

Genetic Alliance UK

Newsletter



Winter 2010/2011

Looking to the year ahead

By the time this reaches you Christmas will be a distant memory, New Year Resolutions will long since have been consigned to the dustbin of good intentions and we will be firmly engaged with responding to the re-organisation of the NHS in England and preparing for Rare Disease Day 2011 on 28th February.

The Government seems determined to stick to the ambitious timetable it has set for itself for re-shaping the NHS and devolving most of the budget to GP Consortia. Some services will be commissioned directly by the new National Commissioning Board for England and we will be working hard to persuade Ministers of the importance of ensuring that the complex needs of those affected by genetic disorders are recognised and responded to appropriately. This should involve a mix of services commissioned through national centres of excellence and those provided at local level through GP consortia. It will be important to ensure that nationally and locally commissioned services and support are integrated if patients and families are to receive the care they need.

It is the need for a strategic approach to service delivery that prompted Genetic Alliance UK to establish Rare Disease UK as a multi stakeholder consortium committed to the development of such a strategy. Expert groups have been working hard to assemble the components into a coherent plan that will be launched at Parliamentary receptions in all four UK Parliaments on and around Rare Disease Day at the end of February. We hope to see many of you joining us at these to press our elected representatives into action, generating the necessary momentum to translate intentions into services and support!

We look forward to continuing our collaboration with officials in the Health Departments and the NHSs across the UK to implement the strategy in line with the commitment given at the Council of the European Union in June 2009.

Important as the merging national strategy for rare diseases is that we continue to find the time and energy to address other policy initiatives. Access to pre-implantation genetic diagnosis is very patchy, with some PCTs apparently confusing it with infertility services. We will be addressing this in the coming months (see the article by Nick Meade on p.3). The European Directive regulating the use of animals in medical research will also be transposed into UK law in 2011 and we are cooperating with officials in the Home Office, Ministers and politicians to ensure that this is done appropriately, so that ethical research involving animals can proceed as smoothly as possible under a regulatory framework that is appropriate and proportionate.

Looking ahead, 2011 promises to be challenging as downward pressures on resources exert their affect and cuts begin to bite in the public sector particularly. The team at Genetic Alliance UK will continue to work hard on behalf of all our members and the individuals and families they support to ensure that their voice is heard, and that those downward pressures are resisted with the utmost vigour we can bring to bear.



Alastair Kent, Director

Alastair Kent, Director
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In this Issue

Director honoured with OBE
Page 2

Our campaign for equal access to PGD
Page 3

Big Lottery grant received
Page 6

Rare Disease UK's Report on patient experience
Page 8

Director of Genetic Alliance UK is honoured in the New Years Honours List

It is with great pleasure and delight that we can report that Alastair Kent, Director of Genetic Alliance UK has been awarded an OBE in this year's New Years Honours List for his tireless work in helping families affected by genetic conditions.

Chris Friend, Chair of Genetic Alliance UK said, "I can't think of anyone more deserving of this honour. Alastair has contributed over the past 20 years to GIG (as we were) and now to Genetic Alliance UK as well as to healthcare policy in general. His contributions have been genuinely outstanding and I, for one, am absolutely delighted. What a fantastic way for us to begin 2011!"

We have also received some lovely emails from our Trustee Board which we would like to share with you all.

"Very well deserved and not before time!"

"Congratulations Alastair, this is very well deserved!"

"Heartiest congratulations. This is so well served and starts 2011 in a great way."

"I am delighted that your efforts have been recognised, you have worked long and hard to help those with genetic disorders and you deserve the congratulations of us all. It's a privilege to work with you."

Alastair commented, "I see this as recognition for the work that we have all achieved as Genetic Alliance UK, not as a personal award, and I would like to thank and congratulate you all for all you have contributed to making this occur".

Disabled children at the heart of Newlife Foundation



Newlife Foundation for Disabled Children is a national charity, founded in 1991, that specialises in making life better and brighter for disabled children and their families. Newlife achieves this through its services and funding of 4 key areas of action.

Nurse Services

The National Helpline is staffed by a qualified team of nurses who listen, advise and offer help and support to all who call or contact them. The nurses use their clinical skills and understanding to provide an invaluable service to families who have a disabled child. Whether you choose to contact them through a call or via the instant-messenger Live Nurse Chat service, Newlife nurses are a confidential and trusted source of support and information to thousands of families every year. With 123 years of combined experience between them, Newlife nurses offer immeasurable support and have recently expanded to respond to the growing number of calls to the service. All calls are free on 0800 902 0095.

Equipment Grant Services

Newlife nurses are the access point for families applying for a grant for equipment relevant to their child's disability. This could be anything from wheelchairs to beds, hoists to seating systems and much more. One free phone call can result in completion of a simple application form for one of Newlife's Equipment Grants or signposting to another funder. Newlife Equipment Grants are non-means tested and often a decision on equipment can be made in days. A new equipment loan scheme 'Just Can't Wait' starts in Jan 2011 and it is targeted to specifically help children with terminal/life-limiting conditions, where the need for equipment at home is urgent. Newlife has spent £5.6 million on Equipment Grants in the last five years.

Medical Research

Newlife believes that medical research holds the key to finding the causes of disabled and life-threatening conditions. By funding this research, Newlife aims to improve understanding, diagnosis, treatment, prevention and improve children's health in the UK. Newlife has spent £11 million on research in the last 19 years.

Campaigns and Awareness

Many parents feel alone in the daily struggles and battles they face when looking after their disabled child so Newlife Foundation's campaign and awareness activities help to give them a voice on important issues.

All this help starts with a phone call. Newlife Nurse Helpline (0800 902 0095) is available Monday - Friday 9.30am-5pm and Wednesdays until 7pm. Live Nurse Chat is available during office hours at www.newlifecharity.co.uk.

Newlife ask for no subscriptions or memberships and all services are free. Newlife's Lead Nurse Karen Dobson says: "Our Nurse's have great real-life experience and so this service does not operate like a Call Centre. We are real nurses helping real families, facing real difficulties, providing information so do give us a call and we will be pleased to help."

Can you help our campaign for equal access to PGD?

We are planning a spring campaign to highlight inequity in access to pre-implantation genetic diagnosis in the NHS. Could you or any of your members help? Nick Meade explains more.

Pre-implantation genetic diagnosis (PGD) is a method of artificial conception that allows a couple to ensure they do not pass a genetic condition on to their child. It is essentially in vitro fertilisation (IVF) where embryos are tested for a genetic condition before they are implanted in their mother.

Though we live in a time of exciting developments in the field of biomedical technology, the fact remains that the vast majority of life-limiting genetic conditions have no cure or treatment and any treatments that do exist do not reverse the full effects of the condition they treat. The opportunity therefore, for parents to avoid the birth of an affected child, without practicing ante-natal diagnosis and possible termination of pregnancy, is clearly valuable and in most cases by far the most powerful tool we have to fight genetic disease.

The same technique as PGD can be used by parents to ensure their child is a tissue match for another child with a condition that would benefit from a bone marrow transplant. Often dubbed a "saviour sibling", this is called pre-implantation tissue-typing (PTT).

At the moment very few genetic conditions can be cured (rather than treated), and probably (it is difficult to be categorical in this fast moving world), the only current cure is bone marrow transplant. For suitable conditions, a group of anaemias and blood disorders, PTT represents the best means by which a suitable match can be found for bone marrow transplant.

Taken together, PGD and PTT are extremely powerful tools that have the potential to rid families of the effects of genetic conditions.

Access to PGD is variable between and within the devolved nations of the UK. We have had experience of PCTs deciding their funding criteria for PGD based upon their criteria for IVF, which is to ignore the many differences in situation between couples wishing to use IVF because of infertility and couples wishing to use PGD to avoid the birth of a child affected by a genetic condition. These are entirely different situations, for which the

intervention is similar, but not identical. Similar issues are most likely occurring with PTT, we wish to investigate this further.

The aim of our campaign is to present an account of the benefits of PGD and PTT to the Departments of Health in England, Scotland, Northern Ireland and Wales. To achieve this, we will gather information from a number of sources. We hope to have statistics of use, success and access across the UK and will make the many arguments in favour of PGD and PTT.

How you can help

We believe that the voice of these families who have benefitted from these treatments or are struggling to access them will be extremely valuable for this account. Real life stories of the effects that these funding decisions have on families will be more powerful than statistics and arguments.

So please, if you or any of your members have any experience of PGD or PTT or have had problems getting PGD or PTT funded on the NHS, then please get in touch with me either by phone on 020 7704 3141 or by email at nick@geneticalliance.org.uk.

Please be assured that any conversations we have will be held in the strictest confidence and will only be used in our campaign if patients and families are happy to do so. Please do contact us by the end of March 2011.

More information on PGD and PTT can be found on our website and on the HFEA's at: www.hfea.gov.uk.

Nick Meade, Policy Analyst
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Familial Hypercholesterolaemia (FH) Cascade Testing Service

A new genetic service that can prevent early onset heart disease and could save thousands of lives is being rolled out across Wales, following a sustained campaign by families working in conjunction with Genetic Alliance UK and clinical colleagues.

The new service, is leading the way as the first of its kind in the UK, will help identify the genetic condition familial hypercholesterolaemia (FH) that affects about 6,000 people across Wales and has already won the backing of affected individuals and their families.



It is hoped this service can be rolled out across the UK and will also help introduce clinical genetic services for other inherited cardiac conditions. The service was officially launched in December 2010 at the National Assembly for Wales by the health minister Mrs Edwina Hart. The

service, is being funded by a partnership between the Welsh Assembly Government and the British Heart Foundation.

Dr Mike Knapp from the BHF and a trustee of Genetic Alliance UK told the gathered audience at the launch event: It is good news to see a new service launched into the NHS that will diagnose people with this inherited condition. The campaign saw groups of people such as families, charities, clinicians, nurses and scientist coming together to influence the introduction of this new service into the NHS. We hope many more people across the UK will benefit in the future.

Health policy changes continue at a rapid pace

With the new government moving forward with plans to restructure the NHS, Nick Meade covers the current consultation documents that we are responding to now and in the near future.

Now entering their ninth month in power, our coalition government continues to deliver new health policy for England at great pace. We have been responding to these documents, explaining the impacts - sometimes good, sometimes negative - that the government's plans will have on those affected by genetic conditions. We have been consulting with you, our members, and we are grateful for the responses we have had so far. Your comments and approval increase the power of our consultation responses.

The Department of Health (DH) has been finding time in between the writing of proposals and consultations to respond to our responses to their consultations. Even after this back and forth, a lot of detail remains to be made clear, partly because of the high degree of autonomy that is being afforded to the new NHS Commissioning Board (NHSCB), which is yet to be formed and will take ultimate responsibility for the performance of the NHS. The NHSCB will be created in shadow form over this year and the beginning of 2012, and become active in April 2012.

We have been pleased to see our words quoted by the DH in their response in a number of places and are satisfied that our comments and suggestions are being taken on board by the Department. The area in which had most concern in the first round of consultations was the proposals for an outcomes framework for the monitoring of performance based on a few key indicators. We felt, and still feel, having read the NHS Outcomes Framework 2011/12, that it is likely that relatively uncommon genetic conditions are likely to lose visibility with the proposed system. There have been a few positive movements in this area, there is now explicit recognition of those with rare diseases as a vulnerable group for example but we need to monitor the success of the NHS Outcomes Framework closely.

Our position on commissioning, that the revision of commissioning arrangements in England is an opportunity to

improve access to services that are currently commissioned regionally (as part of the Specialised Services National Definition Set), is shared with the Department and a number of other respondents.

The Future

As this issue of our newsletter goes to press, we are finalising our responses to the "Information Revolution" and the "Greater Choice and Control" consultations. In both of these responses, we highlight the important contributions that patient groups can make to providing patients and their families with information and advice and urge the Department to embrace this resource.

Coming up on the horizon are consultations on value-based pricing of medicine, to which we will propose a more central role for patients; and on public health, where we will examine the role that screening can have to raise awareness of, improve diagnosis of, and help to prevent genetic disease.

As ever, your voice is valuable to us, so please do get in contact with your views about any component of the new health policies in England.

Of course, this is a year of elections in Northern Ireland, Wales and Scotland, so we will be keeping an eye out for future changes in health policy in the rest of the UK after the votes have been cast.

You can see the open consultations here: www.dh.gov.uk/en/Consultations/index.htm and you can read our responses here: www.geneticalliance.org.uk/policycampaigns.htm

Nick Meade, Policy Analyst
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Genetic Alliance UK has its first ever place in the 2011 Virgin London Marathon

We were all even more delighted that Ali Hillier, who is married to our Assistant Director, Melissa, agreed to run on behalf of Genetic Alliance UK on 17th April, as we were all rather worried that we might have to begin training!

This is the first time we have had a London Marathon place and the first time that Ali has run the marathon, so we are all very excited! We do hope you will be able to support him, and as we know many of you also have runners in the London Marathon should you be attending do look out for Genetic Alliance UK's only runner! The team here will also be out in support on the day cheering him on.

If you would like to sponsor Ali, please go to www.geneticalliance.org.uk/marathon_event.htm

Good luck Ali!



Rare Disease Day

Rare Disease Day 2011 "Rare but Equal"

Stephen Nutt provides more detail on the forthcoming Rare Disease Day events, including the four parliamentary receptions in the each of the home nations.

The fourth international Rare Disease Day will be taking place on 28th February 2011. The theme for this year's day is "Rare but Equal". The aim is to highlight that often patients face an unequal level of care, treatment and support because they have the misfortune of their condition being rare.

The purpose of Rare Disease Day is to raise awareness of rare diseases and to emphasise their importance as a health priority. In the past, policy makers have tended to overlook rare diseases. This is partly due to the mistaken belief that rare diseases affect a small number of people, that there is little that can be done to help patients and families with rare diseases or that what can be done would be unfeasibly expensive. Rare Disease Day provides the opportunity to highlight that there are over 6000 rare diseases that will affect approximately 3.5 million people across the UK (or 1 in 17 people). Collectively rare diseases are not rare! As a result they need to be viewed as a priority.

Rare Disease Day provides the opportunity to bring all the stakeholders involved in rare diseases together. This includes patients, families, carers, policy makers, healthcare providers, clinicians, researchers, health-workers, the pharmaceutical industry and patient organisations. By acting simultaneously and collaboratively, nationally and internationally, the voice of rare disease patients is stronger.

Rare Disease UK (RDUK) will be holding four parliamentary receptions in each of the home nations to mark the occasion. At these receptions, RDUK will be launching its report outlining recommendations for a strategy for rare diseases. This report will be the culmination of nearly a year and a half's work investigating what a strategy for rare diseases needs to include. It is hoped that these recommendations will be taken on board by the Department of Health and the Devolved Administrations in developing a strategy for rare diseases in accordance with the Council of the European Union's Recommendation on an action in the field of rare diseases (June 2009).

These receptions will be taking place on the following dates:
Scottish Parliament – 22nd February
Westminster – 28th February
Northern Ireland Assembly – 3rd March
Welsh Assembly – 16th March

RDUK is also encouraging members to get involved in contact campaigns to their local politician(s) to help raise awareness of rare diseases at a political level.

More information about Rare Disease Day can be found on the website:
<http://www.rarediseaseday.org/>

For more information about RDUK's activities around Rare Disease Day and how you can get involved, please see the website <http://www.raredisease.org.uk> or contact Stephen Nutt.

Stephen Nutt, Public Affairs Officer and Secretariat to Rare Disease UK, stephen@raredisease.org.uk

Syndromes Without a Name (SWAN)

– The Big Lottery Fund will support a groundbreaking five year project to help families with undiagnosed conditions in England.



We are absolutely thrilled to announce that Genetic Alliance UK has been successful in securing Big Lottery Funding, through the Reaching Communities Programme for the “Supporting Families with Children with Undiagnosed Conditions Project”.

This project has been developed to fill a need that was identified when the charity Syndromes Without a Name (SWAN) closed in 2008. SWAN was a member of Genetic Alliance UK and as a successful charity was set up and run by the grandmother of a child with an undiagnosed condition. Over the years it built up a considerable database of families and shared information through its newsletter, the Signet. It provided a place for those families experiencing the difficulties of searching for a diagnosis and for those coming to terms with the fact that a diagnosis may not be found, to share experiences and ideas. Sadly, the charity had to close when its founder and main organiser was advised to take a step back due to ill health. Genetic Alliance UK, has been working hard over the last two years to secure funding to enable us to re-establish the work that this organisation undertook, we have carried out a consultation with families, charities and health professionals to establish if there was a need for such an organisation and if so, what sort of activities people would like it to undertake. The findings showed that 74.4% of parents stated that they would like there to be a specific supportive service for families with children with undiagnosed conditions and 100% of health professionals and charity staff stated that there is a need for a service specifically for these families.

About 50% of children with special needs have no definite diagnosis for their condition. Many of these children have severe physical and/or learning disabilities and without a name for the condition, their families often experience extreme difficulty accessing the information, care and services that their children need. Without support from professionals with expertise in their condition or contact with other families affected by the same condition, families may also face immense isolation. The overall aim of this project is to provide support for families with children with undiagnosed genetic conditions. The project will develop a hub for information and support, with a focus on mutual support, offering families opportunities to share knowledge, experiences, difficulties and solutions.

The project will involve: -

1. Developing a community of families with children with undiagnosed genetic conditions by:
 - Promoting the project widely to attract families that previously belonged to the (no longer operating) support group Syndromes Without a Name (SWAN), as well as to new families.
 - Offering opportunities for families to share knowledge, experiences, difficulties and solutions through a newsletter, an annual meeting and by linking families for mutual support.
 - Encouraging families to become actively involved in the project and in determining its priorities and directions.

EuroGenTest2 – pilot study complete and preparatory work well underway

Genetic Alliance UK has secured a role in the follow on project from EuroGenTest. Here Fransika Severin who worked with us for a few months before Christmas explains a bit about her pilot study which will feed into the EuroGenTest 2 project.

We have completed our pilot study looking at the patient experience of prioritisation of genetic testing as preparatory work for EuroGenTest2. Franziska Severin, who was with us for three months, conducted the study as part of her internship. She interviewed 17 patient group representatives from 8 different European countries including Italy, Finland, Sweden, Netherlands, Germany, UK, Czech Republic and Poland.

2. Gathering and sharing information through:
 - Telephone and e-mail support.
 - A quarterly newsletter.
 - A website containing relevant information and signposting to useful organisations.
 - A moderated online chat forum.
 - An annual meeting.
3. Developing links with health and social care professionals and creating a network of professionals with expertise in undiagnosed genetic conditions.
4. Increasing awareness and understanding of undiagnosed genetic conditions through:
 - Articles in newspapers and journals.
 - Presentations at conferences and meetings.

So why is this project needed, and why should Genetic Alliance UK take on this role? There are many answers to these questions and I have summarised our key thoughts below. We are very excited about starting this new project and will be providing you all with regular updates.

- Without a named condition, families often have extreme difficulty accessing information about their child's conditions and the support and services available to them. In our consultation, 89% of parents felt that having no diagnosis for their child's condition made accessing services and support more difficult. One parent said "it's always been a fight and it's still a fight. Because you don't have that key that opens the door. The autism key or the down's syndrome key."
- In our consultation, 65% of parents reported feeling isolated because of their child's lack of diagnosis. One parent said, "Well it's very isolating I think. Because you just don't know what the future holds."
- There is currently little understanding about undiagnosed conditions amongst health and social care professionals, policy makers, academics, educationalists and the general public. In our consultation, 54% of parents reported feeling misunderstood and more than 96% stated that they would like there to be more awareness and understanding amongst health, social care and education professionals and amongst the general public.

- In our consultation, the main reason that parents stated they would benefit from a specific supportive service were by "being able to share with and meet others in a similar situation" (SWAN Research, 2010). In the words of one parent "being with people who are in the same boat as you and not just going, "Oh I don't know how you cope with this."
- Many families with undiagnosed conditions will eventually receive a diagnosis and so their time interacting with the undiagnosed community may be limited. However, having this network of support and information is invaluable for all the reasons we have listed and many more too.
- A diagnosis will in many cases link the condition to genetics, in fact many families visit Genetics Clinics more than once on their search for a diagnosis. As technologies and research progress so it becomes possible to identify more conditions.
- Genetic Alliance UK has expertise in working with patients and families with all genetic conditions and we often hear through our work of the experiences people have in searching for an initial diagnosis. We feel that a service specifically for children with undiagnosed conditions is vital to gather together this community of people to enable them to access the support and information they require, at a time when it is most needed. Currently there is no such place and we hope that this project will bridge this gap.

To read our SWAN Research Report please ask for a copy from the office or you can download it from our website at http://www.geneticalliance.org.uk/docs/swan_web.pdf

For more information on the Supporting Families with Children with Undiagnosed Conditions Project please contact Melissa Hillier, Assistant Director on melissa@geneticalliance.org.uk

Key findings from the pilot study

- Few patient group representatives had heard of patients being refused genetic tests in their country, although they felt those that were refused were unlikely to join a patient group without a diagnosis.
- Prioritisation criteria acceptable to patient group representatives included the utility of the test results, whether alternative methods for getting a diagnosis existed and the severity of the condition.

- The general stance appeared to be that economic considerations should not hinder access to genetic testing.
- It proved more difficult than expected to engage patients in the subject matter of prioritisation. However, this is a finding in itself that has proved very useful in developing a future work plan for EuroGenTest.2

For further information on EuroGenTest 2, please contact Celine Lewis, Project Officer celine@geneticalliance.org.uk

Rare Disease UK report highlights shocking experiences of patients and families affected by rare diseases.

Rare Disease UK's survey received almost 600 responses from patients and families, the Rare Disease UK team have collated the results to form the basis of the report "Experiences of Rare Diseases: An Insight from Patients and Families". Stephen Nutt and Lauren Limb explain more.

A new RDUK report "Experiences of Rare Diseases: An Insight from Patients and Families" highlights how patients and families affected by rare diseases in the UK face difficulties in accessing the care and support they need. Despite the vast range of rare diseases, the findings demonstrate how patients and families frequently face common problems, regardless of the disease.

The report is based on a survey of nearly 600 patients and family members affected by 119 different rare diseases. The wide-ranging survey dealt with topics including access to information and support, coordination of care, access to treatment, diagnosis and participation in research. The aim of the survey was to gain a better understanding of the issues faced by patients and families affected by rare diseases in accessing the services and support but also to highlight areas of good practice where they exist.

The report, launched on the 6th December, generated media coverage across a range of platforms including national and regional newspapers, the STV news in Scotland and medical and pharmaceutical industry press. RDUK also distributed the report widely to key stakeholders and politicians with the aim of raising awareness of the issues experienced by patients and families affected by rare diseases.

The findings

The survey highlighted diagnosis as a significant area of concern. Without diagnosis patients and families cannot access effective treatment, therapy or manage their condition appropriately. Despite this, almost half (46%) of patients with rare diseases had to wait over one year for a final diagnosis following the onset of symptoms. Of this 1 in 5 (20%) had waited over 5 years and more than 1 in 10 (12%) had waited over 10 years. The results also reveal a striking inequality of experience with 26% of respondents receiving a diagnosis within 3 months on

one hand, whereas 36% waited over 2 years on the other.

Misdiagnosis is a significant problem, 46% of patients were initially incorrectly diagnosed and 30% had received three or more misdiagnoses. Delays in diagnosis and misdiagnosis may involve multiple avoidable hospital appointments and patients receiving inappropriate treatments and tests. Not only is this an inefficient use of NHS resources, it can lead to a deterioration of the condition as appropriate treatment and management of the condition is delayed. Many survey respondents reported the period prior to diagnosis as particularly distressing for both the patient and their family.

There is a significant lack of support for rare disease patients with both their medical and non-medical needs. Only a third of respondents reported receiving sufficient support with their social needs, fewer still (29%) felt that they receive adequate psychological support. Less than a quarter (24%) receive enough support with financial concerns. This is particularly problematic when considering that 61% of those who care for someone with a rare disease reported that their role as a carer affected their ability to hold paid employment.



Anna Pickering



Dame Anne Begg MP

The report highlights a lack of information about all aspects of living with and managing a rare condition. Over half of respondents (52%) felt that they hadn't been given sufficient information on their condition following diagnosis. Even more worryingly, many respondents elaborated that they were given no information at all on diagnosis. These patients are left to their own initiative to find information on their condition even though good quality information about rare diseases can be difficult to find. Nearly two-thirds of patients (64%) were not given details of the relevant patient support groups at the time of diagnosis, despite the fact that in RDUK's experience these organisations are a vital source of information and support, in the survey 52% reported that patient support groups are the main source of information on their condition.

Most rare diseases affect multiple parts of the body and many different professionals often need to be involved in care and treatment. It is essential that there is good coordination and communication between all those involved. However, 75% of respondents do not have a care coordinator. This aggravates a number of other problems experienced by patients and families as a result of poor coordination of care. Some of the problems highlighted included each professional the patient comes into contact

with looking at a specific aspect of the condition, but no one being concerned with the condition as a whole, patients or families having to repeatedly explain their condition to all those involved in their care due to a lack of knowledge about rare diseases; feelings of being lost in the healthcare system; patients' notes being lost or not passed on, patients and families having no one to go to with queries about their condition or care and a lack of continuity of professionals involved in the care of the patient.

These are just some of the findings highlighted in "Experiences of Rare Diseases: An insight from patients and families". What is particularly striking is that despite the vast range of rare diseases, each with different symptoms and prognoses, patients and families face

similar problems in areas such as accessing care, information, support and treatment.

What next?

RDUK will continue to use the report and the findings of the survey to raise awareness of the issues experienced by patients and families affected by rare diseases. The results will also inform RDUK's report outlining recommendations for a strategy for rare diseases, which will be launched to coincide with Rare Disease Day 2011.

Each of the pictured people's stories feature in our report. The report is available to download from the RDUK website: <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>

If you would like a hard copy please contact RDUK.



The Mitchell family
Stephen Nutt, Public Affairs Officer and Secretariat to Rare Disease UK and Lauren Limb, Research Assistant, Rare Disease UK
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Paving the Way

Douglas Caldwell, Project Officer for Paving the Way, provides an update on how the project has been running and future plans for the podcasts.

A lot has been happening since I was first asked to introduce the 'Paving the Way' project to you all back in our last newsletter. 'Paving the Way' will be creating a series of online video interviews of people with rare genetic conditions talking about how they have become experts in managing their condition. This will hopefully help lead to health professionals recognising the expertise of people with genetic conditions and provide a lot of help for people starting out on their own management.

The first thing I want to say is a big thank you to everyone who has put up with my constant e-mailing and phone calls. Most of all I'd like to thank everyone who has invited me into their homes to talk about the conditions they live with and how they've learned to manage them effectively. I'm very pleased that many of you are going to be sharing the stories you've told me with the rest of the Genetic Alliance UK umbrella and beyond.

Right now 'Paving the Way' is on track for its late May/early June launch having had only minor setbacks. We've had what I think are the expected issues surrounding a video based project, namely camera shyness and I think when the project is over there will be a bigger than usual round of applause for the brave souls who put aside their personal misgivings and sat in front of the cameras to tell their tales.

The project has been aided along brilliantly by our Steering Group, who at our first meeting decided the shape and feel of the project and who have since been putting me in touch with many of the people who will be providing you all with not only informative self-management stories but also at times entertaining ones. The recent weather has unfortunately delayed our meeting in December but like troopers they have been working away with

me by e-mail to keep the project from being delayed and I believe that filming of the interviews will commence by mid-January.

The finished interviews will be approximately 10 minutes long and feature a wide range of self-management stories with both carers and the people living with rare genetic conditions. We have a nice range of genetic conditions as well, including some ultra rare ones you may never have heard of and it's going to be fantastic to help raise awareness of these conditions by giving people a chance to talk about them. Don't worry if your particular genetic condition isn't being featured as the people being interviewed definitely have advice for everyone that transcends genetic boundaries and I'm positive everyone will be able to learn something from them. We are also hopeful that there will be at least one interview with a health professional to provide another perspective on self-management and how to get the best results when working with your health professional on managing your condition.

At the moment I'm working on the best placement for the interviews within the Genetic Alliance UK website, arranging shooting schedules and preparing the editing equipment. I think people will be able to look forward to our new YouTube account in the future which I hope the Genetic Alliance UK team will use with our fantastic new video equipment to fill for years to come but don't worry all videos will still be able to be found on the Genetic Alliance UK website.

Douglas Caldwell, Project Officer (Self Management)
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Risks and Benefits Citizens' Jury

– Update and Focus Group Recruitment

We are now over three months into the Risks and Benefits Citizens' Jury project. Amy Simpson provides a brief re-cap on the project's aim and method, outlines what we have achieved so far and our next steps, as well as letting you know how you can get involved!

Aim and Method

The overarching aim of the project is to systematically examine how patients and their families perceive the balance between the risks and the benefits of new biomedical therapies. The study will be based on a Citizens' Jury model. The Citizens' Jury is used as a participatory research method, designed to allow decision-makers to hear the people's voice. It draws upon aspects of a legal trial by jury, bringing together a small group of citizens (in this case, up to 16 patients and family members) to form a jury over a few days, to learn about and deliberate a particular issue in depth. Typically, the jury will reach a 'verdict' or frame a set of recommendations.

Progress and Next Steps

In order to do something useful within the time and resources, we have to narrow the scope of the research. As the jury are not due to first meet until September 2011, a significant amount of time has been dedicated to the careful consideration of the precise focus of the jury and the development of a detailed project proposal. This work is well under way, and for the first three months we have spent time familiarizing ourselves with the relevant research and drug regulatory systems, exploring how

decisions and risk/benefit assessments are made within this system, and scoping the issue to develop a detailed project proposal. We are working closely with key stakeholders and the Steering Committee.

So far we have discussed a number of important elements with the Steering Committee:

- Selected jurors will be patients (or family members of patients) who have severe conditions. The severe condition will have a genetic component and possible be rare.
- Examining the broad principles and values of risks and benefits is important, as attributing all findings to one national system may limit the use and 'shelf life' of the findings.
- Jurors will deliberate the risks and benefits associated with a small number of hypothetical cases, whilst considering some of the complexities and difficulties that occur within the research and regulatory system.
- The results of this research would hopefully be read by the MHRA/EMA and their equivalents in other member states, as well as HTA bodies.

Over the next couple of months, we will be exploring and consulting further with Steering Committee members and other stakeholders to agree and 'sign off' the detailed project proposal.

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Joint RDUK/AMRC Workshop on Rare Disease Research

Secretariat to RDUK, Stephen Nutt, brings us up to date on the recent RDUK/AMRC joint workshop on rare disease research.

During December, Rare Disease UK held a joint workshop with the Association of Medical Research Charities (AMRC) exploring issues around research into rare diseases. The AMRC is a membership organisation of the leading medical and health research charities in the UK with the aim of supporting the sector's effectiveness and advancing medical research.

The aim of the workshop was to provide a forum for those funding research into rare disorders to come together to discuss and share the issues they face. The thoughts and insights discussed during the day were captured to help

inform RDUK's report into a strategy for rare diseases which is being launched on Rare Disease Day 2011.

The workshop brought together over 50 delegates representing patient organisations, industry and research. Guest speakers were on hand to stimulate discussion by setting out the issues from their perspective.

The first session looked at perspectives of both industry and patient organisations funding research to examine whether there are common problems faced by both and whether there are ways to increase collaboration. Dr Susan Walsh, from the CGD Research Trust spoke from the perspective of a patient organisation and Dr Jon Beauchamp, from Alexion gave an industry view.

Dr Anil Mehta began the second session by taking about his experience of conducting research into cystic fibrosis at the University of Dundee and how the EuroCareCF network has facilitated this. The session went on to examine the

challenges of researching without a network and how these can be overcome.

The next session "Getting and Keeping Research Capacity" was introduced by Dr Robin Ali from the UCL Institute of Ophthalmology, who spoke about how his team has managed to attract funding and researchers with an interest and retain this expertise in the development of gene and cell therapy for the treatment of retinal disorders.

Finally, to tie it back into the ultimate aim of influencing policy in this area, Mike Birtwistle, Managing Director of Health Mandate, gave attendees advice on how they can influence decision makers in what has traditionally been considered a "niche" area and against competing voices in the area.

The AMRC's website: www.amrc.org.uk

Stephen Nutt - Public Affairs Officer and Secretariat to Rare Disease UK
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RAPID Annual Conference

An update on the recent RAPID Annual Conference and our work in this project is provided by Celine Lewis.

November 2010 saw the first RAPID (Reliable Accurate Prenatal non-Invasive Diagnosis) annual conference since the kick-off meeting last year. The RAPID programme aims to evaluate all aspects of a new prenatal testing technique called Non Invasive Prenatal Diagnosis (NIPD). During pregnancy, the fetus's DNA circulates in the mother's bloodstream. NIPD allows us to use a normal blood sample, taken from the mother's arm, and look at the fetus's DNA which is circulating in the mother's blood. Currently, we are able to use NIPD to determine the sex of the fetus. Research is also being conducted to enable us to use NIPD to identify whether the fetus has a particular genetic condition. As NIPD is a simple blood test, there is no risk of miscarriage as with invasive tests such as amniocentesis or Chorionic villus sampling (CVS) and testing will be possible earlier in pregnancy, as early as nine weeks. The RAPID programme will confirm laboratory standards, evaluate cost effectiveness, determine parents and health professional views and needs, consider the wider social and ethical issues and develop an education plan for health professionals and the public.

Genetic Alliance UK's role within the RAPID programme is to investigate the preferences and information requirements of service users who have used or may consider using this new test in the future. Over the past five months we have been interviewing women that have used NIPD to determine the sex of the fetus. This is important for X-linked conditions which only affect boys, such as haemophilia and muscular dystrophy. The RAPID annual conference was an opportunity to feedback the findings from these interviews to interested stakeholders, such as midwives, genetic counsellors, screening coordinators, commissioners and patient groups.

The key findings that have emerged from the interviews so far are as follows:

- The value women perceive from NIPD is that it is accurate, does not present any risk to the fetus, and can be done early.
- Identifying the sex of the fetus at nine weeks reduces anxiety early on during pregnancy. If the fetus is female, then there is no further worry, if it is male, the information is useful because it enables invasive testing to be performed as early as possible to see if the fetus is affected.

Alternatively, it gives parents the opportunity to prepare practically and mentally for the birth of a child with a genetic condition.

- Knowing the sex of the baby early on gave women a sense of control over their pregnancy which they valued.
- Women's views about raising a child with a particular genetic condition will be shaped by the information they have about the condition, their religious and personal beliefs and their own experience of the condition in their family.
- One possible negative outcome of NIPD is that parents are more likely to bond with the baby early on when the risk of miscarriage is still elevated.

Over the next year we will be interviewing more carriers, in particular those who might be likely to use NIPD in the future. We will also be developing informed and accessible patient information with the help of patients and families. If you are a carrier of a genetic condition and would like to be involved, please contact.

Celine Lewis, Project Officer (RAPID)
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New Member of the Team

Welcome to Julian Walker – Genetic Alliance UK's new Marketing and Communications Officer

Introducing our newest member of the team, who you will all be meeting and working with at some point over the coming year.

As you will all know we have been very busy over the past couple of years with many new projects and lots of policy and campaigning work. We now need to have a dedicated member of our team who can focus on telling our members, as

well as all our other contacts including researchers, the genetics community, funders and many more about what we have been up to.

We are delighted to have appointed Julian Walker to this newly created role. Julian has a professional certificate in marketing and has been working at the RSPB since February last year as a Marketing Assistant.

Julian will be joining the team in January and will be getting to know you all in the coming months, I hope you are able to welcome him and to be generous enough to spare a little bit of time to let him know about all the interesting and rewarding work that you do as well.

Julian can be contacted by email on julian@geneticalliance.org.uk

UK Europlan Conference – The Rare Disease Community comes together to express views, opinions and solutions into dealing with rare diseases.

Assistant Director, Melissa Hillier, fills us in on the recent Europlan Conference, what the current work packages are working towards and the outcomes of the UK workshop.

In November 2010, the UK held its Europlan Conference. Europlan which is the – “European Project for Rare Diseases National Plans Development” is a three year project which aims to provide recommendations, best practices, case studies and indicators to help each EU Member State define their strategy for rare diseases. The EUROPLAN project includes 25 countries. It aims to assist and encourage member states in developing national health policies to ensure equal access and availability of prevention, diagnosis, treatment for people with rare diseases.

EUROPLAN is looking at each European country to understand how they are currently providing services for those affected by rare conditions. The project will also analyse case studies to identify already successful experiences and is coordinated by Dr. Domenica Taruscio, Director of the National Centre for Rare Diseases, Italy.

Stakeholders within the project include national health and social care authorities as well as health care planners, health care professionals, researchers, patients and patient organisations. EUROPLAN will stimulate discussion and hopefully reach consensus as well as acting as a catalyst to promote the development of national strategies in all European countries.

Work Packages (WP)

The EUROPLAN project is divided into ‘work packages’ and we have outlined five of these below. They look at various aspects of the project including the collection of information on country initiatives as well as the development of indicators for implementing, monitoring and evaluating strategies for rare diseases.

WP4 collects information on the initiatives undertaken by EU Member States on rare diseases. A dedicated survey is aimed at gathering examples of different Member States experiences and lessons learned allowing the sharing of information among Member States.

WP5 develops indicators for monitoring the implementation and for evaluating the impact of National Plans or Strategies for rare diseases.

WP6 analyses case studies to identify relevant experiences.

WP7 develops the content of the EUROPLAN recommendations including a methodological guidance to design comprehensive and integrated strategies for guiding and structuring all relevant actions in the field of rare diseases.

WP8 deals with the organisation of National Conferences where national stakeholders are convened to discuss the EUROPLAN Guidance Document within the specific national context.

The last work package, WP8, is concerned with the organisation of national conferences. These conferences are key to gaining the real-life perspective of services, information and support for those affected by rare conditions within each country.

It will be an opportunity to discuss the transferability of recommendations with UK stakeholders, including health professionals, researchers, patients and industry and will enable the UK to feed into the European debate on the development of national strategies for rare diseases.

The UK workshop

The UK workshop was an opportunity for those working in the field of rare conditions to input into the policy debate around the commissioning, organising and coordinating of services in the UK. We heard from Alastair Kent, Chair of Rare Disease UK, who updated delegates on the work of Genetic Alliance UK and Rare Disease UK into the national strategy and we also heard from Ed Jessop from the National Commissioning Group at the Department of Health, who set the scene from the Department’s perspective.

We had over 80 delegates attending and we certainly had a packed day, we had workshops on various aspects of what should be included into a UK strategy for rare diseases including, governance issues, definitions and codifications, centres of expertise, research and patient empowerment. The discussions were very fruitful and the ideas that were generated and the examples of good practice raised will go into a report for the EUROPLAN project as well as feed into the work that Rare Disease UK is carrying out. The EUROPLAN reports will be gathered, collated and disseminated to EU officials so they can gauge a better understanding of how each EU member state is preparing for their rare disease strategies and also to enable tools to be developed to help this process.

The findings from the EUROPLAN project will have direct relevance to EU Health Ministers and will enable national stakeholders to communicate their experiences of rare conditions to be shared amongst other member states. A report will be available in February from the UK workshop and will be published on the Rare Disease UK and Genetic Alliance UK websites.

For more information about EUROPLAN see <http://www.eurordis.org/content/europlan-guidance-national-plans-and-conferences>

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