# Partnering with patients on their rare disease journey

As more rare diseases are diagnosed, it's becoming vital for patients and their families to receive guidance and advocacy in their search for effective treatments.

INTERVIEW WITH
**Jonathan Whitton**
Senior Medical Director, Bionical Emas

WRITTEN BY
**James Martin**

Rare diseases are defined as affecting less than 1 in 2,000 people in the UK. Due to advances in diagnostics, there are more rare diseases being discovered every day. This presents a challenge for patients and their families, who are seeking the best medical care possible.

**Giving people living with rare diseases a voice**

As a former paediatric surgical registrar and Senior Medical Director at Bionical Emas, Dr Jonathan Whitton and his colleagues can understand the challenges facing people living with a rare disease and their families, and the need to partner with them on their journey, towards effective therapies.

"In many ways, a person living with a rare disease has the deck stacked against them," he explains. "They are often diagnosed late, due to unfamiliarity of healthcare professionals with rare diseases and more common diseases being ruled out first, or their concerns being dismissed. The frequency of rare diseases leads to further isolation, meaning patients and their families can't always reach others to discuss their conditions and learn about the latest therapies available."

"Our medical affairs team include rare disease patient advocacy specialists who have either worked with people living with rare diseases or have a family member diagnosed with a rare disease. This support framework and expertise allows us to partner with patients and represent their needs in the pharmaceutical sector more effectively."

**Improving patient quality of life and drug development**

Bionical Emas combines clinical development, early access programmes and clinical trial supply to bring life-changing medicines to patients around the world. Their vision is to maximise access and evidence generation across the entire clinical development pathway, positively impacting current and future patients. As advocates for rare diseases, they liaise with pharmaceutical companies, regulatory bodies and clinical healthcare authorities.

Dr Whitton explains: "Clinical development plans need to consider the whole patient journey, the overall burden faced by people living with the rare condition and how this impacts societal contribution to healthcare economics. For example, treating the patient may mean they are less likely to require hospitalisation as a child and can gain more from the education systems that they are a part of."

By partnering with the patient at the beginning of the clinical development process, Bionical Emas aims to provide the effective representation of the patient to both the healthcare professionals and the pharmaceutical companies to enable this and benefit both the speed and quality of drug development.

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# From lab to clinic: what charities are doing to power rare disease progress

Charities play a vital role in attracting investment towards rare diseases and supporting research that might not otherwise make it to the clinic.

WRITTEN BY
**Dr Catriona Crombie**
Head of Rare Diseases, LifeArc

The rare disease community has long been underserved, with many people struggling to receive the diagnosis and treatment they desperately need. Finding collaboration, funding and the right support are all hurdles for researchers to overcome in bringing innovations from the lab to the clinic. This is where charities and not-for-profits have a vital role to play.

**How charities encourage investment in rare disease**

While exciting progress has been made in rare disease research, investing in rare conditions — especially ultra-rare conditions — can carry more risk for commercial companies than common conditions. However, charities have been stepping in and partnering with industry to help progress these innovations towards patients.

They can fund early-stage work to help reduce the risk and provide resources and advice to get medicines or new ways to diagnose a rare disease through trickier parts of their journeys. This can make it easier and more appealing for investment from industry partners.

**Moving research from the lab into medical breakthroughs**

Access to new tests and treatments through clinical research is crucial for individuals living with rare diseases. Many times, though, research gets stuck between the lab and the next phase of development due to a lack of investment, resources or knowledge. This 'translation' of science from the lab into results that benefit patients is the key to unlocking innovations. Some medical charities are targeting their efforts in this space, despite its tricky and (at times) risky nature.

They help provide funding, identify the commercial potential of research and assist with the things a commercial company might not: early-stage development; advice on licensing; collaboration; intellectual property protection; and more.

**Advocating for the bigger picture in rare disease**

Addressing the challenges faced by the rare disease community requires not just research but policy change, coordinated investment, collaboration and education. Charities and the UK Government are working to build a nationwide ecosystem of researchers and partners to do just that.

They can also use their neutrality to bring different parties (with different interests and needs) together for a common aim. This can help influence business models and solutions that might not have been found otherwise.

There is no such thing as a disease too rare for investment. By driving innovations from the lab to the clinic, charities are helping to ensure that no rare disease goes unnoticed or untreated.

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