Epilepsy
Genetic Testing

Help your patients find answers

About half of all patients with epilepsy have an identifiable genetic component, including up to 40% of those previously thought to have idiopathic disease.[1] Genetic testing enables diagnosis in many patients and can explain comorbidities, inform prognosis, and define risks to relatives and future pregnancies. Importantly, genetic findings can impact disease management and may influence treatment choices.

Choose the test panel that's right for your patient

Comprehensive Epilepsy Gene Panel (69 genes):
The majority of known genetic etiologies for epilepsy in a single test

The 69 genes in this panel were chosen using guidance from both clinical geneticists and neurologists, and are based on disorders for which epilepsy is a key feature according to the Online Mendelian Inheritance in Man (OMIM) database. The included genes cover a wide phenotypic spectrum, allowing for the evaluation of multiple heritable conditions that present with seizures. Consider using this test for patients with non-specific clinical/EEG findings and a poor prognosis.

Management Impact Epilepsy Gene Panel (16 genes):
Targeted panel of epilepsy genes associated with treatable inborn errors of metabolism

The 16 genes in this panel are associated with treatable metabolic disorders in which seizures can occur (e.g. pyridoxine dependent epilepsy, folic acid responsive seizures, pyridoxal phosphate dependent epilepsy, creatine deficiency syndromes, glucose transporter 1 (GLUT1) deficiency, cerebral folate deficiency). This testing can be considered for epilepsy patients with clinical features suggestive of an inborn error of metabolism, or with any of the following:

- Family history of metabolic disorder associated with epilepsy
- Parental consanguinity
- A "metabolic" epilepsy biochemical marker identified by newborn or metabolic screening

Epilepsy panel genes

Comprehensive Epilepsy Panel: *ALDH7A1, AMT, ARX, ASAH1, ATP1A2, ATP1A3, CDKL5, CERS1, CHD2, CHRNA7, CNTNAP2, CSTB, DNM1, DOCK7, EPM2A, FOLR1, FOXG1, GAMT, GATM, GLDC, GOSR2, GRIN2A, GRIN2B, HCN1, KCNC1, KCNJ10, KCNJ11, KCNQ2, KCNQ3, KCNT1, KCTD7, LMNB2, MBD5, MECP2, MEF2C, MOCS1, NECAP1, NEU1, NHLRC1, NRXN1, PCDH19, PHGDH, PLCB1, PNKP, PNPO, POLG, PRICKLE2, PRRT2, PSAT1, PSPH, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SLC6A8, SLC9A6, SPTAN1, STXBP1, SUOX, SYNGAP1, TBC1D24, TCF4, TSC1, TSC2, UBE3A, ZEB2.*

Management Impact Epilepsy Panel: *ALDH7A1, AMT, FOLR1, GAMT, GATM, GLDC, MOCS1, PHGDH, PNPO, POLG, PSAT1, PSPH, SCN1A, SLC2A1, SLC6A8, SUOX.*

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State-of-the-art testing provides results you can trust

The Epilepsy Gene Panel tests, developed by Dr. Bekim Sadikovic and the molecular genetics team at the London Health Sciences Centre (LHSC), provide highly accurate assessment of both sequence and copy number changes in genes associated with monogenic and polygenic epileptic disorders. Test features include:

- Full-gene sequencing and deletion/duplication analysis using next-generation sequencing technology (NGS) [2,3]
- >99.9% detection of described sequencing and deletion/duplication mutations in included genes, when present (analytic sensitivity)
- Reportable variants confirmed by alternate method, such as Sanger sequencing or MLPA
- Free parental testing for co-segregation studies of variants of uncertain significance (VUS)
- Interpretation by molecular genetics team at LHSC, led by Dr. Bekim Sadikovic
- ISO 15189 Plus, CAP-accredited, OLA-accredited and CLIA-certified testing laboratory

Ordering: Please visit impactgenetics.com for details

Turnaround time: 4-7 weeks

REFERENCES

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2. Kerkhof J, Schenkel LC, Reilly J, McRobbie S, Aref-Eshghi E, Stuart A, Rupar CA, Adams P, Hegele RA, Lin H, Rodenhiser D, Knoll J, Ainsworth PJ, Sadikovic B. *J Mol Diagn*. 2017; 19(6):905-920. PMID: 28818680
3. Schenkel LC, Kerkhof J, Stuart A, Reilly J, Eng B, Woodside C, Levstik A, Howlett CJ, Rupar AC, Knoll JHM, Ainsworth P, Waye JS, Sadikovic B. *J Mol Diagn*. 2016; 18(5):657-667. PMID: 27376475