Long-read sequencing sheds new light on the genomic basis of Indigenous MJD

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**ABSTRACT:**

New technologies for ‘long-read’ DNA sequencing enable efficient, accurate analysis of repetitive genes and genomic structural variation, including short tandem repeat (STR) expansions. These technologies promise to shed new light on our understanding of genetic diseases where repetitive DNA is implicated, and to improve their diagnosis. In this talk, I will first outline our recent efforts to harness long-read sequencing to streamline and improve the diagnosis of patients with STR expansion disorders, including Machado Joseph Disease (MJD) and other hereditary ataxias. To ensure the benefit of these new methods may extend to Australians of Indigenous – as well as non-Indigenous – ancestry, we are currently working with the National Centre of Indigenous Genomics (NCIG) to characterise genomic variation among Indigenous communities around the country. The aim is to establish a base layer of genomics resources to inform genomic medicine applications suitable for Indigenous Australians. I will describe how we are using long-read sequencing data from communities affected by MJD to better characterise the genomic basis, inheritance and population history of MJD in an Indigenous Australian context.