

The Leading Strand Educational Webinar Series



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Diagnostic Strategies in Genetic Neuromuscular Diseases

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Host: Dr. Jim Dowling

Register in advance for this webinar: https://us06web.zoom.us/webinar/register/WN_vXOheKQmTnOAG2Mt1BZbJw

Genetic neuromuscular diseases are complex multisystem disorders, representing hundreds of rare diseases in Canada. Genetic neuromuscular diseases can lead to disability and early death through progressive muscle weakness and cardiac and respiratory failure. Next-generation sequencing approaches, including genome sequencing and RNA sequencing, offer hope for a faster and accurate genetic diagnosis, but a more detailed understanding of muscle and nerve involvement is often required to interpret these tests. We will review the combination of genomics/multi-omics techniques with pathology and cutting-edge imaging techniques to facilitate appropriate sample selection for muscle biopsy and RNA-Sequencing, delineate new disease patterns, and novel gene discovery. The combination of multi-omics with novel imaging techniques integrates these innovative approaches into a diagnostic and discovery pipeline for rare NMDs.