Yonago Acta Medica 2019;62(3):244–252 doi: 10.33160/yam.2019.09.001

Supplementary Table S2. Rank positions of causative variants in patients' data

Patient	Disease (OMIM term)	*Gene*	HGVS notation (GRCh37/ hg19)	Inheritance	HPO ID					Rank				
										hiPHIVE	PhenIX	Phen-Gen	combined max (eXtasy)	order statistics (eXtasy)
1	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1K	*ALG1*	g.5121993 G>A, g.5128843 C>T	AR	0003256	0002014	0001250	0000252	0001263	7	10	13	62	108
2	SOTOS SYNDROME	*NSD1*	g.176715832C>T	AD	0000256	0001548	0011622	0001643	0001263	1	1	-	25	2
3	JOUBERT SYNDROME 3	*AHI1*	g.135754326G>A, g.135784353C>A	AR	0002419	0000639	0001263			1	1	6	-	-
4	NEUROFIBROMATOSIS TYPE 1	*NF1*	g.29664880G>A	AD	0000957					1	1	-	×	×
5	DEAFNESS, AUTOSOMAL DOMINANT 6	*WFS1*	g.6303911 G>A	AD	0000365					1	1	-	×	×
6	HYPOCHONDROPLASIA	*FGFR3*	g.1807371 C>A	AD	0000256	0007359	0002882	0004322		2	1	-	15	1

Supplementary Table S2 – Continued

7	MITOCHONDRIAL SHORT-CHAIN ENOYL-CoA HYDRATASE 1 DEFICIENCY	*ECHS1*	g.135186833G>A, g.135186837T>C	AR	0007146	0002151	0000639	0001263	0001250	-	-	-	-	-
8	SOTOS SYNDROME	*NSD1*	g.176719045C>T	AD	0000256	0001263				1	1	-	16	13
9	AORTIC ANEURYSM FAMILIAL THORACIC 6	*ACTA2*	g.90699299C>T	AD	0002622					1	1	-	×	×
10	TELANGIECTASIA HEREDITARY HEMORRHAGIC TYPE 1	*ENG*	g.130587554A>AG	AD	0000421					3	1	-	×	×
11	JOUBERT SYNDROME 5	*CEP290*	g.884565556C>T, g.88462434A>T	AR	0002419	0001337	0000496	0001249		1	1	-	-	-
12	MITOCHONDRIAL SHORT-CHAIN ENOYL-CoA HYDRATASE 1 DEFICIENCY	*ECHS1*	g.135184174T>C, g.135186833G>A	AR	0007146	0002072	0001250	0001263		-	-	-	-	-

Supplementary Table S2 – Continued

13	MARFAN SYNDROME	*FBN1*	g.4893692 4CG>C	AD	0001 166	0002 622				1	1	-	×	×
14	NEUROFIBROMATOSIS TYPE1	*NF1*	g.2955052 1CAG>C	AD	0000 957					1	1	-	×	×
15	RETT SYNDROME CONGENITAL VARIANT	*FOXG1*	g.2923693 8C>CG	AD	0005 484	0001 263	0007 359	0002 072		1	1	-	-	-
16	MIGRAINE FAMILIAL HEMIPLEGIC1	*CACNA1A*	g.1341900 3C>G	AD	0006 846	0001 249	0001 250			4	14	-	-	-
17	NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR1	*SHOC2*	g.1127241 20A>G	AD	0001 249	0000 316	0004 322			1	1	-	198	97
18	NEUROFIBROMATOSIS TYPE1	*NF1*	g.2966508 0GC>G	AD	0000 957	0001 263				1	1	-	×	×
19	NIEMANN-PICK DISEASE TYPE C1	*NPC1*	g.2112447 0T>TAG, g.2114147 9G>A	AR	0002 505	0001 730	0001 260	0100 543	0000 130	2	16	19	149	18
20	NEUROFIBROMATOSIS TYPE 1	*NF1*	g.2956301 6T>TA	AD	0000 957					1	1	-	×	×

‘-’ - not ranked. ‘×’ - could not analyze.