Appendix 2 (as submitted by the authors): Table S1 Pathogenic SNVs, pathogenic CNVs-SVs and rare coding CNVs

ID	Gene: Accession	Variant	Genomic position	Zygosity	Disease	Inheritance	ACMG interpretation	Evidence	PMID
PGPC-01	RARS2: NM_020320	c.C997G, p.R333G	chr6:88231220G>C	het	Pontocerebellar hypoplasia	AR	Recessive likely pathogenic (II)	PS1, PM2	25289895
PGPC-01	OPTN: NM_001008211	c.1239_1240del, p.K413fs	chr10:13168036delAG	het	Glaucoma, open angle	AR	Recessive likely pathogenic (II)	PS1, PM2	
PGPC-02	CC2D2A: NM_001080522	c.4179delG, p.E1393fs	chr4:15589552delG	het	Meckel syndrome	AR	Recessive pathogenic (Ia)	PVS1, PS4	19466712
PGPC-02	VPS13B: NM_017890	c.11906_11916delCCAGCTGTTCTinsG	chr8:100887731delCCAGCTGTTCTinsG	het	Cohen syndrome	AR	Recessive pathogenic (Ia)	PVS1, PS4	15141358
PGPC-02	HEXA: NM_000520	c.1277_1278insTATC, p.S426fs	chr15:72638921insGATA	het	Tay-Sachs disease	AR	Recessive pathogenic (Ia)	PVS1, PS4	2848800
PGPC-02	ABCA3: NM_001089	c.A875T, p.E292V	chr16:2367764T>A	het	Surfactant metabolism dysfunction	AR	Recessive pathogenic (II)	PS4, PS3	15976379
PGPC-02	CHEK2: NM_145862	c.T470C, p.I157T	chr22:29121087A>G	het	Li-Fraumeni syndrome 2	AD/AR	Risk factor	PS4, PP5	10617473
PGPC-03	PHKB: NM_000293	c.2381_2384del, p.S794fs	chr16:47698840delCCTC	het	Glycogen storage disease IXb	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-03	AMPD1: NM_001172626	c.G1017T, p.M339I	chr1:115221116C>A	het	Myoadenylate deaminase deficiency	AR	Recessive likely pathogenic (II)	PS3, PM3, BP4	15173240
PGPC-03	SLC6A19: NM_001003841	c.G517A, p.D173N	chr5:1212453G>A	het	Hartnup disease	AR	Recessive likely pathogenic (II)	PS1, PP1-M	15286788
PGPC-03	IVD: NM_001159508	c.C851T, p.A284V	chr15:40707653C>T	het	Isovaleric acidemia	AR	Recessive pathogenic (II)	PS3, PS4	9665741
PGPC-04	CLCNKB: NM_001165945	c.118delA, p.R40fs	chr1:16375584delA	het	Bartter syndrome	AR/Digenic	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-04	CYP24A1: NM_000782	c.A667T, p.R223X	chr20:52782346T>A	het	1,25(OH)(2)D-24-hydroxylase deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-05	DTNBP1: NM_183040	c.C853T, p.R285X	chr6:15524715G>A	het	Hermansky-Pudlak syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-05	HOGA1: NM_138413	c.937_939del, p.313_313del	chr10:99371369delGAG	het	Hyperoxaluria	AR	Recessive likely pathogenic (II)	PS4, PM2	20797690
PGPC-05	NHLRC1: NM_198586	c.G436A, p.D146N	chr6:18122402C>T	het	Epilepsy, progressive myoclonic (Lafora)	AR	Recessive likely pathogenic (II)	PS4, PM2, PP5	12958597
PGPC-05	GBA: NM_001171811	c.A965G, p.N322S	chr1:155205634T>C	het	Gaucher disease	AR	Recessive pathogenic (II)	PS4, PP5, PS3	3353383
PGPC-06	ABCA12: NM_173076	c.C2140T, p.R714X	chr2:215876355G>A	het	Ichthyosis	AR	Recessive likely pathogenic (I)	PVS1, PM2	16902423
PGPC-06	NME8: NM_016616	c.G1090T, p.E364X	chr7:37924000G>T	het	Ciliary dyskinesia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-06	ADCK3: NM_020247	c.1746_1748del, p.582_583del	chr1:227174240delCAC	het	Progressive cerebellar ataxia and atrophy	AR	Recessive likely pathogenic (II)	PS3, PM2	18319074
PGPC-06	GJB2: NM_004004	c.167delT, p.L56fs	chr13:20763554delA	het	Deafness	AR	Recessive pathogenic (Ia)	PVS1, PS4	9285800
PGPC-06	MEFV: NM_000243	c.T2177C, p.V726A	chr16:3293310A>G	het	Familial Mediterranean fever	AR	Recessive pathogenic (II)	PS1, PS4, BP4	9288758
PGPC-07	IL1RN: NM_173842	c.C76T, p.R26X	chr2:113885277C>T	het	Osteomyelitis	AR	Recessive likely pathogenic (I)	PVS1, PM2	26100510
PGPC-07	RARS2: NM_020320	c.G773A, p.R258H	chr6:88239365C>T	het	Pontocerebellar hypoplasia	AR	Recessive likely pathogenic (V)	PM2, PM3, PP1, PP3	24047924
PGPC-08	DNAJC6: NM_001256865	c.2281+1G>C	chr1:65871817G>C	het	Juvenile Parkinsonism	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-08	CFTR: NM_000492	c.1520_1522del, p.507_508del	chr7:117199645delTCT	het	Cystic fibrosis	AR	Recessive pathogenic (II)	PS4, PS3	
PGPC-09	XDH: NM_000379	c.930_931insC, p.T311fs	chr2:31605975insG	het	Xanthinuria	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-09	TGDS: NM_014305	c.G298T, p.A100S	chr13:95243122C>A	het	Catel-Manzke syndrome	AR	Recessive likely pathogenic (II)	PS4, PM3, PP3	25480037
PGPC-09	TMPRSS15: NM_002772	c.C2135G, p.S712X	chr21:19685292G>C	het	Enterokinase deficiency	AR	Recessive pathogenic (Ia)	PVS1, PS4	11719902
PGPC-09	CFTR: NM_000492	c.G1040A, p.R347H	chr7:117180324G>A	het	Cystic fibrosis	AR	Recessive pathogenic (II)	PS4, PM2, PP3, PS1	1284538
PGPC-10	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-10	WNK1: NM_001184985	c.3378+1G>T	chr12:978271G>T	het	Neuropathy	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-10	GNE: NM_001190383	c.T1538C, p.I513T	chr9:36219891A>G	het	Inclusion body myopathy	AR	Recessive pathogenic (II)	PS4, PM3, PS1	12497639
PGPC-11	CFTR: NM_000492	c.1520_1522del, p.507_508del	chr7:117199645delTCT	het	Cystic fibrosis	AR	Recessive pathogenic (II)	PS4, PS3	
PGPC-12	ORC6: NM_014321	c.T2C, p.M1T	chr16:46723619T>C	het	Meier-Gorlin syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM3	22333897
PGPC-12	MOGS: NM_006302	c.C370T, p.Q124X	chr2:74691832G>A	het	Congenital disorder of glycosylation	AR	Recessive pathogenic (Ib)	PVS1, PM3, PM2	26805780
PGPC-12	CTH: NM_153742	c.C200T, p.T67I	chr1:70881670C>T	het	Cystathioninuria	AR	Recessive pathogenic (II)	PS1, PS4, PP3	12574942
PGPC-12	AGXT: NM_000030	c.G508A, p.G170R	chr2:241810850G>A	het	Hyperoxaluria	AR	Recessive pathogenic (II)	PS4, PS3, PM3, PP3	1703535
PGPC-13	NCF4: NM_013416	c.C808T, p.R270X	chr22:37271875C>T	het	Granulomatous disease	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-13	USH2A: NM_206933	c.G2276T, p.C759F	chr1:216420460C>A	het	Usher syndrome	AR	Recessive likely pathogenic (II)	PS4, PM3	10775529
PGPC-13	F7: NM_019616	c.C995T, p.A332V	chr13:113772982C>T	het	Factor VII deficiency	AR	Recessive likely pathogenic (II)	PS4, PM3	7981691
PGPC-13	C8B: NM_001278543	c.C1126T, p.R376X	chr1:57406638G>A	het	Complement component 8 deficiency	AR	Recessive pathogenic (Ia)	PVS1, PS4	8098723
PGPC-14	CLCNKB: NM_001165945	c.8_9insCCCTC, p.C3fs	chr1:16375475insCCCTC	het	Bartter syndrome	AR/Digenic	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-14	CLRN1: NM_174878	c.148_149insTGTC, p.S50fs	chr3:150690348insGACA	het	Retinitis pigmentosa	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-14	KCNE2: NM_172201	c.C29T, p.T10M	chr21:35742806C>T	het	Long QT syndrome; Atrial fibrillation	AD	VUS - not enough evidence	PP5, BS1	15840476
PGPC-15	LAMA2: NM_001079823	c.2045_2046del	chr6:129573389delAG	het	Muscular dystrophy	AR	Recessive likely pathogenic (I)	PVS1, PM2	9541105
PGPC-15	ALDH5A1: NM_170740	c.G612A, p.W204X	chr6:24505099G>A	het	Succinic semialdehyde dehydrogenase deficiency	AR	Recessive pathogenic (Ia)	PVS1, PS4	11243727
PGPC-15	NPC2: NM_006432	c.G58T, p.E20X	chr14:74959920C>A	het	Niemann-pick disease	AR	Recessive pathogenic (Ia)	PVS1, PS4	11125141
PGPC-15	ISCU: NM_014301	c.343+382G>C	chr12:108961426G>C	het	Myopathy with lactic acidosis	AR	Recessive pathogenic (II)	PS3, PS4	18304497
PGPC-16	ELN: NM_001278914	c.455-1G>A	chr7:73459551G>A	het	Supravalvular aortic stenosis	AD	Likely pathogenic (I)	PVS1, PM2	
PGPC-16	LZTR1: NM_006767	c.772delT, p.F258fs	chr22:21344795delT	het	Schwannomatosis	AD	Likely pathogenic (I)	PVS1, PM2	
PGPC-16	DPYD: NM_000110	c.G1376A, p.W459X	chr1:98015264C>T	het	Dihydropyrimidine dehydrogenase deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-16	DNAH11: NM_001277115	c.10219_10220del, p.L3407fs	chr7:21847554delCT	het	Ciliary dyskinesia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-16	TCIRG1: NM_006019	c.1674-1G>A	chr11:67816547G>A	het	Osteopetrosis	AR	Recessive likely pathogenic (I)	PVS1, PM2	10888887
PGPC-17	SLC25A20: NM_000387	c.418-1G>A	chr3:48900093C>T	het	Carnitine-acylcarnitine translocase deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-18	CEP135: NM_025009	c.A3211T, p.K1071X	chr4:56885717A>T	het	Microcephaly	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-18	ITGA7: NM_001144997	c.C2989T, p.Q997X	chr12:56078988G>A	het	Muscular dystrophy	AR	Recessive likely pathogenic (I)	PVS1, PM2	

Appendix to: Reuter MS, Walker S, Thiruvahindrapuram B, et al. The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. CMAJ 2018. doi: 10.1503/cmaj.171151. Copyright © 2018 The Author(s) or their employer(s).
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PGPC-18	PEX1: NM_000466	c.2097dupT, p.I700fs	chr7:92132484dupA	het	Heimler syndrome	AR	Recessive pathogenic (Ia)	PVS1, PS4, PP5	10447258
PGPC-18	CFTR: NM_000492	c.1520_1522del, p.507_508del	chr7:117199645delTCT	het	Cystic fibrosis	AR	Recessive pathogenic (II)	PS4, PS3	
PGPC-19	FLG: NM_002016	c.C1501T, p.R501X	chr1:152285861G>A	het	Ichthyosis vulgaris	AD/AR	(Recessive) Pathogenic (Ia)	PVS1, PS4	16444271
PGPC-19	TRIP11: NM_004239	c.2140_2141del, p.L714fs	chr14:92472179delAG	het	Achondrogenesis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-19	AGXT: NM_000030	c.G508A, p.G170R	chr2:241810850G>A	het	Hyperoxaluria	AR	Recessive pathogenic (II)	PS4, PS3, PM3, PP3	1703535
PGPC-19	ABCA3: NM_001089	c.A875T, p.E292V	chr16:2367764T>A	het	Surfactant metabolism dysfunction	AR	Recessive pathogenic (II)	PS4, PS3	15976379
PGPC-19	CHMP2B: NM_014043	c.A85G, p.I29V	chr3:87289899A>G	het	Amyotrophic lateral sclerosis; Dementia	AD	VUS - not enough evidence	PS3, PP5, BP4	20352044
PGPC-20	ASPM: NM_018136	c.7243delC, p.Q2415fs	chr1:197071138delG	het	Microcephaly	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-21	C6: NM_001115131	c.821delA, p.Q274fs	chr5:41181567delT	het	Complement component 6 deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	10632667
PGPC-21	RPGRIP1L: NM_015272	c.T2093G, p.L698X	chr16:53686506A>C	het	Joubert syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-21	TMPRSS3: NM_024022	c.C413A, p.A138E	chr21:43808545G>T	het	Deafness	AR	Recessive likely pathogenic (II)	PM2, PS1	16283880
PGPC-22	AURKC: NM_001015878	c.C744G, p.Y248X	chr19:57746411C>G	het	Spermatogenic failure	AR	Recessive pathogenic (Ia)	PVS1, PS4	22888167
PGPC-22	ABCA4: NM_000350	c.G2588C, p.G863A	chr1:94517254C>G	het	Stargardt disease	AR	Recessive pathogenic (II)	PS1, PS3	9054934
PGPC-23	DHTKD1: NM_018706	c.1897-1G>A	chr10:12148244G>A	het	Charcot-Marie-Tooth disease	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-23	FREM1: NM_144966	c.T3971G, p.L1324R	chr9:14792751A>C	het	Manitoba oculotrichoanal syndrome	AR	Recessive likely pathogenic (II)	PM2, PP3, PS1	21507892
PGPC-23	NAGA: NM_000262	c.G973A, p.E325K	chr22:42457056C>T	het	Kanzaki disease	AR	Recessive pathogenic (II)	PS1, PS4	2243144
PGPC-24	TTC21B: NM_024753	c.2630_2633del, p.P877fs	chr2:166758356delGCAG	het	Short-rib thoracic dysplasia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-24	ARL13B: NM_182896	c.823dupA, p.E274fs	chr3:93761882insA	het	Joubert syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-24	ATP7B: NM_000053	c.A2305G, p.M769V	chr13:52532497T>C	het	Wilson disease	AR	Recessive pathogenic (II)	PS1, PS4	7626145
PGPC-24	LPL: NM_000237	c.A953G, p.N318S	chr8:19813529A>G	het	Dyslipidemia	AD/AR	Risk factor	PS3, PS4	7647785
PGPC-25	LMNA: NM_170707	c.C1748T, p.S583L	chr1:156108328C>T	het	Lipodystrophy, partial	AD/AR	(Recessive) Likely pathogenic (II)	PM1, PM2, PP1, PP3	
PGPC-25	TRIP11: NM_004239	c.5719+2T>C	chr14:92439059A>G	het	Achondrogenesis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-25	COX15: NM_004376	c.396-3C>G	chr10:101486914G>C	het	Leigh syndrome	AR	Recessive likely pathogenic (II)	PM2, PS3	12474143
PGPC-25	F2: NM_000506	c.*97G>A	chr11:46761055G>A	het	Prothrombin deficiency	AD/AR	Risk factor	PS3, PS4	8916933
PGPC-26	FLG: NM_002016	c.2282_2285del, p.S761fs	chr1:152285077delACTG	het	Icthyosis vulgaris	AD/AR	(Recessive) Pathogenic (Ia)	PVS1, PS4	16444271
PGPC-26	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-26	TNFRSF13B: NM_012452	c.C492G, p.Y164X	chr17:16843779G>C	het	Immunoglobulin A deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	27123465
PGPC-26	VWF: NM_000552	c.G2561A, p.R854Q	chr12:6143978C>T	het	von Willebrand disease	AR	Recessive pathogenic (II)	PS3, PS4, PM3	1832934
PGPC-26	GOSR2: NM_054022	c.G430T, p.G144W	chr17:45012488G>T	het	Epilepsy	AR	Recessive pathogenic (II)	PS4, PS3	21549339
PGPC-27	IFT80: NM_020800	c.1213delT, p.S405fs	chr3:160018773delA	het	Short-rib thoracic dysplasia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-27	GJB2: NM_004004	c.35delG, p.G12fs	chr13:20763686delC	het	Deafness	AR	Recessive pathogenic (Ia)	PVS1, PS4	9285800
PGPC-27	CHEK2: NM_145862	c.T470C, p.I157T	chr22:29121087A>G	het	Li-Fraumeni syndrome 2	AD/AR	Risk factor	PS4, PP5	10617473
PGPC-28	TSPYL1: NM_003309	c.745delG, p.A249fs	chr6:116600249delC	het	Sudden infant death syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-28	GJB2: NM_004004	c.35delG, p.G12fs	chr13:20763686delC	het	Deafness	CX	Recessive pathogenic (Ia)	PVS1, PS4	9285800
PGPC-28	VWF: NM_000552	c.G2561A, p.R854Q	chr12:6143978C>T	het	von Willebrand disease	AR	Recessive pathogenic (II)	PS3, PS4, PM3	1832934
PGPC-29	F2: NM_000506	c.*97G>A	chr11:46761055G>A	het	Prothrombin deficiency	AD/AR	Risk factor	PS3, PS4	8916933
PGPC-31	USH2A: NM_206933	c.15297+1G>C	chr1:215807800C>G	het	Usher syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-31	ABCA12: NM_173076	c.C214T, p.Q72X	chr2:215928892G>A	het	Ichthyosis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-32	MT-TV		chrM:1659T>C	7% heteropla	Childhood neurologic disorder		Likely pathogenic		
PGPC-33	RTEL1: NM_001283009	c.C2971T, p.Q991X	chr20:62324615C>T	het	Dyskeratosis congenita	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-34	DHCR24: NM_014762	c.C711A, p.Y237X	chr1:55337188G>T	het	Desmosterolosis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-34	CRB2: NM_173689	c.G3746A, p.R1249Q	chr9:126139229G>A	het	Focal segmental glomerulosclerosis	AR	Recessive likely pathogenic (II)	PS1, PM2, PP3	25557779
PGPC-34	ANO5: NM_213599	c.T2498A, p.M833K	chr11:22297723T>A	het	Gnathodiaphyseal dysplasia	AR	Recessive likely pathogenic (II)	PS1, PM2, PP3	23663589
PGPC-34	VPS11: NM_021729	c.T2536G, p.C846G	chr11:118951899T>G	het	Leukodystrophy	AR	Recessive likely pathogenic (III)	PS4, PP1, PP3	26307567
PGPC-34	CACNA2D4: NM_172364	c.C1882T, p.R628X	chr12:1969369G>A	het	Retinal cone dystrophy	AR	Recessive pathogenic (Ic)	PM2, PVS1	26560832
PGPC-34	MEFV: NM_000243	c.T2177C, p.V726A	chr16:3293310A>G	het	Familial Mediterranean fever	AR	Recessive pathogenic (II)	PS1, PS4, BP4	9288758
PGPC-35	COL4A4: NM_000092	c.C2906G, p.S969X	chr2:227917083G>C	het	Alport syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	12325029
PGPC-35	NCF1: NM_000265	c.73_74del, p.V25fs	chr7:74191613delGT	het	Chronic granulomatous disease	AR	Recessive pathogenic (Ia)	PVS1, PS4, PM2	2011585
PGPC-35	INSR: NM_001079817	c.C3043T, p.R1015X	chr19:7125473G>A	het	Hyperinsulinemic hypoglycemia	AR	Recessive pathogenic (Ia)	PVS1, PS4	2365819
PGPC-35	NHLRC1: NM_198586	c.C205G, p.P69A	chr6:18122633G>C	het	Epilepsy, progressive myoclonic (Lafora)	AR	Recessive pathogenic (II)	PP3, PS4, PS1	12958597
PGPC-36	MUTYH: NM_001048171	c.892-2A>G	chr1:45797760T>C	het	Colorectal adenomatous polyposis	AR	(Recessive) Pathogenic (Ia), Risk factor	PVS1, PS3, PP5, BS1	15180946
PGPC-36	SLC22A12: NM_144585	c.G774A, p.W258X	chr11:64361219G>A	het	Hypouricemia	AR	Recessive likely pathogenic (I)	PVS1, PM2	12024214
PGPC-36	UPB1: NM_016327	c.G977A, p.R326Q	chr22:24919647G>A	het	Beta-ureidopropionase deficiency	AR	Recessive likely pathogenic (II)	PP3, PS3, PM2	22525402
PGPC-37	POLG: NM_001126131	c.C752T, p.T251I	chr15:89873415G>A	het	Progressive external ophthalmoplegia; mitochondrial DNA de	AR	Recessive likely pathogenic (II)	PS4, BP4, PM2	12210792
PGPC-37	CTH: NM_153742	c.C200T, p.T67I	chr1:70881670C>T	het	Cystathioninuria	AR	Recessive pathogenic (II)	PS1, PS4, PP3	12574942
PGPC-37	SPG7: NM_199367	c.G1045A, p.G349S	chr16:89598369G>A	het	Spastic paraplegia	AR	Recessive pathogenic (II)	PS1, PS3, PM2	20186691
PGPC-38	ILDR1: NM_001199799	c.1357delC, p.R453fs	chr3:121712239delG	het	Deafness	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-38	CSPP1: NM_001291339	c.643_646del, p.T215fs	chr8:68030526delACTA	het	Joubert syndrome	AR	Recessive likely pathogenic (I)	PM2, PVS1	
PGPC-38	AP4S1: NM_001254727	c.430delG, p.G144fs	chr14:31552795delG	het	Spastic paraplegia	AR	Recessive likely pathogenic (I)	PVS1, PM2	

PGPC-38	LIMS2: NM_001161404	c.C275T, p.P92L; c.C261G, p.N87K	chr2:128412067G>A; chr2:128412081G>C	het	Muscular dystrophy	AR	Recessive likely pathogenic (II)	PS1, PM2, PP3	25589244
PGPC-38	HPGD: NM_000860	c.175_176del, p.L59fs	chr4:175443136delAG	het	Hypertrophic osteoarthropathy	AR	Recessive pathogenic (Ia)	PVS1, PS4	18500342
PGPC-38	LRRC6: NM_012472	c.598_599del, p.K200fs	chr8:133645040delTT	het	Ciliary dyskinesia	AR	Recessive pathogenic (Ia)	PVS1, PS4	23122589
PGPC-38	GJB2: NM_004004	c.35delG, p.G12fs	chr13:20763686delC	het	Deafness	AR	Recessive pathogenic (Ia)	PVS1, PS4	9285800
PGPC-38	SERPINA1: NM_001002235	c.G1096A, p.E366K	chr14:94844947C>T	het	Alpha-1-Antitrypsin deficiency	AR	Recessive pathogenic (II)	PS4, PS3	6306478
PGPC-39	ST3GAL5: NM_001042437	c.284delA, p.K95fs	chr2:86075293delT	het	Ganglioside GM3 synthase deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-39	ZP1: NM_207341	c.C628T, p.Q210X	chr11:60637319C>T	het	Oocyte maturation defect	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-39	IGHMBP2: NM_002180	c.C1488A, p.C496X	chr11:68701332C>A	het	Charcot-Marie-Tooth disease	AR	Recessive pathogenic (Ia)	PS4, PVS1	14681881
PGPC-39	NPC2: NM_006432	c.441+1G>A	chr14:74947404C>T	het	Niemann-pick disease	AR	Recessive pathogenic (Ia)	PVS1, PS4	23352160
PGPC-39	PAH: NM_000277	c.G898T, p.A300S	chr12:103245479C>A	het	Phenylketonuria	AR	Recessive pathogenic (II)	PS4, PS1	1301187
PGPC-40	SLC7A9: NM_001243036	c.614dupA, p.K205fs	chr19:33353114dupT	het	Cystinuria	AD/AR	(Recessive) Pathogenic (Ia)	PVS1, PS4-M, PP5	12371955
PGPC-40	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-40	PLEKHG5: NM_001265592	c.C2779T, p.R927X	chr1:6528354G>A	het	Charcot-Marie-Tooth disease	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-40	HFE: NM_139004	c.A193T, p.S65C	chr6:26091185A>T	het	Hemochromatosis	AR	Recessive pathogenic (II)	PS1, PS4, PP3	10194428
PGPC-40	ANK2: NM_020977	c.A4373G, p.E1458G	chr4:114269433A>G	het	Long QT syndrome; Cardiac arrhythmia	AD	VUS - not enough evidence	PS3, PP1, BS1	12571597
PGPC-40	CDH1: NM_004360	c.A2343T, p.E781D	chr16:68863604A>T	het	CDH1-related cancer	AD	VUS - not enough evidence	(PS3), PM2	17545690
PGPC-41	LAMB2: NM_002292	c.2344+1G>A	chr3:49163399C>T	het	Pierson syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-41	ALOXE3: NM_021628	c.C1974A, p.Y658X	chr17:8000107G>T	het	Ichthyosiform erythroderma	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-41	VKORC1: NM_024006	c.C292T, p.R98W	chr16:31102655G>A	het	Deficiency of vitamin K-dependent clotting factors	AR	Recessive likely pathogenic (II)	PM2, PS4	14765194
PGPC-41	PAH: NM_000277	c.G814T, p.G272X	chr12:103246621C>A	het	Phenylketonuria	AR	Recessive pathogenic (Ia)	PVS1, PS4	1975559
PGPC-42	ACADSB: NM_001609	c.C443T, p.T148I	chr10:124800121C>T	het	2-methylbutyryl-CoA dehydrogenase deficiency	AR	Recessive likely pathogenic (II)	PS3, PM2	15615815
PGPC-43	BRCA1: NM_007300	c.68_69delAG, p.E23fs	chr17:41276046delCT	het	Breast-ovarian cancer	AD	Pathogenic (Ia)	PVS1, PS3, PS4, PP5	7611277
PGPC-43	IGLL1: NM_020070	c.258delG, p.G86fs	chr22:23917218delC	het	Agammaglobulinemia	AR	Recessive likely pathogenic (I)	PVS1, PM2	25502423
PGPC-43	ACADSB: NM_001609	c.C443T, p.T148I	chr10:124800121C>T	het	2-methylbutyryl-CoA dehydrogenase deficiency	AR	Recessive likely pathogenic (II)	PS3, PM2	15615815
PGPC-44	ACADS: NM_000017	c.C319T, p.R107C	chr12:121174897C>T	het	Deficiency of Acyl-CoA dehydrogenase	AR	Recessive pathogenic (II)	PS1, PS4	1692038
PGPC-46	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-46	SH3PXD2B: NM_001017995	c.2626_2629del, p.F876fs	chr5:171765480delCAAA	het	Frank-ter Haar syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-46	RP1L1: NM_178857	c.G6118T, p.E2040X	chr8:10465490C>A	het	Retinitis pigmentosa	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-46	GNRHR: NM_000406	c.A317G, p.Q106R	chr4:68619737T>C	het	Hypogonadotropic hypogonadism	AR	Recessive likely pathogenic (II)	PS4, PM3	9371856
PGPC-46	NR2E3: NM_014249	c.119-2A>C	chr15:72103821A>C	het	Retinitis pigmentosa	AR	Recessive pathogenic (Ia)	PVS1, PS3, PS4, PM2	10655056
PGPC-46	CFTR: NM_000492	c.2657+5G>A	chr7:117242922G>A	het	Cystic fibrosis	AR	Recessive pathogenic (II)	PS1, PM2, PS3	15994263
PGPC-47	PINK1: NM_032409	c.C1474T, p.R492X	chr1:20975710C>T	het	Parkinson disease	AR	Recessive likely pathogenic (I)	PVS1, PM2	15349870
PGPC-47	CAPN3: NM_173087	c.C1018T, p.Q340X	chr15:42689044C>T	het	Muscular dystrophy	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-48	CCDC40: NM_017950	c.C3307T, p.R1103X	chr17:78073452C>T	het	Ciliary dyskinesia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-48	AMPD1: NM_001172626	c.G1017T, p.M339I	chr1:115221116C>A	het	Myoadenylate deaminase deficiency	AR	Recessive likely pathogenic (II)	PS3, PM3, BP4	15173240
PGPC-48	SUMF1: NM_001164675	c.C58T, p.L20F	chr3:4508872G>A	het	Multiple sulfatase deficiency	AR	Recessive likely pathogenic (II)	PS3, PM2, BP4	15146462
PGPC-49	SZT2: NM_015284	c.C10003T, p.R3335X	chr1:43916026C>T	het	Epileptic encephalopathy	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-49	IMPG1: NM_001282368	c.901+1G>T	chr6:76715003C>A	het	Macular dystrophy	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-49	BRAT1: NM_152743	c.2125_2128del, p.F709fs	chr7:2578041delCAAA	het	Rigidity and multifocal seizure syndrome	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-49	TRIP11: NM_004239	c.1935_1938del, p.E645fs	chr14:92472379delTCTT	het	Achondrogenesis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-49	EXOSC8: NM_181503	c.G815C, p.S272T	chr13:37583420G>C	het	Pontocerebellar hypoplasia	AR	Recessive likely pathogenic (II)	PS3, PP1-M	24989451
PGPC-50	CD36: NM_001127444	c.957dupA, p.S319fs	chr7:80300431insA	het	Platelet glycoprotein IV deficiency	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-50	CARD9: NM_052813	c.1576delA, p.T526fs	chr9:139258789delT	het	Candidiasis	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-50	PHYH: NM_001323083	c.C715T, p.R239X	chr10:13320339G>A	het	Refsum disease	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-50	MYO15A: NM_016239	c.952delC, p.P318fs	chr17:18023066delC	het	Deafness	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-50	HFE: NM_139004	c.A193T, p.S65C	chr6:26091185A>T	het	Hemochromatosis	AR	Recessive pathogenic (II)	PS1, PS4, PP3	10194428
PGPC-51	FLG: NM_002016	c.C6109T, p.R2037X	chr1:152281253G>A	het	Ichthyosis vulgaris	AD/AR	(Recessive) Likely pathogenic (I)	PVS1, PM2	
PGPC-51	EIF2B2: NM_014239	c.G599T, p.G200V	chr14:75472570G>T	het	Leukoencephalopathy	AR	Recessive pathogenic (II)	PS3, PS4, PP3	14566705
PGPC-52	ADAMTS10: NM_030957	c.1374delC, p.P458fs	chr19:8660751delG	het	Weill-Marchesani syndrome	AR	Recessive likely pathogenic (I)	PM2, PVS1	
PGPC-52	EXOSC8: NM_181503	c.G815C, p.S272T	chr13:37583420G>C	het	Pontocerebellar hypoplasia	AR	Recessive likely pathogenic (II)	PS3, PP1-M	24989451
PGPC-53	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-53	IRAK4: NM_001114182	c.C877T, p.Q293X	chr12:44172041C>T	het	IRAK4 deficiency	AR	Recessive pathogenic (Ia)	PVS1, PS4	12637671
PGPC-53	SLC7A9: NM_001243036	c.C997T, p.R333W	chr19:33334838G>A	het	Cystinuria	AR	Recessive pathogenic (II)	PS3, PS4, PP3	11157794
PGPC-54	NEK1: NM_001199400	c.C481T, p.R161X	chr4:170509870G>A	het	Short-rib thoracic dysplasia	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-54	PEX1: NM_000466	c.2097dupT, p.I700fs	chr7:92132484insA	het	Heimler syndrome	AR	Recessive pathogenic (Ia)	PVS1, PS4, PP5	10447258
PGPC-55	WNT10A: NM_025216	c.T682A, p.F228I	chr2:219755011T>A	het	Odontoonychodermal dysplasia	AD/AR	(Recessive) Pathogenic (II)	PS1, PS4, PP3	19559398
PGPC-55	TPO: NM_000547	c.2422delT, p.C808fs	chr2:1507755delT	het	Thyroid dyshormonogenesis	AR	Recessive likely pathogenic (I)	PVS1, PM2	11061528
PGPC-55	ITGA6: NM_001316306	c.371delG, p.R124fs	chr2:173337571delG	het	Epidermolysis bullosa	AR	Recessive likely pathogenic (I)	PVS1, PM2	
PGPC-55	GLYCTK: NM_001144951	c.T615A, p.C205X	chr3:52326361T>A	het	D-glyceric aciduria	AR	Recessive likely pathogenic (I)	PVS1, PM2	

PGPC-55	CYP21A2: NM_000500	c.T518A, p.I173N	chr6:32007203T>A	het	Adrenal hyperplasia due to 21-hydroxylase deficiency	AR	Recessive likely pathogenic (II)	PS4, PP3, PM3	3257825
PGPC-55	NCF1: NM_000265	c.73_74del, p.V25fs	chr7:74191613delGT	het	Chronic granulomatous disease	AR	Recessive pathogenic (Ia)	PVS1, PS4, PM2	2011585
PGPC-56	ADAMTS13: NM_139027	c.1511dupA, p.D504fs	chr9:136302944dupA	het	Thrombotic thrombocytopenic purpura	AR	Recessive likely pathogenic (I)	PM2, PVS1	
PGPC-56	SLC24A1: NM_004727	c.2817delC, p.F939fs	chr15:65944031delC	het	Night blindness	AR	Recessive likely pathogenic (I)	PM2, PVS1	

Note: AD = autosomal dominant, AR = autosomal recessive.

Pathogenic CNVs-SNVs

ID	Gene	Disease	Genomic position	Type	Zygosity	Inheritance	Size (bp)	ACMG interpretation	Evidence	PMID
PGPC-04	DCLRE1C	Severe combined immunodeficiency	chr10:14992001-14999000	deletion	het	AR	7,000	Recessive likely pathogenic	PVS1, PM2	
PGPC-05	PNPT1	Leigh syndrome	chr2:55868001-55884500	deletion	het	AR	16,500	Recessive likely pathogenic	PVS1, PM2	
PGPC-15	NAGA	Kanzaki disease	chr22:42461434-42465500	deletion	het	AR	4,067	Recessive likely pathogenic	PVS1, PM2	
PGPC-22	NA	Susceptibility for unbalanced aberrations	chr20:35583655-44214109	inversion	het	NA	8,630	Risk factor		
PGPC-24	SUGCT	Glutaric aciduria	chr7:40462001-40518606	deletion	het	AR	56,606	Recessive likely pathogenic	PVS1, PM2	
PGPC-27	NBAS	Infantile liver failure syndrome	chr2:15399501-15432184	deletion	het	AR	32,684	Recessive likely pathogenic	PVS1, PM2	
PGPC-27	NA	Mosaic Turner syndrome	46,XX/45,X0	deletion	mosaic (70%)	XL		Pathogenic		
PGPC-37	PTPRQ	Deafness	chr12:80853631-80879093	deletion	het	AR	25,463	Recessive likely pathogenic	PVS1, PM2	
PGPC-48	PCDH15	Deafness; Usher syndrome	chr10:55741501-57660800	deletion	het	AR	1,919,300	Recessive likely pathogenic, Risk factor	PVS1, PM2	
PGPC-56	STRADA	Megalencephaly	chr17:61779476-61786809	deletion	het	AR	7,334	Recessive pathogenic	PVS1, PS4	17522105

Note: AR = autosomal recessive, het = heterozygous.

Rare coding CNVs > 100 kb

ID	Gene(s)	Disease	Genomic position	Type	Zygosity	Inheritance	Size (bp)	ACMG interpretation	Evidence	PMID
PGPC-10	KLHL24|KLHL6|YEATS2	NA	chr3:183191501-183470000	duplication	het	NA	278,500	VUS - not enough evidence		
PGPC-20	VCX3A	NA	chrX:6449501-6761500	duplication	het	NA	312,000	VUS - not enough evidence		
PGPC-21	ADCK5|ARHGAP39|C8orf82|CPSF1|CYHR1|FOXH1|GPT|KIFC2|LRRC14|LRRC24|MFSD3|PPP1R16A|RECQL4|SLC39A4|TONSL|VPS28	NA	chr8:145598001-145837500	duplication	het	NA	239,500	VUS - not enough evidence		
PGPC-23	C17orf112	NA	chr17:50951276-51314632	deletion	het	NA	363,357	VUS - not enough evidence		
PGPC-23	MAP4	NA	chr3:47973001-48173000	duplication	het	NA	200,000	VUS - not enough evidence		
PGPC-44	ANOS1	NA	chrX:8595001-8706000	duplication	het	NA	111,000	VUS - not enough evidence		
PGPC-48	PCDH15	Deafness; Usher syndrome	chr10:55741501-57660800	deletion	het	AR	#######	Recessive likely pathogenic, Risk factor	PVS1, PM2	
PGPC-56	BTBD17|C17orf77|CD300A|CD300C|CD300E|CD300LB|CD300LD|CD300LF|DNAI2|GPR142|GPRC5C|KIF19|LOC100130520|RAB37|RPL38|TTYH2	NA	chr17:71828001-72691000	duplication	het	NA	863,000	VUS - not enough evidence		