Research Offers Options for Families Affected by Rare Diseases

Why Research Matters: Insights From Geraldine Halpin, A Parent Of An Adult Son With A Rare Disease

Every year, thousands of families in Ireland receive a rare disease diagnosis. With limited public awareness and few treatment options, many face an uncertain and isolating path. But research is changing that—offering insights, options, and a sense of community for those impacted.

Understanding the Challenges of Rare Diseases

For parents, receiving a rare disease diagnosis for their child can be the start of a complex journey. People with a rare disease often face challenges accessing medical care, education, employment and the support to live independently. While medical information can explain how the condition affects the body, it may provide little insight into the realities of everyday life or what the future may hold.

"*It's very complex, particularly for parents of young children. One of the things that instantly struck me was the isolation—you feel a lack of community*." says Geraldine, the parent of an adult son living with a rare disease.

Treatment options for rare diseases remain limited. Families and doctors often find themselves working with little information, trying to gauge what is happening and how best to respond. "*There's no clear sense of how to live with this. What challenges will we face? What might this child need, that others in my life don't need? What can we expect moving forward*?"

Research plays a crucial role in advancing our understanding of rare diseases and expanding treatment options, including innovative therapies and clinical trials. To bring more options to Irish patients, there is an urgent need for increased investment—funding for research infrastructure, training for healthcare professionals to become 'rare aware', and resources to translate basic scientific discoveries into real-world treatments.

The Impact of Public and Patient Involvement

Rare disease research faces unique challenges, including small patient populations and a lack of existing data. *"As a parent, you realise many treatments are designed for those without rare diseases. You worry whether they'll be effective, how they'll work, and if they'll address the root cause*."

Actively involving patients and families as research partners through public and patient involvement (PPI) ensures that research is relevant, impactful, and focused

on the issues that matter most. Without these voices, the reality of life for children and adults living with rare diseases often remains overlooked.

"*So much of what we go through is invisible to others, and at the same time, you're battling misconceptions about what your life must be like. PPI brings that experience into the light, bringing awareness to the challenges, the curiosities, and the wonderful things. Then, we can ask the research questions that need to be asked, the ones that won't even come up unless someone makes these hidden realities visible*."

By becoming equal partners in research, those affected by rare diseases can influence the future of treatments and support services. "*PPI is more than just sharing your lived experience—it's about sharing in a way that has an impact. We are experts by experience, and our insights have real value in research. We are the ones most affected, so our needs must be considered, discussed, and met*."

A Network of Support and Progress

The Rare Disease Clinical Trial Network (RDCTN) is leading the way in rare disease research. As well as being a hub for clinical trials, the network creates a space where people can connect over shared experiences, discuss current and future research, and advocate for the issues that matter most—including those that might otherwise be overlooked. By bringing together patients, carers, healthcare professionals, community organisations and researchers, the RDCTN fosters collaboration, knowledge-sharing, and, ultimately, the advancement of research that can enrich the lives of people affected by rare diseases.

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