

# Genetic Alliance UK

## Newsletter



Autumn 2010

## Reforming the NHS?

It is clear that the coalition government has got the bit between its teeth when it comes to reforming the NHS! Within weeks of coming into power the Secretary of State for Health had issued a White Paper-Equity and Excellence: Liberating the NHS, and a raft of other consultation documents which, if adopted in the way proposed, will constitute the most extensive change in the way health care is organised since the establishment of the NHS over 60 years ago in England. Wales, Scotland and Northern Ireland make their own arrangements.

Elsewhere in this edition we explain the changes in more detail - here it is enough to say that the government proposes the abolition of two tiers in the current system. Primary Care Trusts (PCTs) and Strategic Health Authorities are to go. Local commissioning will become the responsibility of consortia of GPs, while a new National Commissioning Board (NCB) will assume responsibility for services currently commissioned at regional or national level. At least that is the theory. The White Paper and its associated consultation documents are woefully short on detail, so it is difficult to work out what this may mean for the members of Genetic Alliance UK, and the individuals and families they support.

If the rhetoric of the White Paper can be translated into action then this represents a real opportunity to improve services and support from the NHS for families with genetic conditions. The new NCB could be a mechanism whereby appropriate models for services could be planned and implemented rationally across England. GP Consortia could prove to be more receptive to arguments based on patient need and the opportunity of health gain than PCTs have sometimes proved to be. And the importance of listening to the patients' voice: "nothing about us with us" is long overdue.

So far, so promising. Genetic Alliance UK has responded in detail to the various consultations and will be vigorously campaigning to ensure that the needs and

expectations of our members and those they represent are promoted in the measures the government takes to implement the changes it wishes to bring about.

The devil, however, is as always in the detail. The commitment to preserve the budget from the NHS is welcome, as is the emphasis on using resources to promote patient care. But the pressure on the NHS budget is substantial and this is likely to increase due to our aging population and increasing possibilities to intervene in serious diseases. This will inevitably generate a situation where hard choices have to be made in order to make the money go round as effectively as possible.

We will be campaigning vigorously to ensure that individuals and families affected by genetic conditions get their fair share of the NHS "cake". We will work alongside doctors, scientists, planners, politicians and all the other vested interests to make the most of the opportunities that this re-organisation of the NHS presents. We will try to ensure that patients and families faced with the daily reality of living with genetic conditions that reduce the quality or quantity (or both) of their lives can be confident that the health service is there for them and able to deliver high quality care based on good clinical practice and sound science in a timely, effective and user friendly way. To join us in this campaign please contact myself, Melissa, Stephen or Nick at Genetic Alliance UK and we will happily share ideas about what to do to move things forward!



Alastair Kent, Director

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# DH Consultations

## **Nick Meade, Policy Analyst, gives an overview of Genetic Alliance UK's response to the DH White Paper – Equity and Excellence: Liberating the NHS released in July this year.**

As Alastair has said on the front page, there are many changes coming our way in the NHS in England. Since the publication of the White Paper in July and its accompanying consultation documents we've been working hard on communicating how these changes will affect our members and the patients and families they support. Our consultation documents are in their final revisions as I write this but by the time you are reading this issue, they will be available to read on our website. I am going to explain some of the key messages we have incorporated into our responses.

### **Outcome measures**

The NHS's performance is going to be measured in a different way in the future. Five domains have been identified, within which performance will be measured by "key performance indicators" and improvement will be targeted in "improvement areas". We were delighted to see that one of these five domains will be "enhancing the quality of life for people with long-term conditions" as many of the conditions our members support or live with would fit into this category.

Unfortunately, the concept behind the system seems likely to let many of our members down. Both "key performance indicators" and "improvement areas" are likely to be very targeted, with a danger of focussing the attention of the health service on specific areas to the disadvantage of those affected by conditions which are not being highlighted.

Taking a closer look at the domain for chronic conditions, we found that of the 24 indicators for "improvement areas", only 1 covers tangible outcomes for a patient population covered by Genetic Alliance UK's umbrella. 16 indicators do not describe any of our members, and the remainder will hide individual conditions behind vague, broad indications.

We have therefore made it very clear in our responses that this system of measuring outcomes and prioritising improvements will neglect the vast number of conditions with small patient populations, which together are a large number of patients.

We hope that appropriate use of patient reported outcome measures will help to counter-balance this tendency towards spotlighting certain areas of the NHS.

### **Commissioning**

One of the biggest changes will be in the method by which health care is planned and paid for in England. You will all have heard much about GP commissioning in the news when the White Paper was first published in the summer. Commissioning at the other end of the spectrum, specialised commissioning: for services which are less commonly used, did not receive as much of the lime light.

The truth is that there is not enough detail in the White Paper and the supporting consultation for us to be able to see exactly how specialised commissioning will change in the future; but one thing is certain: the bodies around which the system is currently based, are changing drastically or disappearing entirely. The days of regional Strategic Health Authorities and the regional Specialised Commissioning Groups seem numbered; and we know that specialised commissioning will become the remit of the new NHS Commissioning Board.

We have taken this opportunity to communicate how this process of change can be used to improve specialised commissioning. The current regional structures contribute to current "postcode lotteries" and we hope that the restructure will be used to best advantage to alleviate these problems.

### **An uncertain future**

Our consultation responses include many more issues of course, please examine them online, we will be pleased to discuss them further. We hope that our responses will influence the legislation that will be coming to Parliament soon and that they will influence future policy in the NHS. Genetic Alliance UK will watch the progress of these changes closely, and will continue to communicate the needs of all those affected by genetic conditions.

### **What is a PROM?**

A Patient Reported Outcome Measure (PROM) is a means by which information can be gathered on the effectiveness of healthcare, by measuring what is important to patients via a small questionnaire. More information can be found here: [www.ic.nhs.uk/proms](http://www.ic.nhs.uk/proms)

# The animal research regulation debate is moving to the UK

**Nick Meade provides an update on where the revision of the Directive 86/609/EEC, this governs animal research within Europe.**

The revision of Directive 86/609/EEC, which governs animal research in Europe, has been a long running story in these pages. I'm delighted to announce the end of a chapter in this story. The European Parliament has voted to adopt the revision at its second reading this month.

As the UK charity for those affected by genetic conditions, we are very aware of the number of conditions affecting our members for which there is no cure, or for which treatment is inadequate. We recognise biomedical research as the route by which these cures and treatments will arrive. Animal research is a necessary component of biomedical research and is a practice which frequently comes under fire. Since our members are the likely beneficiaries of biomedical research, we have decided to add our voice to those that support animal research as a necessary component of biomedical research and have followed this revision closely for the past few years.

The Members of the European Parliament (MEPs) have voted to accept the current proposal for changing the law in Europe and

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soon the processes to convert this into UK law will begin. There is still scope for debate on this issue, as there are a number of areas in the current text which can be interpreted in more than one way. Of particular concern are the interpretation of restrictions on the use of Non-Human-Primates (NHPS - animals which are vital to the understanding of some complex health concerns such as neurological conditions), which are intended to prevent trivial use, but could be misinterpreted to restrict use to a degree damaging to biomedical research.

There will be a Government consultation on the planned implementation of this regulation at the end of this year. Genetic Alliance UK will continue to take an active interest in the issue. If you would like to give your view on this issue, or discuss the matter further, please get in touch with me.

## Opportunity for young people affected by genetic conditions to get involved in research.

Are you 16-21? Do you feel unheard? Do you feel there's enough information available to you about living with a possible genetic condition? Would you like to change that?

MSc Genetic Counselling students from Cardiff University are looking for a group of young people in South Wales to create and produce a short internet film to support other young people living at risk of a genetic condition.

There is a lot of information and research into helping parents talk to their children about genetic conditions but very little that is directly for young people themselves. And NOTHING designed by young people. If you want to change this then join our project.

We are looking for young people who are at risk of a genetic condition. This could mean there is a late-onset condition in your family, or you could be a carrier of a condition, or you have family members with a condition and don't know how this affects you.

We will be holding 3 workshops for you to have basic film making training so you can work together to produce a short film to support other young people in similar situations and have your voice heard. This is a fantastic opportunity, especially for anyone wanting to gain more experience in science or media, and will look great on CVs and UCAS applications. You will receive a certificate of recognition at the end, and a gift voucher as a thank you. Food and refreshments will also be provided at the workshops.

Workshops will be held in South Wales and participants will need to be able to make their own way to them, although the venue will be chosen based on the most convenient location for the majority of the group.

**If you are interested in this study, or would like to know more, please contact Catherine Johnston, MSc student on [johnstoncm1@cardiff.ac.uk](mailto:johnstoncm1@cardiff.ac.uk) or Project Supervisor, Rachel Iredale, on [Rachel.Iredale@wales.nhs.uk](mailto:Rachel.Iredale@wales.nhs.uk) or 02920 746731.**

# RASopathies Network Launch 2010

## Project Officer, Krystle Kontoh, writes about the successful launch of the RASopathies Network at the House of Lords.

On Friday the 10th of September, a 120 strong delegation of patients and their families, healthcare professionals and policy workers descended upon the House of Lords to mark the launch of the 'RASopathies Network' - a patient and clinician partnered initiative.

'RASopathies' denote a class of developmental syndromes that are caused by germline mutations in genes that encode protein components of the Ras/mitogen activated protein kinase (MAPK) pathway. These conditions are attracting an increasing amount of attention from the medical community particularly due to their link to cancer (in six of the nine syndromes). The conditions are:

- Noonan Syndrome
- LEOPARD Syndrome
- Hereditary Gingival Fibromatosis Type 1
- Capillary Malformation-AV Malformation Syndrome
- Neurofibromatosis Type 1
- Legius Syndrome
- Costello Syndrome
- Cardio-facio-cutaneous Syndrome – CFC
- Autoimmune Lymphoproliferative Syndrome

The impressive guest list included well respected health care professionals within the UK, Dr. Bronwyn Kerr, Dr. Edmund Jessop, esteemed international speakers Prof. Bruce Gelb and Dr. Martin Zenker, and Melissa Hillier from the project team.

This momentous event was the first of its kind to be held in the UK and signified a culmination of two years work on behalf of Genetic Alliance UK's Facilitating Networks project. During this time, the project team has successfully harnessed the hard work, passion and commitment of patient representatives from the Costello Syndrome Support Group, Neurofibromatosis Association, BDF Newlife and CFC Syndrome in the UK.

When any of the RASopathy conditions are considered in isolation they can be classified as rare, however, combined it has been estimated that RASopathies account for one in one thousand births in the UK. With an increased collective patient mass, the support groups concerned have each expressed their commitment to creating a concerted approach to sharing information raising awareness and facilitating medical research.

This UK RASopathies based network is a critical first step in connecting the dots for collaborative international research efforts and Genetic Alliance UK is delighted to have played such an instrumental role in its establishment.

For more information please look on the Rasopathies Alliance new website <http://ras-pathway-syndromes.com/>

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*Dr Kenneth Bird, Krystle Kontoh, Lord Ward Atherton*



*Ir Stephen Nutt, Melissa Hillier and Krystle Kontoh*

# Route Maps for Rare Conditions

Jessica Burke, Project Manager, details this new project and the progress made to date on this innovative new approach to developing care models.

The Route Maps for Rare Conditions is an exciting new project that will develop a practical and cost-effective framework for improving information, access and coordination of health and social care services for individuals and families with a wide range of rare genetic conditions. This pilot scheme will support ten user-led support groups to develop a series of condition specific Route Maps. A toolkit and a written methodology will be created alongside the Route Maps so that in the future other groups can develop a Route Map for their own condition.

Each Route Map will provide a comprehensive resource for use by patients, families and health and social care professionals and will have the potential to play a key role in personalised care planning for people with these conditions. This project will run for three years from July 2010.



## The Outcomes of the Project

- The development of a practical methodology for improving information, access and coordination of health and social care services for people with rare conditions
- A comprehensive evaluation of the effectiveness of this approach

### More specifically:

- Improved access to information for patients, families and health and social care professionals, leading to increased awareness and understanding of these conditions.
- Improved access to health and social care services for patients and families, leading to earlier diagnosis, improved care and support.
- The development of a practical tool that will enable patients and families to better understand their condition and take an active role in their care.
- The empowerment of patients and families with rare conditions, by facilitating their direct contribution to improving service provision.
- A written evaluation of the effectiveness of this approach in improving care for patients and families with rare conditions.
- The development of a practical 'toolkit' of resources that can be used to enable other support groups to develop their own Route Map.
- Increased understanding of the mechanisms involved in developing the role of user-led support group and volunteers with personal experience of health conditions in projects of this kind.
- A written evaluation of the project process, including recommendations about how to develop the role of user-led support groups and volunteers in projects of this kind. The ten groups involved are

- The Jennifer Trust for Spinal Muscular Atrophy
- UK Thalassaemia
- The Stuart Strange Trust for Vasculitis UK
- Breathtakers Charity
- Prader-Willi Syndrome Association
- AKU Society
- A-T Society
- Cri du Chat Syndrome Support Group
- ALD Life
- British Retinitis Pigmentosa Society RP Fighting Blindness

An Advisory Committee has been set up to support the project. This group consists of advisors that have been involved in Route Maps of similar projects before. The members are:

- John Devlin, Cystic Fibrosis Trust
- Mike Knapton, Trustee, Genetic Alliance UK
- Alison Metcalf, School of Health and Population Sciences, University of Birmingham
- Greta Westwood, CLRN Senior Manager, National Institute for Health Research (NIHR) Comprehensive Clinical Research Network (CCRN)
- Marion McAlliastair, Genetic Counsellor/MRC Research Fellow, NOWGEN
- Jo Grey, CEO AMEND

Currently, all of the chosen groups are working on developing a plan of action and the initial stages of their Route Map. The project page on the Genetic Alliance UK will be updated on a regular basis so keep an eye on the page for future information.

Jessica Burke, Project Manager  
Route Maps for Rare Conditions [jessica@geneticalliance.org.uk](mailto:jessica@geneticalliance.org.uk)

# AllClear Life Insurance

“AllClear Life - a unique fast online service which allows those with genetically related conditions to obtain an indication of the likely cost of Life Insurance. AllClear Life also offers an 'Expert Referral Service' for those with more complex conditions”

Genetic Alliance UK has been working with AllClear Life for some time to help provide anonymised case studies in order to trial their new online system. This system is now launched. AllClear Life enables individuals to anonymously complete information in relation to 42 medical conditions, the system is then able to provide an indication of the cost that might be involved for life insurance. If the applicant would like to take the application forward AllClear Life can introduce them to an insurance specialist.

This new process can save many months of waiting whilst insurance companies review a Life Insurance application, as it provides the individual with an indication of the potential costs

involved immediately. That individual can then decide if they wish to proceed with an application or not. This can crucially save time and money as well as enabling patients to find out immediately the likely cost of a life insurance policy.

AllClear Life are keen to extend the number of conditions they currently cover, so if your condition does not appear on their list please do contact them.

[www.allclearlife.co.uk](http://www.allclearlife.co.uk)

## Background information on AllClear Life

AllClear Life and its partners have developed a unique on line facility that enables people with medical conditions gain an understanding of the likely cost of term life insurance. Until now, anyone with a medical condition will not know the cost of life insurance until they have applied and the insurance company has completed its underwriting process, which can take up to three months to complete.

The service operates on the principle of initial anonymity, that is to say that an enquirer is not required to divulge identification details to use the system. By inputting details about their medical condition and lifestyle an enquirer can obtain an estimate of the likely cost of life insurance. If, at that stage, the enquirer wishes to make an application for life insurance the system will request contact details, and the enquiry will be referred to our partner All My Plans, a regulated financial intermediary, who will manage the application quickly and efficiently.

In association with our partner All My Plans we also specialise in the provision of estimates of the cost of life insurance for people with more complex or severe medical conditions, and if appropriate can access specialist insurers for situations where the mainstream market is not willing to offer cover.

Currently, the service covers 42 medical conditions, including four which are genetically based. AllClear will be constantly increasing the range of medical conditions and are keen to include additional genetically related medical conditions.





RARE DISEASE | UK

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

# Rare Disease UK Survey of Patient and Family Experiences

**Public Affairs Officer, Stephen Nutt and Lauren Limb, Research Assistant from Rare Disease UK explain more about the results from the recent survey about patient and family experiences.**

Rare Disease UK recently carried out a survey of the experiences of patients and families affected by rare diseases. This survey covered multiple aspects of rare diseases including diagnosis, delivery of care, awareness and participation in research, access to drugs and services, and access to thorough, reliable information and support.

We were delighted to receive almost 600 responses from patients, relatives and carers affected by a wide range of rare conditions and we would like to thank all the Genetic Alliance UK members and the many others who took the time to tell us of their experiences. We will be using the results from this survey along with several case studies to produce a publication to improve awareness of rare diseases amongst the public and policy makers. This will also be used to add weight to the need for a national strategy for rare diseases by highlighting some of the common issues currently faced by patients and their families. Rare Disease UK are working to produce a report as to what this national strategy for rare diseases should include to ensure that patients of rare conditions are able to access equitable, high quality care, information and support in a timely manner, and to reduce the fragmentation of the services they receive.

The full results of this survey will be launched in November but initial findings have confirmed that there are indeed many common issues experienced by patients of rare diseases and their families.

Many respondents reported problems in getting a diagnosis such as delays following the onset of symptoms and numerous misdiagnoses.

"Varying diagnosis and incorrect information given. It was only after the death of our youngest daughter at the age of 6 months that diagnosis was received".

## **Relative of a patient with Gorlin Syndrome**

"Many doctors had no idea and some even said the symptoms were in my head".

## **Patient, Myasthenia Gravis**

"[We received] many incorrect diagnoses - but the worse diagnosis doctors (especially GPs) give is there is nothing wrong with your child - perhaps the problem is with you (the parent)".

## **Mother of a child who died from Haemophagocytic Lymphohistiocytosis.**

Another issue faced by many respondents was the lack of information they were given on their condition and the lack of support they were provided with following diagnosis.

Many commented on the importance of patient organisations in providing reliable information, and in keeping them updated on current research.

"Support in all areas has been poor. Benefits are now coming in because of constant pressure of me and my parents".

## **Patient, Becker Muscular Dystrophy**

"When we heard about our condition we were left to deal with it. I saw an article in the paper about a lady who had the same condition as me and there was a number to ring which was Ataxia UK. I can't thank them enough for bringing us out of the woods".

Patient, Familial Hemiplegic Migraine Type2 with Cerebellar Ataxia

"I only feel I get up to date information because of my involvement with a patient group, otherwise, I would not hear about any ongoing research".

## **Patient, Acute Intermittent Porphyrria**

Many patients need to see multiple different specialists for their condition, but too frequently a lack of communication and coordination of their care was reported. Problems at transition periods, particularly from paediatric to adult services, were also common. These included a lack of support with medical, psychological, social and financial support.

"I was 'forgotten' about by medics when I turned 18." Patient with Ehlers-Danlos Syndrome, Postural Orthostatic Tachycardia Syndrome, Raynauds Syndrome.

"There was no transition help at all".

## **Relative of a patient with Ichthyosis**

"Better co-ordination of care is needed for all affected by long term medical conditions".

## **Relative of a patient with Gorlin Syndrome**

**Stephen Nutt, Public Affairs Assistant and Lauren Limb, Research Assistant, [stephen@raredisease.org.uk](mailto:stephen@raredisease.org.uk) and [lauren@raredisease.org.uk](mailto:lauren@raredisease.org.uk)**

# British Medical Association Medical Book Awards 2010 Award Ceremony

## Genetic Alliance UK was recently successful at the British Medical Association Medical Books Awards 2010, receiving a Commended and Highly Commended.

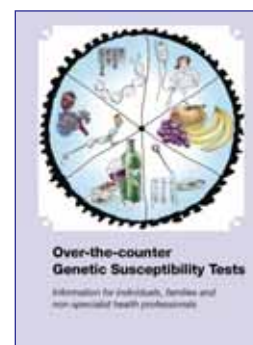
This year Genetic Alliance UK entered three of our patient information leaflets into the British Medical Association (BMA) Patient Information Awards. We were delighted to find out that two of them received a Commended and Highly Commended, 'Living without a Diagnosis – Information for Parents'; and 'Over the Counter Genetic Susceptibility Tests'.

The Patient Information Awards are part of the BMA Medical Book Competition, the awards for patient information were introduced in 1997, with the aim to encourage the production, dissemination and evaluation of accessible, evidence-based and well-designed patient information. This year there were over 50 publications that achieved Highly Commended and we were all invited to attend the Medical Book Awards Ceremony at BMA House in London. Melissa Hillier attended this event with Laura Gilbert, who is a freelance consultant and researched and wrote the publication nominated.

All the Highly Commended publications were entered into various special prizes awards, including the overall award for children, creative design, information for carers, men's health, NHS Trust, online and digital media, people with learning difficulties,

women's health and young people. There was then a prize for the Patient Information Resource of the Year.

We were very pleased to have been nominated for these two awards, amongst other large charity names including the British Heart Foundation, Breakthrough Breast Cancer and the Terrence Higgins Trust. We hope that this recognition of the patient information we provide will enhance the credibility of our work and ensure our resources are disseminated widely.



## Eurogentest 2

### Priority setting in genetic testing

The current genetic testing environment is constantly evolving, here Fransika Severin, an Intern with Genetic Alliance UK, describes a new project aiming to clarify prioritisation process in Europe.

#### Current situation

The intensity of research into molecular genetics is leading to the very rapid invention of new genetic tests. These new tests are often highly specialized and expensive. Simultaneously, there are demands in many European countries to cut the costs of public health care. This creates a situation where it is not always possible to fund all desirable tests (e.g. due to budget constraints), so that some tests must be withheld. The geneticist must decide which patient should be offered a genetic test and which patient must be given alternative options. In professional jargon, you call these issues 'prioritisation decisions'.

Although prioritisation decisions may vary from region to region as well as country to country, decision makers are still faced with situations where explicit criteria for the prioritisation process aren't sufficiently defined. As a result, decisions about whether to test or not are made on an ad hoc basis which leads to an unequal distribution of benefits and harm. This underlines the need for guidance for these situations.

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#### What we are aiming at

Given this situation, Genetic Alliance UK in collaboration with the Helmholtz Center Munich is currently preparing a survey among geneticists and patients groups within the EU. The aim will be to understand further resource scarcity within genetic clinics and the prioritisation criteria currently used in practice. As the views on the value derived from genetic testing may differ between patients and professionals, it is also important for us to hear from the patient perspective. Patients themselves may have experience of situations in which access to genetic tests has been limited because of prioritisation. Moreover, we want to include them in the process of developing prioritization criteria to ensure the criteria will be acceptable to them.

#### What we have done so far

The project began in September 2010. We are currently preparing a questionnaire for patients and clinicians and hope to start piloting them within the next few weeks. These questionnaires will serve as a tool for structured telephone interviews with patients and clinicians. The main challenge will be recruiting clinicians and patient representatives from different European countries so that we can gain an overview of what is happening in Europe.

Nevertheless, we are hoping to have results by Christmas so that we can present these in the next newsletter.

# Risks and Benefits

**The Risks and Benefits Citizen's Jury project has recently started this year, Amy Simpson will be leading this project and provides us with more detail.**

From September 2010, Amy Simpson will be taking on the role of Project Officer for the Risks and Benefits Citizen's Jury project. Amy is a Research Assistant based at the Welsh Institute for Health and Social Care (WIHSC). WIHSC is an academic department at the University of Glamorgan, which works to connect the world of research, policy and practice in Health and Social Care. Amy Simpson and Professor Marcus Longley (Director of WIHSC) will be working in close partnership with Genetic Alliance UK on the project.

Measuring and balancing the risks associated with new therapies against the potential benefits they may offer patients is an immensely complex decision making process, and there is a clear need for a structured and in-depth study into how patients and

families perceive this balance. Based on a Citizens' Jury model, the project will involve up to 16 patients and family members in deliberative dialogue on this issue, and will frame recommendations that can be used as a practical tool by policy makers and regulators.

The Citizens' Juries will meet at the end of 2011. The project Steering Committee met for the first time on 24th September, to begin considering the scope and focus of the research. For more information about the Risks and Benefits Citizen's Jury project, please contact Amy.

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# Familial Cancer Risk in Ethnic Minorities

**A research project has recently begun, led by Anna Allford, Research Assistant, looking into why there is a lack of representation of cancer referrals from minority ethnic groups to genetics services.**

We have begun research into why there is a significant under-representation of cancer referrals to clinical genetics services amongst people from minority ethnic groups. This project is funded by a grant from the Big Lottery Fund Research Programme and aims to inform the development of interventions in order to improve access to genetics services for people from minority ethnic communities in England.

Anna Allford, Research Assistant, met with a group of clinicians at the Medical Genetics Unit of St. George's Healthcare Trust (London) to explore the service experience for people from South Asian, Black Caribbean and White Irish communities. This informed the first interviews with clinicians at the Trust and further interviews with other clinicians and professionals are planned.

This Trust is collaborating with us to select patients from these communities who have already been seen by this genetics service so that we can invite people to participate and understand their experiences. Another regional genetics service in the West Midlands has agreed to be our second research site and an application to the Trust is underway for approval to begin.

## Where you can help

We are developing a leaflet and posters to distribute to support groups and organisations to advertise for research participants from the public who belong to the above ethnic minority communities. The participants may have a significant family history of the following common cancers but will not have had a consultation with any genetics services:

- bowel,
- breast (this is sometimes associated with a family history of ovarian cancer), and
- prostate cancer.

Genetic Alliance UK members will be sent an email with further details about this in the near future but if you are not yet a member and would like more information please contact Anna, email: [anna@geneticalliance.org.uk](mailto:anna@geneticalliance.org.uk) or phone the office: **020 7704 3141**. Please feel free to pass this information on to others who may be interested.

**Anna Allford, Research Assistant/Project Officer**  
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# New Projects

## My Condition, My Terms, My Life campaign

**We have just embarked on a new project “Paving the Way to Self Management” which is finding out how patients living with rare conditions in Scotland self manage their day-to-day lives. This work has been funded by the Long Term Conditions Alliance Scotland (LTCAS). To coincide with the work that LTCAS is carrying out in Scotland on self management, they have recently launched the “My Condition, My Terms, My Life” campaign.**

The LTCAS’s campaign has been developed to share the message that living with a long term condition does not stop you being in charge of your own life.

The campaign has been designed to help improve public understanding of what self management means for people living with long term conditions, and to encourage people living with long term conditions, and the people who support them to adopt a self management approach.

### Why is Self Management valuable?

Self management can put patients in the driving seat, giving them the opportunities to access knowledge and skills in order to help decide what is right for them. LTCA believes that self management can mean that patients and families are better informed about their condition, better prepared for life’s everyday challenges and better supported to deal with issues as they arise.

**Douglas Caldwell, Project officer Paving the Way to Self-Management**  
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Being told you have a condition for which there may be no cure can be devastating. Individuals respond in individual ways. It can often have a profound impact on a person’s well-being and self-esteem.

People try to cope as best they can with the support they have. But frequently they do not have the information or skills to manage their condition well. It is often difficult to make well informed decisions about their life; let alone make plans for the future. Together with health professionals and others who provide support, self management can help people to make decisions that are right for their life.

### Self management working with health services

Self management is not a replacement for good quality health services, it does not mean that an individual should manage their condition alone, but rather it’s about working together with people who can provide the right support, at the right time.

For more information on this campaign do have a look at the website created <http://www.myconditionmylife.org/>

If you would like more information about our Self Management Project please contact Douglas.

## New Members of the Team

### Jessica Burke - Project Manager - Route Map For Rare Conditions

Hi, my name is Jessica and I am the Project Manager working on the Route Maps for Rare Conditions project, this is a project newly started at Genetic Alliance UK. I am pleased to have started work on this exciting new project.

I recently moved to the UK from New Zealand, where I worked in the public health system (similar to the NHS) on various projects and programmes around obesity prevention, mental health, primary care and health of older people.

I have just spent the summer travelling around Europe and hope to see as much of the UK and Europe as I can while I am living over this side of the world. I have a background in Nutrition and Commerce from the University of Otago in New Zealand.

To keep up to date with the progress of the project, keep an eye out on the Route Maps for Rare Conditions project page on the Genetic Alliance UK website - <http://www.geneticalliance.org.uk/projects/routemapsrareconditions.htm>.

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## Natalie Frankish - Scottish Development Officer

I am very pleased to have joined Genetic Alliance UK as the Development Officer for Scotland. My previous role as an engagement advisor for individuals with long term health conditions highlighted the importance of representing patient's views at NHS and government level to ensure the right services are available for all who need them. This, coupled with my background in policy development at the Scottish Government, drew me to Genetic Alliance UK, an organisation I believe to be committed to representing and supporting all those affected by rare or genetic conditions.

My role is to represent the views of patients in Scotland, to influence decision makers in Government and the NHS in improving equity and access to service in Scotland. I will work closely with a large number of patient organisations in Scotland and sit on a number of steering groups, representing the patient voice in the development of new services and strategies relating

to genetic conditions. In addition, I will also be supporting the work of Rare Disease UK in Scotland including the events on Rare Disease Day 2011, an event I am really looking forward to already! I will also be responsible for keeping the Scottish website up to date with the work of Genetic Alliance UK in Scotland, so please check online to see what I am up to!

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## Douglas Caldwell - Project Officer - Paving the Way to Self Management

Hello everyone. I'm very happy to be joining Genetic Alliance UK and getting to work on the 'Paving the Way to Self-Management' project until May next year.

My role in the project is to create a series of 10 Vodcasts (online videos) featuring people with rare conditions from across Scotland describing how they have become experts at self-managing their condition.

We will be aiming to show the range of people that are involved in the management of a rare condition including family and carers as well as providing insight into the problems these people encounter and how they have endeavoured to overcome them. The videos will also show that people with rare conditions often become experts on their condition and can be more knowledgeable about the condition than the healthcare professionals that they come into contact with.



We hope that these videos will become a valuable training resource for health professionals as well as helping to inspire people to become more pro-active in managing their own conditions and even give people a few ideas about where to get started.

Remember to look out for the videos next year on the Genetic Alliance UK website.

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## Amy Simpson – Project Officer – Risks and Benefits

My name is Amy, and I will be taking on the role of Project Officer for the Risks and Benefits Citizens' Jury project. I am a Research Assistant based at the Welsh Institute for Health and Social Care, and I will be dedicating half of my week over the next 18 months, to work closely with Genetic Alliance UK. I graduated from Cardiff University in 2007 with a Joint Honours degree in History and Sociology. Since then I have contributed to a huge variety of research projects in the field of Health and Social Care, locally and across the UK. I am delighted to be part of the team and to be involved in the research at Genetic Alliance UK.

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## New Alliance is launched

The Ciliopathy Alliance UK is an alliance of medical researchers, doctors and patient organisations representing patients and families suffering from ciliopathy diseases.

Ciliopathy diseases are caused by defects in the function or structure of cilia, small hair-like protuberances, like antenna, found on the surface of virtually every cell in the human body. Once thought to be vestigial organs, defective cilia are now understood to underlie a number of devastating genetic conditions including:

Alstrom Syndrome, Laurence-Moon-Bardet-Biedl Syndrome, Nephronophthisis, Orofaciodigital Syndrome, Polycystic Kidney Disease, Primary Ciliary Dyskinesia, Senior-Loken Syndrome, Usher Syndrome.

### The Alliance was formed to:

- Share knowledge and understanding
- Promote awareness of ciliopathies and the respective patient organizations
- Encourage collaborative research and facilitate where possible
- Apply for research funding from government, trusts and foundations

The Ciliopathies Alliance is hosting its inaugural public meeting on 29th November from 12.00 – 4.30 at the Institute of Child Health, Kennedy Lecture Theatre, 30 Gulford Street, London WC1N 1EH. If you would like to attend please do register on their website <http://ciliopathyalliance.org.uk/>

## Genetic Alliance UK – Celebrates its 21st Annual General Meeting

**Genetic Alliance UK held its AGM on 19th October and we were very pleased to welcome three excellent speakers, prior to the formalities of the AGM.**

We heard from Josie Godfrey, from the National Specialised Commissioning Team, who spoke about how the current NHS restructure will affect patients with genetic conditions. She explained how specialised services are currently commissioned in England, the recent changes that they have been implementing and how they will move forward with the outcomes from the Department of Health White Paper and the national strategy for rare diseases.

We then heard from Dr Colin Stewart, a consultant bone marrow transplants for genetic conditions at Bristol Hospital and Michaela

Damin, CEO of the Barth Syndrome Trust, who jointly worked together to apply for national commissioning for Barth Syndrome, a very rare X linked condition that currently is known to affect about 150 boys worldwide.

It was very interesting to hear how the theory of national commissioning has developed and then to hear how it works in practice. All the presentations are available on our website, in our latest news section.