WHEN SHOULD WE REQUEST A GENETIC TEST IN CHILDHOOD EPILEPSIES, AND WHICH TEST?

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Learning Objectives

- To be aware of the broad spectrum of genetic causes of childhood epilepsies.
- To appreciate genetic counselling considerations before and after genetic testing.
- To know the different genetic testing methods available, and their limitations.
- To be able to decide which test should be performed in which childhood epilepsy type.

Key Messages

- Epilepsies with a monogenic cause are currently the primary target for diagnostic genetic testing
- Key indicators for genetic testing (after exclusion of acquired etiologies)
  - Concomitant neurodevelopmental problems, or other systemic comorbidities
  - Brain cortical malformations
  - PME
  - Phenotype consistent with a specific familial epilepsy syndrome (eg. BFNE, BFIS, FFEVF,…)
  - (Non-lesional treatment resistant, during pre-surgical workup?)
- For most genetic epilepsy disorders, wide genetic heterogeneity has been described
- Therefore, NGS strategies are currently recommended as the first line of testing
- Testing for CNVs, if not already part of the NGS analysis, should be considered next
  - Exception: multisystem disorders suggestive of multi-gene pathology
- Genetic re-evaluation should be undertaken for patients with suspected genetic epilepsy without a genetic diagnosis
  - Re-analysis of past sequencing data
  - Consideration of new testing based on availability of new or more sensitive testing strategies

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