

R031: Myotonic dystrophy in Northern Ireland – QUB

Myotonic Dystrophy (DM) is an inherited condition which affects multiple body systems and is associated with muscle wasting. Accumulating evidence suggests that patients with DM have an increased risk of developing cancer.

Northern Ireland offers a unique environment in which to undertake an investigation of the reported associations between DM and cancer as information is available for the entire country. Additionally, Northern Ireland has population-based registers of pre-cancerous conditions that are established or in development in addition to registers of all non-melanoma skin cancer cases which are not captured by most cancer registries.

We plan on developing a population-based database of DM patients which will be linked within the Honest Broker Service to the Northern Ireland Cancer Registry for details on premalignant and cancer diagnoses (date of diagnosis, site, morphology, topography) and to the General Registrar Office death files for date and cause of death. We will also carry out a genome wide association study (GWAS) on pre-collected DM patient samples. The findings from the GWAS study will be linked to the DM database and through meta-analytic approaches combined with other global cohorts; this may enable the identification of genetic susceptibility to cancer in DM patients.

If our study reveals higher incidence of specific cancers in DM patients, screening might be considered to enable earlier detection and treatment to improve prognosis. In future, we plan on using the Northern Ireland Biobank to investigate the molecular and pathological nature of cancers that develop in DM patients, compared to cancers occurring in the general population in order to determine prevention strategies.