Conférence

CRCHUM

Université de Montréal

AXE NEUROSCIENCES

CONFÉRENCIÈRE

Zoom : https://us06web.zoom.us/j/89905079375?pwd=44bpP7si1I1XC6qUlsbsAG8wk5Y3f2.1

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Navigating the Labyrinth of Genetic Ataxias in the Era of Next Generation Sequencing

Childhood-onset cerebellar atrophy is characterized by considerable clinical and genetic heterogeneity, with over 340 genetic conditions associated with cerebellar atrophy listed in the OMIM (Online Mendelian Inheritance in Man) database. A broad-based approach to genetic testing which incorporates clinical and imaging information is often required to confirm the diagnosis. Next-generation sequencing approaches such as whole exome sequencing (WES) are widely available, however the diagnostic rate of WES for our Canada-wide study of patients with cerebellar atrophy was 53% compared with 47% for patients who received gene panels and standard metabolic testing. This may be attributed in part to the high incidence of variants of uncertain significance (VUS), which often require follow-up molecular and functional analyses to aid VUS interpretation. This highlights the need for collaboration between clinicians and basic scientists, and a multi-modal approach to diagnosing patients with complex neurological presentations.

Le vendredi 1er décembre 12 h à 13 h

Amphithéâtre du CRCHUM, R.05.212A/B
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L'AUDACE DE CHERCHER PLUS LOIN

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