

#	Variant	HPO	Specification (spec)	Variant Type	Inheritance	Tool	PVS1	PS1	PS2	PS3	PS4	PM1	PM2	PM3	PM4	PM5	PM6	PP1	PP2	PP3	PP4	PP5	BA1	BS1	BS2	BS3	BS4	BP1	BP2	BP3	BP4	BP5	BP6	BP7	Final Classification	Points					
1	PIEZ01 (NM_001142864.4):c.7367G>A	HP-0001878 Hemolytic anemia	no	missense	AR/AD	Varsome				4	1	4	2			2					4														Pathogenic	10					
						Franklin										2				2		1		2													Pathogenic	12			
						MobiDetails_hg38_intervar									2	2								1			1											VUS	5		
						MobiDetails_GeneBe									2	2				2						8												Pathogenic	16		
						VarChat														2						4												Likely Pathogenic	7		
2	LZTR1 (NM_006767.4):c.1078A>T	HP-0001667 Right ventricular hypertrophy; HP-0005143 Anomalous origin of right pulmonary artery from ascending aorta; HP-0001639 Hypertrophic cardiomyopathy	yes	nonsense	AR/AD	human_1				2	2	1	1			1		4		2		4														Pathogenic	13				
						human_2				1	4	1	1			1	2		2																			Pathogenic	12		
						Varsome	8						1													1													Pathogenic	10	
						Franklin	8				2														1														Pathogenic	12	
						MobiDetails_hg38_intervar	8																		1														Pathogenic	11	
3	NF1 (NM_001042492.3):c.4974dup	HP-0001067 Neurofibroma; HP-0007565 Multiple cafe-au-lait spots	in prep	nonsense	AD	MobiDetails_GeneBe																	2													Pathogenic	12				
						VarChat	8																																Likely Pathogenic	8	
						human_1	8																																Pathogenic	10	
						human_2	8																																Pathogenic	12	
						Varsome	8																				1												Likely Pathogenic	9	
4	KCNQ1 (NM_000218.3):c.832T>G	HP-0001657 Prolonged QT interval	in prep	missense	AD	Franklin	8						2										1														Pathogenic	11			
						MobiDetails_hg38_intervar	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	Pathogenic	12	
						MobiDetails_GeneBe	8																				2												Pathogenic	12	
						VarChat	8																																	Likely Pathogenic	8
						human_1	8																																	Likely Pathogenic	10
5	NF1 (NM_001042492.3):c.6772C>T	HP-0001067 Neurofibroma	in prep	nonsense	AD	human_2	8																														Likely Pathogenic	9			
						Varsome	8																																	Pathogenic	17
						Franklin	8																																	Pathogenic	14
						MobiDetails_hg38_intervar	8																																	Pathogenic	12
						MobiDetails_GeneBe	8																																	Pathogenic	18
6	SLC11A2 (NM_000617.3):c.223G>A	HP-0001903 Anemia; HP-0001935 Microcytic anemia	no	missense	AR	VarChat	8																														Pathogenic	15			
						human_1	8																																	Pathogenic	12
						human_2	8																																	Pathogenic	12
						Varsome	8																																	VUS	5
						Franklin	8																																	Likely Pathogenic	8
7	CYP24A1 (NM_000782.5):c.428_430del	HP-0000787 Nephrolithiasis; HP-0003072 Hypercalcemia	no	deletion	AR	MobiDetails_GeneBe																															Likely Pathogenic	6			
						VarChat																																		Likely Pathogenic	7
						human_1																																		Likely Pathogenic	6
						human_2																																		VUS	4
						Varsome	8																																	Pathogenic	11
8	HOGA1 (NM_138413.4):c.208C>T	HP-0000787 Nephrolithiasis; HP-0003072 Hypercalcemia	no	nonsense	AR	Franklin	8																														Likely Pathogenic	8			
						MobiDetails_hg38_intervar	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	Pathogenic	11		
						MobiDetails_GeneBe	8																																	Pathogenic	10
						VarChat	8																																	Pathogenic	10
						human_1	8																																	Pathogenic	10
9	XDH (NM_000379.4):c.2097del	HP-0000787 Nephrolithiasis	no	nonsense	AR	human_2	8																														Likely Pathogenic	9			
						Varsome	8																																	Pathogenic	10
						Franklin	8																																	Pathogenic	10
						MobiDetails_hg38_intervar	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	Pathogenic	10	
						MobiDetails_GeneBe	8																																	Pathogenic	10
10	CFTR (NM_000492.3):c.1521_1523delN	HP-0012265 Ciliary dyskinesia	no	deletion	AR/AD	VarChat	8																														Likely Pathogenic	9			
						human_1	8																																	Pathogenic	10
						human_2	8																																	Likely Pathogenic	9
						Varsome	8																																	Pathogenic	23
						Franklin	8																																	Pathogenic	11
11	CCDC103 (NM_213607.3):c.461A>C	HP-0012265 Ciliary dyskinesia; HP-0001696 Situs inversus totalis	no	missense	AR	MobiDetails_hg38_intervar	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	Likely Pathogenic	7				
						MobiDetails_GeneBe	8																																	Pathogenic	12
						VarChat	8																																	VUS	3
						human_1	8																																	Likely Pathogenic	9
						human_2	8																																	VUS	5
12	F8 (NM_000132.4):c.608T>C	HP-0003125 Reduced factor VIII activity	yes	missense	XLR	human_1	8																														VUS	3			
						human_2	8																																	VUS	5
						Varsome	8																																	Likely Pathogenic	7
						Franklin	8																																	VUS	5
						MobiDetails_hg38_intervar	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	/	VUS	4
13	AKT1 (NM_001382430.1):c.1099C>T	HP-0001528 Hemihypertrophy	no	missense	AD	MobiDetails_GeneBe	8																														VUS	3			
						VarChat	8																																	VUS	4
						human_1	8																																	VUS	2
						human_2	8																																	VUS	2
						Varsome	8																																	VUS	3
14	SPINK5 (NM_006846.4):c.153del	HP-0008064 Ichthyosis	no	nonsense	AR	human_1	8																															Pathogenic	10		
						human_2	8																																	Pathogenic	14
						Varsome	8																																		

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89	NPR2 (NM_003995.4):c.430C>G	HP-0004322 Short stature	no	missense	ARI/AD	Franklin																							VUS	4					
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
						human_1																													
human_2																																			
90	ALPL (NM_000478.6):c.542C>T	HP-0004322 Short stature	no	missense	ARI/AD	Varsome																								Pathogenic	17				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
91	SLC34A1 (NM_003052.5):c.1223T>A	HP-000121 Nephrocalcinosis	no	missense	ARI/AD	Varsome																								Pathogenic	11				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
92	FLG (NM_002016.2):c.2282_2285del	HP-0001047 Atopic dermatitis	no	nonsense	ARI/AD	Varsome																								Pathogenic	16				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
93	ASS1 (NM_054012.4):c.1168G>A	HP-0001987 Hyperammonemia	no	missense	AR	Varsome																								Pathogenic	17				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
94	RYR2 (NM_001035.3):c.3767C>T	HP-0004756 Ventricular tachycardia	no	missense	AD	Varsome																								Pathogenic	9				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
95	FLNB NM_001457.4):c.7760G>A	HP-0004322 Short stature	no	nonsense	ARI/AD	Varsome																								Likely Pathogenic	9				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
96	ACAN (NM_001369268.1):c.7492G>A	HP-0004322 Short stature	no	missense	ARI/AD	Varsome																								VUS	0				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
97	ANKRD11 (NM_013275.6):c.3931C>T	HP-0025169 Left ventricular systolic dysfunction	no	nonsense	AD	Varsome																								Pathogenic	13				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
98	F11 (NM_000128.4):c.1556G>A	HP-0001929 Reduced factor XI activity	in prep	nonsense	ARI/AD	Varsome																								Pathogenic	13				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
99	SLC5A1 (NM_000343.4):c.188G>A	HP-0001943 Hypoglycemia	no	missense	AR	Varsome																								Likely Pathogenic	6				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
100	GCK (NM_000162.5):c.863+1G>C	HP-0004904 Maturity-onset diabetes of the young	yes	splicing	ARI/AD	Varsome																								Pathogenic	11				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													
human_1																																			
human_2																																			
101	F7 (NM_019816.4):c.995C>T	HP-0001928 Abnormality of coagulation	in prep	missense	AR	Varsome																								Pathogenic	12				
						Franklin																													
						MobiDetails_hg38_intervar																													
						MobiDetails_GeneBe																													
						VarChat																													

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