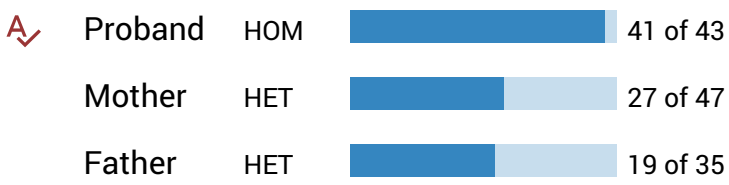
Significant Variants

Variant information:

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

Notes: no note is added
Variant Summaries:



Variant information:

Gene	LMNA
Location	chr1
Mode of inheritance	de novo
CSN	ENST00000368300.4
Consequence	missense variant
dbSNP ID	null
Variant type	snp
Variant quality	1.36779e-10
REVEL	0.560
HGVSc	ENST00000368300.4:c.1694A>C
HGVSp	ENSP00000357283.4:p.His565Pro
gnomAD allele frequency	0
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

Variant information:

Gene	SCN8A
Location	chr12
Mode of inheritance	de novo
CSN	ENST00000354534.6
Consequence	missense variant
dbSNP ID	rs796053228
Variant type	snp
Variant quality	429.716
REVEL	0.901

HGVSc	ENST00000354534.6:c.5614C>T
HGVSp	ENSP00000346534.4:p.Arg1872Trp
gnomAD allele frequency	0
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

Proband HET 24 of 46

Variant information:

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

Notes: no note is added
Variant Summaries:

Proband HET 19 of 44

Variant information:

Gene	ABCA3
Location	chr16
Mode of inheritance	compound het
CSN	ENST00000301732.5
Consequence	missense variant
dbSNP ID	rs45592239
Variant type	snp
Variant quality	1166.76
REVEL	0.113
HGVSc	ENST00000301732.5:c.2296C>T
HGVSp	ENSP00000301732.5:p.Pro766Ser
gnomAD allele frequency	0.001708
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

Variant information:

Gene	ABCA3
Location	chr16
Mode of inheritance	compound het
CSN	ENST00000301732.5
Consequence	missense variant
dbSNP ID	rs775442517

Variant type	snp
Variant quality	1.09117e-06
REVEL	0.508
HGVSc	ENST00000301732.5:c.740A>C
HGVSp	ENSP00000301732.5:p.Tyr247Ser
gnomAD allele frequency	0.001114
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

Variant information:

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

Notes: no note is added
Variant Summaries:

Proband HET 24 of 43

Variant information:

Gene	NOTCH4
Location	chr6
Mode of inheritance	compound het
CSN	ENST00000375023.3
Consequence	missense variant
dbSNP ID	rs72846312
Variant type	snp
Variant quality	1096.51
REVEL	0.204
HGVSc	ENST00000375023.3:c.3361G>A
HGVSp	ENSP00000364163.3:p.Gly1121Arg
gnomAD allele frequency	7.417e-05
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

Proband HET 24 of 35
Father HET 22 of 42

Variant information:

Gene	NOTCH4
Location	chr6

Mode of inheritance	compound het
CSN	ENST00000375023.3
Consequence	missense variant
dbSNP ID	rs71556915
Variant type	complex
Variant quality	1354.34
REVEL	
HGVSc	ENST00000375023.3:c.813_815delinsGGG
HGVSp	ENSP00000364163.3:p.Asp272Gly
gnomAD allele frequency	0
ExAC allele frequency	.
Impact	moderate

Notes: no note is added
Variant Summaries:

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: [object Object] 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.