OEGTP - EPILEPSY TEST REQUISITION

LAB USE ONLY:

Received date:

Notes:

SAMPLE COLLECTION:

Date drawn:
YYYY/MM/DD

◯ EDTA blood (2 lavender top at room temp; min 2ml)

◯ DNA (100ng minimum) Conc:

TEST REQUEST:

NGS PANELS: *See page 2 for mandatory questionnaire and page 3 for panel genes**

◯ Epilepsy Comprehensive panel: 190 genes

◯ Actionable Gene Epilepsy panel: 25 genes

◯ Brain Malformation Epilepsy panel: 45 genes

◯ Childhood Onset Epilepsy panel: 59 genes

◯ Early Infantile Epilepsy panel: 84 genes

◯ Focal Epilepsy panel: 12 genes

◯ Progressive Myoclonic Epilepsy panel: 21 genes

◯ Single gene test:

◯ Carrier Testing/ Known Family Mutation

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

◯ Affected ◯ Unaffected

Date of Birth: Relationship to patient:

Gene: RefSeq:NM

Mutation:

REASON FOR REFERRAL:

Clinical Diagnosis/Presentation:

PATIENT INFORMATION: INCOMPLETE REQUESTS WILL BE BANKED

Name:

Address:

Date of Birth:
YYYY/MM/DD

Health Card No:

Sex: ◯ M ◯ F ◯ Unknown ◯ Unspecified

Birthsex: ◯ M ◯ F ◯ Unknown ◯ Unspecified

REQUEST FOR EXPEDITED RESULT:

◯ Pregnancy (LMP, YYYY/MM/DD):

◯ Medical Intervention (Specify Date)

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 B10-128
800 Commissioners Rd. E.
London, Ontario | N6A 5W9
Ph: 519-685-8122 | Fax: 519-685-8279

London Health Sciences Centre
Pathology and Laboratory Medicine

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.

CLINICAL PRESENTATIONS

PATIENT MUST MEET ONE OR MORE OF THE FOLLOWING

A family history of epilepsy:	◯ Yes	◯ No
Epileptic encephalopathy:	◯ Yes	◯ No
Progression of seizures indicative of a poor prognosis:	◯ Yes	◯ No
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
- ◯ Learning disability
- ◯ Speech delay
- ◯ Autism Spectrum Disorder
- ◯ Intellectual disability
- ◯ Motor delay

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

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)

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)

OEGTP - EPILEPSY TEST PANELS

Patient Identifier:

PAGE 3 FOR REFERENCE ONLY.

COMPREHENSIVE EPILEPSY PANEL: 190 Genes

ABAT, ACTB, ACTG1, ADGRG1, ADSL, AKT3, ALDH7A1, ALG13, AMT, AP3B2, ARFGEF2, ARHGEF9, ARV1, ARX, ASAH1, ASNS, ATP1A2, ATP1A3, ATP6V0A2, ATP7A, ATRX, B3GALNT2, CACNA1A, CACNA1E, CAD, CDKL5, CHD2, CHRNA4, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, CTSF, DCX, DEPDC5, DNAJC5, DNM1, DOCK7, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EPM2A, FGF12, FKRP, FKTN, FLNA, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GMPPB, GNAO1, GOSR2, GPSM2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, HCN1, HNRNPU, ITPA, KANSL1, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH5, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF2A, LAMA2, LARGE1, LGI1, MBD5, MDH2, MECP2, MEF2C, MFSD8, MOCS1, NDE1, NEU1, NEXMIF, NGLY1, NHLRC1, NPRL2, NPRL3, NRXN1, OCLN, PAFAH1B1, PAK3, PCDH19, PHF6, PHGDH, PIGA, PIGG, PIGN, PIGO, PIGT, PIGV, PLCB1, PLPBP, PNKP, PNPO, POLG, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPT1, PRRT2, PSAT1, PSPH, PURA, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RELN, ROGDI, RTTN, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SGCE, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SNAP29, SPATA5, SPTAN1, SRD5A3, ST3GAL5, STX1B, STXBP1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TPP1, TRPM3, TSC1, TSC2, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, UBA5, UBE3A, VLDLR, WDR45, WDR62, WWOX, YWHAG, ZEB2

ACTIONABLE GENE EPILEPSY PANEL: 25 Genes

ALDH7A1, AMT, ATP7A, CAD, FOLR1, GAMT, GLDC, KCNQ2, KCNT1, MOCS1, PHGDH, PLPBP, PNPO, POLG, PSAT1, PSPH, SCN1A, SLC19A3, SLC2A1, SLC6A8, SUOX, TPP1, TRPM3, TSC1, TSC2

BRAIN MALFORMATION EPILEPSY PANEL: 45 Genes

ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ASNS, ATP1A2, ATP6V0A2, B3GALNT2, DCX, DYNC1H1, FKRP, FKTN, FLNA, GMPPB, GPSM2, GRIN1, KATNB1, KIF2A, LAMA2, LARGE1, NDE1, OCLN, PAFAH1B1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SCN3A, SNAP29, SRD5A3, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, VLDLR, WDR62

CHILDHOOD ONSET EPILEPSY PANEL: 59 Genes

ADSL, ARX, ATP1A3, ATRX, CDKL5, CHD2, CLCN4, CNTNAP2, DEPDC5, DNAJC5, DYRK1A, EHMT1, FOXG1, GABBR2, GABRB2, GABRG2, GRIN2A, GRIN2D, KANSL1, KCNJ10, KCNMA1, KCNQ3, KDM5C, MBD5, MECP2, MEF2C, NEXMIF, NGLY1, NRXN1, PAK3, PCDH19, PHF6, PIGA, PIGN, PIGO, PNKP, POLG, PRRT2, RAB39B, ROGDI, SCN1A, SCN1B, SCN2A, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, STX1B, SYN1, SYNGAP1, TBC1D24, TCF4, TRPM3, TSC1, TSC2, UBE3A, WDR45, ZEB2

EARLY INFANTILE EPILEPSY PANEL: 84 Genes

ABAT, ADSL, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, CACNA1A, CACNA1E, CAD, CDKL5, CHD2, DCX, DNM1, DOCK7, DYRK1A, EEF1A2, FGF12, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GRIN2A, GRIN2B, GRIN2D, HCN1, HNRNPU, ITPA, KCNA1, KCNA2, KCNB1, KCNH5, KCNQ2, KCNQ3, KCNT1, MDH2, MECP2, MEF2C, NGLY1, PCDH19, PIGA, PIGG, PIGN, PIGO, PIGT, PIGV, PLCB1, PNKP, PNPO, POLG, PRRT2, PURA, SCN1A, SCN1B, SCN2A, SCN8A, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SPATA5, SPTAN1, ST3GAL5, STX1B, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, UBA5, WDR45, WWOX, YWHAG

FOCAL EPILEPSY PANEL: 12 Genes

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

PROGRESSIVE MYOCLONIC EPILEPSY PANEL: 21 Genes

ASAH1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, EPM2A, GOSR2, GRN, KCNC1, KCTD7, MFSD8, NEU1, NHLRC1, PPT1, SCARB2, SERPINI1, SGCE, TPP1

*Genetic testing may reveal secondary findings which are not related to the referral indication. Secondary findings will be communicated to the ordering provider and may be reported according to ACMG guidelines.