



best of business intelligence

The Chair's Award: Best Approach to Rare Diseases

Supported by:



"A lack of efficient treatment and accurate diagnosis for the majority of rare diseases represents an enormous unmet medical need. The BHBIA is proud of the work done by our members in generating insights that will help drive sustainable access to treatments for these patients. We are delighted to support this year's 'Chair's Award' which will allow you to showcase and celebrate your successes in this field."

Awarded for an approach to rare diseases (in general or a specific rare disease) that has had a positive impact on patient outcomes or the client's UK business

Highly Commended Entries:

Patient Evidence Collection to Support Access to Treatment



Alex Morrison
Rare Disease Research Partners



Marnie Ross
Rare Disease Research Partners

Supporting team:

- Samantha Wiseman**, Rare Disease Research Partners
- Sophie Thomas**, Society for Mucopolysaccharide and Related Diseases
- Vivienne Clark**, MLD Support Association UK
- Georgina Morton**, ArchAngel MLD Trust

Executive Summary:

When NICE evaluated the first treatment for metachromatic leukodystrophy, an ultra-rare disease, the patient community needed to collect robust evidence that demonstrated the true impacts of disease and its consequences on the quality of life of those affected and their families.

We conducted a collaborative study involving patients, parents and caregivers, patient organisations and clinical specialists to develop an evidence package that provided context to the clinical outcomes data under consideration and ensured the patient voice was comprehensively represented. NICE made a positive recommendation and gene therapy is now available for patients with early onset disease.