Speech: We must bring an end to the diagnostic odyssey of rare diseases

It is a such a pleasure to mark Rare Disease Day with you all.

And I want to start by saying thank you to Genetic Alliance, Rare Disease UK and all of you whether you are patients, carers, researchers, clinicians or campaigners – it is down to you that we have come so far in recent years with rare diseases research and care.

We all know that:

- for too long too many parents had to cope with a sick child with a rare disease but with little information as to what the disease is, let alone where they might find treatment

- for too long GPs were frustrated by being unsure where the best centre of expertise is to treat a particular rare disease

- for too long the treatment of those with a rare disease was been seen as a public policy afterthought rather than as a priority

And this is why the establishment of the National Rare Disease Policy Board and Forum was a genuine turning point I think. It sent out a strong message to the whole system that information, diagnosis and treatment of those with a rare disease is not only now at the heart of the policy machine but it was also much more strategic and being developed with patients as its guide.

My own experience as an Ehlers Danlos patient has been entirely typical I think and has led me to my own conclusions about rare diseases policy.

I was undiagnosed for 30 years and went through all the usual experiences of the diagnostic odyssey – getting very sick from childhood and being referred to many doctors who each tried their best, ordering more and more complicated and invasive tests but ended up suggesting a psychiatrist or prescribing me something that just made me sicker.

Finally, a wonderful neurologist with experience of EDS realised what had been going on and referred me to a specialist who diagnosed me in 20 minutes. Twenty minutes – after all those years.

Over the next 18 months I acquired a fleet of specialists. Initially it was a disaster. As they started trying to find the right medical regime for me I got much, much sicker and I found trying to co-ordinate all the tests and appointments and new medications – while still working – impossible.

I do not think there is a flat surface in this building I have not collapsed on – including on this very podium where I tried to give speech a couple of years ago.

Then the NHS stepped in and saved me. The occupational therapist here in Parliament, my GP and the whole team at UCL Autonomic Unit literally picked me up and held my hand – helping co-ordinate my care and getting me the support I needed at work. Gradually, the pieces fell into place and I have clawed my way back to stable health. I will not pretend to any of you it was easy. It was not. And there were many setbacks along the way. But I do know I am incredibly lucky.

Firstly, my battles are nothing compared to many of you here today and for that you have my unending admiration.

Secondly, without my family I simply would not have made it. They have sacrificed beyond measure to care for me and I can never repay them.

Thirdly, I owe so many NHS workers – nurses, doctors, my GP, pharmacist, paramedics and more – my stable health today. I am not sure I will ever be able to communicate to them quite how dramatically they have changed my life.

But this process has also taught me indelible lessons about how urgent it is to improve care for rare diseases for everyone – not just the lucky ones like me.

This is why we must never relent in our campaign to bring an end to the diagnostic odyssey – it is pernicious and even after diagnosis the damage it does to mental health of patients and their families must not be forgotten.

We must press even harder on clinical awareness and groundbreaking research so more patients can be diagnosed and treated earlier.

Finally, co-ordinating your care can feel almost impossible when you are ill and the complexity of services for rare disease must not act as a barrier to access for care.

That is why the publication of the UK Strategy for Rare Diseases in 2013 represented such a significant achievement for everyone here today.

It put the emphasis firmly on raising awareness, improving diagnosis, and enhancing research and patient care and we have come a long way.

Today DHSC and NHS England have made good on the promise to publish annual updates to the implementation plan.

One year on we can celebrate some incredible milestones – let me just highlight a few.

Care co-ordination

Firstly, on the issue of care co-ordination I have asked NHS England to implement a rare disease ‘insert’ from April 2019. This refers to a set of

provider criteria to sit alongside NHS England specifications for services treating patients with rare diseases and allows NHS England to hold providers to account for the way in which they treat patients with rare diseases.

There will be up to 3 criteria (depending on the nature of the service):

1. ensure there is a person responsible for co-ordinating the care of any patients with rare diseases
2. give every patient with a rare disease an ‘alert card’, including information about their condition, treatment regime and contact details for the individual expert involved in their care
3. ensure that every paediatric patient has an active transition to an appropriate adult service, even if that adult service is not the commissioning responsibility of NHS England

I hope that this will make a real difference for patients on the ground. I will be keeping a close eye on whether it does.

Research

Many of you will know that the National Institute for Health Research (NIHR) has established the new BioResource for Translational Research for Common and Rare Diseases.

By March 2018, 37 individual rare diseases had been adopted and by 2022 we expect this number to have increased to 100.

The in-depth phenotyping for rare diseases, linking that to genomic data promises to provide an invaluable research environment for rare diseases discovery.

At Public Health England the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) has made brilliant progress expanding their registries this year.

They have achieved 100% population coverage for conditions diagnosed either antenatally or postnatally, up from 49% in 2017 to 2018.

I truly believe this will improve our understanding of the causes, diagnosis and treatment of rare diseases.

I would like to thank everyone in this room who has played a part in developing these vital research projects.

EU Exit and European Reference Networks

I know though that many here are concerned about the potential impact of Brexit on our research capabilities. I want to stress the government is a strong supporter of the European Reference Networks (ERNs).

We are keen to agree a future relationship with the EU that includes continued participation in the ERNs.

We have made this position clear to the European Commission.

In return, the European Commission have shown that they recognise the significant expertise of UK clinicians. Both sides appreciate the importance of maintaining UK participation.

Refresh of the UK Strategy for Rare Diseases

As we work hard on what our future relationship with the EU will look like, I am also aware that we need to work hard on what the future of the UK Strategy for Rare Diseases post 2020 will look like.

I can reassure you that, post 2020, there will be an overarching framework to improve the lives of all those living with rare conditions. I am sure you will appreciate that it is too early to know exactly what this would look like, but I am personally committed to ensure that the rare disease community are closely involved as we move forward on this.

Genomics

The really big news, of course, is that in December 2018, the 100,000 Genomes Project completed its sequencing phase – a fantastic achievement by NHS England, Genomics England and other partners including Health Education England.

I know that this project has delivered life-changing results for patients – 1 in 4 participants with rare diseases are receiving a diagnosis for the first time – but it wouldn't have happened at all without the support and participation of many of you here today so I want to personally thank you for the historic role you have played in helping us transform life chances for so many others with rare diseases.

Not only that, but this project has paved the way for using genomics in 'everyday' healthcare.

Last year, NHS England launched the Genomics Medicines Service (GMS), making the UK the first in the world to integrate genomic technologies, including whole genome sequencing, into routine clinical care.

As the NHS Long Term Plan in January, seriously ill children who are likely to have a rare genetic disorder, children with cancer, and adults suffering from certain rare conditions or specific cancers, will all be offered whole genome sequencing from 2019 under the new Genomics Medicines Service.

This all speaks to a wider ambition.

We want to lead the world in the use of data and technology to prevent illness, not just treat it.

We want to diagnose conditions before symptoms occur.

And we want to deliver personalised treatment, informed not just by our

general understanding of disease but by our own personal, de-identified medical data – including our genetic make-up.

Now you will know that the NHS Long Term Plan outlines our vision for the NHS over the next 10 years.

The plan focuses on prevention and early detection and has been developed with frontline staff, patients and their families.

The plan also sets out the ambition to focus targeted investment in areas of innovation, particularly genomics.

This will enable more comprehensive and precise diagnosis, and allow patients to access more targeted treatments to reduce the use of harmful medications and interventions.

In order to make this a reality, I am delighted to announce that we will be working with the National Genomics Board, people in this room and the broader genomics community to develop a National Genomic Healthcare Strategy. This will tackle not just rare diseases, but it will be built on the foundations that the rare diseases community helped build with the 100,000 Genomes Project.

The National Genomic Healthcare Strategy will set out how the whole genomics community can work together to make the UK the global leader in genomic healthcare.

It is vitally important that this is not just a government exercise: we will be leading a national conversation and I want to encourage everyone with an interest – patient, carer or professional – to share their views and contribute to a coherent, national vision.

Conclusion

There is so much more I could say but I just want to close with this.

I know living with a rare disease or caring for someone for a rare disease can feel relentless. Unseen.

Please do not lost faith.

In the National Rare Diseases Policy Board, the Forum and in me as your minister, you have people advocating for you right at the heart of the system.

But we know we cannot do it without you. The mountains we have climbed were only conquered when we worked in true partnership – and there is still so much more to do.

That is why Rare Disease Day is all about you.

Thank you for your dedication, your expertise and your sacrifice.

And please – enjoy today, you've earned it.

News story: Ministry of Justice grant to support victims of domestic abuse in family courts

In the draft Domestic Abuse Bill and consultation response published on 21 January 2019, government announced its intention to improve support for victims of domestic abuse who are going through family court proceedings.

The Ministry of Justice is pleased to announce that it has awarded a grant of just under £900,000 to two organisations who provide in-court support to vulnerable victims: the Personal Support Unit (PSU) and the Citizens Advice Witness Service.

PSU has been supporting litigants in person in family courts since 2001 and Citizens Advice has been supporting witnesses in criminal court proceedings since 2014. They both have extensive front-line experience of helping victims and witnesses navigate court processes and feel safe at court.

Citizens Advice will be using the funding to extend their current Witness Service to selected family courts to provide information and practical and emotional support to victims before, during and after the day of the hearing. PSU will be using the funding to invest in further training of their staff and volunteers and to share learnings on best practice with a range of family justice stakeholders.

The funding runs from January 2019 through to 31 March 2020, and will allow Citizens Advice to provide these services in up to 12 family courts across England and PSU in 24 courts across England and Wales.

We are determined to improve the experience of victims of domestic abuse in the family courts, and this grant is part of a wider package of measures across the family justice system. In the draft Domestic Abuse Bill we have committed to giving the family courts the power to stop unrepresented perpetrators of abuse from cross-examining their victims in person in family proceedings. We have also included a package of practical action in the accompanying domestic abuse consultation response.

Related link

Domestic abuse consultation response and draft bill

News story: Parents encouraged to be aware of scarlet fever symptoms

Scarlet fever is a seasonal bacterial illness that is common at this time of year, mainly affects children, and is easily treated with antibiotics.

News story: Parents encouraged to be aware of scarlet fever symptoms

Latest update

The latest report by Public Health England (PHE) on seasonal levels of Group A streptococcal infection, published on Friday 10 May 2019, shows that 9,887 cases of scarlet fever have been reported since mid-September 2018, compared to an average of 14,128 for the same period over the last 5 years.

There were 247 cases reported for the most recent week (29 April to 5 May 2019).

Dr Theresa Lamagni, Senior Epidemiologist at Public Health England, said:

> National levels of scarlet fever this season have remained relatively low compared to the past 5 seasons.
>
> We continue to advise parents to look out for the symptoms such as a sore throat, fever and rash, and to contact their GP or NHS 111 if they spot symptoms or have concerns.
>
> Scarlet fever, which mainly affects young children, is not usually a serious illness and can be easily treated with appropriate antibiotics.

Previous updates

Thursday 28 March 2019

The latest report by Public Health England (PHE) published at midday, Thursday 28 March 2019, shows that 7,854 cases of scarlet fever have been

reported since mid-September 2018, compared to an average of 9,617 for the same period over the last 5 years.

There were 456 cases reported for the most recent week (18 to 24 March 2019).

Thursday 28 February 2019

The latest report by Public Health England (PHE) shows that 6,316 cases of scarlet fever have been reported since mid-September 2018, compared to an average of 6,680 for the same period over the last 5 years. There were 409 cases reported for the most recent week (18 to 24 February 2019).

Scarlet fever is usually a mild illness but it is highly infectious so PHE is advising parents to be on the lookout for symptoms, which include a sore throat, headache and fever with a characteristic fine, pinkish or red body rash with a sandpapery feel. If signs of scarlet fever are suspected, it is important to contact your local GP or NHS 111.

Early treatment with antibiotics is important as it helps reduce the risk of complications such as pneumonia and the spread of the infection to others. Children or adults diagnosed with scarlet fever are advised to stay at home until at least 24 hours after the start of antibiotic treatment to avoid spreading the infection to others.

Dr Theresa Lamagni, Senior Epidemiologist at Public Health England, said:

> It's not uncommon to see a rise in cases of scarlet fever at this time of year. Scarlet fever is contagious but not usually serious and can be treated with antibiotics to reduce the risk of complications and spread to others. We are monitoring the situation closely and remind parents to be aware of the symptoms of scarlet fever and to contact their GP for assessment if they think their child might have it.
>
> PHE is investigating possible reasons for why there has been a rise in scarlet fever cases over the last few years by studying the strains of bacteria causing disease and the spread of infection in different settings and patient groups.

Professor Helen Stokes-Lampard, Chair of the Royal College of GPs, said:

> Patients with scarlet fever usually present with flu-like symptoms including a sore throat, fever and headache, as well as a characteristic rosy rash – usually on the patient's chest initially.
>
> Scarlet fever is a bacterial infection that generally affects children under 10 more than teenagers or adults. It is very contagious but can be quickly and effectively treated with a full course of antibiotics. GPs would also recommend patients to rest,

drink plenty of fluids, and use antihistamine tablets or calamine lotion for relief of symptoms related to the rash.

We have seen more cases of scarlet fever in the last few years than we've been used to – we're unsure why this is, but if a patient thinks that they, or their child, might have symptoms, then they should seek medical advice.

Background

For further information on scarlet fever visit the NHS website.

Guidelines for the management of scarlet fever are also available from PHE.

Government response: UK statement on the Hanoi Summit

A Foreign Office spokesperson said:

We hope the talks between President Trump and Kim Jong-Un will prove a basis for progress. It is clear that there is more work to do and we welcome the US commitment to continue negotiations.

We urge North Korea to engage in this process and take concrete steps towards its complete, verifiable and irreversible denuclearisation.

As a permanent member of the Security Council with close partnerships and a network of economic and security interests in the region, a peaceful resolution of the issues on the Korean

Peninsula is important to the UK.

We will continue to support the United States in their efforts to secure a negotiated outcome, and encourage Kim Jong-Un to choose to prioritise the welfare of his people over the development and preservation of his illegal nuclear weapons and ballistic missile programmes.

Further information

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