GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

Clinical assessment and investigations of an individual with epilepsy including when to consider a genetic etiology.

Initial evaluation of an individual with epilepsy

Clinical assessment

- Epilepsy
  - age at onset, seizure types, known triggers, response to treatment
- Developmental profile
  - developmental delays, intellectual disability, autism, behavioural problems, regression, fluctuating course
- Other neurological features
  - movement disorders, tone abnormalities, hearing or vision disturbances
- Systemic involvement: heart, kidney, liver anomalies
- Unusual facial features or skin findings
- Relevant family history

Investigations

- MRI Brain (Epilepsy Protocol)
- Video-EEG
- Metabolic screening of blood, urine, skin
- and/or CSF (if features are suggestive)

Indication to consider a genetic cause for epilepsy

- Associated developmental delay, intellectual disability, plateauing or regression
- Associated features like dyskinesias, ataxias, or migraines
- Associated dysmorphic features or multiple congenital anomalies
- Refractoriness to medical management (with no apparent acquired cause)
- Presence of familial epilepsy, defined as at least 2 first-degree relatives on the same side of the family with related epilepsy syndromes, unless the epilepsy syndrome is benign
- Features suggestive of an inborn error of metabolism
- Developmental and/or epileptic encephalopathy
- Distinctive patterns of malformations of cortical development identified on neuroimaging studies
- Poor prognosis based on clinical and EEG findings or high likelihood of lethal outcome

Individuals with the following types of epilepsy *do not* require genetic testing:

- Isolated self-limited focal epilepsy
- Typical idiopathic generalized epilepsy (e.g. childhood absence epilepsy, juvenile myoclonic epilepsy)
- Isolated mesial temporal lobe epilepsy with hippocampal sclerosis
- A clearly acquired epilepsy (e.g. occurring secondary to head trauma, CNS infection, hypoxia)

June 2026