

32**Reflections on developments from our Northern Ireland Rare Disease Implementation Plan 2015-2020: helping patients get a final diagnosis and optimised care faster**

Prof. Amy Jayne McKnight¹, on behalf of the NIRDFIG²

¹ Queen's University Belfast, Belfast, United Kingdom. ²Northern Ireland Rare Disease Framework Implementation Group, Belfast, United Kingdom

Abstract

Rare diseases are a major public health concern, cumulatively affecting ~6% of the population. In 2015 the Northern Ireland rare disease implementation plan (NIRDIP)[[1](#)] was published, describing 51 commitments. Key developments for molecular services included:

- The NI Genomic Medicine Centre was funded to facilitate people from NI participating in the 100,000 genomes project (<https://tinyurl.com/UK100KGP>), gaining whole genome sequencing and improved diagnostic yield.
- Local bioinformatics architecture was developed - GenOCEANIC (Genomics Open Core Engine for Accelerating Northern Ireland Care), with patient experience measures and views from participating healthcare professionals supporting the development of genomic medicine for NI.
- Access to highly specialised treatments has been facilitated for multiple patients with specific molecular diagnoses.
- The development of ENCOMPASS introducing a digital integrated care record for Northern Ireland has significant potential to improve rare disease patient care, with customised training for data coding and input of phenotypic information critical to maximise diagnoses.
- An evolving programme of local workforce development, including CPD accredited events, helps health and social care professionals use genomics and bioinformatics.
- Harmonised rare disease molecular teaching in Queen's University Belfast and Ulster University includes patient voice sessions.

The legacy from our NIRDIP includes the foundation of an All-Ireland Rare Disease Research Network. A knowledge exchange workshop was held in 2020 with multidisciplinary stakeholders to help prioritise recommendations for rare disease progress across NI.

[[1](#)] Department of Health, Social Services and Public Safety. Providing high quality care for people affected by rare diseases – the Northern Ireland implementation plan for rare diseases. <https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/ni-rare-diseases-implementation-plan-oct-2015.pdf>.