

Significant 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.

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:

A✓	Proband	HOM	<div></div>	41 of 43
	Mother	HET	<div></div>	27 of 47
	Mother	HET	<div></div>	19 of 35

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

Notes:

Variant Summaries:

A✓

Proband

HOM

20 of 25

Mother

HET

7 of 12

Mother

HET

30 of 50

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	n

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

<div><div>A</div><div>✓</div></div>	Proband	HOM	<div><div></div></div>	41 of 43
	Mother	HET	<div><div></div><div></div></div>	27 of 47
	Mother	HET	<div><div></div><div></div></div>	19 of 35

Genes

GTR

Conditions	Genes
Dejerine-Sottas disease	MPZ,EGR2,PMP22,PRX,GJB1,GDAP1,FGD4,ATP1A4,DYNC1H1,NEFL,MFN2,LMNA,MTMR2,NDR1,HSPB8,TRPV4,GNB4,MED25,LRSAM1,YARS1,PDK3,DHTKD1,PLEKHG5,KARS1,KIF1B,TRIM21,HADHB,IGHMBP2,TFG,GLA,HINT1,DNAJB2,MED12A,TTR,CTDP1,FBLN5,SPTLC1,MORC2,DNMT3A

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	Provider: Intermountain Healthcare	Sp
Gender: Male	Physician: Dr. Jane Smith	Da
Medical Record #: 00123456	Pathologist: Dr. John Brown	Sp
Additional Recipients: John Doe		Sp

Clinical Description

According to information provided to ARUP, the patient is a one year old male. He was delivered by cesarean section. At birth he weighed 6 pounds and 1 ounce, and was 17.5 inches long. He has multiple congenital anomalies including encephalocele, tecto-cerebellar dysraphism, posterior plagiocephaly, relative macrocephaly, left-sided facial cleft closure, optic nerve hypoplasia, prominent nasal bridge and columella, bilateral low-set microtia with ear canal atresia, Mobitz type II atrioventricular block s/p epicardial pacemaker, right torticollis, vertebral segmentation defect (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. He is rolling from front to back at 5 months and babbling since 6 months. He did not fully support his head at 12 months. His diagnostic test results included creatinine, blood urea nitrogen, cytomegalovirus and cytogenomic SNP microarray. His sister has heterochromia. His father has 2-3 syndactyly and history of porencephalic cyst. His family history includes grandfather with unilateral hearing loss at birth, a maternal uncle with macrocephaly, a maternal uncle who died of abnormal ears, a maternal great uncle with an unilateral ear anomaly and bilateral hearing loss, a maternal grandfather with hearing loss, a paternal first cousin with congenital heart valve defect requiring surgery, a paternal uncle who is 80 percent blind in one eye, another paternal uncle with sarcoidosis and a paternal uncle with cleft palate who died with lupus and Crohn's disease and a paternal great uncle has intellectual disabilities and has been institutionalized in adulthood.