Utah Center for Genetic Discovery

iobio

mosaic

Case Description

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Clinical Notes

The patient is diagnosed Dejerine sottas disease. The suspected phenotypes for this patient are hammertoes; distal muscle weakness; pes cavus.

Significant Variants

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:

— *2019-8-30 15:16* *Demo User*

Test note

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

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

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

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

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

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

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

Unknown Significance Variants

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

Genes

Phenotypes searched	Genes (100)
GTR: • Dejerine-Sottas disease Phenolyzer: • Dejerine sottas disease HPO:	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

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.