New strategy to accelerate diagnosis and improve treatment of rare diseases

- Rare Disease Framework sets out vision to improve the lives of more than 3.5 million people with rare diseases in the UK
- The framework will speed up diagnosis, increase awareness and improve treatment and care
- The new strategy has been designed around the views and experiences of those living with rare diseases

Millions of people with rare diseases, like Huntington's disease or cystic fibrosis, will benefit from a new framework to raise awareness of rare diseases, speed up diagnosis and improve care and treatment.

The UK Rare Diseases Framework, signed and agreed by all four nations of the UK, builds upon the successes of the previous strategy and was developed in consultation with those living with rare diseases following the National Conversation on Rare Diseases.

Rare diseases often start through unusual patterns of common symptoms that can be hard for a GP to recognise, there can often be trips to multiple specialists before a final diagnosis is reached. This can take years and can have a big impact on patients, their families and the NHS.

The new framework sets four priorities across England, Wales, Scotland and Northern Ireland including:

- Helping patients get a final diagnosis faster
- Increasing awareness of rare diseases among healthcare professionals
- Better coordination of care
- Improving access to specialist care, treatments and drugs

Health and Social Care Secretary, Matt Hancock said:

> People with rare disease deserve to get the best possible access to care and treatment. Many spend years trying to discover what is wrong so it's essential we ensure we take every step to accelerate diagnosis and our brilliant health and social care workforce have a thorough understanding of those living with rare diseases.
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> The UK Rare Diseases Framework has been developed in close collaboration with people with a lived experience. It will build on the UK's exceptional strength in life sciences, our genomic capability, and of course the huge benefit of having the NHS, to shape our policies on rare diseases in the years to come and improve the lives of so many people.

There are 3.5 million people in the UK with a rare disease, the equivalent of

1 in 17 people.

It is currently estimated there are over 7,000 rare diseases with new conditions continually being identified as research advances. The more well known among them include Huntington’s disease, Ehlers Danlos syndromes, cystic fibrosis and systemic scleroderma, however there are many more which are not as well known or understood, which is why raising awareness is one of the key priorities of this Framework.

Health Minister, Lord Bethell said:

> I want the experiences of those living with a rare disease to shape the priorities of government to make sure our policies work for them. We can harness the potential of new technologies, including genomics, to support earlier detection and faster diagnosis of disease, tailor and target treatments
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> With such a vast range of rare diseases out there, it is hugely important the rare disease community was at the centre of designing the UK Rare Disease Framework.

The National Conversation on Rare Diseases survey, launched by Baroness Blackwood, aimed to identify the major challenges faced by those living and working with rare diseases, and received an amazing 6,293 responses, including from over 5,000 patients, families and patient organisations.

It provided clear evidence diagnosis and awareness of rare diseases, and difficulty in accessing specialist care were some of the challenges consistently seen as the most impactful across patients, their families and patient organisations.

Jayne Spink, CEO of Genetic Alliance UK said:

> We welcome the publication of this Framework and look forward to working with the four nations of the UK to develop action plans to deliver its aims. A framework for rare disease policy is necessary now more than ever.
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> We have powerful genomic tools and exciting research breakthroughs on the horizon that are eagerly anticipated by people living with rare conditions. We hope that this framework can build the pathways that will allow these breakthroughs to realise their full potential in the NHS, across the whole of the UK.

Haseeb Ahmad, President of the Association of the British Pharmaceutical Industry (ABPI) said:

> Today’s strategy sets out a welcome ambition for how people with

rare diseases can get a fast diagnosis and access to treatments they desperately need.

Cutting edge research means that there will be even more exciting, new treatments developed for rare disease patients. We look forward to continuing our work in partnership with Governments across all four nations to make the ambition of this framework a reality for people with rare diseases and their families.