Q&A

Call to arms: how can we improve access to rare disease treatments?

Fleur Chandler, head of market access at Sanofi UK & Ireland and parent to Dominic, a child with a life-limiting disease, Duchenne Muscular Dystrophy, has called on the government and industry to improve how new rare disease treatments are assessed and made available

Q What is it like to have a child with a life-limiting rare disease?

A My child has a life-limiting, disabling, progressive rare disease that has no treatment – exhausting is the overarching sentiment. Three quarters of rare diseases affect children and 30% of patients with rare diseases will not live to their fifth birthday, according to the Department of Health and Social Care. When you're treating adult diseases, by and large you are treating an individual. When you're treating a child with a rare disease, you're treating a whole family. If the progression of the illness can be slowed or stopped, the exhausting experience of children and families like mine would also be slowed or avoided.

For my family, the cost, both financially and mentally, has been enormous.

My husband had to give up his job to help us manage the myriad of hospital appointments and necessary care. We have had to make significant adaptations to our house, to enable wheelchair access, with minimal support. We had to buy a wheelchair-adapted vehicle. As it is difficult to travel, and accessible options are limited, we are unable to go on typical family holidays. Our daughter also makes sacrifices to enable us to function as a family.

Some of that might sound like first-world problems but as somebody who works in the pharmaceutical industry, ensuring the value of Sanofi's medicines are articulated well to groups like the National Institute for Health and Clinical Excellence (NICE) and Scottish Medicines Consortium (SMC), it can be frustrating to know that treatments could be available for certain rare diseases if we approached testing and access in a more innovative and pragmatic way.

Q Why is the current way rare disease treatments are assessed not working?

A There are many medicines in development across a wide range of paediatric progressive conditions. But the UK system for looking at the evidence and saying 'yes' to those medicines is just not set up to accommodate rare diseases, which it is much harder to gather evidence for. This is in part because there are very small patient populations and the type of evidence being sought is difficult to gather in these diseases. As companies go about trying to gather that evidence, they're fishing in the same small pool, which is ultimately a set of patients and families who already have a huge emotional load to contend with, in addition to potentially participating in the clinical trials that help pharma companies generate the right evidence. The approval process is lengthy, particularly when evidence is sparse, and sadly during the time taken to assess a medicine's value, many children become ineligible for treatment as their diseases have progressed beyond the point at which treatment is potentially impactful.

> **Ultimately, we need to see more governmental will towards treating rare diseases. Families don't have the time to wait**

Q What needs to happen for more rare disease treatments to become available?

A We need the government to acknowledge the current system isn't working. I'd like to see more emphasis on access to medicines for rare paediatric progressive conditions, and a clear commitment that the UK will make available those medicines that are developed for paediatric patients. It would also mean the UK demonstrating its commitment to being a global life sciences leader, which the current government has articulated as a priority.

Ultimately, we need to see more governmental will towards treating rare diseases. Families don't have the time to wait. Statistics show one in 17 people in the UK will be affected with a rare disease, which is noted in the Department for Health and Social Care's UK Rare Diseases Framework, and may cause alarm at how much it might cost to tackle. But not all rare diseases are fatal or even impactful – they're just rare – and it wouldn't be difficult to identify the ones which really need attention, particularly in paediatric care.

In a BIA survey earlier this year, 79% of respondents said patients with a rare disease should be able to access medicines on the same basis as people with more common conditions, and 78% agreed the NHS should ensure access on the basis of clinical need even if it is more costly.

Q Which organisations are leading the way in rare diseases?

A It's a fascinating space. There's room for everybody and there are lots of small biotechs and exciting developments coming out of university research. But it's especially important to have larger pharmaceutical companies working in this area. You need the muscle of these companies, which have the expertise in running clinical trials and health technology assessments. The biggest impact, however, will be through collaboration; bringing all stakeholders together to generate the evidence so desperately required.

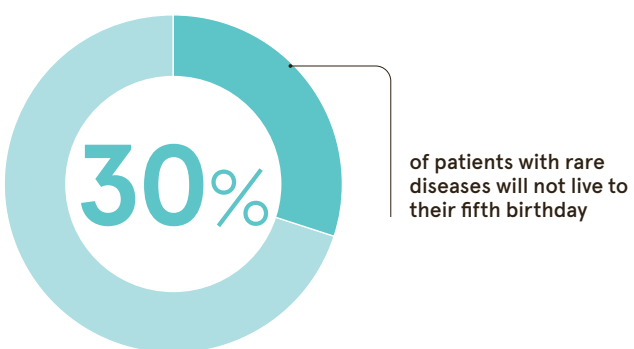
Q How are you seeing organisations collaborating to improve rare disease treatment?

A One thing I've done outside Sanofi, though Sanofi has been very supportive of it, is set up Project HERCULES, a collaborative project in Duchenne evidence generation across ten pharma companies, led by the patient organisation Duchenne UK. This initiative has created a disease-level evidence base that all companies can use when it comes to decisions around access. There is room for that model now in other rare diseases. We need people and policies to work together better to support the rare disease community.

Q What else needs to change to improve rare disease outcomes in the UK?

A Education, support and commitment. I've got the head of a health economist but the heart of a rare disease mum, so I'm in a unique position to help patient organisations understand the needs of those assessing new medicines, and how the pharmaceutical industry meets those needs. I don't know if there will be a treatment available in UK in my son's lifetime, so I'm extremely passionate about ensuring as few UK parents as possible end up in the same situation. We must ensure the voices of patients and families like mine are heard in a way that assessors can understand. For example, there are standard ways of collecting quality of life data, which is a key part of the decision, but they don't address what you're feeling as a family. They don't touch the burden or the psychological hardship. We need to see much more specific methods of data collection to articulate the ultimate value of treating these diseases. It's a privilege to use my professional knowledge and personal experience to support all paediatric progressive conditions, as my experience is similar to many parents of children with rare disease. It's time that access to treatment for rare diseases - especially in paediatric progressive, life-limiting diseases - is prioritised in the UK.

3/4 of rare diseases affect children



of patients with rare diseases will not live to their fifth birthday

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Date of preparation: November 2021
MAT-GB-2105311 (v1.1)