We need national debate on rare diseases to offer the best possible care

Good afternoon everyone.

It is such a pleasure to be here to celebrate and raise awareness of rare diseases with you all – and it is a pleasure to see some of you again who were present during the excellent event hosted in the Houses of Parliament earlier this year. And even more of a pleasure to be celebrating through eating cake!

So, thank you to the British Paediatric Surveillance Unit for inviting me to such a special and enjoyable event.

Today, I really want to address the incredible young people here in the audience. Like many of you, I got ill as a child and for a very long time nobody knew what was wrong with me.

I was undiagnosed and went through all the usual experiences of the diagnostic odyssey – getting very sick from childhood and being referred to many doctors who did their best but just couldn't find out what was wrong with me. This went on for 30 years. Finally, a wonderful neurologist realised what was wrong and referred me to a specialist who diagnosed me in just 20 minutes.

It was such a relief, but as I acquired more and more specialists and we began to try to get the right medical regime for me, I got much, much sicker and found trying to co-ordinate all the tests and appointments and new medications – while still working – impossible.

Then the NHS stepped in and saved me and gradually, the pieces fell into place and I have clawed my way back to stable health.

I still have my battles, you may have seen that the other day I fainted right in the middle of giving a speech in the House of Lords, but I know that this is nothing compared to many of you here today – and for that you have my deepest respect.

I am absolutely thrilled to be the minister for rare diseases again and committed to make a real difference. One thing I know from my own experience is that without my family I simply would not be here. I am sure many of you here feel similarly about your families and carers.

I want to take a moment to thank all clinicians, researchers and others who are working so hard to improve care for people with rare diseases. I'd like to highlight a few recent achievements in this area.

Genomics

Over the past years we've learned more and more about how our individual genetic make-up can lead us to develop a rare disease. And the UK's 100,000 Genomes Project has helped with that. The headline is of course that in December 2018, the 100,000 Genomes Project completed its sequencing phase – a fantastic achievement by NHS England, Genomics England and other partners.

The project has already delivered life-changing results for patients, with 1 in 4 participants with rare diseases receiving a diagnosis for the first time. We are still returning results to some patients and will make sure that this is a priority over the course of 2019.

Let me tell you about one of these participants – a 4-year-old little girl called Jessica. She had a rare condition that caused epilepsy and affected her movement development. From looking at her specific DNA, Jessica received a diagnosis – 'Glut 1 deficiency syndrome' – and as a result her doctor recommended a very specific diet that has helped reduce seizures for others with her condition. Jessica and her family were able to take immediate action, help control her epilepsy and improve her condition.

I'm delighted that based on the amazing achievements from the 100,000 Genomes Project, NHS England launched the Genomics Medicines Service (GMS), making our country the first in the world to integrate genomic technologies, including whole genome sequencing, into routine clinical care. And here's the important part – seriously ill children who are likely to have a rare genetic disorder will be offered whole genome sequencing under the GMS. As demonstrated by Jessica and numerous others, we hope this will bring an end to the diagnostic odyssey for many ill children.

To continue cementing the UK as world leader in genomics, in February I announced that government is developing a UK Genomics Healthcare Strategy. I'm very pleased to say that the work is well underway and the strategy will provide a clear, national vision setting out how the genomics community can work together to make the UK the global leader in genomic healthcare – for the benefit of patients. My colleagues have been inviting key stakeholders and representative groups, including the rare diseases community, to share their views and contribute to the coherent national vision, and are continuing to do so. The strategy will be ready for publication this autumn… so watch this space.

NHS insert

Another very exciting initiative I want to share with you is the NHS insert, which I announced in February and the NHS are now implementing. The insert gives NHS England a way to hold providers to account and improve services for rare diseases. I have met with NHS England just this week, who have assured me that the insert has been included and will be monitored through the Quality Surveillance Systems, with trusts reporting on it for the first time this September.

There will be up to 3 criteria providers report on:

- care co-ordination
- an alert card
- transition

Let me explain these each in turn.

Firstly, the provider must ensure that there is a person responsible for co-ordinating the care of any patient with a rare disease. Secondly, the provider must give every patient with a rare disease an ‘alert card’. This will include information about their condition, treatment regime and contact details for the individual expert involved in their care. Finally, the provider must ensure that every child has an active transition to an appropriate adult service, even if that adult service is not the commissioning responsibility of NHS England.

It is my sincere intention that changes like these make a difference to the lives of patients with rare diseases, people just like you.

Post-2020 framework

There is still much to be done to improve the experience of patients with rare conditions. That’s why I want to lead a national conversation on rare diseases, and how we care better for people.

We want to talk to staff, patients, experts and researchers.

We want to use in-person and online methods to capture views on the big strategic issues that affect you.

We recognise there are a range views, and that’s why the department and I will be canvassing views – drawing them together into a big, compelling strategic framework to improve care for people with rare diseases.

It won’t address every challenge – but it will be an ambitious attempt to build on our world-leading commitments, set out in the 2013 strategy.

We will be working with patient organisations and experts to develop a mechanism for capturing honest views on your experience. This is the start of the process towards a future framework, an opportunity to gather evidence and identify common themes and proposals to help us develop the vision for a post-2020 rare disease framework.

Closing

There is so much more I would like to tell you today, but time is running away and I just want to close with this.

In the UK Rare Disease Policy Board, the forum and me as your minister and fellow patient, you have people advocating for you at the heart of the system. To the children and young people in the audience here – I am fighting

as your minister so that you and future generations may grow up in a world where:

- the diagnostic odyssey is no longer an odyssey
- you know more about your rare conditions through significant research efforts and breakthroughs
- you may grow up in a system where you have a smooth transition to appropriate adult services

I assure you that government hopes to make a real difference on these matters and remains as committed as ever to improving the lives of those living with rare diseases.

I’m looking forward to hearing from the patients, researchers and advocaters here today, and we can all look forward to discussing the important issues raised over some tea and cake later. So, let me finish by saying thank you very much for your patience and attention. Please enjoy today.