Living well: shifting the narrative

Increasingly the rare disease community is being defined by the pursuit of cures and treatments. While access to medical care is critical, this narrow focus fails to address the full spectrum of needs for those living with rare conditions. A growing movement, spearheaded by Metabolic Support UK, is calling for a fundamental shift—one that prioritises quality of life and empowers the rare disease community to live well regardless of medical prognosis. Kirsty Hoyle, CEO at Metabolic Support UK, joined me for a chat to share their work, and to discuss what 'living well' means in our own households as rare mums

Written by Nicola Miller,
RARE Revolution Magazine

Interview with Kirsty Hoyle,
CEO, Metabolic Support UK

Kirsty Hoyle, CEO of Metabolic Support UK, is at the forefront of their 'Living Well' movement. After her son's rare disease diagnosis, Kirsty was struck by the "disaster narrative" that pervaded the rare disease landscape. "The first thing everyone talked about was diagnosis, burden and sad faces," she recalls. "There can be a heavy focus on cures and treatment, rather than the lived experiences of those with rare diseases." Kirsty's organisation decided to take a different approach. They commissioned in-depth qualitative research to understand what "living well" truly means for the rare disease community. The findings were eye-opening.

Contrary to assumptions, accessing treatments was not the top priority. Instead, people emphasised the importance of joy, employment, social connections, identity and feeling accepted by society. "It's not having a rare disease that causes a low quality of life," Kirsty explains. "It's the gaps in the 'net' of support that prevent people from living well." This realisation led Metabolic Support UK to overhaul their own practices, diversifying funding sources and championing positive narratives. Recognising the importance of this work, they soon realised the value of this work outside of metabolic disorders and have subsequently partnered with the UK based charity Beacon, to expand the remit of the movement to one of a cross-condition nature.

Kirsty's personal journey into the rare disease space was a steep learning curve prompted by her son, William's, diagnosis with ectodermal dysplasia.

Kirsty pictured with her son William

Prior to entering the space, she had limited knowledge of how the rare disease ecosystem worked—from drug development to the inner workings of the NHS. But as the previous CEO of a disabled people's organisation, she was well-versed in progressive disability rights movements and the social model of disability, and as she transitioned into the rare disease space she observed some notable contrasts.

In the disability rights movement, Kirsty found the focus to be an identity and needs-based model. "Asking about someone's specific diagnosis or impairment was often irrelevant, as the emphasis was on supporting their overall needs and ensuring accessibility." However, when Kirsty entered the rare disease space, she was struck by how the narrative was very different. The first things people talked about were diagnosis, "burden," and parental needs—it was very focused on the medical aspects and the impact on caregivers, rather than the lived experiences of those with rare conditions.

Kirsty also noticed that the rare disease community seemed to be more competitive, with patient organisations vying for funding and influence. This contrasted with the more collaborative approach she was used to in the disability rights movement. Additionally, Kirsty felt there was a "false narrative around patient centricity" in rare diseases—the reality being that commercial interests from pharmaceutical companies often took precedence over the true needs of the community. Whereas in the disability rights space, the focus was on identity, rights and ensuring accessibility, the rare disease landscape appeared to be dominated by a medical model centred on diagnosis, treatment and caregiver burden.

"I started seeing the 'GoFundMe' attitude with a rare diagnosis. There was this understandable 'must get a cure' focus which I hadn't seen previously. But drug development processes are long so families need to be supported to live their lives in the here and now" This stark difference prompted Kirsty and her organisation to embark on their research project to truly understand what "living well" means for the rare disease community, with the goal of reframing the narrative and advocating for more holistic, person-centred support systems.

But Kirsty knows systemic change is needed to truly transform the landscape. That's why Metabolic Support UK are advocating for rare diseases to be recognised as a distinct protected characteristic under the Equality Act.

They also want the upcoming Rare Diseases Framework to include a dedicated, fifth priority on "living well", ensuring health and social care systems work in tandem to address the holistic needs of the community. "We're not rejecting the importance of treatments and cures," Kirsty clarifies. "But we have to acknowledge the reality that many rare diseases still lack effective therapies. In the meantime, we have to empower people to thrive despite their circumstances, not just survive them."

This shift in mindset is already resonating within the rare disease community, certainly in my personal household where my own son, Eddison, to me, exemplifies the concept of the "living well" ethos. Though he faces significant challenges due to his condition, xeroderma pigmentosum, he remains active, athletic and proud of his identity.

"When the sun goes down, I'm just like everyone else," he often recounts, but by day he lives by his own personal mantra of "push your limits and be the best you can—today." For Eddison it's about finding creative solutions, not just focusing on the barriers resulting from his condition."

This is the very heart of living well. Similarly, Kirsty's son William, who has developed a wonderful and fun Living Well hand gesture, demonstrates that in the face of the challenges of his condition of ectodermal dysplasia, living well and thriving within his personal constraints is possible. Thanks to creativity and resilience in abundance to overcome their challenges and a strong support system in their respective school and social settings, both boys are indeed thriving and wonderful role models for the concept.

What is xeroderma pigmentosum?

Xeroderma pigmentosum (XP) is a DNA repair disorder characterised by extreme sensitivity to ultra-violet (UV) light, which increases risk of developing skin and internal cancers. Many patients also present with progressive neurological impairment. There is no known treatment or cure for XP and disease management requires adoption of an adapted lifestyle for complete shielding and avoidance of natural and artificial sources of UV lighting.

Learn more at: actionforxp.org/what-is-xp.html

What is ectodermal dysplasia?

Ectodermal dysplasias are a diverse group of genetic disorders that involve defects of the hair, nails, teeth, skin and glands. Other parts of the body, such as the eyes or throat, may be affected as well. The combination of physical features a person has and the way in which it is inherited determines if it is an ectodermal dysplasia.

Learn more at: nfed.org/learn and edsociety.co.uk/what-is-ed/

Stories like Eddison's, William's and the million others like theirs are powerful reminders that one's quality of life is highly subjective, and we each have the power to determine what that means to us.

With the right support systems and societal understanding, the rare disease community can thrive. By shifting the narrative, this movement aims to ensure that opportunities to live our best life are not missed by our focus being so guided toward pursuit of therapies and cures—cures that for many will not materialise within our own lifetimes.

An action plan to drive change

1. **Shift the narrative from just cures and treatments to a holistic focus on quality of life.** The research by Metabolic Support UK found that accessing treatments was not the top priority for the rare disease community. Instead, they emphasised the importance of joy, employment, social connections, identity and feeling accepted by society. By reframing the conversation to prioritise these "living well" factors, we can better address the full spectrum of needs.

2. **Advocate for rare diseases to be recognised as a distinct protected characteristic under the Equality Act.** Currently, rare diseases are not explicitly included in frameworks like the Equality Act. Pushing for rare diseases to have this legal recognition would help ensure their specific needs are considered in policies, assessments and service provision.

3. **Integrate health and social care support through a dedicated "living well" priority in the Rare Diseases Framework.** The Rare Diseases Framework presents an opportunity to create a new priority focused on enabling the rare disease community to thrive, not just survive. This would help bridge the gap between medical care and the holistic support needed for quality of life.

4. **Revise disability benefit assessments to include rare disease-specific considerations.** Current assessment criteria often fail to capture the nuanced needs of the rare disease community. Updating these processes to better reflect the diverse challenges faced, beyond just physical mobility, would help ensure appropriate support is provided.

5. **Empower the rare disease community to understand and exercise their rights.** Many in the rare disease community are unaware of the legal frameworks and rights that could support them. Providing education and resources to navigate these systems would give them greater agency in advocating for their needs.

6. **Showcase positive, inspiring stories of "living well" with rare diseases.** By amplifying the experiences of those thriving despite their circumstances, we can counter the pervasive "disaster narrative" and demonstrate the possibilities when the right support systems are in place. The overarching goal is to shift the focus from just surviving a rare disease to enabling the community to truly live well and thrive, with their needs and identities at the centre of the conversation.

William pictured 'living well' and enjoying sports

Kirsty's vision is for Living Well to become a movement, backed up by William's empowering hand gesture, which the rare disease community will get behind and find great value in. Metabolic Support UK, in partnership with Beacon, will be hosting a Living Well Symposium in March 2025, shortly after Rare Disease Day. An event designed to be an "alternative Rare Disease Day" that brings together a wide range of stakeholders, including rare disease advocates and individuals, policymakers and government representatives, healthcare professionals, pharmaceutical companies and legal experts.

The symposium is an opportunity to help shape the Living Well movement, refine policy asks, and build a consortium of partners to drive systemic change. Attending this event is crucial for anyone interested in supporting the rare disease community in living well. In the lead-up to the symposium, Metabolic Support UK will be hosting a series of webinars to educate the rare disease community about disability rights, legal frameworks and strategies for living well. Signing up for these webinars is an important way to build knowledge and prepare for the symposium. Metabolic Support UK is actively seeking partnerships and support to amplify the Living Well message. Organisations can work with Metabolic Support UK to share stories and promote the campaign. By attending the symposium, engaging with the webinar series and collaborating with Metabolic Support UK, stakeholders can play a direct role in shaping the future of the rare disease community and ensuring they are empowered to live well—to thrive not just survive.

The overarching call to action is for the broader community to follow William and Eddison's lead by continuing **to live your own version of well today and every day.**

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LIVING WELL

SYMPOSIUM

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beacon for rare diseases

Living with a rare disease can mean being surrounded by disaster narratives focused on the 'burden' of our conditions. Let's change the conversation and live well NOW.

The Living Well symposium explores what it truly means to live well with a rare disease-join us and be part of shaping the future!

CLICK HERE TO REGISTER YOUR INTEREST

STOKE-ON-TRENT, 7TH OF MARCH, 2025