Dante Labs Inc
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Phone:
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Patient Information

Name
Gender Unspecified
Id
2228

Sample Information

Sample Site
Saliva
Sample Type
Germline DNA
Collection Method
Oragene•DNA (OG-500)
Panel Coverage
Full DNA

Avg. Read Depth
30X
Collection Date

Receipt Date

Report Date

Results

Positive: Pathogenic variant with an establish link detected.

Affected Genes

ADSL (0)	*ALDH5A1* (0)	*ALDH7A1* (0)	*ALG13* (0)	*ARHGEF9* (0)	*ARX* (0)	*ATP1A2* (0)	*ATP1A3* (0)	*ATRX* (0)	*BRAT1* (0)	*C12ORF57* (0)
CACNA2D2 (0)	*CASK* (0)	*CDKL5* (0)	*CHD2* (0)	*CHRNA2* (0)	*CHRNA4* (0)	*CHRNB2* (0)	*CLN3* (0)	*CLN5* (0)	*CLN6* (0)	*CLN8* (0)
CSTB (0)	*CTNAP2* (0)	*CTSD* (0)	*DEPDC5* (0)	*DNAJC5* (0)	*DNM1* (0)	*DYRK1A* (0)	*EEF1A2* (0)	*EFHC1* (0)	*EHMT1* (0)	*EPM2A* (0)
FOLR1 (0)	*FOXG1* (0)	*FRRS1* (0)	*GABRA1* (0)	*GABRB3* (0)	*GABRG2* (0)	*GAMT* (0)	*GATM* (0)	*GLRA1* (0)	*GNAO1* (0)	*GOSR2* (0)
GRIN1 (0)	*GRIN2A* (0)	*GRIN2B* (0)	*HCN1* (0)	*HNRNPU* (0)	*IER3IP1* (0)	*IQSEC2* (0)	*ITPA* (0)	***KANSL1*** **(1)**	*KCNA2* (0)	*KCNB1* (0)
KCNC1	*KCNH2*	*KCNJ10*	*KCNQ2*	*KCNQ3*	*KCNT1*	*KCTD7*	*LGI1*	*LIAS*	*MBD5*	*MECP2*

(0)	(0)	(0)	(0)	(0)	(0)	(0)	(0)	(0)	(0)	(0)
MEF2C (0)	*MFSD8* (0)	*NEXMIF* (0)	*NGLY1* (0)	***NHLRC1*** **(1)**	*NRXN1* (0)	*PACS1* (0)	*PCDH19* (0)	*PIGA* (0)	*PIGN* (0)	*PIGO* (0)
PLCB1 (0)	*PNKD* (0)	*PNKP* (0)	*PNPO* (0)	*POLG* (0)	*PPT1* (0)	*PRICKLE1* (0)	*PRRT2* (0)	*PURA* (0)	*QARS* (0)	*ROGDI* (0)
SATB2 (0)	*SCARB2* (0)	*SCN1A* (0)	*SCN1B* (0)	*SCN2A* (0)	*SCN3A* (0)	*SCN8A* (0)	*SCN9A* *SERPINI1* (0)	*SGCE* (0)	*SLC13A* (0)	*SLC19A3* (0)
SLC25A22 **(1)**	*SLC2A1* (0)	*SLC35A2* (0)	*SLC6A1* (0)	*SLC6A8* (0)	*SLC9A6* (0)	*SMC1A* (0)	*SNX27* (0)	*SPATA5* (0)	*SPTAN1* (0)	*STX1B* (0)
STXBP1 (0)	*SYN1* (0)	*SYNGAP1* (0)	*SYNJ1* (0)	*SZT2* (0)	*TBC1D2* (0)	*TCF4* (0)	*TPP1* (0)	*TSC1* (0)	*TSC2* (0)	*UBE3A* (0)
WWOX (0)	*ZDHHC9* (0)	*ZEB2* (0)								

Primary Findings

Gene	Zygosity	Variant	Exon	Pathogenicity
SCN9A	Homozygous Variant	NM_002977.3:c.1828C>A(NP_002968.1:p.Pro610Thr)	12	Pathogenic

OncoMD Drug Summary

OncoMD Trial Summary

Individual Variant Interpretations

NP_002968.1:p.Pro610Thr in Exon 12 of *SCN9A* (NM_002977.3:c.1828C>A) Pathogenic

Incidental Findings

3 Prime UTR Variant in *NHLRC1* (NM_198586.2:c.*326C>T)

Variant of Unknown Significance.

5 Prime UTR Variant in *SLC25A22* (NM_024698.5:c.-141G>A)

Variant of Unknown Significance.

NP_056258.1:p.Ala7Thr in Exon 2 of *KANSL1* (NM_015443.3:c.19G>A)

Variant of Unknown Significance.

Additional Information

Test

Epilepsy

Indication

Virtual Panel from WGS 30X

Background

Idiopathic generalized epilepsy is a broad term that encompasses several common seizure phenotypes, classically including childhood absence epilepsy (CAE, ECA; see 600131), juvenile absence epilepsy (JAE, EJA; see 607631), juvenile myoclonic epilepsy (JME, EJM; see 254770), and epilepsy with grand mal seizures on awakening (Commission on Classification and Terminology of the International League Against Epilepsy, 1989). These recurrent seizures occur in the absence of detectable brain lesions and/or metabolic abnormalities. Seizures are initially generalized with a bilateral, synchronous, generalized, symmetrical EEG discharge (Zara et al., 1995; Lu and Wang, 2009). See also childhood absence epilepsy (ECA1; 600131), which has also been mapped to 8q24. Of note, benign neonatal epilepsy 2 (EBN2; 121201) is caused by mutation in the KCNQ3 gene (602232) on 8q24. Genetic Heterogeneity of Idiopathic Generalized Epilepsy EIG1 has been mapped to chromosome 8q24. Other loci or genes associated with EIG include EIG2 (606972) on 14q23; EIG3 (608762) on 9q32; EIG4 (609750) on 10q25; EIG5 (611934) on 10p11; EIG6 (611942), caused by mutation in the CACNA1H gene (607904) on 16p; EIG7 (604827) on 15q14; EIG8 (612899), caused by mutation in the CASR gene (601199) on 3q13.3-q21; EIG9 (607682), caused by mutation in the CACNB4 gene (601949) on 2q22-q23; EIG10 (613060), caused by mutation in the GABRD gene (137163) on 1p36.3; EIG11 (607628), caused by variation in the CLCN2 gene (600570) on 3q36; EIG12 (614847), caused by mutation in the SLC2A1 gene (138140) on 1p34; EIG13 (611136), caused by mutation in the GABRA1 gene (137160) on 5q34; and EIG14 (616685), caused by mutation in the SLC12A5 gene (606726) on 20q12.

Method

Next-Generation (NGS)/Massively parallel sequencing (MPS)

Limitations

Nothing in the genetic report should be used for medical diagnosis or treatment. The information provide d should not be considered complete, nor should it be relied on to suggest diagnosis or treatment of a particular in dividual. Material in the genetic report should not be relied upon for personal, medical, legal, technical, or financial decisions.

This genetic report should not be used in place of a visit with or advice from your doctor or other qualified healthcare professional. You should always get the advice of your doctor or other appropriate health care professional if you have any question about diagnosis, treatment, prevention, mitigation, or cure of any medical condition, phenotype, condition, impairment, or the status of your health. Do not stop any medications you have been prescribed, start any new medications, or modify any medical treatments ordered by your healthcare provider without first talking with your provider.

DRAFT REPORT