Utah Center for Genetic Discovery

iobio

Sample:
16012132564

Significant

Variant information:

Gene	PRX
Mode of inheritance	recessive
Consequence	stop gained
HGVSp	ENSP00000326018.6:p.Arg953Ter
HGVSc	ENST00000324001.7:c.2857C>T
rsId	rs104894714
gnomAD allele frequency	3.183e-05
ExAC allele frequency	.
REVEL	
Impact	high
Location	chr19: 40901402-40901403
Variant type	snp
Variant quality	1933.95

Notes: No note is added

Variant information:

Gene	SCN8A
Mode of inheritance	de novo
Consequence	missense variant
HGVSp	ENSP00000346534.4:p.Arg1872Trp
HGVSc	ENST00000354534.6:c.5614C>T
rsId	rs796053228

gnomAD allele frequency	0
ExAC allele frequency	.
REVEL	0.901
Impact	moderate
Location	chr12: 52200884-52200885
Variant type	snp
Variant quality	429.716

Notes: No note is added

Variant information:

Gene	DLL4
Mode of inheritance	de novo
Consequence	missense variant
HGVSp	ENSP00000249749.5:p.Cys653Trp
HGVSc	ENST00000249749.5:c.1959T>G
rsId	rs533126562
gnomAD allele frequency	0
ExAC allele frequency	.
REVEL	0.785
Impact	moderate
Location	chr15: 41229631-41229632
Variant type	snp
Variant quality	383.909

Notes: No note is added

Variant information:

Gene	ABCA3

Mode of inheritance	compound het
Consequence	missense variant
HGVSp	ENSP00000301732.5:p.Pro766Ser
HGVSc	ENST00000301732.5:c.2296C>T
rsId	rs45592239
gnomAD allele frequency	0.001708
ExAC allele frequency	.
REVEL	0.113
Impact	moderate
Location	chr16: 2345709-2345710
Variant type	snp
Variant quality	1166.76

Notes: No note is added

Variant information:

Gene	ABCA3
Mode of inheritance	compound het
Consequence	missense variant
HGVSp	ENSP00000301732.5:p.Tyr247Ser
HGVSc	ENST00000301732.5:c.740A>C
rsId	rs775442517
gnomAD allele frequency	0.001114
ExAC allele frequency	.
REVEL	0.508
Impact	moderate
Location	chr16: 2369715-2369716
Variant type	snp
Variant quality	1.09117e-06

Notes: No note is added

Variant information:

Gene	DVL3
Mode of inheritance	de novo
Consequence	missense variant
HGVSp	ENSP00000316054.3:p.Arg221Gly
HGVSc	ENST00000313143.3:c.661C>G
rsId	rs76594728
gnomAD allele frequency	0
ExAC allele frequency	.
REVEL	0.296
Impact	moderate
Location	chr3: 183882962-183882963
Variant type	snp
Variant quality	487.882

Notes: No note is added

Variant information:

Gene	NOTCH4
Mode of inheritance	compound het
Consequence	missense variant
HGVSp	ENSP00000364163.3:p.Gly1121Arg
HGVSc	ENST00000375023.3:c.3361G>A
rsId	rs72846312
gnomAD allele frequency	7.417e-05
ExAC allele frequency	.
REVEL	0.204
Impact	moderate

Location	chr6: 32170247-32170248
Variant type	snp
Variant quality	1096.51

Notes: No note is added

Variant information:

Gene	NOTCH4
Mode of inheritance	compound het
Consequence	missense variant
HGVSp	ENSP00000364163.3:p.Asp272Gly
HGVSc	ENST00000375023.3:c.813_815delinsGGG
rsId	rs71556915
gnomAD allele frequency	0
ExAC allele frequency	.
REVEL	
Impact	moderate
Location	chr6: 32188640-32188643
Variant type	complex
Variant quality	1354.34

Notes: No note is added

Unknown Significance

Variant information:

Gene	LMNA

Mode of inheritance	de novo
Consequence	missense variant
HGVSp	ENSP00000357283.4:p.His565Pro
HGVSc	ENST00000368300.4:c.1694A>C
rsId	Not found
gnomAD allele frequency	0
ExAC allele frequency	.
REVEL	0.560
Impact	moderate
Location	chr1: 156107530-156107531
Variant type	snp
Variant quality	1.36779e-10

Notes: No note is added

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.

Genes

Phenotypes searched	Genes (100)
GTR: Dejerine-Sottas disease , ,DSD , ,ABCNSJN Phenolyzer: [object Object]	MPZ , EGR2 , PMP22 , PRX , GDAP1 , FGD4 , DNM2 , HSPB1 , FIG4 , DYNC1H1 , NEFL , MFN2 , LMNA , MTMR2 , NDRG1 , SH3TC2 , SBF2 , RAB7A , LITAF , HSPB8 , TRPV4 , MED25 , LRSAM1 , BSCL2 , SBF1 , SPG11 , HK1 , PEX7 , PHYH , SPAST , GAN , GJB1 , AARS1 , GARS1 , GNB4 , YARS1 , AIFM1 , COX6A1 , PRPS1 , PDK3 , DHTKD1 , PLEKHG5 , KARS1 , KIF1B , TRIM2 , INF2 , C12orf65 , ATP1A1 , HADHB , IGHMBP2 , TFG , GLA , HINT1 , DNAJB2 , MARS1 , HARS1 , KIF5A , TTR , CTDP1 , FBLN5 , SPTLC1 , MORC2 , DNMT1 , HOXD10 , SCN9A , SPTLC2 , SLC12A6 , ATL1 , REEP1 , TSEN54 , ALMS1 , RARS2 , FA2H , L1CAM , SEPT9 , SMCHD1 , PLP1 , ALS2 , DUX4 , ETS1 , SPG7 , FGF3 , ISCU , MARS2 , GARS , TSEN2 , TSEN34 , VRK1 , GRN , SPG20 , KIAA0196 , AARS , EXOSC3 , OPA3 , GATA2 , SEPSECS , FRG1 , PRKCA , YBX1 , AKT1

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.