OEGTP - EPILEPSY TEST REQUISITION

LAB USE ONLY:

Received date:

Notes:

PATIENT INFORMATION:

Name:

Address:

Date of Birth: YY/MM/DD

Sex: M F

Health Card No:

TEST REQUEST:

See page 2 for gene list for each of the panels below

- Epilepsy Comprehensive panel: 167 genes
- Focal Epilepsy panel: 14 genes
- Progressive Myoclonic Epilepsy panel: 20 genes
- Early Infantile Epilepsy panel: 51 genes
- Childhood Onset Epilepsy panel: 45 genes
- Brain Malformation Epilepsy panel: 44 genes
- Actionable Gene Epilepsy panel: 22 genes
- Single gene test:

Carrier Testing/ KnownFamily Mutation

Name of index case in the family (include copy of report)

Affected Unaffected

Date of Birth: Relationship to patient:

Gene: RefSeq:NM

Mutation:

SAMPLE COLLECTION:

Date drawn: YY/MM/DD

EDTA blood (lavender top) (5ml at room temp)

REASON FOR REFERRAL:

Diagnostic Testing

Clinical Diagnosis:

Clinical Presentation:

REFERRING PHYSICIAN: Authorized Signature is Required

Physician Name (print):

Signature: Email:

Clinic/Hospital:

Address:

Telephone: Fax:

CC report to:

Name:

Address:

Telephone: Fax:

Molecular Genetics Laboratory
Victoria Hospital, Room B10-123A
800 Commissioners Rd. E.
London, Ontario | N6A 5W9
Ph: 519-685-8122 | Fax: 519-685-8279

London Health Sciences Centre
St Joseph's Health Care London
Pathology and Laboratory Medicine

OEGTP - EPILEPSY TEST PANELS

Patient Identifier:

COMPREHENSIVE EPILEPSY PANEL: 167 Genes

ACTB, ACTG1, ADSL, AKT3, ALDH7A1, AMT, AP3B2, ARFGEF2, ARHGEF9, ARV1, ARX, ASAH1, ASNS, ATP1A3, ATP6V0A2, ATP7A, ATRX, B3GALNT2, B3GNT1(B4GAT1), CACNA1A, CAD, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CPA6, CSTB, CTSD, DCX, DENND5A, DEPDC5, DNAJC5, DNM1, DOCK7, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EPM2A, FGF12, FKRP, FKTN, FLNA, FOLR1, FOXG1, FRRS1L, GABRA1, GABRB3, GABRG2, GAMT, GLDC, GMPPB, GNAO1, GOSR2, GPR56 (ADGRG1), GPSM2, GRIN2A, GRIN2B, GTDC2(POMGNT2), HCN1, HNRNPU, ITPA, KANSL1, KATNB1, KCNA2, KCNB1, KCNC1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7(CLN14), KDM5C, KIAA1279(KIF1BP), KIF2A, LAMA2, LARGE, LGI1, LMNB2, MBD5, MDH2, MECP2, MEF2C, MFSD8, MOCS1, NDE1, NEU1, NHLRC1, NPRL2, NPRL3, NRXN1, OCLN, PAFAH1B1, PAK3, PCDH19, PHF6, PHGDH, PIGA, PLCB1, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PPT1, PRICKLE1, PROSC(PLPBP), PRRT2, PSAT1, PSPH, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RELN, ROGDI, RTTN, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SGCE, SGK196(POMK), SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP29, SPTAN1, SRD5A3, STX1B, STXBP1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TPP1, TSC1, TSC2, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, UBA5, UBE3A, VLDLR, WDR62, WWOX, YWHAG, ZEB2

FOCAL EPILEPSY PANEL: 14 Genes

CHRNA2, CHRNA4, CHRNB2, CPA6, DEPDC5, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PRRT2, SCN1A, SCN1B, SLC2A1

PROGRESSIVE MYOCLONIC EPILEPSY PANEL: 20 Genes

ASAH1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, EPM2A, GOSR2, KCNC1, KCTD7(CLN14), LMNB2, MFSD8, NEU1, NHLRC1, PPT1, PRICKLE1, SCARB2, SGCE, TPP1

EARLY INFANTILE EPILEPSY PANEL: 51 Genes

AP3B2, ARHGEF9, ARV1, ARX, CACNA1A, CAD, CDKL5, CHD2, DENND5A, DNM1, DOCK7, EEF1A2, FGF12, FRRS1L, GABRA1, GABRB3, GNAO1, GRIN2A, GRIN2B, HCN1, HNRNPU, ITPA, KCNA2, KCNB1, KCNQ2, KCNT1, MDH2, MECP2, MEF2C, PCDH19, PIGA, PLCB1, PNKP, PRRT2, SCN1A, SCN2A, SCN8A, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC35A2, SPTAN1, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, UBA5, WWOX, YWHAG

CHILDHOOD ONSET EPILEPSY PANEL: 45 Genes

ADSL, ARX, ATP1A3, ATRX, CDKL5, CHD2, CNTNAP2, DEPDC5, DNAJC5, DYRK1A, EHMT1, FOXG1, GABRG2, GRIN2A, KANSL1, KCNJ10, KCNMA1, KCNQ3, KDM5C, MBD5, MECP2, MEF2C, NRXN1, PAK3, PCDH19, PHF6, PIGA, PNKP, PRRT2, RAB39B, ROGDI, SCN1A, SCN1B, SCN2A, SLC2A1, SLC9A6, STX1B, SYN1, SYNGAP1, TBC1D24, TCF4, TSC1, TSC2, UBE3A, ZEB2

BRAIN MALFORMATION EPILEPSY PANEL: 44 Genes

ACTB, ACTG1, AKT3, ARFGEF2, ARX, ASNS, ATP6V0A2, B3GALNT2, B3GNT1(B4GAT1), DCX, DYNC1H1, FKRP, FKTN, FLNA, GMPPB, GPR56(ADGRG1), GPSM2, GTDC2(POMGNT2), KATNB1, KIAA1279(KIF1BP), KIF2A, LAMA2, LARGE, NDE1, OCLN, PAFAH1B1, POMGNT1, POMT1, POMT2, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SGK196(POMK), SNAP29, SRD5A3, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, VLDLR, WDR62

ACTIONABLE GENE EPILEPSY PANEL: 22 Genes

ALDH7A1, AMT, ATP7A, CAD, FOLR1, GAMT, GLDC, KCNQ2, KCNT1, MOCS1, PHGDH, PNPO, POLG, PROSC(PLPBP), PSAT1, PSPH, SCNA1, SLC2A1, SLC6A8, SUOX, TSC1, TSC2

OEGTP - EPILEPSY TEST QUESTIONNAIRE

Patient Identifier:

This Epilepsy panel test is a deep sequencing NGS assay designed as a rule out sequencing and copy number analysis test for all coding sequences of all genes tested. Content is designed by a panel of clinical experts Ontario MOHLTC Genetic Epilepsy Working Group to include majority of genes associated with epilepsy as the cardinal clinical presentation. In patients where epilepsy is not the cardinal clinical feature, and genetic etiology is suspected, other genetic and genomic analyses and clinical genetics referral may be considered.

MUST CONFIRM THAT THE AGE OF ONSET, SEIZURE TYPE, AND ELECTROCLINICAL SYNDROME IS CONSISTENT WITH A GENETIC EPILEPSY

AGE OF ONSET:

- ○ < 1 year
- ○ 1 - 16 years
- ○ >16 years

SEIZURE TYPES: *(Check all that apply to your patient)*

- ○ Focal Onset
  - ○ Aware
  - ○ Impaired Awareness
    - ○ Motor Onset
      - ○ Automatisms
      - ○ Atonic
      - ○ Clonic
      - ○ Epileptic spasms
      - ○ Hyperkinetic
      - ○ Myoclonic
      - ○ Tonic
    - ○ Non motor Onset
      - ○ Autonomic
      - ○ Behaviour arrest
      - ○ Cognitive
      - ○ Emotional
      - ○ Sensory
  - ○ Focal to bilateral tonic-clonic
- ○ Generalized Onset
  - ○ Motor Onset
    - ○ Tonic-clonic
    - ○ Clonic
    - ○ Tonic
    - ○ Myoclonic
    - ○ Myoclonic-tonic-clonic
    - ○ Myoclonic-atonic
    - ○ Atonic
    - ○ Epileptic spasms
  - ○ Non motor Onset
    - ○ Typical absence
    - ○ Atypical absence
    - ○ Myoclonic absence
    - ○ Absence with eyelid myoclonia
- ○ Unknown Onset
  - ○ Motor Onset
    - ○ Tonic-clonic
    - ○ Epileptic spasms
  - ○ Non motor Onset
    - ○ Behaviour arrest
- ○ Unclassified

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ELECTRO CLINICAL SYNDROME:

EEG or semiology that is consistent with a distinct electroclinical syndrome (excluding those with polygenic inheritance patterns)

- Yes
- No

If yes check the most applicable electroclinical syndrome

Neonatal or Infantile Onset

- Self-limited neonatal seizures and self-limited familial neonatal epilepsy
- Self-limited familial and non-familial infantile epilepsy
- Early myoclonic encephalopathy
- Ohtahara Syndrome
- West Syndrome
- Dravet Syndrome
- Epilepsy of infancy with migrating focal seizures
- Myoclonic encephalopathy in non-progressive disorders
- Febrile seizures plus, genetic epilepsy with febrile seizures

Childhood Onset

- Epilepsy with myoclonic-atonic seizures
- Lennox Gastaut Syndrome
- Photosensitive occipital lobe epilepsy
- Atypical childhood epilepsy with centrotemporal spikes
- Epileptic encephalopathy with continuous spike and wave in sleep
- Landau-Kleffner Syndrome
- Autosomal dominant nocturnal frontal lobe epilepsy

Adolescent or Adult Onset

- Autosomal dominant epilepsy with auditory features
- Other familial temporal lobe epilepsies

Any Age

- Familial focal epilepsy with variable foci
- Progressive myoclonic epilepsies

PATIENT MUST MEET ONE OR MORE OF THE FOLLOWING CLINICAL PRESENTATIONS. CHECK ALL THAT APPLY.

A family history of epilepsy — Yes / No

Epileptic encephalopathy or progression of seizures indicative of a poor prognosis for seizure control with or without a high likelihood of lethality, or treatment resistant epilepsy — Yes / No

Epilepsy is associated with neurodevelopmental impairment — Yes / No

If yes, check those that apply

- Early developmental impairment (global developmental delay)
- Developmental regression
- Motor delay
- Speech delay
- Learning disability
- Autism Spectrum Disorder
- Intellectual disability

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Patient Identifier:

Epilepsy is associated with a structural brain malformation ○ Yes ○ No

If yes, check those that apply

- ○ Focal cortical dysplasia
- ○ Hemimegalencephaly
- ○ Cortical Tuber(s)
- ○ Double cortex
- ○ Lissencephaly
- ○ Polymicrogyria
- ○ Agyria
- ○ Pachygyria
- ○ Holopresencephaly
- ○ Hippocampal malrotation
- ○ Periventricular nodular heterotopia
- ○ Grey matter heterotopia
- ○ Subcortical band heterotopia
- ○ Agenesis of the corpus callosum
- ○ Dysmorphic corpus callosum
- ○ Cerebellar hypoplasia
- ○ Septo-optic dysplasia
- ○ Hydrocephalus
- ○ Vascular malformation
- ○ White matter abnormalities
- ○ Basal ganglia abnormalities
- ○ Brainstem abnormalities
- ○ Leukodystrophy

Other, please specify:

Epilepsy is associated with systemic involvement ○ Yes ○ No

If yes, check those that apply

- ○ Cardiac malformation
- ○ GI malformation
- ○ Renal malformation
- ○ Ophthalmological anomalies

Other, please specify:

Epilepsy is associated with syndromic features ○ Yes ○ No

If yes, check those that apply

- ○ Macrocephaly
- ○ Microcephaly
- ○ Growth delay
- ○ Overgrowth
- ○ Dysmorphic facial features

Epilepsy is associated with neurodegeneration ○ Yes ○ No

Epilepsy is associated with paroxysmal neurological features ○ Yes ○ No

PATIENTS EXCLUDED FROM THE ONTARIO EPILEPSY GENE TESTING PROGRAM

○ I confirm that the patient does NOT have:

- Mesial temporal epilepsy with hippocampal sclerosis and no relevant family history
- Myoclonic Epilepsy of Infancy
- Epilepsy with eyelid myoclonias
- Childhood Absence Epilepsy (unless atypical, ex: presenting prior to age 4)
- Epilepsy with myoclonic absences
- Panayiotopoulos syndrome
- Childhood Occipital Epilepsy (Gastaut Type)
- Childhood epilepsy with centrotemporal spikes
- Juvenile Absence Epilepsy
- Juvenile Myoclonic Epilepsy
- Epilepsy with Generalized tonic seizures alone
- Reflex epilepsies
- An acquired epilepsy as other causative circumstances (e.g., environmental exposures, injury, and infection) do NOT explain the patient's clinical presentation, based on the most complete clinical history
- A genetic diagnosis based on the previous targeted testing that explains the history of epilepsy
- A phenotype highly specific to a known genetic condition for which an optimized genetic panel exists. If so, then the targeted gene panel should be given priority assuming it is more sensitive (e.g. Tuberous Sclerosis Complex)

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REQUIRED PREREQUISITES

- I confirm that pretest genetic counseling has been completed
- I confirm that the following conditions for the affected individual have been met.
  Check all that apply.
  - A Medical Genetics consultation if family history or syndromic features are present
  - A metabolics evaluation with a geneticist or a biochemical geneticist if there is evidence of developmental regression and features suggestive of an inborn error of metabolism
  - Diagnostic procedures including EEG with or without EEG video monitoring and brain imaging [MRI] have been completed
  - I confirm that adequate post-test counseling will be provided or the patient will be referred to a Genetics clinic if needed
- I confirm that a completed management impact, post-test questionnaire will be returned to the laboratory

REQUIREMENTS FOR ORDERING THE EPILEPSY GENE PANELS

- I confirm that I am a physician in Ontario.
  Check all that apply.
  - Who is affiliated with a Provincial District Epilepsy Centre, a Regional Epilepsy Surgery Centre of Excellence, or Thunder Bay Regional Health Sciences Centre
  - Who is an FRCP neurologist in active clinical practice who has had a minimum of six months of training in epilepsy and EEG
  - Who has completed the Continuing Medical Education (CME) certified epilepsy curriculum in Project ECHO Ontario – Epilepsy Across the Life Span.
  - Who is practicing in the area of Medical Genetics (RCPSC or CCMG certified)

Molecular Genetics Laboratory
Victoria Hospital, Room B10-123A
800 Commissioners Rd. E.
London, Ontario | N6A 5W9
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