USTAR Center for Genetic Discovery

iobio

Patient:
16012132564

Diagnosis:
Undiagnosed

Date:
November 10, 2016

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

Specimen ID #: 1234567
Date Collected: 09/09/2016
Specimen Site:
Specimen Grade:

Clinical Description

According to information provided to ARUP, the patient is a one year old male. He was delivered by cesarean section at 39 weeks and 5 days. At birth he weighed 6 pounds and 1 ounce, and was 17.5 inches long. He has multiple congenital anomalies including a large occipital encephalocele, tecto-cerebellar dysraphism, posterior plagiocephaly, relative macrocephaly, left-sided facial weakness, unilateral lack of eye closure, optic nerve hypoplasia, prominent nasal bridge and columella, bilateral low-set microtia with ear tags, bilateral mixed hearing loss, Mobitz type II atrioventricular block s/p epicardial pacemaker, right torticollis, vertebral segmentation defects (C2-3 fusion, abnormal T2-3 and T12), fused right first and second rib and rudimentary left rib, mild scoliosis, long and narrow left thumb, polysplenia, transverse liver and horseshoe kidney.. His weight and height are less than 1st percentile but show normal growth velocity. He was socially smiling at 8 weeks,

rolling from front to back at 5 months and babbling since 6 months. He did not fully support his head at nine months. Previous normal diagnostic test results included creatinine, blood urea nitrogen, cytomegalovirus and cytogenomic SNP microarray. He has one healthy older sister with heterochromia. His father has 2-3 syndactyly and history of porencephalic cyst. His family history also includes a maternal grandfather with unilateral hearing loss at birth, a maternal uncle with macrocephaly, a maternal uncle who died with congenital anomalies and abnormal ears, a maternal great uncle with an unilateral ear anomaly and bilateral hearing loss, a maternal second cousin with an unilateral ear anomaly and hearing loss, a paternal first cousin with congenital heart valve defect requiring surgery, a paternal uncle with sarcoid disease who is 80 percent blind in one eye, another paternal uncle with sarcoidosis and a paternal uncle with cleft palate. His paternal grandmother died with lupus and Crohn's disease and a paternal great uncle has intellectual disabilities and has been in assisted living since early adulthood.

Pathogenic variants

Gene	Location	CSN	Consequence	dbSNP ID	Inheritance
PRX	chr19	ENST00000324001.7	stop gained	rs104894714	recessive

Variants of unknown significance

Gene	Location	CSN	Consequence	dbSNP ID	Inheritance

SLC9B1P1	chrY	ENST00000331172.6	splice region variant, intron variant	rs763588005	recessive

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 including sample preparation, sequencing and data analysis.

Variant summaries

PRX : G > A snp

Mother HET 19 of 35

Genes

GTR

Conditions	Genes
Dejerine-Sottas disease	MPZ,EGR2,PMP22,PRX,GJB1,GDAP1,FGD4,AARS1,DNM2,GARS1,HSPB1,FIG4,DYNC1H1,NEFL,MFN2,LMNA,MTMR2,NDRG1,SH3TC2,SBF2,RAB7A,LITAF,HSPB8,TRPV4,GNB4,MED25,LRSAM1,YARS1,AIFM1,COX6A1,PRPS1,BSCL2,PDK3,DHTKD1,PLEKHG5,KARS1,KIF1B,TRIM2,SBF1,INF2,C12orf65,ATP1A1,HADHB,IGHMBP2,TFG,GLA,HINT1,DNAJB2,MARS1,SPG11,HARS1,HK1,KIF5A,TTR,CTDP1,FBLN5,SPTLC1,MORC2,DNMT1,HOXD10,PEX7,PHYH,SCN9A,SPAST,GAN,SPTLC2,SLC12A6,ATL1,REEP1

Phenolyzer

Phentypes	Genes
dejerine sottas disease	EGR2,PMP22,MPZ,TSEN54,GDAP1,NEFL,MTMR2,GAN,MED25,PHYH,ALMS1,RARS2,LMNA,PEX7,FIG4,FA2H,FGD4,LITAF,SBF2,PRX,NDRG1,L1CAM,SH3TC

2,DNM2,SEPT9,SMCHD1,PLP1,ALS2,DUX4,ETS1,HSPB1,SPG7,FGF3,RAB7A,ISCU,SBF1,MARS2,GARS,TSEN34,TSEN2,VRK1,SPAST,GRN,SPG11,SPG20,KIAA0196,DYNC1H1,HK1,AARS,EXOSC3,OPA3,LRSAM1,MFN2,GATA2,HSPB8,TRPV4,SEPSECS,FRG1,PRKCA,YBX1,AKT1,AR,YWHAG,UBC,CREBBP,TP53,ITGAV,BSCL2,CANX,YWHAB,AKT2,AKT3,HSP90AA1,CTNNB1,EGF,MYC,CDKN2A,PRKACA,RXRA,CDK1,SRC,FOS,UBB,UBA52,RPS27A,PIK3CA,PIK3R1,YWHAZ,PSMD2,PSMC3,PSMC2,PPP2R1A,HCFC1,CLTC,EGFR,EP300,CALM1,CDH1,ITGB1,PPP2R1B,TBL1XR1,MAPK3,NFKB1,PIK3R2,YWHAE,GSK3B,PRDM14,PIK3R3,PIK3CB,PIK3CD,PSMC1,PPP2CA,HDAC1,FGFR2,FGFR3,CYCS,CSNK2A1,PTEN,MAPKAPK2,AP2M1,PPP2CB,JUN,MIR1281,SEH1L,NUP43,NUP37,CSNK2B,PSMD6,PSMD12,PSMD11,PSMD7,PSMD3,PSMD1,PSMD14,PSMD13,RANBP2,PSMB1,PSMB7,PSMA5,PSMA2,PRKAA2,ERBB2,RARA,HRAS,KIT,GRB2,TBL1X,APAF1,YWHAQ,PSMB3,CALM3,CALM2,CLTA,RAC1,PSMC6,PSMC4,PSMD8,PSMC5,SHFM1,PSMD4,PSMA7,TSC1,CDK8,PSMA3,PSMD10,PSME2,PRKACB,PSME4,PSMA6,PSMB11,PSMB10,PSMB9,PSMB4,PSMA4,LOC100506136,PSME3,PSMB2,PSME1,PSMD5,PSMB8,PSMA1,PSMB6,PSMB5,PSMD9,PSMF1,PSMA8,HDAC3,YWHAH,SMAD3,MAPK14,MDM2,RAE1,AAAS,MED1,FGFR1,ACTB,GATA3,CASP1,HIF1A,PRKACG,NCAN,PLCG1,SP1,CDKN1A,RPS6,HDAC2,ITGB2,CD36,SMAD2,CSNK2A2,AP2A1,IGF2,MAPK11,HSPA8,PIP5K1C,CDKN1B,SREBF1,RHOA,MED15,MED6,RAB5A,NCOA2,INS,MED8,SUZ12,BIRC5,RBPJ,MET,NUP85,FYN,PDGFRA,AP2A2,TNF,MAPK1,NUP133,NCOR1,NUP98,TSC2,ESR1,NUP107,NUP160,MED31,PDPK1,FOXP3,AP2S1,AP2B1,WASL,PRKCD,MED16,PRKCB,PRKAB2,PRKAG2,NCOA1,NUP214,HIST2H3D,SHC1,HLA-A,RORA,PPP2R5C,RELA,SPTAN1,CDK2,TPR,SLC2A4,NCOR2,CREB1,PDGFRB,HIST1H3H,HIST1H3B,HIST2H3C,HIST1H3A,HIST1H3F,HIST1H3D,HIST1H3I,HIST1H3G,HIST1H3E,HIST1H3J,HIST2H3A,HIST1H3C,CDC42,PPARA,ICAM1,NUP153,TGFB1,POM121,NUP155,NUP50,DNM1,HLA-B,VEGFA,MTOR,NUP62,NUP93,POM121C,NUP188,NUP205,NUP210,NUP88,NUP54,NUP35