At Shire we believe that patient organisations are fundamental to the support and care of patients and their families affected by rare and orphan diseases.

Shire values and respects the experience of patient organisations and their deep understanding of the conditions they represent and we are committed to help them achieve their mission for patients.
At some point in their lives, 3.5 million people in the UK will be affected by a rare disease. This equates to 1 person in every 17, says Alastair Kent, director at Genetic Alliance UK.

Breaking boundaries in diagnosis and treatment

This statistic may be shocking but it highlights a frequent misconception of rare diseases. Whilst, individually, rare diseases may be classified as uncommon, collectively, they affect a large proportion of the population and represent one of the biggest areas of healthcare expenditure in the UK. They are also, paradoxically, an area which is often overlooked and underinvested in; both by policy makers and researchers. Today is Rare Disease Day; an international event created to raise awareness of diseases and treatment.

Rare disorders without borders

Often patients affected by rare conditions face long delays in obtaining a diagnosis and can be misdiagnosed (often multiple times) with more common conditions. Once patients have been given a diagnosis, they often struggle to find trusted sources of information or anyone who understands the condition to talk to. Reaching out across borders can help them find common solutions and remind them they are not alone.

Equally, health professionals struggle to get information on how the conditions should be managed. This is aggravated by a lack of scientific knowledge of the disease — often a lack of research means that there will be little understanding about how a disease affects the body, or what the outcome for the patient will be. There are no effective treatments for most rare diseases and very few conditions can be cured. It is these patients and health professionals who rely heavily on the international community for support and information on rare conditions as well as research to develop their understanding.

International rare disease research has a greater chance of success when teams of researchers from different countries work together to increase our understanding of the disease and the chance of finding successful therapies.

The need for a strategy and the rare disease plan

Each country in the European Union has committed to developing a plan to address the needs of patients living with rare diseases by the end of 2013, as well as facilitating much-needed research into these conditions. Through national rare disease plans, examples of good care and practice can be exchanged internationally to raise the quality of care and services available to those affected by rare conditions in the UK and internationally.

Rare Disease UK has been campaigning vigorously to ensure that this commitment is acted upon in the UK and we look forward to seeing the UK plan published later this year. We hope that by coming together on Rare Disease Day we can reinforce to the governments and health departments in each of the UK’s four home nations the need for a UK wide plan to make a real and significant difference to the lives of people living with rare diseases and their families.
Supporting families with rare disorders

Every time a child is diagnosed with a rare genetic disorder, there are parents left devastated, grandparents and extended family whose lives are turned upside down. I know. In 2005, I was one of those parents.

When it happens to you, you feel so utterly alone. Later, when I learned that my child is born with a genetic disorder in the UK each year it was astounding. I never imagined there would be so many.

Before my eldest son was diagnosed with hypothyroidism due to cystic fibrosis, I barely gave a second thought to what genes did and what happened when they didn’t do what they should do. Rare medical conditions happened in other people’s families, not mine. But as I found out, a genetic disorder can affect any family.

David Cameron’s son, Ivan, had a rare genetic disorder, Dravet syndrome; Gordon Brown’s son, Fraser, has cystic fibrosis; Katie Price’s son, Harvey, is affected by a number of disorders, including Prader-Willi syndrome; and Coleen Rooney’s sister, Rosie, sadly passed away this year from Rett syndrome.

It is a little-known fact that genetic disorders and their related symptoms are the main cause of death for more than half of the children who die under the age of 14 in the UK.

The truth is that rare genetic disorders aren’t actually that ‘rare’. Some genetic conditions affect fewer than 10 families in the UK but others affect the lives of thousands. When you add together everyone whose lives are touched by an estimated 6,000 genetic disorders, you are talking about hundreds and thousands of children, adults and families in the UK living each day with their ‘rare’ disorder.

Since the 1990s, Jeans for Genes Day has been raising money to support these families by asking employees and pupils to wear their jeans to work or school for one day in exchange for a small donation. The money raised is given as grants to genetic disorder-specific charities invested in developing educational resources for schools and, more recently, has been used to fund the formation of Genetic Disorders UK.

But Jeans for Genes Day is more than just a fundraising event. It also offers an opportunity to bring the subject of rare genetic disorders into the open.

Today, the media carry as many stories about genetic disorders as they do about breast cancer or Alzheimer’s but the stories are rarely identified as such. Lots of genetic disorders have unpronounceable names, such as rhabdomyolysis punctata, and so instead of highlighting the disorder, the article might be titled ‘The girl who sheds her skin every day’ or ‘The boy who can’t stop eating’.

The effect this has is to hide just how many children and adults affected by rare genetic disorders are facing challenges on a daily basis and this makes our fundraising a monumental hill to climb. In times of recession people like to give their limited donations to the causes they know and understand. Jeans for Genes Day competes in a world of major health charities, televised marathons and disaster appeals. It can be very difficult to make our voice rise above the parapet. It can be almost impossible to make people see the magnitude of our challenge. I am so pleased to support Rare Disease Day and the opportunity it gives to us to be heard.

So if you have read all of the above and are wondering what you could do now, why not sign up for the next Jeans for Genes Day and help change lives?

Oh, and if you happen to be a Premier League footballer wondering how to spend your next million, please give me a call.

Kerry Leeson-Beevers, Acting CEO

AFFECTED TWIN GIRLS, KATIE AND HANNAH

This is highlighted by the annual events ASUK organise such as the Family Conference now attracting worldwide interest and the ASUK sponsored cycle ride, which last year involved patients, family members and medical professionals cycling from Torbay hospital to the Queen Elizabeth Hospital, Birmingham. ASUK also hold a number of Marathon places and we are currently seeking runners for these places.

ASUK attributes its success to positive patient power, giving patients a voice and enabling patients to develop the services they need.
The rare condition that can turn out to be far more than a headache

When Gail Weingartner started getting head pains at age 25, she initially blamed jet lag resulting from her job as air crew. But when she developed greasy skin, exhaustion, menstrual and eye problems, she knew it was something more serious.

“It was only after eight years of consulting doctors, acupuncturists and hypnotherapists that I diagnosed myself with adrenal insufficiency,” says Gail, from Harlemere, Surrey.

What is AI?
Adrenal insufficiency (AI) is a relatively rare condition in which the adrenal glands fail to produce enough steroid hormones.

There are two types of AI. Firstly, Primary AI also known as Addison’s Disease, results from the degradation of the adrenal glands which means that they become incapable of producing enough hormones. In up to 80 per cent of cases the cause is usually due to the destruction of the adrenal gland by the body’s own immune system.

The more common secondary AI results from the adrenal glands being inadequately stimulated by the hypothalamus or the pituitary gland. There are numerous potential causes, such as damage to the pituitary gland following a tumour or surgery.

Despite treatment for her secondary AI, Gail still sometimes gets symptoms. “When my hormone levels are depleted it’s like having an almighty hangover, with a heavy head, exhaustion and nausea. Sleep doesn’t help,” says Gail, now working in photography.

Risk factors
Untreated or poorly managed AI can lead to a potentially fatal crisis. Symptoms include severe back and abdominal pain, diarrhoea and vomiting, high heart rate, low blood pressure, fever, shock and coma. However, AI can be hard to spot early on because symptoms such as exhaustion, nausea or dizziness can be confused with common ailments.

“A blood test can indicate AI but many doctors overlook it because it is such a rare condition,” says Pat McBride of The Pituitary Foundation, a charity which spreads awareness.

Treatment options
Once diagnosed, AI can be treated with hydrocortisone which replaces the natural adrenal hormone cortisol.

Taking regular tablets means most AI patients can live a relatively normal life. New treatment methods mean taking hydrocortisone only once a day instead of several times, but still dosage must be carefully managed.

“Even taking hydrocortisone tablets daily I used to get AI symptoms two or three days a week,” says Gail. “Then I got help to change the time and level of dose and now I get symptoms less often.”

Gail says: “If you think you may have AI push for a test.” McBride advises: “AI is a rare condition but it pays to be aware of it because early treatment saves lives.”

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New treatment could extend lives

A new treatment for adrenal insufficiency (AI) could extend patient’s lives, says endocrinologist Professor Paul Stewart, Dean of Medicine at Birmingham University and leader of a team studying the effect of the hormone cortisol on the body.

“The standard treatment for AI uses a drug called hydrocortisone to replace natural cortisol produced by the adrenal gland. It’s a treatment regime that has changed little since the 1950s, and it provides a normal life for up to a half of our patients — but for the rest it is less effective,” says Professor Stewart.

The problems arise because it is hard to replace the natural pattern of cortisol production — high in the morning dropping to very low at night — with drugs taken at intervals throughout the day.

“Too high a drug dosage results in Cushing’s Syndrome, in which fat cells increase and patients get a bigger midriff, so they are at higher risk of diabetes and cardiovascular disease,” says Professor Stewart.

“As a result, the life expectancy of patients on existing hydrocortisone therapy is five to ten years lower than normal.”

Hope for the future
However, a new drug has been developed that more closely mimics the natural pattern of cortisol secretion, avoiding the peaks that cause Cushing’s Syndrome and the troughs that characterise Addison’s disease (AI).

The new drug has recently been licensed for use in the UK but as yet only a handful of patients have been treated. Professor Stewart advises people with AI: “If your existing hydrocortisone therapy is not causing problems such as tiredness, lethargy or metabolic complications such as obesity or diabetes stay on your present regime. But if you are having problems, ask your endocrinologist about changing your drug therapy.”

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EVERY PATIENT DESERVES HOPE.

ViroPharma is proud to take part in Rare Disease Day.

At ViroPharma, we’re committed to helping people with rare and potentially life-threatening conditions, including hereditary angioedema and adrenal insufficiency. Our mission is simple: Providing hope to patients.

ViroPharma is a growing global specialty biopharmaceutical company dedicated to developing novel treatment solutions to meet patient and physician needs, in an effort to restore health and save lives.

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NEW DRUGS MAY BRING NEW DAWN FOR IPF PATIENTS

There are exciting new developments on the horizon for patients with idiopathic pulmonary fibrosis (IPF).

“We are beginning to see how pulmonary fibrosis works, which has resulted in several drug discoveries. At present, the five-year survival rate for people with pulmonary fibrosis is 25 per cent, but I think we could see a big difference in ten years time,” says Dr Gisli Jenkins, reader in pulmonary biology at the University of Nottingham.

“Forty years ago, the five-year survival rate for breast cancer was around 50 per cent, but today it is 85 per cent,” says Dr Jenkins. “I think we may be on that same path with IPF.”

IPF causes scarring and hardening (fibrosis) of the lung tissue. Currently incurable, about 4,000 to 5,000 people are diagnosed in the UK each year. Many major pharmaceutical companies are working on IPF drugs because an effective drug could also work for other fibrotic diseases, which affect 200 million patients worldwide.

Currently available but only on a named patient basis is a drug that can slow down the formation of lung scar tissue. Dr Jenkins says: “I see it as the first step.”

Other drugs currently in trials include one which has been shown to slow decline in lung function and the antibodies STX-100 and AB0023 targeting fibrotic cells and fibrotic molecules to help prevent fibrosis.

“The steady arrival of new drugs could mean huge changes in IPF treatment,” says Dr Jenkins.

Meanwhile, Dr Helen Parfrey, consultant chest physician at Addenbrookes Hospital is calling for increasing awareness of IPF.

“The symptoms of dry cough and breathlessness can be confused with other lung diseases, and by the time IPF is diagnosed there may be significant amount of scarring in the lungs,” says Dr Parfrey.

Existing treatments include high doses of the antioxidant N-acetylcysteine, available in health food shops. A European study has shown that it slows the development of IPF.

Meanwhile, Dr Parfrey advises: "Patients should stay active and try to avoid respiratory infections by leading a healthy lifestyle. They may be able to join regional patient groups if available, but should also look out for the new organisation Pulmonary Fibrosis UK, which will be launched later this year.

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At InterMune we are committed to patients with idiopathic pulmonary fibrosis

Supporting patients and the public

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References

INTERMUNE

Around 5,000 patients will die of IPF each year in the UK.

IPF is a progressive and fatal lung disease.

The cause of IPF is unknown.

Survival rates for IPF are worse than several types of cancer.

IPF is hard to diagnose because symptoms are similar to other lung diseases.

Speak to your healthcare professional if you are concerned that you may be suffering from IPF or lung disease.
Primary immunodeficiencies (PIDs) can be silent killers. PIDs are a group of around 200 rare conditions characterised by failure of the immune system. Causes vary but include gene abnormalities.

Consultant immunologist Dr David Edgar, Chair of the UK Primary Immunodeficiency Network says: “There are probably between 4,000 and 5,000 people in the UK with PIDs. Conditions range from the relatively mild such as selective immunoglobulin, a deficiency, which often produces no problems, to severe combined immunodeficiency (SCID).”

SCID results in severe infections. Babies with undiagnosed SCID are likely to die in their first year. Many patients are treated with antibiotics or immunoglobulin, but more serious conditions may require bone marrow transplants or gene therapy.

Challenges
PIDs are often undiagnosed for years. Common symptoms such as recurrent infections that respond poorly to antibiotics mimic less serious conditions.

Diane Hammond, 50, says: “My daughter Rachel had constant chest infections from age one. Between infections she was lethargic and her development was delayed. It mystified doctors until she was diagnosed with PIDs aged seven. Immunoglobulin treatment transformed her — it was as if she woke up.”

Now Rachel, 19, is preparing to take part in a Channel swim. Diane was later diagnosed with a similar form of PIDs at 45.

Screening for all
The good news is that there is now a campaign to screen all UK newborns for the most severe form of SCID. “This should reveal SCID earlier so parents can be offered bone marrow transplants. There is a 90 per cent chance of success if this is done early enough,” says Dr Edgar.

People who suspect PIDs should research the condition and keep a diary of symptoms to show doctors,” advises Susan Walsh. “Parents should trust their instincts and push for tests.”

People with PIDs should carry details of PIDs specialist centres and websites to prove to medical staff their need for prompt specialist treatment.

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How Paralympic swimmer Jack Bridge turned a rare disease into an opportunity

At 18 months old, Jack Bridge was diagnosed with haemophilia. Now, at 18, he is one of Britain’s Paralympic swimmers.

How I made it

“As a child I had bleeds into my left elbow, left knee and right ankle joints,” says Jack. “It was painful at the time but, on the plus side, it left me able to qualify as a Paralympian.”

Jack was put on the standard treatment for haemophilia: regular injections of clotting factors, which protect against potentially lethal brain bleeds and painful bleeding into joints. But his body manufactured antibodies (inhibitors) which neutralised the activity of the clotting factors.

“As a result, I had a few difficult years — though I don’t really remember them — until I was put on a desensitisation programme around age five to six. That succeeded in clearing the inhibitors, and I have been stable ever since,” says Jack.

Swimming as therapy

Despite his health problems, Jack started swimming aged four and now trains with Preston Swimming Club. He has now been competing for eight years, and made the Paralympics GB swimming team for the first time in 2012, where he competed in the 200m individual medley, 100m backstroke, 100m breaststroke and 4x100m medley relay.

During the Games, he set a personal best time in the 100m backstroke at 1:09.38 and reduced his personal best time in the 100m breaststroke by more than two seconds, finishing in 1:10.01 and qualifying for his first Paralympic final in which he finished in fourth place.

Jack also competed in the last event in the pool at the Paralympic Games, as part of the men’s 4x100m medley relay where he helped the team secure a British record time of 4:20.54 and finish fifth.

“It was painful at the time but, on the plus side, it left me able to qualify as a Paralympian”

Jack Bridge
Paralympic swimmer, living with haemophilia

Jack’s TYPICAL DAY

3.45am Get up, have a breakfast of bran cereal and self-administer an injection for my haemophilia
4.30am Into the water for training
6.30am Gym training
7.30am Home and eat another portion of breakfast before college
9am Go to college to study A level Law and Business Studies
3.30pm Home and eat more food
5pm Back to the pool for more training

“I don’t get bleeds now,” says Jack. “I have a theory that if I keep fit and keep my muscles and joints working, the muscles are strong enough to resist the bleeds. There is no scientific proof, but my coach believes it too, and it seems to work for me.”

Jack takes a prophylactic dose of 2,000 units of clotting factor 8 four times a week, which he injects himself. He fits his doses around a rigorous training schedule and life as an A level student.

“It’s a tough regime, but Jack says he does not let his haemophilia get in the way of his ambitions.”

“My consultant thinks my inhibitors are dormant, and my haemophilia is not a problem at all at present, but I know that my inhibitors could come back at any time, and I think developing my fitness combats bleeds and pain.”

“My next target is the World Championships in Canada this August, then the Glasgow Commonwealth Games in 2014, and ultimately the 2016 Paralympics in Rio.”

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AMBITIOnS
Jack is training hard with his sights set on competing in the 2016 Paralympic Games in Rio

PHOTO: MARK WAUGH

How Paralympic swimmer Jack Bridge turned a rare disease into an opportunity

The main treatment for haemophilia is preventive of on-demand factor concentrate therapy, which many patients self-administer at home. Gene therapy is a possibility for the future.

There is no cure, but modern treatments mean patients can lead normal lives.

Read more on the web:
www.haemophilia.org.uk

Haemophilia is an inherited bleeding disorder where clotting factors required for blood to clot normally are lacking.

People with haemophilia A lack clotting factor VIII and represent 90 per cent of all those affected by haemophilia.

People with haemophilia B lack clotting factor IX.

Around 6,000 people in the UK have haemophilia, almost all male. Symptoms can take years to appear and many people don’t know they have it.

Symptoms include bleeding into muscles and joints, especially the knees, elbows, and ankles; prolonged bleeding after a cut, tooth removal, surgery, or an accident; big bruises and internal bleeding into vital organs, most commonly after a serious trauma.

Bleeding into joints can lead to disabling arthritis, and bleeding in the brain can lead to serious complications.

Symptoms include difficulty walking, frequent vomiting, behaviour changes, sleepiness, neck pain or stiffness, double vision and convulsions or seizures.

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Paralympian Jack Bridge is an inspiration to us all, and we are proud to announce that this year Novo Nordisk will be working with Jack on a number of swimming programmes, designed to further understanding of haemophilia in the broader society and promote the importance of safe exercise for people with haemophilia.

Following on from the successful Buddy Awards, launched with the Haemophilia Society to celebrate the vital support provided by family, friends, teachers and healthcare professionals to children with haemophilia and bleeding disorders, our swimming programmes are another innovative way of putting the spotlight on haemophilia and supporting the community and their families.

At Novo Nordisk, we are working towards a future where all people living with haemophilia have the opportunity to lead the lives they deserve.

Our programme, Changing Possibilities in Haemophilia®, represents our commitment to the whole of the haemophilia and bleeding disorder community. We recognise that improving the lives of people with haemophilia and bleeding disorders is about much more than just medicines.

So, as well as having almost a century of innovation and product development behind us, we take a holistic approach; listening to patients, their families and healthcare professionals about the challenges they face so we can meet their varied and complex needs.

We are proud to support Rare Disease Day to help drive awareness and understanding of haemophilia and bleeding disorders.

This year’s theme of ‘Rare Disorders without Borders’ is particularly relevant for the haemophilia community as people living in developing countries make up 75% of the global patient population.

The Novo Nordisk Haemophilia Foundation, a not-for-profit organisation, works to improve access to care for people in developing countries, where many patients remain undiagnosed or do not receive adequate treatment.

Novo Nordisk is looking forward to a future of working together, here in the UK and across the globe, to Change the Possibilities for everyone living with haemophilia.

Further information is available at [www.novonordisk.co.uk](http://www.novonordisk.co.uk)
After decades of research, largely funded by the tireless efforts of those affected and their families through charities like the Muscular Dystrophy Campaign, pioneering genetic treatments for Duchenne muscular dystrophy are now in clinical trials. The potential to change the fate of hundreds of young men and boys living with this very severe, life-limiting form of muscular dystrophy, may be within our reach.

However, as we have seen with other rare and complex diseases, successfully developing an effective treatment is far from the end of the battle because all rare diseases are not the same. Some rare diseases affect only around 1,000 people or less and treatments are termed ‘ultra orphan’, which is in contrast to ‘orphan’ drugs which may treat up to 31,000 patients in the UK. This difference in magnitude becomes evident in cost effectiveness assessments, owing to it being virtually impossible to generate sufficient data, even on a worldwide basis, from very small patient populations. In Scotland, we have seen how the Scottish Medicines Consortium (SMC) has a tendency not to approve ultra orphan drugs. This has had a devastating impact for some patients and their families.

**Low-volume, high-cost drugs**

Later this year, the new National Institute for Health and Clinical Excellence (NICE) takes over the cost effectiveness assessment for low-volume, high-cost drugs. A concern among many is that NICE uses a similar assessment for both orphan and ultra orphan drugs, which the evidence suggests, could prevent patient access to potentially lifesaving treatments simply on the basis of rarity. It is essential that NICE’s assessment process fully recognises ultra orphan drug treatments but also balances this with affordability to the wider NHS, as simple economics tells us that there is a tradeoff between low-volume and high-cost. This week, the All Party Parliamentary Group for Muscular Dystrophy, of which I am Chair, launched a new inquiry into the provision of low-volume, high-cost drugs in England. Over the coming months we will be hearing evidence from NHS commissioners, the pharmaceutical industry, patients and their families, the Muscular Dystrophy Campaign and NICE itself. Together we will examine how we create a sound infrastructure around the assessment and delivery of orphan and ultra orphan drugs.

As genetic medicine progresses we will see an increasing number of successful therapies available to treat very small patient populations. Many families have gone through the agony of diagnosis of a severe life-limiting rare condition. We must ensure this is not doubled by an effective, life-changing, market-ready treatment being denied.

Dave Anderson  
Chair of the All Party Parliamentary Group for Muscular Dystrophy

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**NEWS**

**TREATMENTS FOR ULTRA RARE CONDITIONS MUST REACH THOSE THEY WERE DEVELOPED FOR**

There are over 60 different forms of muscular dystrophy and many more related muscle-wasting conditions. All are very rare, and some so much so that just a handful of people are affected in the UK. What they all have in common is a devastating effect on the body’s muscles, increasingly compromising mobility, and often breathing and heart function. They all lead to progressive disability and in all too many cases, immeasurable stress and misery. This is something I myself understand, having lost my brother, sister, a nephew and a niece to myotonic dystrophy.

After decades of research, largely funded by the tireless efforts of those affected and their families through charities like the Muscular Dystrophy Campaign, pioneering genetic treatments for Duchenne muscular dystrophy are now in clinical trials. The potential to change the fate of hundreds of young men and boys living with this very severe, life-limiting form of muscular dystrophy, may be within our reach.

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Chair of the All Party Parliamentary Group for Muscular Dystrophy

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**NHS investigates ultra rare conditions**

The results of Muscular Dystrophy Campaign’s 2013 survey of people in the UK affected by severe muscle wasting conditions made for difficult reading. Some children born with Duchenne muscular dystrophy can live into their 30s, even early 40s, and yet some parents are still being told their child will not live past their teens. Many people with facioscapulohumeral (FSH) muscular dystrophy are being told nothing can be done for them, yet physiotherapy and hydrotherapy are known to alleviate symptoms and improve mobility. Others are not receiving heart and
How patient power can transform access to treatment

Christine Lavery, chief executive of The Mucopolysaccharide Society, for people affected by Mucopolysaccharide disease (MPS), is a firm advocate of patient power.

"Over 95 per cent of people with MPS are members of the society, which offers an advocacy service to individual patients," says Lavery.

Its work benefits doctors and researchers too. "We inform patients about research and clinical trials and provide financial support for patients who otherwise could not take part," says Lavery.

The 2010 Equity and Excellence White paper included the phrase "Nothing about me without me". However, there have been concerns that the patient voice hasn’t yet been hard-wired into the new NHS structures. The MPS Society is part of a broader coalition of LSD Patient Associations representing all 50 lysosomal storage diseases which have been lobbying Government.

Patient involvement

Lavery continued: "In partnership with our clinical and commissioning colleagues we have contributed significantly to the shaping of the current service for the very rare, ultra orphan diseases. During this period of change within the NHS we feel it as important as ever that the patient perspective has strong representation. The patient must be central to all decisions not only into existing and new services but into the evaluation of new technologies."

The MPS Society was instrumental following the last major NHS re-organisation. "We won the battle in 2005, so patients with LSDs in England can access the same level of treatment from the Department of Health code lottery," says Lavery.

"Recently, we also won a commitment from the Department of Health that enzyme replacement therapy, used to treat many LSDs, will continue to be offered to patients England wide under the new NHS commissioning board."

Robert Meadowcroft

Chief Executive, Muscular Dystrophy Campaign

SPEAKING OUT

Christine Lavery, chief executive of The Mucopolysaccharide Society, for people affected by Mucopolysaccharide disease (MPS), is a firm advocate of patient power. "Over 95 per cent of people with MPS are members of the society, which offers an advocacy service to individual patients," says Lavery.

Its work benefits doctors and researchers too. "We inform patients about research and clinical trials and provide financial support for patients who otherwise could not take part," says Lavery.

The 2010 Equity and Excellence White paper included the phrase "Nothing about me without me". However, there have been concerns that the patient voice hasn’t yet been hard-wired into the new NHS structures. The MPS Society is part of a broader coalition of LSD Patient Associations representing all 50 lysosomal storage diseases which have been lobbying Government.

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How a drug trial gave TSC patient his life back

At 18, Jack Royall had to be accompanied 24/7 because he suffered frequent seizures as a result of tuberous sclerosis complex (TSC). Now, at 20, he goes out alone, has recently moved into his own flat and does voluntary work in an office and hospital.

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Treatment with epilepsy drugs did not stop Jack’s seizures, which could be life-threatening, and he had to be accompanied at all times. “At five he was very intelligent but the tumours and brain surgery left him with a mind like an eight year old, with behavioural problems and symptoms similar to those of people with Asperger’s or on the autism spectrum,” says Ally. He was often in hospital. He had repeated brain surgery for non-cancerous tumours and had more tumours in his eye, kidneys and liver. At 17 he faced the prospect of a kidney transplant.

Then he was put on the drug trial. “He has not had a seizure in the two years since,” says Ally. Scans showed his tumours were shrinking and in three months the facial rash that is a common symptom of TSC disappeared. The threat of a kidney transplant has receded. “Now he has the mind of a 20 year old and is getting his memory back,” says Ally.

Gaining independence
Now in an assisted living flat, he makes his own breakfast, shaves himself and goes out alone. “I had a hard time adjusting to that because I had always lived in fear of him having a seizure and dying if he was alone,” says Ally. “The first time he went out on the bus, I secretly followed him in my car.” Jack now spends much of his time running a Skype networking group for young people with TSC and hopes to go to the USA to meet some of his online friends for his 21st birthday.

Positive changes
Jack says: “I had learning difficulties and many disabilities, but now I have a condition controlled by medication. I’m seeing improvements in my condition all the time.” Ally says: “I have got my life back, too.” She advises: “Families affected by TSC should educate themselves about it — the Tuberous Sclerosis association is a good source of advice support and information. Fight for the best treatment and never give up hope.”

■ How can I get it?
TSC clinics can apply for individual funding requests, but only about 10 per cent of TSC patients attend a specialist clinic. We need a national network. Meanwhile we are lobbying for the drug to be an option for all suitable TSC patients.

■ What is TSC?
TSC is a genetic condition that causes non-malignant tumours to grow in the brain, heart, eyes, skin, kidneys, or lungs. It can lead to epilepsy, learning disabilities, autism and kidney problems.

■ How many people are affected?
Two babies are born with TSC daily and there are about 8,000 people with it in the UK. People with milder symptoms may go undiagnosed.

■ Are there new treatments?
An existing drug has been found to turn off the ‘switch’ which causes the growths, and shows few side effects. In trials it was effective at shrinking the potentially fatal brain growths called SEGA tumours, which occur in 10 per cent of people with TSC, and it is now licensed in the UK to treat over-threes with TSC. SEGA tumours. Previously the only treatment has been potentially risky brain surgery.

Trials also show it helped shrink kidney tumours — important because 80 per cent of people with TSC have kidney problems — and reduced the skin problems caused by TSC in over 90 per cent of cases. It is also the first treatment to arrest the lung growths caused by TSC. Ultimately it could help everyone with TSC.

■ How does voluntary work in an office and hospital.

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When science and passion connect, innovation happens.

Novartis is proud to be working with the Tuberous Sclerosis Complex (TSC) community and we are committed to working to provide patients, families and caregivers with treatment hope for the future.
Lysosomal Acid Lipase (LAL) Deficiency: what is it?

LAL Deficiency (also known as Wolman disease and Cholesterol Ester Storage Disease) is a rare, autosomal recessive lysosomal storage disorder that is caused by a marked decrease of the LAL enzyme.

- The reduction of LAL activity in patients with LAL Deficiency leads to the accumulation of lipids in the body. This leads to a range of abnormalities.

- The early onset phenotype (Wolman) is very rare (1:500,000 births) and presents shortly after birth with predominant liver involvement. This form of the disease is rapidly fatal with death usually within the first year of life.

- The late onset phenotype (CESD) affects cases as young as two years old and has predominant liver involvement and high cholesterol. This is the more common form of LAL Deficiency with an estimated prevalence of 1:40,000–1:300,000.

- The liver and spleen are commonly affected with an elevation of liver enzymes and severe liver fibrosis that leads to cirrhosis.

- Cardiovascular involvement is characterised by high cholesterol with early onset vascular disease due to accumulation of lipid deposits in artery walls.

- The presentation of late onset LAL Deficiency is highly variable with some patients going undiagnosed until complications manifest in late adulthood, while others can present with liver dysfunction in early childhood.

- As with other rare conditions, the number of patients diagnosed with LAL Deficiency tends to be lower than the estimated frequency as many are frequently misdiagnosed as Nonalcoholic Fatty Liver (NAFLD) or hyperlipidemia.

- Presentation with gallstones is common, but often the cause of gallstones is not investigated which leads to a missed opportunity to diagnose this rare disorder.

- LAL Deficiency can be diagnosed with a simple blood test.

**Patients groups do more than provide information.**

The Gaucher Association, which represents patients with the lysosomal storage disease Gaucher disease, has provided grants to get research started.

“We provided money for the viral vectors that carry gene therapy treatment into cells. That research showed it was worth pursuing, and led to a further grant from the Medical Research Council,” says Tanya Collin-Histed, chief executive of the Gaucher Association.

The charity is also looking at ways to speed diagnosis, which can be slow. For instance, Henry Jameson, 11, had extensive tests for a swollen spleen, a Gaucher symptom, and was diagnosed with possible leukaemia and then glandular fever before a test revealed Gaucher.

“It was a long, fraught journey,” says mum Lucy, but now Henry has regular enzyme replacement therapy at home and lives a normal life. Henry is a UK champion junior sailor in his boat’s class and came fourth in the world championships in Italy.

“Gaucher does not stand in my way — I’m hoping to make the Olympic sailing team,” says Henry.

**Screening at birth**

Patient groups also mount campaigns. The Save Babies through Screening Foundation UK (SBUK) is lobbying to extend newborn screening in the UK to include more conditions, including Krabbe disease.

“In some US states Krabbe patients identified just after birth have been treated with umbilical stem cell transplants. If successful they go on to develop apparently normally,” says SBUK executive director Pat Roberts. Untreated babies born with Krabbe usually die within two years.

“The UK screens for only five rare disorders nationwide, compared to 28 in Germany and up to 60 in the USA,” says Roberts.

SBUK is also lobbying for improved registers of patients with rare disorders, as they are currently scattered and incomplete.

**NEWS**

Together, everyone at Synageva shares a passion and commitment to improve the lives of patients with rare diseases.

**GET THE SUPPORT AND INFORMATION YOU NEED**

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Linda Whitney
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New therapies hold out hope for Pompe patients

New therapies could help people with Pompe disease within ten years.

Allan Muir, development director of the Association for Glycogen Storage Diseases-UK (AGSD-UK), which supports families living with Pompe, says: “One gene therapy for Pompe disease has already been effective in a few patients. Studies starting soon may produce viable therapies in five to ten years.”

Pompe is a genetic disorder that leads to progressive muscle weakness affecting breathing, mobility and the heart. Untreated, severely affected babies die within a year, but people with less severe cases can live much longer.

There are also new developments in the enzyme replacement therapy (ERT) that is commonly used to treat Pompe.

Recent research has shown that a formulation combining enzymes with a pharmacological chaperone resulted in greater enzyme uptake into muscle, a key tissue of disease, holding out hope for a combination therapy that could further reduce symptoms.

“Before ERT, parents were told to go home and enjoy what little time they had with their child — usually only a few months,” says Muir.

“ERT means we now we have over 20 children who are mostly in mainstream schools. Some have problems but all are bright children with a future.”

ERT is available to all patients in England but in Scotland some patients have been denied treatment. AGSD-UK is calling for ERT to be made nationally available in Scotland. Unfamiliarity with Pompe means babies are typically not diagnosed until four to six months, by which time they are weak and vulnerable to respiratory infection. Adult diagnosis can take ten years.

“Newborn screening for Pompe disease is necessary,” says Muir.

“There are two children in the UK who were treated in their first three weeks of life and they are almost indistinguishable from unaffected children.”

Screening could reveal the ‘missing’ Pompe patients. About 1 in 120,000 and is 1 in 250,000 people, are being actively researched.

“Niemann-Pick diseases are genetically caused lysosomal storage disorders (LSDs), which as a group are common,” says Mathieson. Research into the complex LSDs may result in a therapy that works for many diseases, both rare and common,” says Mathieson.

The group is also funding a national history study of the disease, with results due next year.

New Niemann-Pick test could save years of agony

A test that takes two days to diagnose Niemann-Pick type C (NPC) is coming to the UK soon.

“From early symptoms, it can take years to obtain a diagnosis,” says Toni Mathieson, executive director of the Niemann-Pick Disease Group (UK).

Helen, mother of Hollie who was diagnosed aged two, says: “The diagnosis took ten weeks — the worst ten weeks of our lives. We would have given anything for this process to have been speeded up.”

Mathieson says: “There is no routine UK screening for NPC, and in some cases, children die before diagnosis.”

NPC affects one in 250,000 and is characterised by a build-up of toxic materials in the body's cells, which can lead to intellectual decline, loss of motor skills, seizures and dementia.

The Niemann-Pick Disease Group (UK) supports those affected by all types of the disease and contributes to the research effort. Treatments including enzyme replacement therapy for Type B, which affects one in 250,000 people, are being actively researched.

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Amicus Therapeutics is proud to support Rare Disease Day and continue its ongoing development of novel treatments for the rare disease community.

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More care co-ordinators for muscular dystrophy patients could improve care and save money

Care co-ordinators can transform the lives of people with muscular dystrophy.

THE NEED FOR CHANGE

Muscular dystrophy is a muscle wasting condition affecting many body parts, so the UK’s 70,000 patients face appointments with many different specialists, repeating their story many times and risking poor communication between specialists.

Often no-one is overseeing their condition as a whole, reducing continuity of care. This is where care co-ordinators come in. They are trained professionals who ensure a care plan is carried out smoothly.

Finding support

Rebecca Kinge’s son Ethan, who has Duchenne muscular dystrophy (DMD) which causes progressive muscle weakness says: “We can call our care co-ordinator whenever we need her. She knows Ethan’s condition, what he needs and when. She has built a good relationship with him so as he grows up, he knows he can rely on her as I do.

“She has also been a shoulder to cry on when things aren’t going well. Life would be far more difficult without her. She has talked to Ethan’s school, helped get specialist equipment, and supported the family through appointments.”

Pammy Malhotra, a neuromuscular care pathway co-ordinator and patient advocate for the Muscular Dystrophy Campaign, visits patients, attends clinics with them, provides advice and signposts them to specialist centres.

Malhotra says: “It is very satisfying to see how beneficial my involvement is. Many patients or families say that managing the condition on their own would be impossible.”

She adds: “Many people are lost in the system. A recent audit of unplanned hospital admissions showed a large proportion of patients were unknown to service providers, and are not accessing a care co-ordinator.”

Increasing care co-ordinators

The Muscular Dystrophy Campaign is calling for the number of neuromuscular care co-ordinators to be increased to 70. The UK now has only 31 with full NHS funding.

Chief executive of the Muscular Dystrophy Campaign, Robert Meadowcroft, says: “Neuromuscular care advisors are a lifeline and they can save the NHS money by helping reduce emergency hospital admissions.

“Unfortunately, around half of all patients in the UK still do not have access to them. We want to see more additional care advisors in post as soon as possible.”

FARHANA ALI

Rare Disease UK

OxThera

Hyperoxaluria is one leading risk factor for recurrent kidney stone disease. In patients with a primary form (endogenous oxalate overproduction), severe hyperoxaluria leads to frequent kidney stones and may eventually lead to End Stage Renal Disease (ESRD). Oxalobacter formigenes, an anaerobic microbe normally colonising the intestinal tract, uses oxalate as it sole source of energy. Oral administration of Oxalobacter formigenes may be a potential treatment option for patients with Primary Hyperoxaluria (PH).

PH is a rare autosomal recessive inborn error of the metabolism, with an incidence rate of 0.1-0.2 per million. PH is characterized by severe hyperoxaluria that is present from birth. High levels of oxalate in plasma and urine damage the renal cells both as free oxalate and as calciumoxalate crystals. Patients experience frequent kidney stones, calcification of the kidneys and impaired renal function, which eventually may lead to systemic deposition of calciumoxalate crystals and premature death. There is currently no approved pharmaceutical therapy for PH. The only curative therapy in patients who have reached ESRD is a combined kidney and liver transplantation.

OxThera AB is developing Oxabact®, (capsules containing Oxalobacter formigenes) as a chronic treatment for PH. Administration of Oxalobacter formigenes is believed to increase degradation of oxalate in the gastrointestinal tract and to promote enteric elimination of oxalate, thereby reducing the oxalate burden on the kidneys.

Phase II studies evaluating an initial drug product showed a significant and clinically relevant reduction in urinary oxalate excretion in subjects with PH. There are also case reports supporting clinical benefit in patients with ESRD. During 2013, OxThera will conduct further studies to confirm the efficacy of Oxalobacter formigenes administration.

In order to mark Rare Disease Day 2013, Rare Disease UK (RDUK) has launched a number of reports, including rare disease care co-ordination.

This report shows that poorly co-ordinated care is a major issue for the majority of patients affected by rare diseases.

In a nationwide survey by RDUK, some of the problems experienced by respondents included: professionals only looking at specific elements of the condition; patients or families having to repeatedly tell their story; feelings of being lost in the healthcare system; patients and families not knowing who to go to with queries.

Care co-ordinators can help to alleviate these issues, yet only 13 per cent of respondents with a rare disease had a designated care co-ordinator.

There is a strong case for investment in care co-ordinator posts, not only because of the benefit to patients and families but evidence shows that care coordinators also represent good value for money for service providers.

Read more on the web: www.raredisease.org.uk
The diet that can help you control a rare genetic condition

**Question:** Why is it essential that babies are screened for PKU at birth?
**Answer:** Because early detection means this genetic condition can be controlled by a special diet.

For some babies a special diet can be a lifesaver. “Parents of babies who test positive for phenylketonuria (PKU) are initially shocked and fearful,” says Paula Hallam, dietitian advisor to the National Society for PKU (NSPKU). “Fortunately a special low-protein diet means the child can live a completely normal life.”

**What is PKU?**
PKU is a genetic condition in which the body is unable to break down phenylalanine, one of the building blocks of protein. Untreated, phenylalanine build-up leads to brain damage, learning disabilities, behavioural problems and epilepsy. If untreated the damage is irreversible. Since 1969 all UK-born babies have been tested for PKU.

The treatment is a restricted protein diet, so foods such as meat, fish, and dairy products are banned. Wheat and selected vegetables can only be eaten in measured quantities.

“The diet must be carefully balanced — too little protein will affect normal growth, but too much will raise phenylalanine levels,” says Hallam.

**A PKU DIET**

**Breakfast:** 15 grams of ordinary cornflakes and low-protein milk, a slice of low-protein toast with spread and honey.

**Lunch:** Low-protein pasta, couscous or rice with roasted vegetables or baked potato and coleslaw.

**Dinner:** Low-protein pasta and tomato sauce, or roasted butternut squash and vegetables with vegan cheese.

**Fruit and protein substitute drinks with each meal.**

**Read more on the web:**
www.nspku.org

**DISCIPLINE:** Hannah Bridges, 13, has PKU and needs to stick to a strict diet. It means taking her own food to parties and knowing exactly what is in the food she is eating. She manages it well and lives a normal life.”

For over 20 years Vitaflo International Ltd has been at the forefront of developing a comprehensive range of innovative specialised clinical nutritional products for a wide range of rare metabolic conditions including Phenylketonuria (PKU) and Tyrosinaemia.

Working in partnership with health care professionals and families our aim has been to create products that help to deliver personalised nutritional therapy by combining the best of cutting edge research with the lifestyle demands of modern living.

Our products are specifically designed to provide the simple solution with the most up-to-date nutritional profile and the most acceptable consumer friendly packaging.

**LINDA WHITNEY**
info.uk@mediaplanet.com

**It’s the difference we make...**

...that makes us different
How Cyril and Janet are fighting myelofibrosis

“People with myelofibrosis must be positive and fight it all the way,” says Cyril Onyejekwe, 56, who was diagnosed with myelofibrosis in 2006.

It causes scarring and fibrous tissue build-up inside the bone marrow, which then cannot produce blood cells effectively.

Myeloproliferative disorders such as myelofibrosis affect one person in 100,000. A fifth have no apparent symptoms but others suffer excessive tiredness, weakness or shortness of breath, enlarged liver and spleen, bone pain, night sweats, infections, weight loss and heavy periods.

Cyril says: “For me it means constant tiredness, aching muscles and joints, and bone pains. I struggle to talk because I’m so short of breath and even going upstairs is difficult.” Cyril’s spleen was removed because it was so swollen that it affected his breathing.

Engineer Cyril, from Mitcham, Surrey, works from home mentoring apprentices and working on industrial relations.

“I’m on maintenance treatment. The only cure is a bone marrow transplant but for me the odds against a match are 100,000 to one as against one in four for white caucasian people. There are too few people of ethnic origin — I’m from Nigeria — on the UK transplant register,” he says.

Meanwhile, he tries to stay positive. “My wife Janet and I have six children and grandchildren, so I have lots to fight for. I treat every day as a bonus.”

Janet says: “When we found out it was a major shock. We’d never heard of myelofibrosis. Cyril changed from an outgoing, fun loving person to a man who is short of breath and hurting all over. He’s often shattered by 8pm.”

Janet knew it would be difficult to find a bone marrow match for Cyril because she worked for the African Caribbean Leukaemia Trust (ACLT), a charity that campaigns for more people of ethnic origin to register as bone marrow, blood and organ donors.

Through ACLT Janet found out about a new treatment for myelofibrosis and Cyril is now in transition to the drug. “It causes fewer side effects than the chemotherapy drugs he was on before,” says Janet.

She advises affected families: “Find out as much as you can about the condition, join support groups such as MPD Voice and ask consultants about the latest treatments.”

LINDA WHITNEY
info.uk@mediaplanet.com

When science and passion connect, innovation happens.

Novartis is investing in research to help to bring forward treatment solutions for myelofibrosis patients. We are proud to be working with the myelofibrosis community to help to provide patients, families and caregivers with treatment hope for the future.

www.novartis.co.uk

DOP: 12th February 2013 Job Bag No. JAK13-C014
CAMPAIGNING FOR EQUAL TREATMENT TO BE AVAILABLE TO ALL

Huge strides have been made over the last decade in the management of hereditary angioedema (HAE).

“With the right treatment, patients can achieve good control of their symptoms and enjoy a good quality of life,” says Ann Price, patient representative at the charity HAE UK. But getting the right care can be hard.

“HAE patients still commonly face delay in diagnosis and variable access to appropriate medications,” says Price.

HAE is a genetic condition affecting one in 50,000 people in the UK. Symptoms include recurrent swelling (oedema) in areas such as the hands or feet, or more seriously, the abdomen, where it causes severe pain, diarrhoea and vomiting. Swelling in the airway can result in death.

Attacks can be halted or minimised by prompt injections of C1 Inhibitor, but patients do not always get them in time.

Problems with diagnosis

“Lack of recognition of HAE means some patients wait hours in A&E while the condition worsens,” says Price. Diagnosis can take years. “Some people have had exploratory abdominal surgery and appendectomies in a bid to identify the cause of symptoms,” says Price.

MSUD: when a cold can kill

For people with Maple Syrup Urine Disease (MSUD) a cold is an emergency.

MSUD patients are unable to control levels of the amino acid leucine. High leucine levels cause irreversible neurological damage leading to paralysis, or death.

Symptoms include urine that smells of maple syrup, floppy muscle tone and fluid on the brain. Babies born with MSUD must be treated fast, so MSUD is included in the UK’s pilot expanded newborn screening programme.

“A diet very low in natural protein keeps the child well,” says Farzana Khan, specialist metabolic dietitian at Bradford NHS Foundation Trust.

Meat, dairy, fish, pulses, wheat and some kinds of vegetables are forbidden. Fruit, low-protein vegetables, pure fats and sugars can be combined with special low protein products and protein substitute gels or drinks.

“It is a balancing act, based on the child’s growth, health and results of regular blood tests,” says Khan.

Common problems like colds, infections or vomiting mean rising leucine. “Children must take emergency glucose drinks every two to three hours, day and night,” says Khan.

The diet must continue on special occasions and holidays. Dietitians provide frequent checks, educate families, provide culinary training, and visit schools.

“The risk is always there but motivated families do well,” says Khan.
The Society for Mucopolysaccharide Diseases (the MPS Society) is the only UK registered charity providing support to individuals and families affected by MPS and related diseases including Fabry. These are a group of rare genetic conditions which cause physical and in some cases mental disability. Progressive in nature, there is no cure at present although there is treatment for some of the diseases.

The MPS Society works collaboratively with a number of other patient groups within the field of lysosomal storage diseases. We also work in partnership with other MPS groups across the world creating an international MPS network of patient societies. Whilst our primary focus is on providing a support and advocacy service to all UK MPS and Fabry families, we continue to encourage and facilitate contact with our colleagues within the global international network.

On 15th May 2013 we will be celebrating International MPS Awareness Day, an annual event designed to raise the profile of MPS and related diseases.

For further information please visit www.mpssociety.co.uk

Registered charity 1143472. Charity registered in Scotland SCO41012.