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An integrative multi-omic approach to expanding rare disease research in Northern Ireland: crystal award

Kerr, K., McAneney, H., Smyth, L., Bailie, C., McKee, S., & McKnight, A. J. (2020). *An integrative multi-omic approach to expanding rare disease research in Northern Ireland: crystal award*. Poster session presented at Rare Disease Day 2020: Joint North South Event, Belfast, Belfast.

Document Version:

Publisher's PDF, also known as Version of record

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An Integrative Multi-omic Approach to Expanding Rare Disease Research in Northern Ireland

Katie Kerr, Helen McAnaney, Laura Smyth, Caitlin Bailie, Shane McKee, AJ McKnight

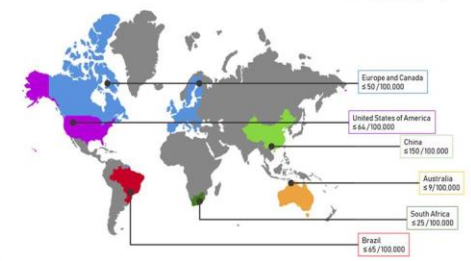
Queen's University, Belfast

AN INTEGRATIVE MULTI-OMIC APPROACH TO EXPANDING RARE DISEASE RESEARCH IN NORTHERN IRELAND

Katie Kerr, PhD Student, Queen's University Belfast.



Definitions of a rare disease vary globally, but in Europe we define rare diseases as one which affects fewer than 5 in 10,000 people.

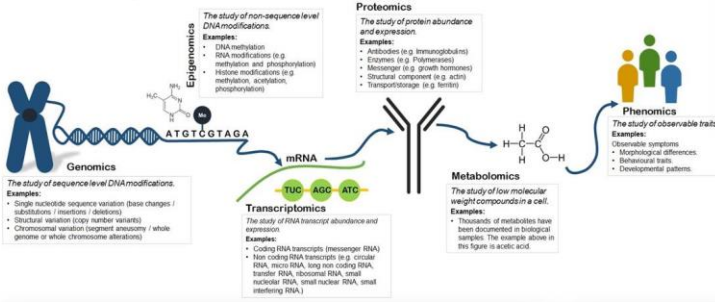


- In the UK, 1 in 17 people are affected by a rare disease. In NI more than 110,000 people have a rare disease, that's the population of the city of Derry/Londonderry!
- The 100,000 Genomes Project sequenced the entire genome of approx. 72,000 rare disease patients and their family members. This provided a diagnosis for up to 50% of patients.



But what about the other 50%?

Multi-omics may provide additional insights for rare diseases



Research Aim

To improve rare disease research and diagnosis for patients living in Northern Ireland by development of a frame-work for studies of multi-omics and rare disease.



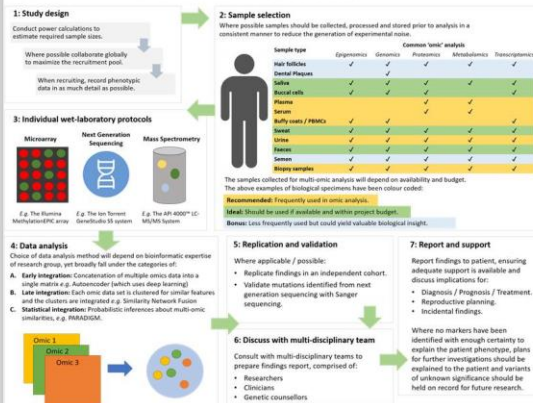
Scoping review of multi-omics and rare disease

Sixty-six full text articles included across 13 multi-omic combinations.

Key findings:

- Technologies advances have caused an explosion of multiomic research
- Multi-omics can be used to identify diagnostic and prognostic biomarkers
- Multi-omics can identify drug repurposing opportunities
- There is a need for a standardised approach to improve study quality

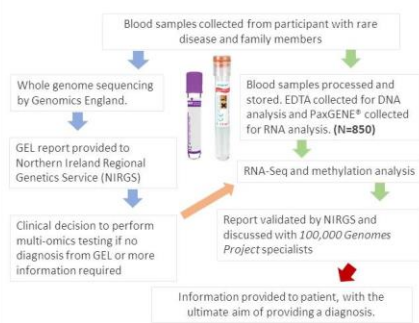
Frame-work for multi-omic rare disease studies...



K. Kerr, H. McAnaney, U. Smyth, C. Bailie, S. McKee, A.J. McKnight. *The multi-omic landscape of rare diseases: a scoping review.* Under review, January 2020.

Multi-omics in the 100,000 Genomes Project

1. Multi-omic analysis of undiagnosed participants:



2. Report on variant analysis and interpretation to identify areas for improvement (e.g. IT pathways) compared to other regional genetics hubs across the UK.

3. Evaluation of patients, carers and healthcare providers perspectives on multi-omics (surveys/interviews)

This research is supported by a Department for the Economy Co-operative Awards in Science and Technology (DfE-CAST) studentship award, the NI Kidney Research Fund, and the SFI-DfE Investigator's program (15/IA/3152).

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