We need to cut time to diagnosis and treatment and make sure that people have access to innovative medicines.

Working to deliver better outcomes for **rare disease patients**

Everyone with a rare disease has the right to early diagnosis and access to specialist care and treatment, according to Ben Whitehouse and Carla Starita, Sanofi UK leaders in rare diseases.

INTERVIEW WITH
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Head of Rare Diseases, Sanofi Genzyme UK&IE

INTERVIEW WITH
Carla Starita
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What's happening in rare diseases today?
Ben: The short answer is 'a lot'. Work is being done by the life science sector that will hopefully offer the prospect of life-altering and perhaps even curative treatments. The Government has an increased focus on life sciences and acknowledges the opportunity that it presents for the UK to be a science superpower. This year we have had the publication of the Rare Disease Framework, the NICE Methods and Process Review and the unveiling of NHS England's Innovative Medicines Fund.

However, rare disease treatments can struggle when assessed for treatment as part of the current NICE methodology due to small patient numbers and limited data, and so there is no question that we need to build on this momentum to bring about further changes that will benefit patients.

How did your organisation come to lead the way in rare disease research and development?
Ben: We've been working in this space for more than 30 years and now have scientists in areas such as genomic medicine, synthetic biology and multiple types of biologics. Alongside research and development we've invested in multinational disease registries, which allow physicians to access clinical real-world evidence and information to identify unmet medical needs and make better decisions.

What drives you forward?
Ben: Two things. First, working with passionate people; and second, working in an area where you can make a real impact on people's lives. The fact is that every person with a rare disease — 75% of whom are children — should have the right to access treatment.[1] At the end of the day, the strength and courage of these patients and their families is a huge source of inspiration.

What needs to change to improve outcomes in rare diseases?
Ben: We need to cut time to diagnosis and treatment and make sure that people have access to innovative medicines. Stakeholders — industry, academia, researchers, governments and patient groups — must work better together to build a sustainable model to deliver that outcome. It's important to place patients at the centre of everything we do, act with urgency, collaborate, listen and be open to new ideas.

What changes will make the biggest difference to patients?
Carla: There's a big role for technology to play. For example, video offers possibilities for home consultations, while apps can offer tracking and monitoring opportunities. This will make a big difference to patients' lives because it will reduce the amount of time they need to spend travelling to specialist centres.

Innovative new treatments are essential, too, which is why our researchers are working together across geographies and specialties to accelerate discovery. As a result, we have several new potential treatments in clinical development.

How do you think data and technology will evolve in this space?
Carla: It's a really exciting time. Technology will continue to boost diagnostic screening, provide patient support and facilitate communication and collaboration between patients, healthcare providers and researchers.

Because reducing time to diagnosis and treatment is so important, we're hoping to use big data to try to predict who may have a rare disease but who has not yet been diagnosed. AI machine learning can generate algorithms which will facilitate early diagnosis in clinical practice. What's more, use of this type of tech and data is widespread and growing.

What does the future hold for rare diseases?
Ben: Rare disease is a highly dynamic area and we are approaching a period of change that has the potential to create real benefits for patients.

As a business, we will continue to build on our leadership position to discover, develop and launch new diagnostic approaches, treatments and support to help people living with rare disease.

Find out more at **sanofi.co.uk**

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1.Rare Disease UK: Understanding Children and Young People's Experiences . Available from: https://www.raredisease.org.uk/our-work/research/understanding-children-and-young-peoples-experiences/

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