS will be a central issue of the campaign, with all the major parties trying to position themselves as the only real guardians of patient access to high quality healthcare. Yet if you talk to those who work in the Department of Health or in the NHS (and in the equivalent bodies in Wales, Scotland and N. Ireland) there is a strong expectation of hard times ahead, with increasing downward pressure on resources, and the need to do more with less.

All this is happening at a time of increasing possibilities for patients and families. Novel therapies are starting to appear on the market for an increasing number of genetic diseases. This is very encouraging and it gives hope to all of us, but we must not lose sight of the fact that the vast majority of genetic diseases remain intractable and sustained research efforts will be needed to change this situation.

In the coming months GIG will be pressing politicians from all parties to state clearly how, if elected, they propose to address the issue of patients' access to health care including novel treatments where these are available. For example, the role and responsibilities of NICE (the National Institute for Health and Clinical Excellence) has recently been expanded substantially, and politicians need to address how NICE; decisions and recommendations will be implemented, and what their priorities will be in allocating resources to address patient and family needs. In times when belts are tightened it is often the research budget that is among the first to be squeezed. The National Institute for

Health Research (the NHS's research and development arm) has made substantial strides in improving the opportunity for doctors and scientists in the NHS to do research relevant to patients needs, and this progress needs to be continued.

Patient needs and healthcare issues, and making our voice heard amidst the clamour of interest competing for politicians interests, will be a key focus for GIG's Trustees and staff as the general election looms. The present "stirrings" are an indication of things to come, and we are actively developing plans and strategies to secure a positive outcome for all our members no matter which party emerges as victor after the Election.

We will keep you informed about our activities and alert you when we need your help and support - whether nationally or locally - in order to protect and enhance the rights of patients and families with genetic diseases to benefit from high quality healthcare that is effective, timely and user-friendly in the years to come.

Alastair Kent, Director
alastair@gig.org.uk



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CaverHubs 2009

Animal Research – A closer look at the 3Rs

The European Genetic Alliances Network (EGAN) of which GIG is a member, has for four years had an annual meeting with Roche, the pharmaceutical organisation, in which the two organisations can share information and learn from each other's initiatives. At this year's meeting in January, I presented the work GIG has done on lobbying on Animal Research legislation.

For many years now, GIG has regarded the issue of research governance as an important issue for patients. Since such a large proportion of GIG's members work for patients and families affected by conditions for which there is neither a cure nor treatment; research is of course a vital issue for us. Our members need that research to be safe and well governed, but they also need that research to be pushing hard at the boundaries of current knowledge. Legislation has the potential to either stifle or foster this kind of research. Following successes regarding the Human Fertilisation and Embryology Act in the UK and the Advanced Therapy Medicinal Products regulation in the EU, we have been working on the review of the regulation of animal research in the EU.

Our main aim in this work is to prevent any reduction in the scope of the research currently being performed in the EU. Research into neurodegenerative diseases using non-human primates (NHPs), and basic research that can teach us how health conditions act on our bodies are particularly at risk. My presentation in January focussed on this area, and I argued that patients, as the beneficiaries of research should be making this case, as we do. I also discussed the 3Rs.

The 3Rs are Refinement, Replacement, and Reduction. The aim is to Refine animal experiments by improving the experience of the animal, Replace animal experiments with

alternatives, and Reduce animal experiments with greater efficiencies. At the EGAN/Roche meeting, I explained that this movement, acknowledging as it does that in an ideal world no animals would be experimented upon, greatly helps patients to support animal research. I explained to Roche that patients would perhaps be more willing to support animal research if they knew more about how the pharmaceutical industry and the research world tried to implement the 3Rs. As a result of this, I was invited to represent GIG and EGAN as the patient representative on the judging panel of Roche's 3Rs Award. This is a global award, available to researchers and laboratory staff who have made the best contribution to 3Rs either in animal care, or in scientific progress in any of Roche's sites. There were many remarkable stories of excellence in this area. I will describe two.

NHPs are social animals; living in groups is good for them. However, received wisdom has it that adult males are too aggressive to live with groups. The group which won the prize for animal care were those that looked after adult male monkeys. They devised a socialisation scheme which allowed the monkeys to get closer to each other, whilst still allowing for privacy if the animals were scared. Once the monkeys were happily living in pairs, they were allowed to socialise further. Over a period of months, this scheme allowed the adult male monkeys to form part of a large social group, directly contradicting knowledge about the species and improving their wellbeing.

The replacement of animal research is the holy grail of the 3Rs. It is unlikely to happen anytime soon, but the winner of the scientific progress prize shows steps towards this. It is a tool for understanding the metabolism of cells in culture, or in vitro. Fine changes in the cell's electrical properties are measured by this tool, allowing the scientists to learn about the drug's effect on the cell. This work allows scientists to eliminate drug candidates from development without testing them on animals, thereby reducing animal use.

Supporting research using animals is something patients must do, to ensure that all health conditions however rare or complex are eventually treatable. Supporting this work is a great deal easier knowing that a great effort is being made by those involved with the animals to make sure their care is as good as it can be and to ensure they are used only where necessary.

The review of regulation of animal research in the EU is now being examined by the Council of Member States, we hope that they will listen to the patient voice and deliver a proportionate regulation that continues to foster excellent biomedical research in the EU. If you would like any more information about contributing to this patient voice please contact me: [Nick Meade, Policy Analyst. nick@ig.org.uk](mailto:nick@ig.org.uk)



Media Coverage - dealing with sensitive topics

Many of you may recall the media coverage in September this year around the world class champion runner Caster Semenya (who won the gold medal for 800 metres – in Berlin) and the debate surrounding her gender. GIG was contacted by various broadcasters looking for people to interview who are affected by intersex conditions, in order to put a real story to the discussions that were taking place.

I spoke to one of GIG's members Margaret Simmonds who runs the AIS Support Group (AISSG) and she said "We are embroiled in a media circus at the moment and it has been a bit frantic". She also commented that it was often hard for small patient groups run by one or two people, and with no media spokesperson to mobilise resources in sufficient time, to counteract insensitive media coverage when an issue like this one breaks. "We have found that all too often the media, who can react quickly, will grab the first medic who comes along claiming to know something about a topic and then credit him/her with being an 'expert'. This can result in outdated information being given and offensive terminology being used. This is frustrating to patient groups, like ours, who have spent years quietly campaigning for more enlightened attitudes."

For example, in the recent coverage the BBC, without consulting AISSG, used an 'expert' on the Radio 4 Today programme (11th September) who was in fact a general cosmetic plastic surgeon and not a specialist in the field of intersex conditions, even though he was talking about intersex in relation to Caster Semenya. This prompted quite a bit of dialogue within AISSG as people registered their concerns and outrage at the misinformation that was given. The BBC agreed to broadcast another report on the issue and as an AISSG member commented, "with seven million people listening to the programme a good report to re-address the balance will be very useful."

AISSG suggested the BBC should contact Dr. Gerard Conway (consultant endocrinologist, 'Middlesex Centre' UCLH, and a specialist in this field). He reported back that "Finally the Today program did break through [via the UCLH Press Office] and they were clearly conscious that earlier reporting had been offensive." As a result another interview was broadcast on the Today programme (18th September) in which two AISSG members (mother and daughter) and Dr. Conway presented a more human and sensitive view of living with an intersex condition.

Another AISSG member, Sarah

Graham, also managed to put forward a balanced article, "I really feel for Caster.. I've been through it", that appeared in the Mirror on 14th September. She and Margaret had managed to dissuade the Mirror from printing too many quotes from the same cosmetic surgeon in an explanatory box alongside Sarah's article. Sarah said of her experience "I've been getting lovely messages from people via my website [following the Mirror article] - including parents of intersex children[...]. Initially I was reluctant to do this intersex media work - before my identity as therapist and my company is established - but I am also aware that I am in the very lucky position of having the skills and support around me to be out there in the way that most can't."

AISSG members have been hard at work writing to newspapers and journalists to counter their stories and claims and directing people to some excellent information and advice which has been added to the AISSG website <http://www.aissg.org/PDFs/aissg-caster-semenya.pdf>. This document outlines the terminology that should, and should not be used and gives clear guidance for journalists and editors.

GIG has lots of experience of dealing with the media over sensitive issues, particularly the work we did surrounding the Human Fertilisation and Embryology Bill last year. It can often be difficult to convey complex information quickly and easily and all too often the 'experts' that come forward are not the true specialists in the field. However, it is encouraging that the media are looking in the right places for good quality information, coming to patient groups is a good start. Clearly, as Margaret has noted it can become rather frantic so please do remember that if you are unable to speak to journalists you may always signpost them to GIG as we can often provide quotes and information on genetic topics.

Melissa Hillier,
Assistant Director
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What is AIS?

Androgen Insensitivity Syndrome (AIS) is one of a number of biological intersex conditions. Intersex results from a variation in the embryological development of the reproductive tract, often determined by a known genetic mutation.

www.aissg.org



RARE DISEASE | UK

The National Alliance for people with rare diseases & all who support them

Update on Rare Disease UK

It has been a busy summer for Rare Disease UK. On 9th June the Council of the European Union's Recommendation on an action in the field of rare diseases was adopted unanimously by each health Minister of the European Union's 27 Member States. Rare Disease UK had been campaigning hard to ensure the UK Government adopted the Recommendation so this was great news for our campaign. Amongst other things the Recommendation calls on Member States to develop plans or strategies for rare diseases. Whilst the Recommendation carries political weight, it is not binding so we now have to ensure that effective strategic planning for integrated service delivery for rare diseases is implemented in the UK.

Over the summer we have been meeting with key figures in the Department of Health, devolved governments in Scotland, Wales and Northern Ireland and the four NHSs in the UK. Generally we have received positive responses and the need to improve the service for people affected by rare diseases has not been disputed. We will continue to keep the contacts that we have made updated and we are looking to meet with more key figures in the near future. Hopefully we will be able to work together to develop an effective strategy to deal with rare diseases.

We have set up five Working Groups to look at various aspects of a strategy for rare diseases in the UK. Each group has multi-stakeholder representation including academia, clinicians, industry, patient organisations and commissioners.

The five Working Groups cover:

- Co-ordination of research
- Prevention and diagnosis
- Commissioning and planning
- Patient care and information
- Delivering co-ordinated care

The aim of the Working Groups is to highlight the key issues that need to be addressed and to come up with a set of measures to evaluate any strategy that is put in place. Hopefully this will assist the

implementation of a strategy in the UK. The Groups will be having their first meetings in October and November.

Over the summer one of the things that became apparent was the need to increase awareness and membership of Rare Disease UK. As a result we have had new promotional material printed (included with this newsletter). We have also made plans to attend a number of conferences and events over the coming months with the aim of raising awareness in health circles. In order to keep our members and other contacts updated on our work we started a monthly e-newsletter. We have also been preparing for our fringe debate at the Conservative Party Conference which is taking place on the 6th October.

Please note that as a member of GIG, you are not automatically a member of Rare Disease UK – to join please visit <http://www.raredisease.org.uk> Membership is open to everyone including, individuals, patient organisations, clinicians and academics.

Stephen Nutt, Public Affairs Officer
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Self Management Project for Young People Launched in Scotland

Living with a long term health condition can be challenging for a young person. These issues can include remembering medication routines and medical appointments, interruption to schooling and coping with the effects of illness amongst others. In addition to all that, young people have to cope with the physical and emotional changes of adolescence. They are moving from primary to secondary school; worrying about relationships; getting on with their parents or carers; while of course wanting to do all the same things as their mates. These changes and challenges can seem insurmountable and many young people diagnosed with long term health conditions can experience a mix of emotions ranging from loss and grief - 'I'm not the same as my pals' - to anxiety about the future, - 'Will this affect me getting to university?'

Stay Well Lanarkshire is our innovative new three year project which aims to empower young people to live well and healthily and to manage their lives during this period of change, so that their health condition is only a part of and not the main factor in their life. Project Officer Karen Martin and trained volunteers will work with groups of young people between the ages of 11 and 18 who have a chronic condition or illness. Our project is not disease specific and the self management programme will be suitable for and open to any young person whatever their chronic condition. It aims to promote resilience, coping resources and to develop life skills in young people. Specifically we will look at developing skills in communication, decision making and problem solving. All self management sessions will be held in community venues easily accessible by public transport, and will take place over an eight week period. Each session will also include a fun activity chosen by participants. At the end of the programme we hope that young people will go on and contribute to further self management/ peer support activities and there will be an opportunity to meet again six months after the initial course to catch up with each other.

There will also be a chance for participants to take part in our awareness raising work with schools and fellow peers. This will inform people of their own age and those who work with young people about the realities of living with long term health problems and how they can help the young person cope. Information about the project is being developed and will be distributed initially, through outpatient clinics, schools and youth projects throughout Lanarkshire.

Young people interested in taking part can get in touch with the Project Officer for further information about self management sessions etc. In the meantime for more about the project, or about volunteering as session worker, contact:

Project Officer Karen Martin on
ascs@btconnect.com
Phone 07935 305930.



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Asking Relevant Questions - Update

There have been some interesting developments in the exciting world on insurance recently. Firstly The UK Forum for Genetics and Insurance (UKFGI) commissioned a research document looking at the current Moratorium and Concordat. The research is made up of interviews from key stakeholders and the results have made for intriguing reading, demonstrating the continued support not only from the Government and patient support organisations but also from the industry itself.

GIG has also been involved in consultations looking at the wording of Critical Illness Cover policies and the possible insurance implications of obtaining a direct to consumer genetic test. Alongside all of this, GIG has been working to improve information for consumers in relation to insurance and genetics and is working on a revamp of the document provided to consumers by the Association of British Insurers. As part of this ongoing work GIG has been working with Which? to develop a handy guide to buying Travel Insurance. "The 5P's Guide To Travel Insurance" can be found on the projects section of the GIG website.

Ben Francis, Project Officer
Insurance Templates ben@gig.org.uk



FEATURE ON NANOMEDICINE

Nanomedicine – Patients Views and Understanding of this new area



As reported in previous GIG Today's, we are the Patient Lead on the

Nanomed Round Table Project. This is a "Coordination and support action" project funded for one year by the European Commission in its Seventh Framework Programme (FP7).

Nano-medicine is a newly developing field in research and development and genetic conditions stand at the forefront of those who stand to benefit from this new technology. This project has been funded to gain a broader understanding of various stakeholders' knowledge, views and opinions.

The Round Table is Chaired by Professor Sir John Beringer, formerly Pro Vice-Chancellor, University of Bristol and who led the independent review of the UK Government's response to the 2004 Royal Society & Royal Academy of Engineering nanotechnology report. The Working Groups will be focusing on the following five areas: -

- Regulation
- Economic Impact
- Communication
- Patient Needs
- Ethics & Societal Impact

GIG is leading on the Patient Needs Work Package and as such carried out a short pilot study of patient views. Readers may be familiar with the recent survey request that appeared on the GIG website and was also sent to patient members of GIG. Coupled with the information we gained from this short survey GIG also carried out a series of 12 patient interviews in order for us to learn more about individual's knowledge and views on nano-medicine, particularly if people had heard of the use of nano-medicine in relation to their condition.

The full findings from this pilot study will be presented to the European Commission as part of the final Nanomedicine Round Table Report in December 2009. I am very pleased to be able to share a few of the preliminary findings from the Patient Needs Work Group in GIG Today.

SUMMARY of Nanomedicine Pilot Study

- There are high levels of support among patients for nanomedicine products and research.
- Patients have relatively low levels of knowledge and awareness of nanomedicine, but they would like more information.
- Patients have clear views on how and from whom they would like to receive this information.
- Patients do not think that nanomedicine is inherently unsafe, but there is a lack of clear understanding about the potential safety aspects.

- Nanomedicine is an opportunity that patients want to see embraced. Equally, there is a significant and time-critical opportunity to inform patients about nanomedicine.

Patient Survey and Interviews

In the patient survey we carried out 60% of respondents had not heard of nanomedicine research being carried out in their condition area, however nearly half of respondents were supportive of nanomedicine research being undertaken in general. Within current funding levels people were supportive of nanomedicine being allocated increased funding (60%) and nearly 80% of respondents would like further information on nanomedicine. To add to this survey data GIG carried out semi structured interviews with 12 patients to gain more detailed information. Each participant was asked a series of questions, and their responses were gathered and analysed.

The interview questions were as follows:

- What is your exposure to nanomedicine or nanotechnology?
- Have you heard of nanomedicine being used in your area of interest before?
- Knowing this information do you think that nanomedicine research should be continued in this area?
- Do you have any concerns about the use and safety of this technology?
- Do you think that nanomedicine will be useful in the treatment of your disease?
- What type of information would be useful to you about nanomedicine and where would you go for the information?
- In your opinion is nanomedicine completely different from current medicine, or is it the next step in medical research?

What is your exposure to nanomedicine or nanotechnology?

This question gave the most consistent answer. Eight of the patients surveyed had never heard of nanomedicine and four had heard of nanomedicine, but didn't realise that it was nanomedicine. All of the genetic disorder groups interviewed in this survey had the same prior exposure, which was very little to no exposure to nanomedicine.

Participant A had read about the protein sets that are removing the defective genes in Huntington's patients, "but I had not realised that it was nanomedicine research."

Participant H had heard of both nanomedicine and research into Parkinson's: "Pretty much I knew of it and knew the implications in Parkinson's, but I thought that the application would be last on the list."

Have you heard of nanomedicine being used in your area before?

The responses to this question revealed a lack of knowledge in nanomedicine amongst patients and their carers. Half of the patients interviewed had never heard of nanomedicine being used in their area, and only four interviewees had heard of it in a general sense.

Participant D, a Parkinson's patient, had not heard of nanomedicine in Parkinson's but had heard of its use in cancer treatment. Another Parkinson's patient, Participant F, said: "I am aware of the interest and the general sense of the research, but didn't know that it was nanomedicine."

This question revealed that many of the patients had not heard of the research being conducted in their interest area. It also showed that some have heard of the research, but did not realise that it was nanomedicine research.

Knowing this information do you think that nanomedicine research should be continued?

All but one of the patients and carers interviewed believed that research into nanomedicine should be continued based on the information presented in the PowerPoint presentation that was made prior to questions being asked, and which explained what nanomedicine is and gave an example of its current use.

Participant J said: "It sounds as though it has a lot of potential." Participant G said that he thought that, "It would be interesting to see which phase the research is currently in," while Participant B also supported nanomedicine research but noted that: "public opinion research needs to be done to get information out on both patient feelings and knowledge of the subject."

In general, however, the participants of this patient survey believed that nanomedicine research should continue. Many had concerns about the development of the research, but made a risk judgment and felt that with appropriate regulation nanomedicine was just another avenue of research.

Do you have any concerns about the use and safety of this new technology?

This question elicited a wide array of answers with some patients not having any concerns about nanomedicine, whereas others did.

Participant J had concerns about the risks to the patient but believed that: "There are always risks when it comes to testing new technology, but you can't get away from it and you can't let it make you shy away from research."

Participant B said, "I have less concerns for this than gene therapy," but was nonetheless concerned with "the public understanding on nanomedicine. I would like for someone to get across what the advantages and disadvantages are to patient groups, and am concerned with the use of the name nanomedicine. Is it too late to change the name?"

Do you think that nanomedicine will be useful in the treatment of your disease or area of interest?

The majority of patients interviewed thought that nanomedicine would be useful in the treatment of their disease or condition.

Participant D thought that: "It is impossible to tell really because the underlying cause of Parkinson's is still unknown, so until the cause is discovered I cannot see its possible helpfulness in Parkinson's treatment."

Participant G, a biochemist and Parkinson's patient, believed that: "It would be useful in the treatment of most diseases, but a problem could arise when there are multiple genes responsible for a disorder." Participant E was more sceptical: "I hope so, but I am not convinced."

What type of future information would be useful to you about nanomedicine and where would you go to get the information?

Over half of the patients interviewed would like to get the information from patient organisations, while one quarter would like to get their information from the internet, and just one patient would like to go to a physician to get the information.

In your opinion is nanomedicine completely different from current medicine, or is it the next step in medical research?

Over half of the patients believed that nanomedicine is the next step in medical research, while patients like participant D believed that it is "Completely different. It's studying something very small that we still don't know much about and will open new worlds in medicine." Participant H echoed this: "No, I think it is something completely different in terms of technology."

To the participants, nanomedicine is both something new and a continuation of medical research. It is new concerning the size and characteristics that the medicine is exhibiting, but it is a continuation of the research because the tools used in medicine are constantly getting smaller and the scope of treatment is getting more focused on the diseased area.

In summary, this pilot study has demonstrated that there is very little knowledge about nanomedicine and its research in the patient population. All the patients interviewed wanted research to continue in not only their field, but in all areas of medicine. They also wanted more information to be presented to them through either their patient organisations or on the internet on nanomedicine, but in a much more lay friendly format, with the pros and cons presented to enable them to make informed decisions on the treatments that they have.

Melissa Hillier, Assistant Director
melissa@gig.org.uk

GIG Welcomes a New Trustee



Richard West
Behçet's Syndrome Society

Since 1995, I have been living with Behçet's Syndrome Society, a rare, complex and lifelong illness - it is an inflammation of the capillaries and can affect any part of your body internally and externally. The medical profession believe there are genetic links with our illness but it has no cure, no diagnostic test and like a lot of others suffering from a rare disease, it is

difficult to obtain the correct diagnosis and treatment. My condition is suppressed by medication.

I am a Trustee and volunteer Secretary of the Behçet's Society which has about 600 members in the UK. It is known as the Old Silk Route Disease and is much more common in Turkey, China and Japan.

I have just been elected as a Member of the National Assembly for National Voices which is the main voice for patients in relation to Health and Social care in England.

I have come into contact with GIG over the last two years and I have seen at first hand the excellent job they do for people with Genetic conditions and Rare Diseases and I would like to help them in any way I can with their work.

I am looking forward to working with others at GIG to:

1. Increase our influence on policy makers about living with genetic and rare conditions that can have such a devastating affect on your life and those around you.
2. To help GIG improve their services and information to members.

I am an investigator and I work for Insurance companies and Solicitors dealing in all types of accident investigation work.

Richard West
Secretary Behçet's Syndrome Society

14th International Behçet's Disease Conference

The ISBD (International Society for Behçet's Disease) holds a medical conference for clinicians every 2 years, and it is London's turn to host the event in 2010. Running alongside the medical conference is a patients' conference that next year will be organised by the Behçet's Society. The Society were invited to join the organising committee in 2008 and have met regularly to make the event as meaningful as possible for clinicians and patients alike. The opportunity for patients to hear from specialists in Behçet's disease from all around the world is a unique one, and with the last two meetings being held in Portugal and Austria and the 2012 one to be in Japan, we do hope to have as many UK visitors as possible next year.

We have already had enquiries for details from patients in the USA and Japan, so clearly people are willing to travel many miles to get the latest information. The venue is the Queen Mary University complex near Mile End in the East End of London, and the dates are 8-10 July 2010. The university itself has gone through a major rebuild recently, and the facilities are very comprehensive and of a high standard. Of particular interest to patients is the onsite accommodation, which although billed as student accommodation is of excellent quality and offers various combinations of single and twin rooms in flats with self-catering kitchens that will enable delegates to produce food to their own liking and to keep costs down. The accommodation will be charged at under £50 a night, which is very reasonable considering the London venue.

Programme and events

The medical programme is still being agreed but will certainly include immunology (including therapy), vasculitis, inflammation, paediatrics and genetics, as well as debates about autoimmunity versus auto inflammation and the geographical differences in Behçet's disease. Medical speakers from around the world will join the patients' conference to offer various topics, but we will certainly have sessions on new drug treatments and research and the psychological effects of a chronic disease, as well as topics covering every aspect of the disease itself. Many poster contributions are also expected, so that the latest ideas can be presented and discussed directly with the person who has conducted the research or who had the original thought. The Society will host a social event on the Friday night with a buffet and some entertainment for those who wish to chat with friends old and new from around the world.

There is also a formal Gala dinner being planned to be held at St Bartholomew's Great Hall on the Wednesday for those who would like to experience a wonderful venue with excellent food. A sightseeing tour will be organised for the Saturday, and we will also involve carers and partners, a group that is sometimes taken for granted and whose input is not always catered for. The website (www.icbd2010.com) is under development and will include booking for patients in due course.

www.behçets.org.uk | info@behçetsdisease.org.uk

Putting Patients at the Heart of Clinical Trials across Europe



The PatientPartner Project started a little more than a year ago. It is a three year funded project, and is part of the 7th Framework Program of the EU. GIG will be working on the project for one year; the main end goal from GIG's participation will be to identify the main areas that need attention to create guides for Patient Organisations, and for organisers/sponsors of clinical trials, also, to develop a list of recommendations and a thematic website.

The PatientPartner Project has made several advances in the last couple of months. The start-up workshop which allowed us to identify the main areas that need attention took place in the beginning of June. With the information gathered, we managed to take a few steps forward, for instance, an online forum for the European Network of Patients Partnering in Clinical Research (ENPCR) was created so that Patient Organisations may communicate with each other, and discuss issues that are of importance to them, and also to get advice from one another. The information gathered from the start up workshop was also very helpful and important for the next workshops to follow, as the information collected would lead the topics of the next workshops.

The Surveys and interviews that were conducted across Europe the previous year by Kim Wever from the VSOP in the Netherlands are being finalised and will be posted on the PatientPartner website

www.patientpartner-europe.eu This is of great help to the project, as with the interviews and surveys, one can identify the key issues concerning clinical trials, both from the clinicians'/researchers' perspective, and the patients'/patient organisations' perspective.

At the present time more interviews are being conducted with researchers, pharmaceutical companies and doctors across Europe, so that a general understanding as to the situation in different parts of Europe, concerning the difficulties they are facing with clinical trials can be identified.

The countries taking part in the project have been grouped into three; Southern Europe, Central Eastern Europe, and North-Western Europe.

For the time being, preparations for the coming workshop are being made. We at GIG will be giving a presentation on Patients' Rights Charters for the different countries within the EU.

This workshop will be for North-West Europe, and will be taking place in London, on the 12th-13th October. One can sign up to attend the workshop by going to the PatientPartner website www.patientpartner-europe.eu. The workshop will focus on what role Patient Organisations should play in clinical trials, Patients' Rights Charters across Europe with a focus on the North-West, Academia's and Industry's future actions in fulfilling their relationship with Patient Organisations.

Ariadne Stamatopoulou
Patient Partner Project Officer
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EACH Child and Young Person's Health Matters



Action for Sick Children Scotland, is the only charity dedicated to informing, promoting and campaigning on behalf of the needs of ALL sick children in Scotland. Action for Sick Children is hosting a Garden Lobby event at the Scottish Parliament on Wednesday 2nd December, 2009 from 6 to 8pm EACH Child and Young Person's Health Matters, hosted by Christine Grahame, MSP and Convener of the Health & Sports Committee. This event will mark the start of a year-long campaign on the healthcare rights of children and young people at times of illness, promoting the work of the organisation through the medium of the EACH charter by showcasing our local projects as practical examples of how the EACH charter standards apply in practice. We will be sending out invitations and further information nearer the time but in the meantime please note this for your diary. We hope that as many people as possible will be able to come along and hear about our work and our campaign.

If you would like your name to be added to the invitation list please contact Action for Sick Children (Scotland) national office at or Tel: 0131 553 6553

Medical Advisory Service - The Year of The PIG!

Celebrating over 20 years of nurse run helplines and explaining the MAS interest in the Genetic Interest Group:

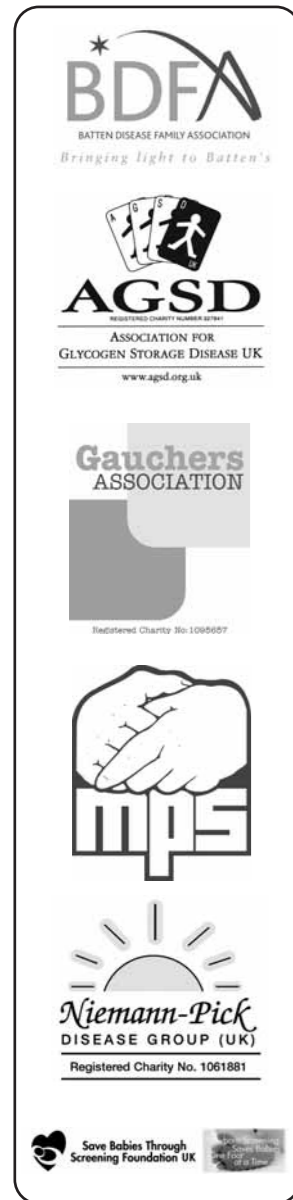
Media brouhaha surrounding the real and/or predicted threat of swine flu to consumers, the NHS, industry and patient groups continues to fill column inches and create anxiety. This is especially so when it comes to contingency in providing much needed services and support. At MAS we are putting in place special training for our work injured helpline staff and strategies for communicating both enquiries and the individual health needs of our work force. This is particularly important that this time as the Charity currently runs several support/helplines for other organisations and is developing more projects month on month.

A positive advantage of the Medical Advisory Service recruitment and work model, is, the isolated nature of our helpline staff ñ hopefully unlikely to move among contagious communities and, as at risk groups due to illness or disability, more likely to receive prompt preventive care.

All this adds up to a positive approach to a possible calamity! In communicating this, should any GIG members or recipients of GIG communications need telephone support or back up services ñ do please bear us in mind. We are currently recruiting nursing advisors through the RCN (Royal College of Nursing) and the NMC (Nursing and Midwifery Council) we hope our next triage training will take place in West London in March next year.

For information or help please visit our website: medicaladvisoryservice.org.uk Or call: 020 8995 8503

UK LSD Patient Organisation Collaboration



Lysosomal storage disorders (LSDs) are a group of approximately 40 rare inherited metabolic disorders that result from defects in lysosomal function. Lysosomal storage disorders result when a specific organelle in the body's cells – the lysosome – malfunctions. All lysosomal storage disorders share a common pathogenesis: a genetic defect in a specific lysosomal enzyme, receptor target, activator protein, membrane protein, or transporter, leading to accumulation of substrates in cell lysosomes.

Patient Organisations representing those affected by Lysosomal Storage Disorders have joined together to form a new action group to work and lobby on behalf of LSD patients and their families in the UK.

The Group is made up of representatives from the Association for Glycogen Storage Disease, Batten Disease Family Association, The Gauchers Association, The Society for Mucopolysaccharide Diseases (the MPS Society), and The Niemann-Pick Disease Group (UK).

Our Mission Statement: To undertake joint promotion and a shared understanding of LSDs, to advance standards of care and to enhance the wellbeing of those affected. To stimulate interest and work in partnership to establish a Forum in which members can discuss together matters of common interest and dissemination of good practice amongst them.

Objectives:

- To enable the development of a stronger voice with which to influence national policy, stimulate interest in and further the knowledge and understanding of, Lysosomal Storage Disorders.
- To encourage close working relationships between Member Associations, in order to facilitate the sharing of information and the effective use of resources.
- To oversee the provision of clinical care for those affected by LSDs; to promote clinical efficiency and best practices in the health service.
- To share information regarding potential therapies and treatments for LSDs.
- To consider the cost implications of potential treatments and therapies; to support and encourage all those affected by LSDs in accessing treatments and therapies.
- To seek to provide a positive influence, leading to increased engagement in the delivery of government policy on LSDs and other rare diseases.

- To consider ways of encouraging new initiatives; including the commissioning of further research into the causes and possible treatments of LSDs.

To be achieved by:

- The dissemination of ideas, examples of good practice and other relevant information to researchers/health care professionals in the UK.
- The provision of considered and co-ordinated advice and information regarding LSDs to all interested parties.
- The establishment of strong links with industry, researchers, clinicians and individuals with an interest in LSDs.
- The implementation of a well-organised campaign to highlight the need for effective, accessible and safe therapies for LSDs and better need for health and social care.
- Lobbying the government and other relevant agencies for improvement in clinical standards of care for those affected by LSDs, including; the early and accurate diagnosis of, and screening programs for, LSDs.
- Through combined activity, the provision of an effective contribution to the updating and ongoing improvement of the highest possible standard of clinical care.
- Working in partnership to raise awareness of LSDs amongst health and social care professionals and the general public.

LSD UK Patient Organisation Secretariat Tanya Collin-Histed Executive Director Gauchers Association Ltd. 3 Bull Pitch, Dursley, Gloucestershire, GL11 4NG E: tanya@gaucher.org.uk T: 01453 549231

In 2009/2010

- In 2009/2010 our work programme will include the following:
- To assist in the development of a patient group for Metachromatic Leukodystrophy (MLD)
 - To organise a family day to bring families together and then support the development of a Metachromatic Leukodystrophy / Leukodystrophy Patient Organisation
 - The development of a Clinical Study Group for Inherited Metabolic Disorders in partnership with the Medicines for Children Research Network (MCRN)
 - To develop an action plan for the continued designation of LSDs as a specialised service post 2012
 - To organise a half day workshop on Transition for LSD patients and their families
 - To organise an LSD patient meeting in Scotland and Northern Ireland

Who are the Members of the UK LSD Collaborative?

- Association for Glycogen Storage Disease: Allan Muir
- The Batten Disease Family Association: Jan Sablitzky
- The Gauchers Association: Tanya Collin-Histed
- The Society for Mucopolysaccharide Diseases (MPS Society): Christine Lavery
- The Niemann-Pick Disease Group (UK): Toni Mathieson
- Save Babies UK - Pat Roberts

Join Us: We are keen to open membership of the Group to other LSD conditions.

If you would like more information contact: Tanya Collin-Histed, Executive Director, The Gauchers Association, by telephone: 00 44 1453 549231, or e-mail at Tanya@gaucher.org.uk

Unique study days

Honestly, we didn't expect it - but 2009 has been an amazingly good year for Unique, the Rare Chromosome Disorder Support Group. We've won a string of awards - a GlaxoSmithKline Impact award, a Jeans for Genes award and generous backing from the Grocers' Charity.

We've decided to use the funding awarded by Jeans for Genes and Grocers' to run a series of groundbreaking study days in 2010 for families affected by a rare chromosome disorder. Most of these families, and the professionals who care for them, have never before met another person with their particular disorder. The disorders aren't unique but they are extremely rare. Look at the list below, how many of the disorders have you heard of? Just take a moment to imagine the isolation and uncertainty that that brings to families and carers.

Rare chromosome disorders usually involve the loss, gain or rearrangement of genetic material. This genetic disruption has a lifelong impact: these disorders are never curable. There is no replacement therapy. Some of the disorders are severely life-limiting while others scarcely affect longevity. The range of effects is huge and confusing. But almost everyone with a significant rare chromosome disorder has special needs – a pick-and-mix selection of intellectual disabilities, birth defects, medical problems, challenging behaviours and mental health issues.

The five Unique study days will bring families and professionals together to share their knowledge and experience and to learn from each other. Geneticists, paediatricians, behaviour experts and a range of therapists will tell families what they know, and families will reciprocate. World-renowned experts will be there: for example, Professor Peter Hammond, famous for his 3-D facial constructions of various genetic and chromosomal syndromes, will be with us. Research studies will be stimulated and new findings reported for the first time. Older families will be able to share longer-term outcomes. New families will be helped to take the first tentative steps towards the understanding and acceptance that eventually follow the shock and grief that the diagnosis of a rare chromosome disorder brings.

Unique's study day series is as follows:

Deletions from chromosome 4q
9q34.3 deletions
8p23.1 deletions
2q37 deletions
Pallister-Killian syndrome

If you would like any more information about the study days, please contact Prisca Middlemiss at Unique on . If you would like to know more about Unique please visit our website at . If you have a specific query about a rare chromosome disorder, email us at . You can also follow us on Twitter (or #rarechromo).

Great news for GIG Member - Alstrom Syndrome UK

A £90,385 grant from BBC Children In Need is to fund a three-year post for a Child Development Worker for ASUK.

This will increase children's opportunities to socialise through activity weekends, liaising with schools to increase understanding of their needs and working with children themselves to ensure they achieve their potential. Due to the rarity and complexity of Alstrom syndrome, children's development is often hindered through late diagnosis. Children are often lonely, bullied and mistreated through lack of understanding. Kay Parkinson, Chief Executive and Founder of Alstrom Syndrome UK, said "Children with Alstrom Syndrome need a lot of help and support to develop fully. This new post will enable us to ensure we meet all their needs - rare diseases are often overlooked so we are very grateful to BBC. BBC grant opens up more opportunities Children in Need for funding this vital post." The move has been welcomed by Dr Richard Paisey from Torbay Hospital and Professor Tim Barrett from Birmingham Children's Hospital. The new Child Development Worker is expected to start in the New Year.

What is Alstrom Syndrome?

Alstrom Syndrome is characterised principally by a number of key conditions:

- Retinal degeneration (inherited progressive eye disease)
- Sensorineural hearing loss (disorders of the cochlear part of the ear)
- Obesity
- Insulin resistance

Additional features can include:

- Renal and hepatic dysfunction (affecting the kidneys and liver)
- Type 2 diabetes mellitus
- Hypertriglyceridaemia and tryglycerides (elevation of fatty substances found in the bloodstream)
- Cardiomyopathy (poor cardiac function where the heart muscle is weakened and enlarged)

