NeuroSURE℠: EPILEPSY GENE PANEL

Many **patients with epilepsy have an identifiable genetic component,** including up to 40% of those previously thought to have idiopathic disease.[1]

Genetic testing for epilepsy enables diagnosis in many patients and can explain comorbidities, inform prognosis, and define risks to relatives and future pregnancies. Genetic insights can impact disease management and may influence treatment choices.[1]

NeuroSURE℠: Epilepsy Gene Panel (481515)

This assay was developed through an industry-academic collaboration that used guidance from clinical geneticists, neurologists, scientific literature, and curated online genetic databases in which epilepsy is a key feature to assemble a panel of 69 genes. 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/electroencephalogram (EEG) findings and a poor prognosis.

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.*

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

16 genes in this panel are associated with treatable metabolic disorders in which seizures can occur and may directly impact patient management. This test can be helpful for epilepsy patients with infantile onset, epilepsy refractory to treatment, epilepsy plus developmental delay, or families that may choose to have prenatal testing in future pregnancy.

Clinical Disorder	Genes
Pyridoxine dependent Seizures[1-4]	*ALDH7A1*
Nonketotic hyperglycinemia[1]	*AMT, GLDC*
Cerebral folate deficiency[2,3,5]	*FOLR1*
Creatine deficiency syndromes[1,3,4,5]	*GAMT, GATM, SLC6A8*
Molybdenum cofactor deficiency[1]	*MOCS1*
Serine Biosynthesis (Deficiency) Disorders[3, 4]	*PHGDH, PSAT1, PSPH*
Pyridoxal Phosphate Deficiency[2,3,5]	*PNPO*
Alpers-Huttenlocher Syndrome[1,4,5]	*POLG*
Dravet Syndrome[1,2]	*SCN1A*
Glucose 1 Transporter Deficiency[1-5]	*SLC2A1*
Sulfite Oxidase Deficiency[5]	*SUOX*

LabCorp

State-of-the-art testing

The NeuroSURE℠: Epilepsy Gene Panel provides highly accurate assessments 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).
- >99.9% detection of described sequencing and deletion/duplication mutations in included genes, when present (analytic sensitivity).[6,7]
- Reportable variants confirmed by alternate method (Sanger sequencing or MLPA).
- Parental testing for co-segregation studies of variants of uncertain significance (VUS) is included.
- Impact Genetics is ISO 15189 Plus, CAP-accredited, OLA-accredited and CLIA-certified testing laboratory.
- Insurance prior verification with patient consent for out of pocket expenses eases the workload in your office.

LabCorp benefits

- 1900 patient service centers nationwide available for specimen collection; courier services are available for in-office specimen pickup.
- Genetic counselors are available for result interpretation. To schedule an appointment call 855-GC-CALLS or 855-422-2557.

References

1. Pong AW, Pal DK, Chung WK. Developments in molecular genetic diagnostics: an update for the pediatric epilepsy specialist. *Pediatr Neurol.* 2011;44:317–327.
2. Ream M. A. & Patel A. D. Obtaining genetic testing in pediatric epilepsy. *Epilepsia,* 2015; 56(10) :1505-1514.
3. Tein, I. Vitamin and cofactor responsive encephalopathies and seizures. JICNA. 2015(15);105:1-10. Available at: http://jicna.org/index.php/journal/article/view/2/2015-105-pdf. Date accessed March 15, 2018.
4. Sharma S., Prasad A.N. Inborn errors of metabolism and epilepsy: Current understanding, diagnosis, and treatment approaches. *Int. J. Mol. Sci.* 2017;18:1384.
5. Rahman S., Footitt E.J., Varadkar S., Clayton P.T. Inborn errors of metabolism causing epilepsy. *Dev. Med. Child Neurol.* 2013;55:23–36.
6. 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.
7. 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.

Please direct any questions regarding this test to Impact Genetics at **877-624-9769.**

Test Name	Test No.
NeuroSURE℠: Epilepsy Gene Panel	**481518***

Epilepsy-related Testing	
Test Name	**Test No.**
Amino Acid Profile, Glycine Encephalopathy, Plasma and Cerebrospinal Fluid	700200
Amino Acid Profile, Quantitative, Cerebrospinal Fluid	700180
Carbamazepine, Serum or Plasma	007419
Carbamazepine-10,11 Epoxide	716803
Coenzyme Q10, Total	120251
Creatine	002402
Creatine, 24-Hour Urine	003475
Ethosuximide, Serum or Plasma	007443
Felbamate, Serum or Plasma	716530
Folate (Folic Acid)	002014
Gabapentin, Serum or Plasma	716811
Glucose	001032
Glucose, Cerebrospinal Fluid	002048
Lacosamide	007012
Lamotrigine, Serum or Plasma	716944
Levetiracetam, Serum or Plasma	716936
Oxcarbazepine, Serum or Plasma	716928
Phenobarbital, Serum or Plasma	007823
Phenytoin, Serum or Plasma	007401
Pregabalin, Urine	809623
Primidone, Serum or Plasma	007856
Topiramate, Serum or Plasma	716285
Uric Acid	001057
Valproic Acid, Serum or Plasma	007260
Vitamin B12	001503
Vitamin B6, Plasma	004655
Zonisamide, Serum or Plasma	007915

* Testing performed at Impact Genetics Inc. (Ontario, Canada), a wholly-owned subsidiary of DynaCare Gamma Laboratory Partnership, a subsidiary of Laboratory Corporation of America Holdings.

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