TRANSCRIPT OF RARE DISEASE STRATEGY ADJOURNMENT DEBATE
30TH APRIL 2012

Motion made, and Question proposed, That this House do now adjourn.—*(James Duddridge.)*

Mr Edward Timpson (Crewe and Nantwich) (Con): As ever, I am grateful, and indeed delighted, to have the opportunity to address the House in this timely and much-needed debate. Just eight days ago, along with 37,000 other hardy souls, I stood, full of trepidation and excitement, at the start line for the 2012 London marathon. I was running with my wife, Julia, and am pleased to report that we finished the course together in just under four hours. Far more importantly, our run raised over £6,000 for the national charity CLIMB.

That is all very interesting, you might say, Mr Deputy Speaker, but what has this rare accomplishment for a Member of Parliament got to do with a rare disease strategy for the UK? To explain, CLIMB stands for "children living with inherited metabolic diseases" and is a charity based in Crewe in my constituency. It is one of an important network of rare disease charities that strive to raise awareness of rare diseases, not least through Rare Disease day, which takes place on the last day of February each year. Under the stewardship of Steve Hannigan, CLIMB provides vital advice and support to many children and families affected by one of the around 730 known metabolic diseases. One of those children is my nephew Leo.

Leo was born in 2001 with an inherited metabolic disease known as MCADD. Approximately one in 10,000 babies born in the UK has MCADD, which means that they are missing an enzyme that helps break down the fats that provide energy for the body, particularly after long periods without food. This inability to break down fat leads to the build-up of medium-chain fats that can produce toxic substances and lead to severe symptoms, including seizures and possibly coma and death. As Leo was born before the introduction of routine baby screening for MCADD in 2008, neither medical professionals nor his parents knew that he had the disease.

For Leo, all was well until he was 10 months old, when he contracted pneumonia that was coupled with three days of severe hypoglycaemic episodes. Because the knowledge of rare diseases among medical staff was and, indeed, remains limited, and because he had not been screened for MCADD, no-one picked up on this underlying rare condition and the need to treat him accordingly. More by luck than by informed professional judgment, thankfully Leo survived.

When Leo suffered a further hypo episode at 14 months, triggered by nothing more than a routine cold, the doctor on duty, who had some knowledge of MCADD, realised that his low blood sugar was unexplained, and subsequent relevant tests revealed a diagnosis of the metabolic disease.

Leo is now 11 and living a full and active life, but, with the mortality rate of children under the age of one who have undiagnosed MCADD being approximately 25%,

the routine screening that CLIMB long campaigned for, and which is soon to be introduced throughout all four nations of the UK, is a vital tool in saving lives.

In the past year alone, more than 80 newborn babies have been diagnosed with MCADD—80 babies whose disease would otherwise have remained undetected and who would have been at significant risk. That is why I wanted this debate: to be able to speak up for all those, like Leo, with a rare disease, to put a proposition to the Minister and to question him about, how we can improve the services, information, treatment and support that those with rare diseases receive in order to ensure that we maximise their quality of life.

We are not talking about a handful of isolated cases. With close to 7,000 distinct rare diseases having been discovered, and with five new diseases described every week in medical literature, rare diseases are collectively far from rare. In the UK it is estimated that 3.5 million people, or one in 17, will be affected by a rare disease at some point in their life, with 30% of patients dying before their fifth birthday.

So what is a rare disease? The European commission on public health defines rare diseases, sometimes known as "orphan diseases", as

"life-threatening or chronically debilitating diseases which are of such low prevalence"—

affecting fewer than one in 2,000 people—

"that special combined efforts are needed to address them."

About 80% are genetic in origin, but many are auto-immune, such as Crohn's disease, and others are infectious in nature.

Motor neurone disease, cystic fibrosis, muscular dystrophy and Huntington's disease may be some of the more widely known rare diseases, whereas acquired non-histamine induced angiodema and Adair-Dighton syndrome are just two of the many thousands of others that I have come across in my own research.

Jim Shannon (Strangford) (DUP): One rare disease that I am aware of is Prader-Willi syndrome, which affects the muscular build-up, eating habits and sleep patterns of some 2,000 people in the United Kingdom and 50 people in Northern Ireland. Does the hon. Gentleman feel that, if there is to be a strategy for the United Kingdom, it must involve all those regions where health matters are devolved to the Administration? Does he feel also that along with such a strategy and, given all the different areas that there are, we need to have something for respite care, because parents, and the way in which they play their part, are vital?

Mr Timpson: I am grateful to the hon. Gentleman for raising a particular issue in his constituency in Northern Ireland. He is absolutely right that we need to ensure that the strategy we develop for rare diseases supports not just those who suffer from the

disease, but the families who every day, for every hour, have to cope with it and support them. We need to ensure that that is a central part of the strategy.

Such diseases—especially those, including the one to which the hon. Member for Strangford (Jim Shannon) refers, that are particularly rare—are no less important, however, and therein lies the problem. Most rare diseases are under the medical and public radar—too exceptional to attract the attention, recognition and resources required and, above all, the coherent plan needed to tackle the fragmented, inefficient and often inequitable services on offer.

That is why I welcome the Government's current consultation on the development of a UK strategy for rare diseases and why this debate is so apt. For too long, rare diseases have been placed in the "too difficult to do" pile, but with the onset of changes to health care commissioning and the refocus on putting patients at the heart of the NHS, there is a real opportunity to do things more effectively and much smarter. That means that the consultation must be ambitious, provide strong leadership, and be unambiguous about how the core vision that it reveals can be successfully implemented practically and realistically on the ground across all four home nations.

I have taken the time to read the consultation document, and although it makes many of the right noises, there is a nagging doubt as to whether it meets all the challenges faced by those with rare diseases. I am told by those working in the field of rare diseases that Lord Howe, the Minister who, together with his colleagues in the devolved nations, is responsible for the document, genuinely appears to understand the importance of getting this right. I trust, therefore, that he and they will listen carefully to the submissions they receive, including mine this evening and those of other hon. Members.

So what needs improving? It is clear from my many conversations and my correspondence with clinicians, patient groups and charities working on and living with rare diseases that the ad-hoc nature in which services have developed has led to the principal problems of delays in diagnosis; misdiagnosis; lack of information, communication and awareness; limited research; scarce and unequal access to orphan medicines; poor commissioning and care co-ordination; and a failure to monitor outcomes.

That is not to say that it is all bad news. There is, as ever, some fantastic best practice already taking place. The TREAT-NMD—Translational Research in Europe: Assessment and Treatment of Neuromuscular Diseases—network for neuromuscular diseases, the European Huntington's Disease Network's REGISTRY, and the Tay Sachs walk-in clinic at Guy's hospital are all good examples of innovative, effective and efficient service delivery on which any strategy should seek to build.

Margot James (Stourbridge) (Con): I congratulate my hon. Friend on this very important debate. Does he agree that some rare diseases at least have a high profile, which is advantageous in garnering research and funding? I should like to

draw his and the Minister's attention to a rare disease that has a very low profile—FOP, or fibrodysplasia ossificans progressiva, which is a tragic condition where bone grows in muscle. The funding and research that is required for this condition is set back due to its low profile. Will my hon. Friend join me in wishing to raise the profile of this rare condition and many others with such a low profile?

Mr Timpson: My hon. Friend has done just that. I know that she has recently become patron of the charity FOP Action. I believe that this disease affects only one in 2 million people. I congratulate her on taking up that task and wish her and the charity well in raising awareness and the profile of that disease so that even the very small number of people who are affected by it receive the best possible care and support throughout their lives.

In the short time that this debate allows, it is not possible to dig down into the detailed analysis of where the system is currently failing to meet patients' needs, but I urge the Minister to concentrate on six specific areas, the first of which is diagnosis and screening. As I have said, diagnosis is a major issue. Rare Disease UK has found that nearly half of all patients wait over a year for an accurate diagnosis following the onset of symptoms, with 20% waiting over five years and 12% over 10 years. Misdiagnosis is also a key problem, with almost half of patients being misdiagnosed and almost a third being so three times or more—an avoidable waste on many levels.

One lady who contacted me was Kay Parkinson. Kay set up Alstrom Syndrome UK, a support group for people affected by the same rare genetic disorder that so tragically took the lives of her two children, Charlotte and Matthew. Their desperate story of misdiagnosis, unnecessary and delayed medical interventions and ultimately the most terrible heartache lays bare the consequences of a failure to diagnose early and to diagnose accurately. Through Kay's dedication, Alstrom Syndrome UK has set up multidisciplinary clinics, funded by the NHS national specialised commissioning group, to help inform patients and professionals of the specialised clinical services available. Its frustration is that it is still unable to find out who and where diagnoses of Alstrom syndrome are made so that it can link them to the specialised NHS services available.

To improve diagnosis, there needs to be: an increase in health care professionals' knowledge and awareness of rare diseases through initial and ongoing training, particularly for paediatricians; improved links between specialist centres and local services to help with that education, and consideration of the inclusion of appropriate rare diseases in newborn screening, which has proved so successful in the case of MCADD.

The criteria that the National Screening Committee uses should be reviewed to ensure that rare diseases are not being treated unfairly. It is disappointing that the UK lags well behind many other countries in the number of rare diseases for which it screens. For example, in India, the figure is 39; in Poland and China, it is 25, and in the

UK, it is just five. Improved access to diagnostic and carrier tests is necessary for even provision across the UK.

Secondly, we need better information. I have spent time between late night votes productively, you will be pleased to hear, Mr Deputy Speaker, in navigating my way around Orphanet in the Library. Orphanet is the European portal for rare diseases and orphan drugs. It provides pretty comprehensive information for patients, professionals, the public and the industry, but is not widely known. Lack of reliable, up-to-date information that helps rather than hinders the prospects of diagnosis remains a significant barrier.

The development of a UK-wide, trusted single portal of information, which has listed against each condition a named clinician who can act as a source of advice and information, would be a major step forward. It would be further enhanced by implementing the international classification of diseases—ICD-11—in 2015. That will bring about the capture of data on the incidence and natural history of rare diseases that are currently poorly understood. The Government should be preparing for such implementation as we speak. I hope that the Minister can give me and other hon. Members assurances that that is the case.

Thirdly, we need research. According to Sir John Burn, professor of clinical genetics at Newcastle university, who was kind enough to contact me, the 80% of rare diseases that have a genetic basis can now be solved using the latest DNA techniques. However, until the 23 regional genetics centres are honed into a single approval structure, huge barriers to research will continue.

Without clinical research networks, the problems of duplication and the perceived lack of impact of research will remain. The knock-on effects are continuing poor relative funding levels and holding back the development of diagnostic tests and treatments.

To ensure that clinical research networks are effective, disease registries need to be established that bring together all clinical information from all patients with a particular condition or type of condition. That will help to deliver more robust research as well as providing more co-ordinated planning and service delivery for the patient.

Fourthly, we need co-ordination of commissioning and care. I have already alluded to the disconnect between the pockets of expertise at regional level and the lack of any real cadre of experts in commissioning locally. That leaves many patients being bounced around the system, with no tangible results or benefits.

Developing a hub-and-spoke model between centres of excellence will help bridge that gap and create meaningful clinical networks. However, ultimately, the new national commissioning board provides a perfect vehicle for ironing out disparities in provision throughout the country. I therefore ask the Minister to explain exactly how

the national commissioning board will help create models for cluster-type service delivery for rare diseases.

To ensure that that objective is reached, a national champion for rare diseases, with the necessary clinical clout, is essential in the form of a national clinical director. Bearing in mind that that was a recommendation by the former chief medical officer in his 2009 annual report, I ask the Minister to set out as far as he can the Government's thinking on that.

The consultation also talks about each patient having a designated care co-ordinator in the same way as cancer patients have now. That is eminently sensible as it fulfils the objective of delivering patient-centred care, and I would be amazed if the Minister felt unable to agree to it in principle.

Fifthly, on access to orphan medicines, the evaluation and appraisal of orphan medicines is different from that of most others. Decisions on whether or not to fund treatments are often made on an individual basis and are very much dependent on which home nation, or indeed which primary care trust, the patient is from. To improve equality of access to orphan medicines requires a proper and consistent appraisal based on the issues specific to them. At the moment, that is lacking, and it follows that a reassessment of the criteria for access is necessary.

Finally and sixthly, on implementation and outcomes, ultimately any strategy is not worth the paper it is written on if it does not deliver significantly improved outcomes for patients. In the case of rare diseases, that could not be more relevant. Processes are important, but the outcome for the patient is the lasting legacy. Effective implementation of the strategy and the monitoring of outcomes flowing from it are crucial. Clear lines of responsibility for delivering the UK plan must exist and the national commissioning board has a big role to play. Without that accountability, we may never truly know whether this has all been worth our effort.

My greatest personal challenge so far this year has been to haul my body round 26 miles, but it is small beer compared with the challenges faced by the 3.5 million people in the UK who continue their battle with a rare disease. However, perhaps the greatest challenge is to our NHS, which over the years has had no choice but to adapt to the changing health needs of, and treatments available to, its patients. For people with rare diseases, the NHS needs to adapt once more. It can do it, and I hope this strategy will ensure that it does.

The Minister of State, Department of Health (Mr Simon Burns): May I begin by congratulating my hon. Friend the Member for Crewe and Nantwich (Mr Timpson) on securing this debate on what is a most important topic for a great number of people, and on the sensitive way in which he outlined his concerns, particularly those that affect his family? May I also congratulate him on running the London marathon an hour and a half quicker than the shadow Chancellor?

As we have heard, anybody, at any stage in life, can be affected by a rare disease, which can range from manageable conditions that do not affect daily living to debilitating conditions that have a significant impact on one's quality and length of life. The Government are committed to providing the best quality of care to people with rare conditions, and the importance that we attach to services for people with rare conditions is clearly demonstrated in the reforms we set out in the Health and Social Care Act 2012. Through the Act, specialised services, which are currently provided at both national and regional level through a range of NHS organisations, will be brought together under one roof. From April 2013, the new NHS Commissioning Board will directly commission services for people with rare diseases on a national basis.

My hon. Friend asks for an explanation of how the NHS Commissioning Board will operate to ensure cluster-type service delivery in respect of rare diseases. Moving to a national standard system of commissioning but maintaining a regional focus gives the geographical and speciality oversight that he describes. National specifications will lead to services being defined once for England, allowing clear planning to take place across the country.

Mr David Anderson (Blaydon) (Lab): I congratulate the hon. Member for Crewe and Nantwich (Mr Timpson) on a fine speech. As the chair of the all-party parliamentary group on muscular dystrophy, may I ask the Minister about two connected points in respect of what he has just said? There has been a great development within neuro-muscular services and work by the House and the Department. Will the Minister meet the all-party group and the muscular dystrophy campaign to discuss the progress of the national neuro-muscular work plan? Will he also give us an assurance on the positive advantages in the south-west region—he mentioned regional development—and confirm whether there will be strategic clinical networks for neuro-muscular services across the country?

Mr Burns: I am grateful to the hon. Gentleman. I recognise the tremendous work he does in this area of health care and congratulate him on his efforts. With regard to a meeting, I will pass on his comments to my noble Friend the Earl Howe, who has responsibility for this area of health care. On the hon. Gentleman's second point, I am more than happy to give him the assurances he seeks.

The proposed operating model for specialised commissioning links national service knowledge and expertise with local contract knowledge of providers and pathways of care, cementing the new system together in the interests of patients. The benefits to patients with rare conditions are clear: a single national commissioning policy and better planning and co-ordination will result in improved consistency across the country.

My hon. Friend asked me to set out the Government's thinking on the suggestion from the former chief medical officer for a national clinical director for rare diseases. I can assure him that there will be a director within the NHS Commissioning Board

with lead responsibility for specialised services for people with rare conditions. The board will also consider the most suitable form of clinical advice covering the domains of the NHS outcomes framework. Rare diseases come under domain 2: long-term conditions.

Our commitment to people with rare conditions is demonstrated through our recently published, "A UK Plan for Rare Diseases". The consultation was launched on 29 February—rare disease day—and was produced jointly by the four nations of the United Kingdom. The consultation will continue until 25 May and is an important step on the way to producing the final plan. I urge everyone with an interest in this area of health care to contribute to the consultation process.

This will be the first time that the UK has developed a plan to tackle rare diseases, and the consultation represents collaboration across the four nations of the UK. It brings together a number of recommendations designed to improve the co-ordination of care and to lead to better outcomes for people with rare diseases. We suggest that improvements can be made through earlier diagnosis, better co-ordination of services, stronger research and better engagement with patients and their families. Many of these recommendations will be of direct benefit to patients and can help the NHS to be more efficient and co-ordinated and to save money.

Earlier diagnosis through clear care pathways to expert centres can prevent disability, and in some cases save lives, by allowing an earlier start for effective treatment. It will also save money by avoiding more intensive or emergency treatment. More co-ordinated care saves patients' time, money and stress by avoiding multiple visits to various clinics. As many rare diseases are of genetic origin, we must also embrace advances in genetics and genomic medicine and ensure that the NHS is ready to support and take full advantage of these developments.

My hon. Friend has already mentioned that people with rare diseases need to be able to access orphan medicines. Our priority is to give NHS patients better access to the innovative and effective drugs that their doctors recommend for them, including those designated as "orphan drugs". The new system of value-based pricing will bring the price the NHS pays for drugs more in line with the value it delivers. Notwithstanding this, we know that there may be instances where an individual medicine should not be assessed under value-based pricing. We will keep the situation under review. If, as we begin to implement value-based pricing, it becomes clear that some treatments would be better dealt with through separate arrangements, we will explore alternative options.

The consultation document sets out a coherent approach to tackling rare diseases. It recognises existing developments while setting out a number of further developments, such as on better information for patients to help them manage their condition. My hon. Friend asked for reassurance that we are putting steps in place in preparation for the introduction of the international classification of diseases—ICD-11. I can assure him that the NHS is moving towards widespread use of systematized

nomenclature of medicine clinical terms—Snomed CT—in preparation for the introduction of ICD-11.

The consultation will inform the final UK plan for rare diseases. We hope that the final plan will offer a framework for managing rare diseases wherever they occur. Each nation of the UK will then take forward implementation of the plan in accordance with its own priorities and patterns of service. In England, much of the implementation of the final plan will be for the new NHS Commissioning Board to take forward, in close dialogue with Public Health England.

As my hon. Friend will appreciate from my comments in this relatively short but important debate, he has raised an extremely important issue that all too often is forgotten in the mainstream of the NHS, where people concentrate on more acute services, rather than this highly specialised area. My hon. Friend the Member for Stourbridge (Margot James) raised a particular illness or condition. May assure her that following her intervention, I will ask my noble Friend the Earl Howe to write to her about the issue she raised?

In conclusion, the development of the first ever UK plan is an important signal of our continuing commitment to providing good quality services to people with rare conditions. The consultation is aimed at a wide audience, including not just clinicians and NHS specialised commissioners, but patients, their carers and families, support groups, specialist organisations, researchers, academics and colleagues from across social care. I call on all hon. Members to encourage their constituents with an interest in rare diseases to take part in this important consultation.

Question put and agreed to.

10.55 pm

House adjourned.