



The Health and Social Care Bill continues to show promise

Getting the law right plays a central role in determining whether or not a patient or a family will be able to get services and support in a timely and effective manner. Making law is more of an art than a science, and it tends to be reactive rather than proactive in its approach to problem solving.

The Health and Social Care Bill which, at the time of writing has completed the Commons stage of its route to the statute book and is shortly to go to the House of Lords is a case in point of this backwards looking tendency on the part of legislators. While there is considerable agreement about the challenges facing the NHS - the pressures that are created by financial constraints, new possibilities and rising public demand - there is much less of a consensus about what to do to resolve this.

Elements in the Bill incorporate developments that Genetic Alliance UK has been calling for for many years.

The possibility for creating a uniform pattern for commissioning and providing specialised services across the whole of England and the eventual elimination of the postcode lottery that the current system has created is a significant advance. We have welcomed this development and will be campaigning to make sure that it is not lost or watered down when the Bill is debated in the House of Lords – especially in the light of some calls for a greater local say in planning that run the risk of marginalising specialised services. But we need to press for a proper allocation of resources to this aspect of the NHS or there will be a levelling down of the best to the average, not a raising up of the weakest through the adoption of national standards.

This is good, and it addresses a long-standing problem but the only radical element to the proposed changes is the recognition that for some very special aspects of healthcare the level for planning is the country as a whole (and this does not include Wales, Scotland and Northern Ireland, each of whom is grappling with similar issues, with the added complication of having smaller populations on which to base decisions). But the Bill misses a real opportunity to turn the NHS into a future facing organisation. Advances in our understanding of genetics and genomics will radically transform our understanding of the processes of health and disease in the next 15 or 20 years. Unless we recognise this and act now the NHS will not have the right infrastructure to benefit from these advances, and the staff who work in it will lack the relevant skills and knowledge to bring new clinical possibilities into their everyday clinical work.

The Human Genomics Strategy Group in the Department of Health, under the chairmanship of Sir John Bell is soon to produce a report on the potential implications of this new knowledge for the NHS and for healthcare related R&D. The UK's Health Departments are shortly to launch a consultation on a UK plan for providing services for patients with rare diseases.

Continued on page 2...



Alastair Kent, OBE, Director,
alastair@geneticalliance.org.uk

In this Issue

The NHS
Commissioning Board
and NHS reform
Pages 2 and 3

Facilitating
Networks
Pages 6 and 7

R.A.P.I.D
project update
Page 8

Risks and Benefits:
citizens jury
Page 4

The NHS Commissioning Board and NHS reform

The NHS in England is changing. Evidence of this can be seen as Clinical Commissioning Groups spring up all over the country, and Primary Care Trusts cluster together.

Even before MPs and Peers have finished debating how our healthcare system will be described in the legislation, the reorganisation is well under way.

We have been following the progress of the Health and Social Care Bill closely as it progresses through Parliament: it is now at the House of Lords with the next scheduled reading being the second reading in October; but we would be remiss if we did not seek to influence the restructuring of the NHS as it happens.

The NHS Commissioning Board (as it is described in the draft legislation - this name is likely to change) will be in charge of many important components of the new NHS. We have therefore written to its architect and future Chief Executive, Sir David Nicholson, to outline our key concerns. Our letter was sent at the end of July, and we have now received a response.

The NHS Commissioning Board (NHS CB) has responsibility for developing the whole commissioning network, defining the culture of the new NHS and many

Duties of the NHS Commissioning Board

- To agree and deliver improved outcomes and account to Ministers and Parliament for progress. There will be a clear mandate, setting out expectations for the Board and the broader commissioning system;
- To oversee the commissioning budget, ensuring financial control and value for money;
- To develop and oversee a comprehensive system of clinical commissioning groups with responsibility for commissioning the majority of healthcare services;
- To commission directly around £20bn of services including specialised services and primary care services (including holding around 35,000 contracts for primary care services);
- To support quality improvement by promoting consistent national Quality Standards, a culture which promotes research and innovation and providing world class support for clinically led service improvement and leadership;
- To promote innovative ways of demonstrating how care can be made more integrated for patients;
- To promote equality and diversity and the reduction of inequalities in all its activities;
- To develop commissioning guidance, standard contracts, pricing mechanisms and information standards;
- To engage with the public, patients and carers, champion patient interests and ensure patients have access to a wider range of information about services;
- To develop a framework to make choice a reality for patients, setting out guidance in consultation with Monitor about how choice and competition should be applied to particular services;
- To oversee planning for emergency resilience and lead the NHS operational response to significant emergencies; and
- With its partners, develop a medium term strategy for the NHS, which alongside the local priorities developed through health and wellbeing boards, helps form the basis for local commissioning plans.

Taken from "Developing the NHS Commissioning Board" 8th July

<http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/>

Introduction continued from page 1...

Both of these are hugely significant developments, but they will be released after the legislative stable door has been closed on the current Bill.

At Genetic Alliance UK we are working closely with colleagues in the Department of Health and the NHS across the UK to keep options open so that the introduction of new clinically significant possibilities is made easier rather than more difficult by the frameworks currently being put onto the statute book. It will be a shame if the rush to re-organise results in a framework which is less good than it might have been, given all the upheaval that is currently underway, when lifting the gaze from the here and now and looking at what is coming down the track might have produced a better result for patients, families and the NHS in the years to come.

Key demands of our letter to David Nicholson of 28th July 2011

- For a designated national director at the highest level of the NHS Commissioning Board (NHSCB) with responsibility for specialised services;
- For the board of the NHSCB to include a non-executive member to champion this aspect of the organisation's work;
- That clinical senates should include representatives with rare disease knowledge;
- Address the risk of patients with rare and complex genetic conditions falling through the gaps in a system measured with high-level overarching indicators and improvement areas;
- That the budget for specialised services be protected and allocated specifically to specialised commissioning;
- That the regional structures outlined in the "Developing the National Commissioning Board" document are for delivery of service rather than for regional commissioning;
- That as staff numbers are cut from approximately 8000 to approximately 3500, it will be essential that sufficient expertise is preserved to ensure the continuation of the complex commissioning roles that they currently perform.

other things described in the box to the left including, vitally, £20 billion of direct commissioning. Some of this goes on primary care, such as dentists and GPs. (Yes, though GPs will be commissioners in the new NHS, they will not be allowed to commission the healthcare they provide.) Of the rest of the £20 billion, we expect about half, will be spent on specialised commissioning. This is commissioning of services for small populations of patients currently carried out at either a national or a regional level.

As we stated in our letter to Sir David, the creation of NHS CB, in a position within the NHS to have oversight of the commissioning of all services, and the power to directly commission appropriate services, provides an opportunity to cut through many of the issues of access and integration that face patients with rare, complex genetic conditions.

We felt though, that we should make our wishes clear on how we see the

NHS CB best functioning, and that we should try to get some assurances on how specialised services will be delivered in the new NHS. You can see a list of our points in the box above.

Sir David's reply is encouraging. He states that specialised services will be a clear focus of the NHS CB and that one of the eight national executive directors of the NHS CB will be identified as the 'lead' for specialised services. Regarding the budget, he explains that the figures are still being calculated, and explains that this will be based on actual spend for current services.

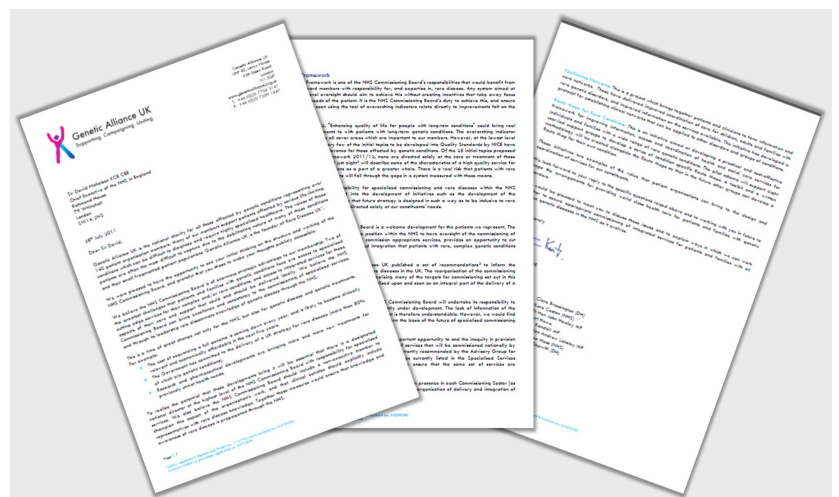
We will continue the dialogue regarding this budget issue, as we believe that the budget should be based on the cost of providing the best service patients are currently receiving in some regions, not a total of what is being spent currently as in some parts of England the expenditure on specialised services is significantly less than others.

You can read the whole of both of these letters and follow our campaign on our website at www.geneticalliance.org.uk/policycampaigns.htm

If you have any comments about this article or about our policy work, please contact Nick Meade.

**Nick Meade,
Policy Analyst,
nick@geneticalliance.org.uk**

To read the letter in full please visit www.geneticalliance.org.uk/policycampaigns.htm



Project Update

Risks and Benefits: Citizens Jury

Come Along and See it Happen!

The *Risks and Benefits* project team have been very busy over the past few months preparing for the Citizens Jury events later this year.

To re-cap...the *Risks and Benefits* project will examine how patients and their families perceive the balance between the risks and the benefits of new biomedical therapies for people with serious and/or rare conditions. Based on a Citizens Jury model, a group of patients and family members will meet, over a number of days, to learn about and deliberate this complex issue. The project began in September 2010, and the main jury events will take place across two week-ends in November and December.

The Aim

The primary aim of the Citizens Jury is to clarify the views and perspectives of patients and their families in relation to this issue, generating informed opinion and identifying what they consider to be the key factors in defining an acceptable balance between risk and benefit for a new therapy. The jury will address the following questions:

- How do patients perceive the balance between the risks and benefits of new biomedical therapies?
- What should the balance be in the risk/benefit calculus? How should these decisions be made?

Jurors will get the opportunity to listen to, and ask questions of, a number of

expert witnesses from pharmaceutical companies, regulatory bodies, patient groups and others. The Jury will then produce a framework of recommend actions to be used as a practical tool in the analysis of risk and benefit. We were pleased that the recruitment campaign received lots of interest, and we had many people applying to be a juror. Between May and July, a total of 72 individuals registered their interest via an online survey, 51 of whom responded in the second stage of the recruitment campaign.

Unfortunately, we could only choose 16 people to take part, but we have tried to select a jury which is as diverse as possible. The venue we have booked for the Citizens Jury events is the Radisson Blu Hotel in Central Birmingham.

The selected jurors met for the first time on Saturday 17th September for the Planning Day. It was a great opportunity for the jurors to meet each other and to meet the project team. Alastair Kent joined us at the beginning of the day to introduce the project and give jurors some background information. We received great feedback from the jurors, and we are looking forward to the 4th November when the jury begins!

How you can get involved

In the next few weeks we will be finalising the programme and preparing the first weekend event. We are developing hypothetical case studies for the jurors to discuss, as well as booking some interesting speakers for the jurors to listen to and 'cross examine'.

We would like to invite you to observe the jurors in action! You can come along

and watch some or all of the Citizen Jury sessions in November (4-6th) or December (2nd -4th). You may want to hear one of the expert witness presentations, or listen to the jury discuss one of the issues. The event is free to attend, however you will not be able to participate in any of the sessions. If you would like to find out more, or reserve a place, then please get in touch with the Project Officer, Amy Simpson Please note that we have limited spaces for observers at each session, so get in touch soon!

If you cannot attend any of the events, don't worry - Keep an eye out for project updates and findings on the Genetic Alliance UK website, newsletters and e-update.

Amy Simpson,
Project Officer, Risks and Benefits,
University of Glamorgan,
asimpson@glam.ac.uk
01443 483078

Editor's note:

Thank you to all of you who registered your interest in being on the citizens jury and to all of you who have shown an interest in the results of this project. We hope that the findings will help to inform future research and help patients to be better understood by those offering new biomedical therapies.



RARE DISEASE | UK

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

Rare Disease UK awaits launch of consultation on UK rare disease plan

Rare Disease UK is the national alliance for people with rare diseases and all who support them. It was established by Genetic Alliance UK and has been campaigning vigorously for the past three years for a plan or strategy* for rare diseases in the UK. This was on the back of the Council of the European Union's Recommendation on an action in the field of rare diseases which was signed by the UK government in June 2009.

*please note we now understand that in the UK there will be a "plan" as opposed to a "strategy" – just to avoid confusion!

Over the summer, RDUK continued to maintain dialogue with the UK's health departments and we are encouraged to hear of the positive progress that is being made. The UK's four health departments have been working together to develop one UK plan for rare diseases. This plan will go out for consultation in each of the four home nations for the mandatory 12 week public consultation period. At the time of writing (September 2011) we cannot say for sure when this consultation will be launched (by the time you are reading this, maybe it will be out already!) Once the consultation has been launched, RDUK will be working hard to inform our members and the wider stakeholder community of the content of the plan, and to gain feedback to inform our responses to the health departments of England, Scotland, Wales and Northern Ireland individually.

As reported in the Spring 2011 issue of the Genetic Alliance UK newsletter, RDUK launched a report *Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy* at four parliamentary receptions at the House of Commons, the Scottish Parliament and the Welsh and Northern Ireland Assemblies to mark Rare Disease Day 2011 (the report is available here: <http://www.raredisease.org.uk/documents/RD-UK-Strategy-Report.pdf>). This report outlines RDUK's recommendations for a plan for rare diseases which were developed over a year and a half with contributions from over 1000 organisations or individuals. We are encouraged with the re-assertions we have received from the health departments that our recommendations have been instrumental in assisting the development of the UK plan.

Whilst this progress is encouraging, we are maintaining the

pressure. Until we have seen the proposed plan, we do not know how many of our recommendations will have been incorporated. No single nation can address all the challenges posed by rare diseases alone, so the collaboration we have seen between the four health departments is encouraging. However, there is also the danger that, as the plan has to be accepted by all four health departments, it might get "watered down".

There is also the possibility, of course, that the development of the plan is merely seen as a tick-box exercise; RDUK has been clear through the entire process that a plan for rare diseases must be implemented in practice. We will be pushing for clear timelines for delivery and outcome measures. Following the consultation period and the subsequent development and launch of the final plan, RDUK is hoping to work with our members, health departments and NHSs to implement it into practice. This will of course necessitate scrutiny of the actions being taken.

We have also been working to secure political support for a plan for rare diseases. This has been particularly necessary in the devolved nations where elections took place in May 2011 meaning that there are new politicians to make links with, new members of health committees who need to be informed about the need for a plan, new Ministers etc. Over the summer and autumn we have been meeting with a number of politicians and we are very encouraged by the support we have been receiving. We will, of course be keeping our members informed of any updates with the rare disease strategy, and hopefully by the next edition of this newsletter we will have some exciting progress to report.

In other news, RDUK bid a fond farewell to Membership and Communications Officer, Lauren Limb. Lauren was instrumental in developing our 'Improving Lives' report as well as a number of other RDUK activities. Whilst she will be missed by our members and staff alike, Lauren will continue to support people affected by rare and genetic diseases as she is currently pursuing a Masters in Genetic Counselling at the University of Manchester. We wish her the best of luck.

Stephen Nutt, Executive Officer – Rare Disease UK
stephen@raredisease.org.uk



Facilitating Networks - getting ready to launch

In the closing months of the Facilitating Networks project, work with each of its constituent patient support groups has gained momentum and revealed strong potential for sustainability far beyond the project's lifespan; culminating with the launch of the project at our AGM on 1st November.

During this time, our efforts have focussed on advancing network plans generated through consultations with stakeholder groups, and approved by patient representatives. This led to the implementation of various work streams, all of which share the common goal of bridging the gaps between the knowledge base and experience of patients with the expertise of health care professionals in order to improve the health services patients and their families access.

The project has been working with four condition areas, grouping some of our smaller member groups together to benefit more than one condition at a time. The condition groups we have been working with are:

- Rare dermatological conditions, including Ectodermal Dysplasia and Hypomelanosis of Ito.
- Rare cancer conditions, Familial Adenomatous Polyposis of the Colon (FAP).
- Rare neurological conditions, including Rett Syndrome, Cavernous Angiomas and the inherited ataxias.
- Conditions on the Ras-mapk pathway; including Costello Syndrome, CFC, Neurofibromatosis and Noonan Syndrome.

Here is a brief summary of recent achievements:

Ataxia study day

In close collaboration with the Ataxia UK research team, the Facilitating Networks project organised a study day for health professionals interested in the care of ataxia patients. The programme incorporated the multi disciplinary perspectives required to achieve a holistic approach to care for people living with the condition. Delegates included an impressive range of professionals including neurologists, geneticists, physiotherapists, speech and language therapists, occupational therapists, and nurses. Many of The health professionals who attended the day emphasised the value of being able to hear the patient perspective on clinical matters and took away much benefit from what they learned. Many also expressed a keen interest in developing an affiliation with the patient support group Ataxia UK by joining the support group's Medical Registry.

Europacoln conference

Over the past three years the project has worked closely with the FAP Support Group to generate a body of research examining FAP patients' experiences of accessing healthcare. In line with plans of disseminating these findings, the Facilitating Networks project officer attended the Europacoln conference. Established in 2005 Europacoln represents the only European organisation dedicated to colorectal cancer. With its aims to unite patients, carers, healthcare professionals and politicians - our presence was pivotal in contributing to the active lobbying for access to best treatments and care for all who are in need.

Hypomelanosis of Ito

Working closely with members of the HITS support group to facilitate a patient-led initiative, the group's patient information resource has been completed! Modelled on Genetic Alliance UK's template for patient 'Route Maps' (<http://geneticalliance.org.uk/projects/familyroutemap.htm>), the resource has been designed to signpost individuals and families affected by Hypomelanosis of Ito to relevant information and services that could improve their quality of life. As well as disseminating these booklets via the patient support group, copies have also been sent to a select number of clinical genetic centres to ensure those who should have access to this information do so.

The International Rasopathies Symposium

Last year's House of Lords reception not only marked the establishment of the UK based Rasopathies Network, but also served as a catalyst for its international counterparts. The International Rasopathies Symposium in Chicago saw one hundred researchers, scientists, parents and affected people gather to learn the latest about Rasopathies. The Facilitating Network's project offer attended workshops focused on formalising international collaborations between interested parties and was able to offer guidance about network development informed by the work that has been achieved to date.

Rasopathies Study Day

Continuing the momentum gained from the Rasopathies International Symposium, the project team decided to facilitate a UK based professional meeting. The event was successful in galvanising the interests of the Rasopathies' UK-based medical community with the enthusiasm of patient support groups. Based on feedback obtained from the day, we are confident that the knowledge shared will go towards energising clinical and basic science research, and ensuring that such developments are translated into the clinic.

With these recent successes in mind, the project team are eager to share the learning obtained from undertaking this work with a view to disseminating it amongst Genetic Alliance UK members in an accessible format. Our project report, complete with protocols for establishing networks of expertise, and guidance relating to cultivating effective relationships between patient groups and health professionals will be made available at the end of the year.

We are delighted to have been instrumental in facilitating the growth of networks of clinicians and professionals with an interest in rare conditions which will make such a difference to patients and families affected by the conditions involved.

Krystle Kontoh, Project Officer Facilitating Networks,
krystle@geneticalliance.org.uk

This project is being launched at our AGM on 1st November 2011. More information about this free event can be found on page 10.



Image captions on this page. From Left to Right:

- Project Officer Krystle in Lisbon raising awareness amongst professionals
- Delegates listening intently at the Ataxia Study Day
- A panel of patient representatives at the Rasopathies network Study Day.

Image captions on page 6 from left to right:

- Krystle Kontoh represented Facilitating Networks at many different European events; pictured here in Barcelona
- The Ataxia Study Day
- The Rasopathies network Study Day

R.A.P.I.D project update

RAPID (reliable accurate non-invasive prenatal diagnosis) is an National Institute for Health Research funded project that has been running since the end of 2009. Currently, couples who know they are at risk of having a child affected by a genetic condition can opt to take a diagnostic test such as an amniocentesis or Chorionic Villus Sampling during pregnancy, to collect a sample of the baby's genetic material.

These invasive tests carry a small but significant risk of miscarriage (up to 1%) and can only be performed after 11 weeks of pregnancy. Recently, we have found out that from 4 weeks of pregnancy the baby's DNA circulates in the mother's blood. Non-Invasive Prenatal Diagnosis (NIPD) will allow us to use a normal blood sample taken from the mother's arm to determine whether the baby has a particular genetic condition. Because it is a simple blood test, there is no risk of miscarriage and testing will be possible earlier in pregnancy.

Currently, we are able to use NIPD to identify the sex of the baby and the test can be done from 7 weeks of pregnancy. This new test is particularly useful for women who are carriers of X-linked conditions (conditions which only affect boys) as it reduced the need to consider invasive testing for about 50% of pregnancies i.e. those which are identified as female. At Genetic Alliance UK, we have been interviewing women who are carriers of x-linked conditions such as Haemophilia and Duchenne muscular dystrophy, who have used this new test during pregnancy, to identify their views on this new technology.

So far, our findings have been very interesting. Womens' views and experiences of NIPD have been overwhelmingly positive with words including "brilliant", "exciting" and "incredibly lucky" being used to summarise their experience. Safety was found to be a key value of NIPD, with most women acknowledging that unlike invasive testing, the procedure posed no risk to the mother or baby. Additionally, there were no feelings of guilt associated with the test as there is no risk of miscarriage with a blood test. The test also had the benefit of being "easy", "quick" and "simple" to conduct.

Knowing the sex of the baby early in pregnancy was also valuable for a number of reasons. For those women with male pregnancies, they could prepare themselves either for going ahead with invasive testing, or for the possibility of having a

child affected by the condition for which they were a carrier. Knowing the sex early also gave women a sense of control over their pregnancy early on which was found to be very important to them, and the information enabled them to make decisions about what their next steps would be. For those women with female pregnancies, they could relax and enjoy their pregnancy early on, and their pregnancy went from being 'medicalised' or 'different to most people's' to feeling more like a 'normal' pregnancy. It was particularly valuable having this information within the first trimester (before 12 weeks) as parents could then tell family and friends about their pregnancy after 12 weeks which is when most other people disclose their pregnancies.



Most women and their partners had already made decisions prior to receiving the NIPD test results as to what they would do if the result was male. There were however a small number of people who had not made a decision about what they would do next, and wanted to find out the sex of the baby before deciding what to do. The NIPD test was useful for this group as it helped them to make a step-by-step approach to making a decision, informed at each stage of the process.

Concerning the possible disadvantages of NIPD, some people were worried that the test might be used for social sexing purposes and there was a strong feeling that it would be unethical to use this test in such a way. In the UK NIPD is only used for clinical reasons and this is tightly regulated by the NHS. Therefore it is highly unlikely that NIPD would ever be used in this country for this reason.

In conclusion, participants' views about using NIPD were overwhelmingly positive with many more advantages than disadvantages being identified. A number of practical and psychological benefits of a test that is early in pregnancy, easy to conduct, safe and highly accurate were identified. This information is useful for both healthcare practitioners as well as policy makers in ensuring NIPD is provided alongside appropriate counselling and support. Until diagnosis using NIPD is available, using the test to identify the sex of the baby is a good interim measure which has a number of notable advantages over traditional methods.

Celine Lewis, Project Officer for R.A.P.I.D
celine@geneticalliance.org.uk

Synageva and Lysosomal Acid Lipase Deficiency



Synageva BioPharma is a biopharmaceutical company dedicated to discovering and developing therapies for patients with rare diseases.

The lead programme (SBC-102) involves patients diagnosed with the rare, autosomal inherited disorder, Lysosomal Acid Lipase Deficiency (LAL). This is currently in clinical trials for patients with either early or late onset disease.

LAL deficiency is an autosomal recessive disorder, caused by a marked decrease or almost absence of the LAL enzyme. Although a single disease, LAL deficiency presents as a clinical continuum, including early onset LAL affecting infants and frequently referred to as Wolman's disease, and late onset LAL. Where patients are affected by LAL post infancy, this is often described as CESD (cholesteryl ester storage disorder). The reduction of this important enzyme leads to a range of effects and symptoms, including enlargement of the liver and spleen, liver dysfunction, liver fibrosis and cirrhosis.

Early onset LAL deficiency presents shortly after birth with mainly gastrointestinal and liver involvement, and is the more rapid form of the disease. Infants diagnosed with LAL deficiency often have challenges with growth failure, poor absorption of nutrition, and liver enlargement.

Late onset LAL deficiency presents across all spectrum of ages, from patients as young as two years and ranging to patients who aren't diagnosed until their late 60s. This is

the more common presentation of the disease, with marked enlargement of the liver and spleen.

A hallmark of the disease is that large amounts of lipids accumulate in cells and tissues throughout the body, with predominant liver involvement and dyslipidemia. The liver is typically the most severely affected organ with marked hepatomegaly, elevation of transaminases and liver fibrosis, often progressing to cirrhosis. Additionally, dyslipidemia is common with type II hyperlipidemia and decreased HDL levels, and these abnormalities are associated with accelerated atherosclerosis.

Synageva is conducting a number of important research studies for patients with LAL deficiency. There is no treatment approved for LAL Deficiency. At present, medical care focuses on managing the symptoms. Intravenous nutritional support is sometimes used for early-onset LAL Deficiency if bone marrow transplant is being considered. For late-onset LAL Deficiency, combining drugs that reduce blood cholesterol with a low cholesterol diet has been effective at reducing some of the symptoms (<http://www.climb.org.uk/pdf/synageva.pdf>).

Why is this research study being done?

SBC-102 and LAL deficiency

SBC-102 is a new investigational drug being developed for the treatment of LAL Deficiency.

SBC-102 is not approved by regulatory authorities like the FDA (U.S. Food and

Drug Administration) or the MHRA (Medicines and Healthcare Products Regulatory Agency). This means that SBC-102 can only be used in research studies at this time.

What is the purpose of the study?

- Synageva are conducting research studies to find out if SBC-102 can help children with Wolman's disease.
- This is the first time SBC-102 is being studied in children and it may also be the first time SBC-102 is studied in humans.
- Another study using SBC-102 in adults with LAL deficiency is also ongoing.

Which patient support groups are involved in the study?

Advocacy and support for families diagnosed with LAL Deficiency is provided by CLIMB

Climb, Climb Building, 176 Nantwich Road, Crewe, CW2 6BG, Tel: (freephone): 0800 652 3181, Tel: 0845 241 2172
<http://www.climb.org.uk>

More information

For more information about the study please contact Julie Kelly,

julie.kelly@synageva.com
www.synageva.com

Our AGM and the launch of Facilitating Networks

On 1st November Genetic Alliance UK will be holding our 22nd AGM. This year's AGM will be particularly special as we will be launching our project, Facilitating Networks. Over the past three years, Krystle Kontoh has been working hard to fulfil the main aim of the project which was to work with small patient support groups to facilitate the development of networks of health and social care professionals in order to improve information, care and services for patients and families affected by, or at risk of, rare genetic disorders. These networks are designed to help those affected by rare dermatological conditions, rare cancer conditions, rare neurological conditions and conditions on the Ras-mapk pathway.

All members are welcome to attend the AGM. This free event is going to be held at the Diana, Princess of Wales, Memorial Fund (within 10 minutes walk of Waterloo) in central London.

On the day, we will be delighted to welcome Kate Caston, Department of Health lead for specialised services transition and Mark Kroese who is the Public Health Advisor for the UK Genetic Testing Network. They will be talking around the NHS reorganisation and how it is going to affect the services and information provided. Kate Caston will be talking about the future for specialised services in the NHS and Mark Kroese will be talking about the changes to the Public Health system in the NHS.

Full details of the day can be found at www.geneticalliance.org.uk/latest-events.htm. If you would like to attend, please fill out the application which can be found on the webpage above with the full details of the day and return it to Julian. If you would like to contact me for more information about the day either contact Julian on the email address below, or on 020 7704 3141.

Julian Walker, Marketing and Communications Officer, julian@geneticalliance.org.uk

National Update- Scotland

Supporting Patient Groups Project

The Supporting Patient Groups Project in Scotland continues to grow and we are delighted to have received a commitment from Genzyme Therapeutics to continue funding this project, through an unrestricted grant, until 2013. This funding, in addition to that already received by Alexion pharmaceuticals, will ensure that many more patients with rare and genetic conditions can receive support to start their own patient group.

PNH Scotland, the first group assisted by the project, has seen a growth in membership over the last few months and will be celebrating its official launch on the 19th of November. The group has already been active in campaigning for equal access to treatment for PNH in Scotland and has started exciting fund-raising activities, including producing a recipe book.

The project is also supporting the Scottish Paediatric & Adult Haemoglobinopathy (SPA) Managed Clinical Network to establish a patient support group for patients with sickle cell anaemia and thalassaemia. Natalie, the Development Officer for Scotland, will be speaking at the SPAH family day on October 8th about the project and how to get the most out of a patient support group.

RDUK Petition to the Scottish Parliament

RDUK have submitted a Public Petition calling on the Scottish Parliament to urge the Scottish Government to review the mechanism and methodology used by the Scottish Medicines Consortium to appraise the value of medicines for orphan diseases. The petition also calls for the Scottish Parliament to instruct the Chief Medical Officer to revise the criteria for accessing Individual Patient Treatment Requests (IPTR) by removing the term 'exceptional' from all health boards IPTR requests in relation to orphan diseases. The petition was launched to address the current inequalities patients with rare diseases in Scotland are facing when trying to access medicines for rare conditions. The Petition will come before the Scottish Parliament's Public Petition Committee on the 4th of October. The full petition can be viewed online at: www.scottish.parliament.uk/business/petitions/docs/PE1398.htm.

If you would like to find out more about the petition or to find out how you can lend your support - please contact Natalie.

Natalie Frankish, Development Officer for Scotland, natalie@geneticalliance.org.uk

National Update - Wales

It has been a busy and enjoyable few months since I took up the position of Development Officer for Wales in June and it seems as though it was just last week that I was writing to introduce myself in the summer newsletter! I have been involved in a number of areas of Genetic Alliance UK's work in Wales which has included canvassing the views of members of the South Wales Familial Hypercholesterolaemia (FH) Family Forum and of specialist FH healthcare professionals on two consumer information leaflets, *Genetic Test Results and Insurance* and *Familial Hypercholesterolaemia and Insurance*, being produced by the Association of British Insurers (ABI). The feedback was well-received by the ABI and incorporated into their final leaflets.

In promoting the work of Rare Disease UK in Wales, and in particular the recommendations published in our recent *Improving Lives, Optimising Resources* report, we met with the Medical Director of NHS Wales and the Welsh Government's Head of Health and Social Care Strategy, and also the Medical Director of the Welsh Health Specialised Services Committee to discuss the forthcoming publication of the draft UK plan for rare diseases which is expected in autumn 2011. We have also been busy contacting many of the Welsh Assembly Members, including the new Minister for Health and Social Services and members of the Health and Social Care Committee, to discuss progress in Wales towards developing a strategy for rare diseases.

In other work, Genetic Alliance UK and Rare Disease UK provided written responses to the public consultation on the revised All Wales decision making policy on Individual Patient Funding Requests published by NHS Wales in July. I was pleased to attend the Action Duchenne lobbying event held at the Welsh Assembly in Cardiff Bay, where stakeholders including families and healthcare professionals gathered to raise awareness and lobby Welsh politicians on the issues facing those affected by Duchenne muscular dystrophy (DMD) in Wales including funding for research into a cure for DMD, integrated and accessible healthcare for those affected, and commissioning for a specialised centre of excellence. I also attended the Alkaptonuria (AKU) symposium held in Cardiff, which included some excellent presentations on the latest scientific research, clinical studies and work of the AKU society.

I have certainly enjoyed my first few months in post and am looking forward to a busy autumn! To find out more about our work in Wales, you can visit the Welsh page of the Genetic Alliance UK website (<http://www.geneticalliance.org.uk/wales/index.html>) or email Rhian.

Rhian Morgan, Development Officer for Wales, rhian@geneticalliance.org.uk

Masters Pharmaceuticals

Improving lives through patient access



The challenges faced by families living with genetic disabilities should never be underestimated and patient advocacy groups are often the best source of guidance and support for patients, their families and physicians, seeking to find the best treatment and care options available. Masters, a

global healthcare company focused on providing patient access to quality medical solutions, has over 25 years experience of developing and delivering Early Access Programmes in more than 40 countries worldwide, to patients with special treatment needs that cannot be fulfilled through traditional prescription; usually in one of the following situations:

- A special requirement for a medicine awaiting a license or still undergoing clinical trials
- A licensed medicine is available, but the dose or form is not suitable for the patient.
- A previously prescribed product has been discontinued or there are supply chain problems with a licensed product.
- The product is identified for off-label use to treat a rare condition, often termed an orphan disease.

Early access is often attractive to pharmaceutical companies and patients alike. For patients, with serious life threatening or debilitating illness, access to pre-licensed medicines where there is no satisfactory alternative can offer benefits. This access, in some cases, can help to alleviate symptoms or reduce difficulties which patients face in day to day life. Early access is also attractive to pharmaceutical companies, facilitating the collection of data outside of the traditional clinical trial setting and expanding the body of knowledge about the drug are advantageous. Most countries legally allow early access programmes, and in order to ensure that patient protection is not compromised they are closely regulated.

Masters' experience of managing patient and physician needs in this area has resulted in close links with several patient advocacy groups, one example of which is the collaboration with Parent Project Muscular Dystrophy. PPMD, established in 2000, is the advocacy group for Duchenne muscular dystrophy in the US. In recent years doctors in the US specialising in child neurology and neuromuscular disorders have selected a currently unlicensed drug in the US for the treatment of Duchenne muscular dystrophy. This is supplied by Masters on a named patient basis. Having worked closely with PPMD for 10 years, and, as a consequence of providing this important medication, Masters have established strong relationships with both physicians and patients' families. For more information about the services Masters provide please visit www.mastersglobal.com

Editor's note

The Haemochromatosis Society held an event for clinicians interested in the group and the condition at the Royal Free Hospital in London. As part of the build up to this event, they have asked to put an article in our newsletter to raise awareness of the condition and to generate extra interest surrounding the event.

If your group would like to write something for our newsletter, please contact Julian on julian@geneticalliance.org.uk.

A note from one of our members: Haemo-chroma- what?

A few weeks ago the Metro newspaper ran a story mentioning Haemochromatosis as a newly identified genetic link to hypertension, which is a risk factor for heart disease and strokes. Whilst we were surprised to see Haemochromatosis mentioned, we were not by the resulting enquiries we received: zero. Genetics isn't an easy subject for a morning newspaper, and because genetic Haemochromatosis (GH) is not well known it would be easy to skip over the complicated word, which means iron overload.

Unfortunately heart problems are one of many possible symptoms of GH, such as chronic fatigue, joint pain (especially in the fingers), poor liver function, permanent skin tan, type II diabetes, and diminished sex drive. The only symptom strongly suggestive of GH is the arthritis in the hands, causing the 'iron fist', so GH is often overlooked as a cause of these symptoms. Members of the Haemochromatosis Society on average have seen 5 doctors over 5 years from the onset of symptoms to diagnosis.

GH is one of the most common genetic recessive disorders. 1 in 200 people of European descent carry two copies of the gene, and 1 in 8 is a carrier. It causes the body to absorb more iron from the diet than required, the iron deposits build up slowly in different areas of the body and early symptoms may not appear until the age of 30-40, or even later. Diagnosis is by simple blood tests and confirmed by the genetic test. Once diagnosed, treatment can begin. This involves the gradual removal of excess iron which prevents the development of further complications and improves many symptoms, with a dramatic positive impact on the quality of life.

It is vital to ensure that family members are tested. Encouraging those diagnosed to take up the offer of genetic counselling, and to get from the counsellor or their consultant a letter to pass on to first degree relatives. Only 1 in 5 members had seen a genetic counsellor, and we have heard from many people that even with diagnosed 1st degree relatives doctors are still unwilling to order the genetic test for them.

Raising awareness of the condition amongst professionals and the public is one of The Haemochromatosis Society's key aims. A recent study found that 1/3 of junior doctors were unable to diagnose GH correctly, and sadly this was from a pool of

doctors who have been lectured on the subject in their training. Of those that are aware of it, there are still misconceptions that it does not affect people until later life, and that women will not store excess iron until they are post menopausal.

A member wrote to us recently of his experiences; before he was diagnosed he was on the heart transplant list because of severe cardiomyopathy, he had had right ankle fusion, was bronzed, diabetic and forgetful. Unfortunately by the time he was diagnosed the stored iron in his system meant he needed the other ankle and five knuckle joints replaced, all of this compounded by the fact that he is a GP. Once the high overload of iron was removed, his heart function improved and he was able to resume his GP practice. Once he understood what had caused his symptoms, he recognised that his father had had them too.

Thank you for reading. If you have any questions about Haemochromatosis please contact Sarah Fernau, sarah@haemochromatosis.org.uk

Metro link: <http://goo.gl/FcaGk>
The Haemochromatosis Society site: www.haemochromatosis.org.uk
CPD conference information www.haemochromatosis.org.uk/cpd

Linking to our website

Every member of Genetic Alliance UK has a link placed on our website. Members can view this link at www.geneticalliance.org.uk/members.htm and can always request that the information accompanying the link is updated by contacting us. As part of our ongoing development of the website I would like to ask you to add a link to our website on your website. This would help to ensure that patients looking for information surrounding genetic conditions are able to find our website quickly.

If you would be willing to add our link to your website please visit <http://www.geneticalliance.org.uk/linktous.htm> which contains everything you need. If you would like more information or any help please contact me on julian@geneticalliance.org.uk