Utah Center for Genetic Discovery

iobio

Sample:
16012132564

Inputs

No clinical note is added

Significant

Gene	undefined
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* *Tonya Lee Disera*

asdf

Genes

Phenotypes searched	Genes (100)
GTR: • undefined Phenolyzer: • undefined 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.