Ethical Legal and Social Issues in Rare Conditions Research and Clinical Practice (ELSI)

Rare Disease Research UK
Ethical Legal and Social Issues in Rare Conditions Research and Clinical Practice - ELSI

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Charity Partners: VOCAL, UNIQUE, Genetic Alliance UK

Background and Aims

The Ethical Legal and Social Issues in Rare Conditions Research and Clinical Practice (ELSI) Node is a partnership between researchers at The University of Manchester, University of Oxford, and Cardiff University. ELSI works with patients, families, clinicians, and academics to explore ethical, legal, and social issues related to advances in genetic technologies with five main aims:

- Improve the design of clinical trials for rare conditions (RCs).
- Develop proposals for good clinical practice.
- Understand the psychosocial impact of living with RCs.
- Identify barriers to effective mainstreaming access to genomic testing.
- Overcome these barriers & support delivery of equitable high-quality genome-based services.

Key Successes

- Strengthened RC clinical trials through systematic reviews and qualitative interviews.
- Built understanding of psychosocial and lived experiences via large-scale qualitative datasets.
- Addressed mainstreaming genomics through data collection & workshops.

Project 1: Impact & Outcomes in Clinical Trials

Ramona Moldovan, Tara Clancy, Faye Johnson, University of Manchester.

Key Project Aim:
To develop capacity for a meaningful set of core outcomes sets (COS) and patient reported outcome measures (PROs/PROMs) that capture the needs of patients and family members.

Key Updates:
- 20 qualitative interviews with parents/carers of children and one interview with a young person enrolled on clinical trials for rare genetic conditions conducted so far.
  - Identified significant psychosocial impact of participation in gene therapy clinical trials.
  - Parental insights could better inform the development of patient-centred clinical trials.
- Completed systematic review on PROs/PROMs in orphan clinical trials (Figure 1).
- A review of the Core Outcome Measures in Effectiveness Trials (COMET) database, containing 480 COS, found only 15 COS for 12 RCs, thus identifying a great need for improvement (2).
- Liaising with clinicians/investigators re qualitative interviews with patients (adults/children) and parents/carers on a wide range of clinical trials for various conditions.

2026 Pipeline Plans:
- Conducting qualitative interviews with adults enrolled on clinical trials for RCs and professionals working in clinical trials.
- Understand individuals who do not take part in, or withdraw from, clinical trials.
- Organise a consensus generating workshop to agree on a set of methods for the development of COS/PROMs for RCs in clinical trial design and policy decision making.

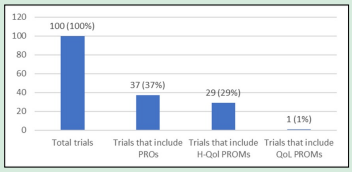
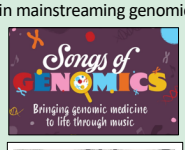


Figure 1. Overview of the quantity and quality of PROs and PROMs used in orphan trials (1). Review of COMET database identified a great need to improve RC COS.

Networking Activities

The ELSI Node has delivered a range of creative networking activities. Some highlights are:
- Presented ELSI work at over **20 external conferences**, including Genethics Forums, RDRUK, European Society for Human Genetics and British Society for Genetic Medicine events.
- Hosted the **1st ELSI Conference** (26th March 2025) (Figure 2).
- Delivered the **'Rare Diseases, Genomics and Justice' Workshop** funded by the Brocher Foundation (22nd–24th January 2025), Geneva, Switzerland. A multidisciplinary event featuring patient speakers that will lead to the publication of a set of academic papers.
- Organised the **'Songs of Genomics: A musical exploration of genomics in healthcare'**, in collaboration with HIVE Choir (4th November 2024), Oxford, UK (Figure 2).
- Produced **'A Day of Rare Conditions' short film** (29th February 2024) (Figure 2).
- Produced a series of **'Genetic Sounds' podcasts.**
- **'Barriers to Mainstreaming' Workshop** (19th November 2025). Provided a forum to identify gaps and address inconsistencies in mainstreaming genomic testing.



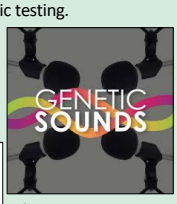


Figure 2. Overview of ELSI networking activities.

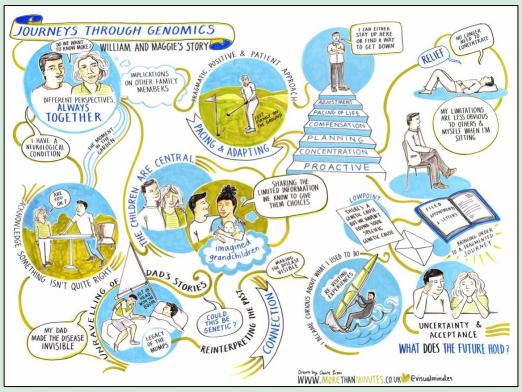
Project 2: Life with Undiagnosed Rare Conditions

Susie Weller, Anneke Lucassen, Rachel Horton, Naomi Marshall, University of Oxford.

Key Project Aim:
To understand the experiences of those living without a diagnosis and consider ways in which people and families could be better supported.

Key Updates:
- Ethical approval received.
- Completed the sourcing and collation of datasets for qualitative secondary analysis.
- Presentations relating to the substantive focus (e.g. *Festival of Genomics and Biodata, Excel*, London, January 2026) and methodological approach (e.g. *Computational Social Science Meets Qualitative Research Workshop*, November 2025; *The Pew Charitable Trusts/DC-AAPOR Washington DC*, November 2025).

2026 Pipeline Plans:
- Large-scale 'big qual' secondary analysis of around 150 interview transcripts.
- Workshops attended by key individuals and support organisations.
- New empirical research with ~20 families 'living in limbo'.
- Synthesis of qualitative secondary analysis and new empirical work .



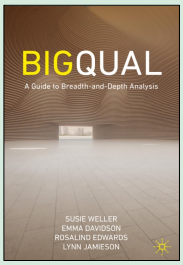


Figure 3. Artistic representation of patient perspectives on RCs. The 'Big qual' textbook presents a new approach to large scale qualitative analysis.

Project 3: Mainstreaming Genomic Investigations

Angus Clarke, Shane Doheny, Cardiff University

Key Project Aim:
To research the ethical, communication and counselling aspects of incorporating genome-based investigations into the practice of mainstream specialties.

Key Updates:
- Ethics application completed with 4 active recruitment sites.
- 160+ lab reports collected, 50+ interviews held (clinical professionals & support groups).

2026 Pipeline Plans:
- Conduct small number of focus groups with clinicians in key clinical professions in early 2026.
- Contribute to European Society of Human Genetics (ESHG) process for developing its strategy report in relation to the mainstreaming of genomics.

Patient and Public Involvement and Engagement

ELSI connects individuals and empowers patient voices by ensuring all our activities feature presentations and forums delivered by patients. Some of our biggest PPIE achievements are:

- Hosted **'Newborn Screening Forum'**, Centre for Personalised Medicine, Oxford (25th February 2026). Discussed current newborn screening programmes and improvements.
- Published **'A Day of Discussion' online article** which provided a summary of the 1st ELSI Conference and reflected on the key themes, priorities and challenges of RC clinical trials.
- Presented at **'Wales Genomics Café'** (2nd October 2025). This online event was organised by the Wales Gene Park and had over a 100 researchers and patient group representatives attend.
- Contributed to **'Involvement by Design' Report** published by Genetic Alliance UK with RDRUK and LifeArc. Explored why PPIE remains difficult to deliver in research and set out 8 recommendations for change (Figure 4).
- Delivered **patient-led plenary session** at 1st ELSI Conference.

Figure 4. Involvement by Design report.

Publications

Scan QR code above for full list of 8 publications

(1) Ciuca, A., Banka, S., Clancy, T., Jones, S., Kirkham, J. J., Newman, W. G., Payne, K. & Moldovan, R. 2025. Patient-reported outcomes and measures are under-utilised in advanced therapy medicinal products trials for orphan conditions. *J Clin Epidemiol*, 178, 111617.
(2) Ciuca, A., Banka, S., Newman, W. G., Moldovan, R. & Kirkham, J. J. 2023. Assessment highlights need for improvement in standards of development of core outcome sets for rare genetic diseases. *J Clin Epidemiol*, 161, 84-93.

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