

'We knew James had problems, we just didn't know the name for them – and nor did anyone else for 13 years'

It is estimated that 175,000 people living in Wales have a rare disease. As Wales marks Rare Disease Day this week, health editor **Madeleine Brindley** spoke to one mother about her 13-year fight to get her son diagnosed

WHEN Chris and Phil Humphreys met their new three-week-old foster son, they were told that his club feet were the only health issues he faced.

But after his first night with them, the couple knew James' problems went beyond the necessity of having his plaster casts changed every week.

It would be 13 years before James was finally diagnosed with a rare disease that few clinicians in Wales had even heard of.

Now 28, James is facing blindness – he will receive a guide dog called Wally this week – and he has been told that he, like the handful of other people with Laurence Moon Bardet Biedl (LMBB) syndrome in Wales, will not be allowed to attend a new half-day clinic once a year held in either London or Birmingham.

His mother Chris, through her work as national co-ordinator of the Laurence Moon Bardet Biedl Society, helped to set up the clinics but the NHS in Wales has said that patients will not be able to attend the clinics.

James has not had any ongoing medical care from a clinician with expertise in LMBB, which is why he was referred from paediatric health services.

Chris, who lives in Rogerstone, near Newport, said: "It took 13 years to get a diagnosis because no-one had heard of it in Wales, these were years when he could have had so much extra help.

"We can't look back and say, 'What if?' – we have to look at what we're going to do in the future.

"I've been involved in all the planning stages for this new clinic and now I've been told that my son isn't going to be allowed to go.

"People with rare diseases need to be able to access clinics that have the clinical experts who know how to correctly diagnose and can best manage their condition.

"For many rare conditions there are no designated clinics available.

"But for those conditions where these clinics exist and are available, people like my son James should be able to and allowed to access them, regardless of where they live in the UK.

Laurence Moon Bardet Biedl syndrome is a group of features which that occur together to characterise a medical disorder. The pattern of characteristics was first described in 1865 by two ophthalmologists – Laurence and

Moon. In the 1920s Bardet and Biedl independently added to the original description of the syndrome.

It is a rare inherited disorder which affects about one in 100,000 babies born – it is thought that there are about 25 to 30 people with LMBB syndrome in Wales.

The syndrome causes visual impairment, which will ultimately lead to blindness; obesity; developmental delays and problems with speech and co-ordination and kidney abnormalities.

Some babies with the syndrome are also born with extra fingers or toes, or with partially fused digits. High definition ultrasound scans mean that some of the symptoms, such as kidney problems or extra digits, can be picked up while babies are still in the womb, which can in turn lead to an earlier diagnosis.

But when James was a child his symptoms were too often put down to – or even dismissed as – the unfounded concerns of a worried, older mother.

The first sign that James' problems extended beyond his club feet were his difficulties year after year of his life he did not sleep for more than an hour-and-a-half a night, spending the rest of the time screaming. James didn't walk until he was two-and-a-half and he showed some behaviours, which would today be classed as autistic.

After suffering difficulties in school, a psychiatrist said she thought James had a chromosomal abnormality, but no-one knew what.

As he grew older James developed bruises along the tops of his arms as he repeatedly bumped into door handles; he was tired all the time and would develop very high temperatures – Chris says it was possible to fry an egg on his legs – accompanied by rapid massive weight gain over the space of a weekend.

Chris, now 62, who has five children and who used to run a residential home for teenagers, said: "He was just an enigma. A lot of the professionals in the early days thought that I was a mature mum who couldn't cope."

At the age of 12, the Humphreys were contacted by social services who told them James' natural sister may have had an unnamed hereditary condition.

"I got in touch with the paediatrician who wasn't certain and then we went down the route



ENIGMA: Chris Humphreys with her foster son James at home in Newport. His rare disease, Laurence Moon Bardet Biedl syndrome, was finally

diagnosed when he was a teenager – and then only thanks to Chris' persistence and determination

of different syndromes that he could have. He had bloods taken, then the bloods were lost," Chris said.

"I look it upon myself to get in touch with different support groups and I was given a helpline to ring and that was the start – everything James had begun to fit with LMBB.

"The consultant was also thinking along this line. I went to the LMBB society's conference in Coventry and by the time I saw the geneticist I knew more about it than she did."

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to mark Rare Disease Day on Wednesday, added: "After the euphoria of diagnosis came the depression of what was ahead.

"I should have felt pleased when told that James would now have the crème-de-la-crème of education from the visually impaired education service, but all I, and the rest of our family, could feel was desolation that we needed such a service.

"I have to admit Gwend did and still does have the best service in the country, especially when I listen to parents in other parts of the UK.

"Chris, who will speak at an event at the Senead, in Cardiff,

"The thing that I and 99% of

other families who have children diagnosed with LMBB can't cope with is the fact that they lose their sight, because we can't comprehend it ourselves.

"For everything else in this syndrome some form of medical intervention will help – years ago kidney problems could result in death. But with the eyes, once the back of the eye has deteriorated there is no replacing it.

"James is virtually blind but having the guide dog will make such a difference to his life."

"Yes he has his problems – learning difficulties, orthopaedic,

neurological spasticity, obsessive compulsive disorder, is on the autistic spectrum, registered as blind and one or two others – but he is a young man with a wonderful very sense of humour, who has raised several thousand pounds for his society, achieving two tandem parachute jumps, and quad biking with a lead bike, followed by back-up bike just in case.

"On one of these very wet occasions, I said to a friend: 'How is he going to manage with the rain splattering the mud in his face?' Her reply was: 'Chris, he's blind, and with that

he disappeared over Eglwysilan mountain with his head to one side, listening to the sound of the lead bike."

More recently, out in Northern Ireland, James was taken out on a speed boat. To the horror of his father and friend who were sitting – hanging on – in the back, James was allowed to take full control at 40mph.

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"James is a remarkable young man who will do things that his brothers

"If anyone had said to me 28 years ago that my life would have taken on what it has done, I would never have believed them.

"This is something that I am passionate about and there's nothing I

wouldn't do to help James or anyone else in a similar situation."

A Welsh Assembly Government spokeswoman said about access to the specialist clinic: "The Welsh Assembly Government provides funding and support for specialised services for rare diseases such as Laurence Moon Bardet Biedl Syndrome (LMBB).

"Our annual plan for specialised services in Wales includes provision for Welsh patients suffering from LMBB and significant funding for these patients has already been allocated in our budget.

"This money is available to enable patients who require treatment at the LMBB diagnostic and management

services in Birmingham and London to access these services.

"We will be in touch with the clinical directors of these services to ensure they fully understand that the services they provide are available for patients living in Wales, especially children requiring early diagnosis and family support, and that their treatment will be fully funded by the Assembly Government."

"Patients who have concerns about accessing these services should, in the first instance, contact Health Commission Wales."

For more information about the Laurence Moon Bardet Biedl Society visit www.lmbbs.org.uk

Rare disease day

Rare Disease Day, on Wednesday, is a global event designed to raise awareness of rare diseases and improve services for those affected.

A rare disease is one which affects about one in 2,000 people. They are surprisingly common – 175,000 people in Wales (3.5 million in the UK) – live with a rare condition at some point in their lives.

There are more than 6,000 different rare conditions and they include Huntington's Disease, Myasthenia Gravis, all childhood cancers and other conditions, which are less familiar, such as Mal de Debarquement Syndrome, Juvenile Batten Disease and Behçet's Syndrome.

Rare diseases vary in severity and symptoms, but people living with these conditions report similar issues around diagnosis and treatment.

It can take patients years to get a firm diagnosis, which can often result in their conditions deteriorating, inappropriate treatment and medication, and significant distress.

Rare Disease UK is now campaigning for a strategy in Wales and for integrated services to speed up diagnosis and ensure there is effective treatment and care for people living with rare conditions.

Alastair Kent, chair of Rare Disease UK, said: "There are tens of thousands of people in Wales and millions of people in the UK living with rare conditions.

"For many, the information available on their conditions is scarce and scientific research is lacking.

"Raising awareness of rare diseases and the need for a co-ordinated strategy for the diagnosis, treatment and research of rare diseases is hugely important."

Rare disease facts

■ 75% of rare diseases affect children and 30% of patients will die before their fifth birthday, in 10% of 17 people will develop a rare condition at some point in their life.

■ 80% of rare diseases have identified genetic origins. Other rare diseases are the result of infections, allergies and environmental causes, or are degenerative and proliferative.

■ Rare diseases are often chronic, progressive, degenerative, and often life-threatening.

■ Rare diseases are disabling – quality of life is often compromised.

■ Patients and their families can suffer high levels of pain.

■ Rare disease patients face a number of problems, including a lack of access to correct diagnosis and a lack of quality information.