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

Supplementary Table S1. Variants details, HPO term and rank positions of causative variants in simulated disease data

ID	Disease (OMIM term)	*Gene*	HGVS notation (GRCh37/ hg19)	Inheritance	HPO ID					Rank				
										hiPHIVE	PhenIX	Phen-Gen	combined max (eXtasy)	order statistics (eXtasy)
1	Usher syndrome 1D/F	*CDH23*	g.73558142T>G	AD,DR	0000365	0000510	0001751			1	1	-	-	-
2	DEAFNESS, AUTOSOMAL DOMINANT 3A; DFNA3A	*GJB2*	g.20763323C>T	AD	0000407					1	1	-	×	×
3	DEAFNESS, AUTOSOMAL DOMINANT 3A; DFNA3A	*GJB2*	g.20763442C>T	AD	0000407					1	3	2	×	×
4	DEAFNESS, AUTOSOMAL DOMINANT 3A; DFNA3A	*GJB2*	g.20763587C>T	AD	0000407					1	1	5	×	×
5	ALZHEIMER DISEASE 3; AD	*PSEN1*	g.73640375C>T	AD	0001260	0001332	0002395	0002354	0002185	1	1	5	1	2
6	DEAFNESS, AUTOSOMAL DOMINANT 20; DFNA20	*ACTG1*	g.79478214C>T	AD	0008619	0000408				1	1	-	279	334

Supplementary Table S1 – Continued

7	PARKINSON DISEASE, LATE-ONSET; PD	*MAPT*	g.44087739A>C	AD,Mu	0000726	0001300	0000751	0002172	0001621	1	2	-	5	2
8	PULMONARY HYPERTENSION, PRIMARY, 1; PPH1	*BMPR2*	g.203329555T>C	AD	0000822	0005308	0005312	0001708	0001009	1	1	-	66	35
9	PULMONARY HYPERTENSION, PRIMARY, 1; PPH1	*BMPR2*	g.203383587_203383588delinsAAGG	AD	0011353	0004964	0005308	0005312	0001667	1	1	-	-	-
10	PULMONARY HYPERTENSION, PRIMARY, 1; PPH1	*BMPR2*	g.203384858T>C	AD	0001977	0005317	0004964	0005308	0001667	1	1	8	73	14
11	FEINGOLD SYNDROME 1; FGLDS1	*MYCN*	g.160859401C>T	AD	0004691	0004692	0000958	0000437	0000232	1	1	-	270	178
12	DEAFNESS, AUTOSOMAL RECESSIVE 9; DFNB9	*OTOF*	g.26682895A>G	AR	0008529	0004463	0000407			1	1	-	×	×
13	DEAFNESS, AUTOSOMAL RECESSIVE 9; DFNB9	*OTOF*	g.26683617T>C	AR	0008529	0004463	0000407			1	1	4	×	×

Supplementary Table S1 – Continued

14	DEAFNESS, AUTOSOMAL RECESSIVE 9; DFNB9	*OTOF*	g.2668386 5C>T	AR		0004 463	0000 407			1	1	4	×	×
15	NOONAN SYNDROME 4; NS4	*SOS1*	g.3926258 1C>A	AD	0000 028	0000 689	0000 494	0000 368	0003 010	1	1	2	2	2
16	DEAFNESS, AUTOSOMAL RECESSIVE 8; DFNB8	*TMPRSS 3*	g.4380408 8G>A	AR	0000 407					1	1	-	×	×
17	GLYCINE ENCEPHALOPATH Y; GCE	*AMT*	g.4945957 8G>A	AR	0001 298	0002 154	0000 737	0001 336	0100 247	1	1	5	8	1
18	METHYLMALONI C ACIDURIA, cblA TYPE	*MMAA*	g.1465607 24C>T	AR	0001 944	0001 987	0002 154	0001 250	0002 013	1	1	-	58	26
19	SOTOS SYNDROME 1; SOTOS1	*NSD1*	g.1766946 45G>A	AD	0002 280	0000 218	0001 943	0000 347	0001 319	1	1	-	17	9
20	CARDIOFACIOCU TANEOUS SYNDROME 1; CFC1	*BRAF*	g.1405013 36C>G	AD	0000 280	0002 750	0000 768	0004 322	0002 217	1	1	5	1	2

Supplementary Table S1 – Continued

21	LONG QT SYNDROME 1; LQT1	*KCNQ1*	g.150648632A>G	AD	0000598	0001657	0001279	0001664	0001663	-	-	-	-	-
22	ANTLEY-BIXLER SYNDROME WITH GENITAL ANOMALIES AND DISORDERED STEROIDOGENESIS; ABS1	*POR*	g.75615163delG	AR	0000818	0012385	0000047	0000054	0002650	1	1	-	14	21
23	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.107303802C>T	AR	0011387	0000376	0000407			1	1	-	×	×
24	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.107323771C>A	AR	0011387	0000376	0000407			1	1	2	×	×

Supplementary Table S1 – Continued

25	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.107334899G>A	AR	0011387	0000376	0000407			1	1	-	×	×
26	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.107350576C>T	AR	0011387	0000376	0000407			1	1	-	×	×
27	CHARGE SYNDROME	*CHD7*	g.61654385G>T	AD	0002023	0002410	0000717	0001511	0000394	1	1	-	-	-
28	BRANCHIOOTORENAL SYNDROME 1; BOR1	*EYA1*	g.72211430G>C	AD	0004742	0009798	0008551	0000410	0000104	1	1	-	5	42
29	BECKWITH-WIEDEMANN SYNDROME; BWS	*CDKN1C*	g.2906409_2906410delinsC	AD	0002060	0004742	0009795	0009797	0000113	7	5	-	-	-

Supplementary Table S1 – Continued

30	WIEDEMANN-STEINER SYNDROME; WDSTS	*KMT2A*	g.118344021delC	AD	0009796	0001374	0010628	0000691	0000384	2	13	-	-	-
31	HYPOURICEMIA, RENAL, 1; RHUC1	*SLC22A12*	g.6436035 5G>A	AR	0001919	0003537	0012611	0000791		1	1	-	×	×
32	DEAFNESS, AUTOSOMAL RECESSIVE 21; DFNB21	*TECTA*	g.120998773G>C	AR	0000407					1	1	-	×	×
33	KABUKI SYNDROME 1; KABUK1	*KMT2D*	g.49420304_49420305delAA	AD	0000164	0000175	0000218	0010314	0000508	1	1	-	-	-
34	KABUKI SYNDROME 1; KABUK1	*KMT2D*	g.49438647G>A	AD	0000592	0001680	0004736	0001212	0000403	1	1	-	-	-
35	KABUKI SYNDROME 1; KABUK1	*KMT2D*	g.49444463C>A	AD	0000365	0001382	0000252	0000358	0000535	1	1	-	-	-
36	KABUKI SYNDROME 1; KABUK1	*KMT2D*	g.49444682_49444683insGAGGCCATCCA	AD	0001007	0001249	0001382	0008897	0002650	1	1	-	-	-

Supplementary Table S1 – Continued

37	KABUKI SYNDROME 1; KABUK1	*KMT2D*	g.49447088_49447089delAG	AD	0000164	0004736	0000400	0000358	0004467	1	1	-	-	-
38	LISSENCEPHALY 3; LIS3	*TUBA1A*	g.49580430G>A	AD	0001251	0010864	0000252	0001250	0002510	2	1	-	126	31
39	DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A	*GJB2*	g.20763691dupC	DR,AR	0000407	0001751				1	1	2	×	×
40	PITUITARY HORMONE DEFICIENCY, COMBINED, 6; CPHD6	*OTX2*	g.57268761C>A	AD	0011755	0004322				1	1	-	×	×
41	CARDIOFACIOCUTANEOUS SYNDROME 1; CFC1	*BRAF*	g.25380280C>G	AD	0000463	0001622	0002019	0000486	0001276	-	-	-	-	-
42	NIJMEGEN BREAKAGE SYNDROME; NBS	*NBN*	g.90983445_90983449delGTTTT	AR	0000957	0000175	0000444	0006532	0003202	1	1	-	-	-

Supplementary Table S1 – Continued

43	LOEYS-DIETZ SYNDROME 2; LDS2	*TGFBR2*	g.3071386 6C>G	AD	0002 308	0001 363	0004 955	0100 259	0002 650	1	1	4	1	36
44	MARFAN SYNDROME; MFS	*FBN1*	g.4871292 3G>A	AD	0000 494	0007 676	0007 800	0001 548	0003 179	1	1	-	3	68
45	ANGELMAN SYNDROME; AS	*UBE3A*	g.2561693 7delC	IC	0001 344	0002 286	0001 347	0005 484	0010 808	1	1	-	-	-
46	ANGELMAN SYNDROME; AS	*UBE3A*	g.2562083 7A>G	IC	0001 344	0002 019	0000 272	0000 639	0010 808	1	1	-	62	21
47	RUBINSTEIN-TAYBI SYNDROME 1; RSTS1	*CREBBP*	g.3801726 C>A	AD	0000 589	0000 028	0005 895	0002 788	0000 736	1	1	-	-	-
48	LI-FRAUMENI SYNDROME; LFS	*TP53*	g.7574031 delG	AD	0002 488	0006 744	0030 078	0002 667	0002 669	1	1	-	-	-
49	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.1073405 60_10734 0561insT	AR	0011 387	0000 376	0000 407			1	1	5	×	×
50	CHARGE SYNDROME	*CHD7*	g.6165459 5C>T	AD	0001 719	0200 021	0000 252	0000 347	0000 054	1	1	-	-	-

Supplementary Table S1 – Continued

51	CHARGE SYNDROME	*CHD7*	g.61654604C>T	AD	0000717	0001156	0000324	0001539	0004496	1	1	4	-	-
52	CHARGE SYNDROME	*CHD7*	g.61655016delA	AD	0000834	0100736	0003974	0010751	0008551	1	1	-	-	-
53	CHARGE SYNDROME	*CHD7*	g.61655027A>T	AD	0000048	0000204	0000378	0002937	0003022	1	1	3	-	-
54	CHARGE SYNDROME	*CHD7*	g.61693850_61693851delCC	AD	0000682	0010669	0000160	0000388	0001629	1	1	-	-	-
55	CHARGE SYNDROME	*CHD7*	g.61728951_61728955delATCTT	AD	0000772	0000324	0000044	0000394	0001561	1	1	-	-	-
56	CHARGE SYNDROME	*CHD7*	g.61749422C>T	AD	0000772	0002015	0000501	0000568	0004058	1	1	3	-	-
57	CHARGE SYNDROME	*CHD7*	g.61749529C>A	AD	0000772	0001360	0002901	0000066	0001171	1	1	-	-	-
58	CHARGE SYNDROME	*CHD7*	g.61763080dupT	AD	0001305	0000821	0001601	0001252	0001883	1	1	6	-	-
59	CHARGE SYNDROME	*CHD7*	g.61769239T>A	AD	0009738	0001679	0000488	0001883	0001629	5	1	3	-	-
60	GLYCINE ENCEPHALOPATHY; GCE	*GLDC*	g.6536188A>C	AR	0001274	0000718	0000737	0001336	0000711	1	2	-	191	85

Supplementary Table S1 – Continued

61	GLYCINE ENCEPHALOPATHY; GCE	*GLDC*	g.6588688G>C	AR	0003108	0001249	0000737	0100247	0001250	1	1	4	219	159
62	CORNELIA DE LANGE SYNDROME 3; CDLS3	*HDAC8*	g.71787820G>A	AD	0001263	0000218	0000527	0000545	0200055	1	1	3	60	94
63	FOCAL DERMAL HYPOPLASIA; FDH	*PORCN*	g.48368278C>T	XLD	0001274	0004334	0100559	0010622	0009381	1	1	2	340	198
64	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, TYPE 2; HHT2	*ACVRL1*	g.52307534C>T	AD	0001342	0002249	0000434	0001901	0001694	1	1	-	163	76
65	MOWAT-WILSON SYNDROME; MOWS	*ZEB2*	g.145158826_145158827delCT	AD	0040082	0009748	0004415	0004322	0001636	-	-	-	-	-
66	MOWAT-WILSON SYNDROME; MOWS	*ZEB2*	g.145158871G>A	AD	0004298	0008572	0000768	0002558	0000076	-	-	-	-	-

Supplementary Table S1 – Continued

67	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, TYPE 1; HHT1	*ENG*	g.130579483delC	AD	0002094	0001901	0001694	0001250	0004406	1	1	-	-	-
68	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, TYPE 1; HHT1	*ENG*	g.130592008delG	AD	0001903	0100858	0002573	0001722	0011934	1	1	-	×	×
69	PHENYLKETONURIA; PKU	*PAH*	g.103234288A>C	AR	0000718	0002514	0004923	0001347	0000737	-	-	-	-	-
70	ALAGILLE SYNDROME 1; ALGS1	*PAH*	g.103245474C>A	AD	0000518	0000490	0002155	0000110	0001328	10	4	7	8	3
71	ALAGILLE SYNDROME 1; ALGS1	*PAH*	g.103246680C>G	AD	0000585	0000490	0002937	0003022	0000486	10	14	3	7	3
72	ALAGILLE SYNDROME 1; ALGS1	*PAH*	g.103246696C>G	AD	0002910	0002937	0000400	0004969	0001947	9	10	-	1	1
73	ALAGILLE SYNDROME 1; ALGS1	*PAH*	g.103306582A>G	AD	0000772	0001394	0000490	0005280	0001629	28	14	-	55	92

Supplementary Table S1 – Continued

74	MUCOPOLYSACCHARIDOSIS, TYPE II; MPS2	*IDS*	g.148564339G>A	XLR	0002159	0002180	0000648	0001085	0001761	1	1	-	95	79
75	EXOSTOSES, MULTIPLE, TYPE II	*EXT2*	g.44151637C>A	AD	0003068	0003276	0003406	0000896	0000918	1	1	2	×	×
76	MARFAN SYNDROME; MFS	*FBN1*	g.48729201G>A	AD	0002647	0000494	0100775	0000565	0003088	4	1	-	22	88
77	MARFAN SYNDROME; MFS	*FBN1*	g.48729982C>A	AD	0001659	0000268	0007676	0000189	0004927	1	1	3	1	6
78	MARFAN SYNDROME; MFS	*FBN1*	g.48757764C>T	AD	0001166	0000268	0002816	0000501	0004927	1	1	2	-	-
79	MARFAN SYNDROME; MFS	*FBN1*	g.48780384T>C	AD	0001635	0001002	0000494	0000275	0000278	1	1	-	1	1
80	Loeys-Dietz syndrome 2	*TGFBR2*	g.30691785C>T	AD	0001156	0000175	0000316	0001634	0000278	1	1	-	1	2
81	Loeys-Dietz syndrome 2	*TGFBR2*	g.30713345C>T	AD	0002631	0001647	0001519	0009473	0002650	1	1	-	2	114
82	Loeys-Dietz syndrome 2	*TGFBR2*	g.30713705T>C	AD	0000766	0005807	0004955	0001388	0000347	1	1	3	1	1
83	Loeys-Dietz syndrome 2	*TGFBR2*	g.30732918C>T	AD	0000766	0004944	0001634	0001643	0100259	1	1	3	3	5
84	Loeys-Dietz syndrome 2	*TGFBR2*	g.30732969C>T	AD	0001631	0004955	0009473	0000939	0001762	-	-	-	-	-

Supplementary Table S1 – Continued

85	Loeys-Dietz syndrome 2	*TGFBR2*	g.30732970G>A	AD	0001166	0002308	0012385	0004944	0001363	1	1	6	12	108
86	Loeys-Dietz syndrome 1	*TGFBR1*	g.101911535G>A	AD	0005182	0001363	0000272	0000977	0001762	1	1	3	58	12
87	Mental retardation, X-linked, syndromic, Christianson type	*SLC9A6*	g.135080642_135080643delGA	XLD	0000717	0100543	0001263	0000400	0000020	-	-	-	-	-
88	Schizophrenia	*NOS1AP*	g.162337012G>A	AD	0000746	0002353	0000738	0100753	0007086	5	36	-	964	1111
89	Pseudoxanthoma elasticum	*ABCC6*	g.162569404G>A	AR	0001635	0001034	0004417	0001718	0000573	1	1	3	134	39
90	Pseudoxanthoma elasticum	*ABCC6*	g.162596709A>G	AR	0004943	0007663	0000083	0100817	0000573	1	1	-	-	-
91	Pseudoxanthoma elasticum	*ABCC6*	g.162919600C>T	AR	0000153	0004417	0000608	0001718	0001723	1	1	-	25	6
92	Hypertrophic osteoarthropathy, primary, autosomal recessive, 2	*SLCO2A1*	g.133672567C>T	AR	0002829	0001217	0030314			1	1	-	496	382
93	Ciliary dyskinesia, primary, 3	*DNAH5*	g.13777315delC	-	0012265	0002205	0001696			1	1	-	-	-
94	Ciliary dyskinesia, primary, 3	*DNAH5*	g.138307804G>A	-	0012265	0002205	0001696			1	1	-	3	1

Supplementary Table S1 – Continued

95	DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT; DFNB4	*SLC26A4*	g.107340538C>G	AR	0011387	0000376	0000407			1	1	5	×	×
96	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL; TTP	*ADAMTS13*	g.136307568A>T	AR	0011387	0000376	0000407			12	20	-	×	×
97	Fabry disease	*GLA*	g.100653458A>G	XL	0003119	0002376	0000962	0005144	0003326	1	1	-	1	1
98	Fabry disease	*GLA*	g.100653816A>G	XL	0001131	0100820	0003394	0003401	0000407	1	1	3	10	5
99	Fabry disease	*GLA*	g.100653930T>C	XL	0001369	0000708	0003326	0001658	0000093	1	1	-	1	2
100	Fabry disease	*GLA*	g.100656798delC	XL	0001681	0001014	0001369	0100543	0012702	1	1	-	-	-

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