Genetics of Epilepsy

Initial Evaluation of an Individual with Epilepsy[1]

Clinical Assessment

- Epilepsy: age at onset, seizure types, known triggers, response to treatment
- Developmental profile: developmental delay, 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

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Investigations to Consider

EEG
MRI Brain (Epilepsy Protocol)
Metabolic screening (if features are suggestive)

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Red Flags for epilepsy with a possible genetic cause

- Associated developmental delay, intellectual disability, plateauing or regression
- Associated features such as 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 a similar epilepsy
- 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

Epilepsies that do not require genetic testing

- Benign focal epilepsy
- Typical idiopathic generalized epilepsy
- Isolated mesial temporal lobe epilepsy with hippocampal sclerosis
- Clearly acquired epilepsy
- Simple febrile seizures

For a concise, evidence-based resource on the genetics of epilepsy including benefits and limitations of genetic testing, please see the GEC-KO *on the run*. For a more comprehensive review and complete reference list please see the GEC-KO Messenger.

[1] This evaluation tool was adapted from Jain P, Andrade D, Donner E, Dyment D, Prasad AN, Goobie S, et al. Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Can J Neurol Sci. 2018; 00:1-7. doi:10.1017/cjn.2018.341.

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