
GENETIC TEST:
Progressive Myoclonic Epilepsy (PME) (gene panel)

FULL NAME:	Progressive Myoclonic Epilepsy (PME) (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Sequence analysis: entire coding region Whole Exome Sequencing (WES) Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	01 Aug 2019 - 12:01
CHANGED:	16 Dec 2022 - 13:08

RELATED CONTENT

Related Laboratories

- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [ACD shelterin complex subunit and telomerase recruitment factor](#)
- [N-acylsphingosine amidohydrolase 1](#)
- [ATPase cation transporting 13A2](#)
- [BSCL2 lipid droplet biogenesis associated, seipin](#)
- [calcium voltage-gated channel subunit alpha1 A](#)
- [ceramide synthase 1](#)
- [CLN3 lysosomal/endosomal transmembrane protein, battenin](#)
- [CLN5 intracellular trafficking protein](#)
- [CLN6 transmembrane ER protein](#)
- [CLN8 transmembrane ER and ERGIC protein](#)
- [cystatin B](#)
- [cathepsin D](#)
- [cathepsin F](#)
- [DnaJ heat shock protein family \(Hsp40\) member C5](#)
- [EPM2A glucan phosphatase, laforin](#)
- [phenylalanyl-tRNA synthetase 2, mitochondrial](#)
- [glucosylceramidase beta 1](#)
- [golgi SNAP receptor complex member 2](#)
- [granulin precursor](#)
- [potassium voltage-gated channel subfamily C member 1](#)

- [potassium channel tetramerization domain containing 7](#)
- [kinesin family member 5A](#)
- [lamin B2](#)
- [major facilitator superfamily domain containing 8](#)
- [mitochondrial methionyl-tRNA formyltransferase](#)
- [neuraminidase 1](#)
- [NHL repeat containing E3 ubiquitin protein ligase 1](#)
- [palmitoyl-protein thioesterase 1](#)
- [prickle planar cell polarity protein 1](#)
- [prion protein](#)
- [sacsin molecular chaperone](#)
- [scavenger receptor class B member 2](#)
- [serpin family I member 1](#)
- [TBC1 domain family member 24](#)

Related Gene Panels

- [Progressive Myoclonic Epilepsy - UGent](#)

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