USTAR Center for Genetic Discovery

iobio

Patient:
16012132564

Signigicant variants

Gene: PRX

Variant information:

Location	chr19
Disease mode of inheritance	recessive
Reference allele	G
Alternate allele	A
CSN	ENST00000324001.7
Consequence	stop gained
dbSNP ID	rs104894714
Type	snp
REVEL	
HGVSc	ENST00000324001.7:c.2857C>T
HGVSp	ENSP00000326018.6:p.Arg953Ter
Gnomad allele frequency	3.251e-05
ExAC allele frequency	4.1e-05
Impact	high

Notes:

Here is a note created from Tony.

— *Tonya Lee Disera*

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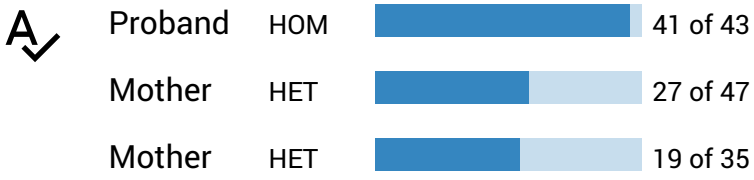
Here is a second longer note. one of the Vuetify.js Vue CLI packages (based on the official examples) to get your project started in no time. Vuetify.js supports SSR (server-side rendering), SPA (single page application), PWA (progressive web application) and standard HTML pages.

— *Tonya Lee Disera*

Variant Summaries:



Unknow significance variants

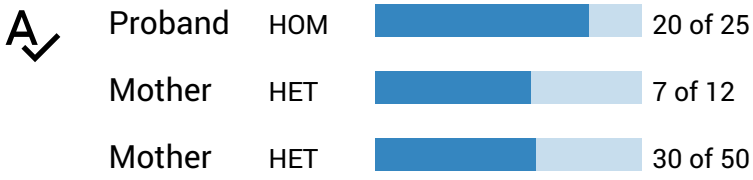
Gene: SLC9B1P1

Variant information:

Location	chrY
Disease mode of inheritance	recessive
Reference allele	C
Alternate allele	T
CSN	ENST00000331172.6
Consequence	splice region variant, intron variant
dbSNP ID	rs763588005
Type	snp
REVEL	

HGVSc	ENST00000331172.6:c.678+5G>A
HGVSp	
Gnomad allele frequency	0
ExAC allele frequency	0.194
Impact	low
Impact	low

Notes: no note is added
Variant Summaries:



Pathogenic variants

Gene	Location	CSN	Consequence	dbSNP ID
PRX	chr19	ENST00000324001.7	stop gained	rs1048947

Variants of unknown significance

Gene	Location	CSN	Consequence	
SLC9B1P1	chrY	ENST00000331172.6	splice region variant, intron variant	

Disclaimers

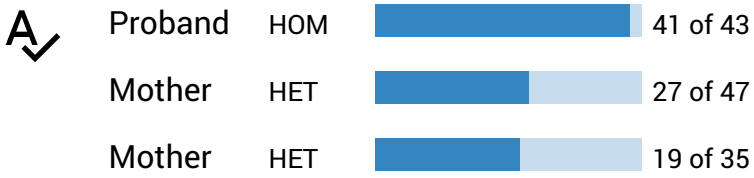
Genetic testing information has caveats and should not be considered a definitive diagnosis.

References/Methodology

DNA sequencing was performed in accordance with established Utah Genome Project (UGP) methodologies
sequencing and data analysis.

Variant summaries

PRX : G > A snp



Genes

GTR

Conditions	Genes
Dejerine-Sottas disease	MPZ,EGR2,PMP22,PRX,GJB1,GDAP1,FGD4,A 4,DYNC1H1,NEFL,MFN2,LMNA,MTMR2,NDR HSPB8,TRPV4,GNB4,MED25,LRSAM1,YARS PDK3,DHTKD1,PLEKHG5,KARS1,KIF1B,TRIM HADHB,IGHMBP2,TFG,GLA,HINT1,DNAJB2,M A,TTR,CTDP1,FBLN5,SPTLC1,MORC2,DNMT PAST,GAN,SPTLC2,SLC12A6,ATL1,REEP1

Phenolyzer

Phentypes	Genes
dejerine sottas disease	EGR2,PMP22,MPZ,TSEN54,GDAP1,NEFL,MT RARS2,LMNA,PEX7,FIG4,FA2H,FGD4,LITAF,S 2,DNM2,SEPT9,SMCHD1,PLP1,ALS2,DUX4,E SCU,SBF1,MARS2,GARS,TSEN34,TSEN2,VRK AA0196,DYNC1H1,HK1,AARS,EXOSC3,OPA3, RPV4,SEPSECS,FRG1,PRKCA,YBX1,AKT1,AR AV,BSCL2,CANX,YWHAB,AKT2,AKT3,HSP90A PRKACA,RXRA,CDK1,SRC,FOS,UBB,UBA52,R PSMD2,PSMC3,PSMC2,PPP2R1A,HCFC1,CL GB1,PPP2R1B,TBL1XR1,MAPK3,NFKB1,PIK3 3R3,PIK3CB,PIK3CD,PSMC1,PPP2CA,HDAC1 PTEN,MAPKAPK2,AP2M1,PPP2CB,JUN,MIR K2B,PSMD6,PSMD12,PSMD11,PSMD7,PSMD NBP2,PSMB1,PSMB7,PSMA5,PSMA2,PRKAA TBL1X,APAF1,YWHAQ,PSMB3,CALM3,CALM MD8,PSMC5,SHFM1,PSMD4,PSMA7,TSC1,C KACB,PSME4,PSMA6,PSMB11,PSMB10,PSM 36,PSME3,PSMB2,PSME1,PSMD5,PSMB8,PS MF1,PSMA8,HDAC3,YWHAH,SMAD3,MAPK1 R1,ACTB,GATA3,CASP1,HIF1A,PRKACG,NCA AC2,ITGB2,CD36,SMAD2,CSNK2A2,AP2A1,IG KN1B,SREBF1,RHOA,MED15,MED6,RAB5A,N BPJ,MET,NUP85,FYN,PDGFRA,AP2A2,TNF,M SC2,ESR1,NUP107,NUP160,MED31,PDPK1,F D,MED16,PRKCB,PRKAB2,PRKAG2,NCOA1,N A,RORA,PPP2R5C,RELA,SPTAN1,CDK2,TPR,S HIST1H3H,HIST1H3B,HIST2H3C,HIST1H3A,H HIST1H3G,HIST1H3E,HIST1H3J,HIST2H3A,H NUP153,TGFB1,POM121,NUP155,NUP50,DN B,VEGFA,MTOR,NUP62,NUP93,POM121C,NU UP54,NUP35

Summary

Date of Birth: 04/15/1950
Gender: Male
Medical Record #: 00123456
Additional Recipients: John Doe

Provider: Intermountain Healthcare
Physician: Dr. Jane Smith
Pathologist: Dr. John Brown

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Clinical Description

According to information provided to ARUP, the patient is a one year old male. He was delivered by cesarea
At birth he weighed 6 pounds and 1 ounce, and was 17.5 inches long. He has multiple congenital anc
encephalocele, tecto-cerebellar dysraphism, posterior plagiocephaly, relative macrocephaly, left-sided facia
closure, optic nerve hypoplasia, prominent nasal bridge and columella, bilateral low-set microtia with ear
Mobitz type II atrioventricular block s/p epicardial pacemaker, right torticollis, vertebral segmentation defec
T12), fused right first and second rib and rudimentary left rib, mild scoliosis, long and narrow left thumb
horseshoe kidney.. His weight and height are less than 1st percentile but show normal growth velocity. H
rolling from front to back at 5 months and babbling since 6 months. He did not fully support his head
diagnostic test results included creatinine, blood urea nitrogen, cytomegalovirus and cytogenomic SNP mi
sister with heterochromia. His father has 2-3 syndactyly and history of porencephalic cyst. His family
grandfather with unilateral hearing loss at birth, a maternal uncle with macrocephaly, a maternal uncle who c
abnormal ears, a maternal great uncle with an unilateral ear anomaly and bilateral hearing loss, a maternal s
anomaly and hearing loss, a paternal first cousin with congenital heart valve defect requiring surgery, a p
who is 80 percent blind in one eye, another paternal uncle with sarcoidosis and a paternal uncle with cleft
died with lupus and Crohn’s disease and a paternal great uncle has intellectual disabilities and has b
adulthood.