

Background:

- Genomic technologies hold great promise to bring benefits for individuals and families living with rare conditions (RC) in terms of diagnosis and treatment
- **Ethical Legal and Social Implications (ELSI)** have received considerable attention yet their **integration into RC research has lagged behind**
- **Our Vision** is to **integrate knowledge, build capacity and drive innovation** in ELSI cross-cutting issues ranging from referral pathways, to the psychosocial impact of RC and the design of clinical trials for treatments of RC

Our aims:

- **Build a national ELSI powerhouse** to serve the needs of the UK RDRP
- **Accelerate research** in areas of long-standing concern for RC

Our priorities:

- **Understand families' hopes and expectations** for diagnosis and treatment
- **Explore the lived experience** of RC patients without a diagnosis
- **Improve therapeutic trial design** for RC
- **Train specialists** in mainstreaming specialties

Our projects:

Project 1. Impact and Outcomes in RC clinical trials, Lead: UoM

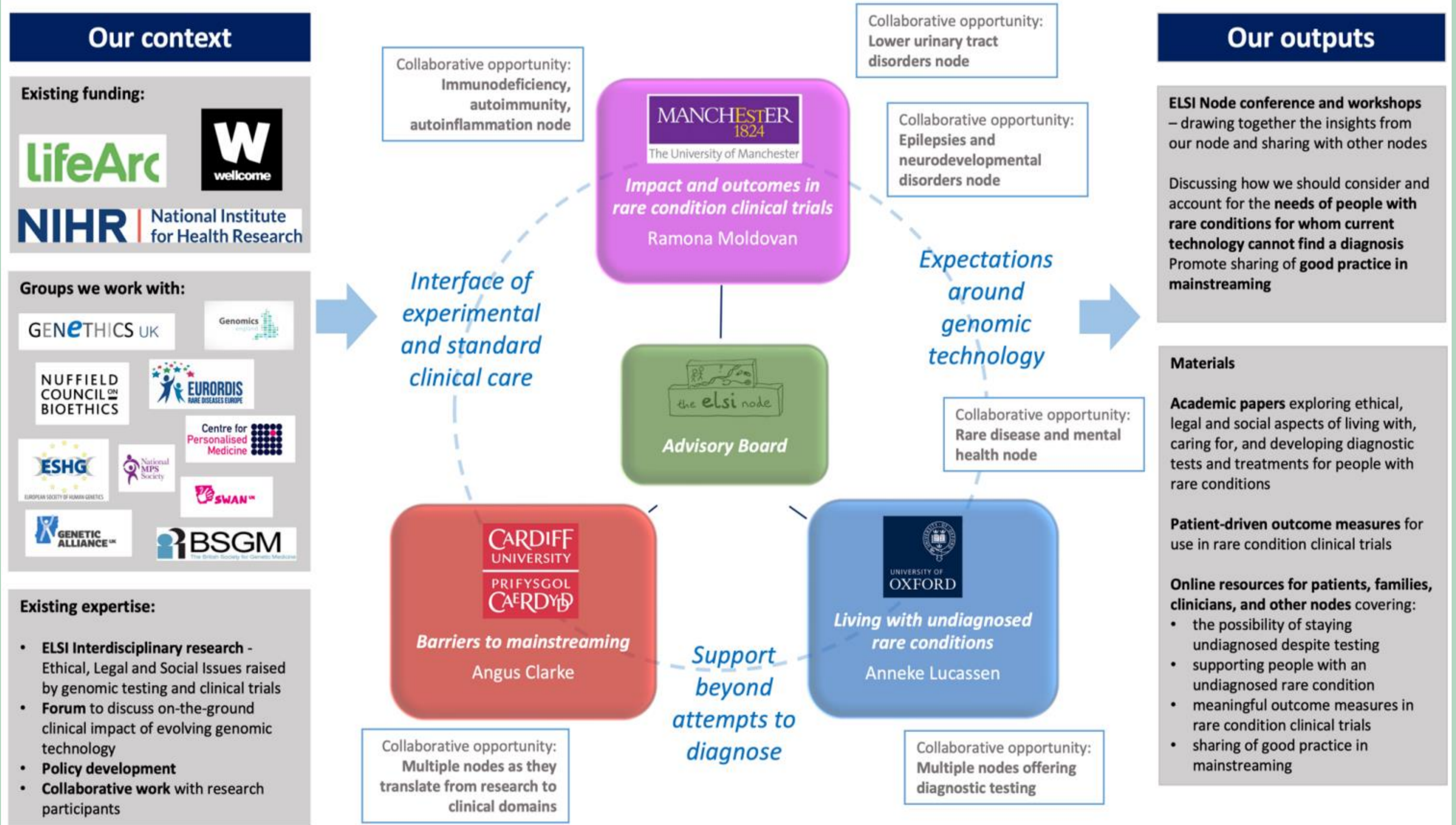
- Understand Patient Reported Outcomes (PROs) & Patient Reported Outcome Measures (PROMs) used in RC research
- Build capacity for developing Core Outcome Sets (COS) in RC

Project 2. Living in limbo: life with an undiagnosed RC, Lead: UO

- Understand the experiences of those living with an undiagnosed RC
- Develop resources for patients, families and clinicians

Project 3. Barriers to Mainstreaming Genome-based Investigations Across Specialties, Lead: CU

- Identify ELSI barriers to mainstreaming of genome-based investigations
- Identify ELSI training needs to incorporate mainstream genomics into clinical specialties



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