Supplementary material

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Table S1. Genes with significant (q-value <0.05) differences in pathogenic variant burden among Polish population (Thousand Polish Genomes database) compared with non-Finnish European (gnomAD)

Gene	Position	Allele	Variant ID	Allele frequency in Polish population	Allele frequency in gnomAD NFE	q-value	OR	IMPACT	Consequence	Clinical significance
APC	chr5:112837834	C>T	rs773020689	0.00041	0	0.04	N/A	MODERATE	missense variant	Conflicting interpretations of pathogenicity
APC	chr5:112838128	G>A	rs776878597	0.00081	8.8e-05	0.02	21	MODERATE	missense variant	Conflicting interpretations of pathogenicity
APC	chr5:112842318	A>G	rs201375478	0.0016	0.00016	<0.001	23	MODERATE	missense variant	Conflicting interpretations of pathogenicity
APC	chr5:112843372	A>G	rs367676584	0.0012	1.5e-05	<0.001	190	MODERATE	missense variant	Conflicting interpretations of pathogenicity
APC	chr5:112843452	T>A	rs587781816	0.00041	0	0.04	N/A	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ARSB	chr5:78885603	C>T	rs200040980	0.0024	0.00065	<0.001	8.6	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ARSB	chr5:78969026	C>T	rs1196325597	0.00041	0	0.04	N/A	MODERATE	missense variant	Pathogenic/Likely pathogenic
ARSB	chr5:78985513	C>T	rs72764913	0.026	0.03	<0.001	2	MODIFIER	upstream gene variant	Conflicting interpretations of pathogenicity
ATM	chr11:108235718	CA>C	rs587781831	0.00041	0	0.04	N/A	HIGH	frameshift variant	Pathogenic/Likely pathogenic
ATM	chr11:108235814	TATCTC>T	rs587780624	0.00041	0	0.04	N/A	HIGH	frameshift variant	Pathogenic/Likely pathogenic
ATM	chr11:108282704	G>A	rs56006345	0.00041	0	0.04	N/A	LOW	splice polypyrimidine tract variant,splice region variant, intron variant	Conflicting interpretations of pathogenicity
ATM	chr11:108293469	C>T	rs35962982	0.0012	0.00035	0.03	7.9	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ATM	chr11:108304660	G>C	rs3092828	0.0081	0.0054	<0.001	3.4	LOW	splice polypyrimidine tract variant,intron variant	Conflicting interpretations of pathogenicity
ATM	chr11:108304736	A>T	rs1801673	0.013	0.0073	<0.001	4.2	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ATM	chr11:108315883	G>A	rs11212587	0.0028	0.0023	0.04	2.8	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ATM	chr11:108326110	G>C	rs1800061	0.0016	0.00056	0.01	6.7	MODERATE	missense variant	Conflicting interpretations of pathogenicity
ATM	chr11:108331877	A>C	rs587779866	0.0012	2.9e-05	<0.001	95	HIGH	splice acceptor variant	Pathogenic
ATM	chr11:108365744	C>T	rs3092834	0.0012	0.00046	0.04	6.1	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
ATM	chr11:108368115	T>C	rs879796523	0.0032	0.001	<0.001	7.4	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
BRCA1	chr17:43057062	T>TG	rs80357906	0.0016	5.9e-05	<0.001	63	HIGH	frameshift variant	Pathogenic
BRCA1	chr17:43067677	C>A	rs80357087	0.0028	0.00038	<0.001	17	MODERATE	missense variant	Conflicting interpretations of pathogenicity

BRCA1	chr17:43091826	GTTTAC>G	rs80357609	0.00041	0	0.04	N/A	HIGH	frameshift variant	Pathogenic
CHEK2	chr22:28725099	A>G	rs17879961	0.028	0.0053	<0.001	12	MODERATE	missense variant	Conflicting interpretations of pathogenicity|risk factor
CHEK2	chr22:28725240	T>C	rs587781279	0.00041	0	0.04	N/A	LOW	splice donor region variant,intron variant	Conflicting interpretations of pathogenicity
CHEK2	chr22:28725242	C>T	rs121908698	0.0036	0.00018	<0.001	47	HIGH	splice donor variant	Conflicting interpretations of pathogenicity
CHEK2	chr22:28725277	C>T	rs368570187	0.0012	0.00019	0.007	15	MODERATE	missense variant	Conflicting interpretations of pathogenicity
DICER1	chr14:95099771	C>CAA	rs1555368535	0.0028	0.039	<0.001	0.14	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
DICER1	chr14:95105250	C>G	rs1595371340	0.00081	0	0.001	N/A	LOW	splice polypyrimidine tract variant,splice region variant,intron variant	Conflicting interpretations of pathogenicity
DICER1	chr14:95115562	G>A	rs2275182	0.2	0.2	<0.001	2.3	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
DICER1	chr14:95133439	T>C	rs117358479	0.0069	0.0032	<0.001	4.9	MODERATE	missense variant	Conflicting interpretations of pathogenicity
GPD1L	chr3:32140231	A>G	rs72552293	0.015	0.0028	<0.001	12	MODERATE	missense variant	Conflicting interpretations of pathogenicity
INSR	chr19:7125507	C>T	rs1799816	0.0081	0.0071	<0.001	2.6	MODERATE	missense variant	Conflicting interpretations of pathogenicity
INSR	chr19:7142970	C>G	rs78433961	0.002	0.00068	0.005	6.9	MODERATE	missense variant	Conflicting interpretations of pathogenicity
INSR	chr19:7152840	G>T	rs142391704	0.00081	0.00019	0.048	9.7	MODERATE	missense variant	Conflicting interpretations of pathogenicity
INSR	chr19:7184640	G>GGAGA	rs3835070	0.023	0.021	<0.001	2.5	LOW	splice polypyrimidine tract variant,splice region variant,intron variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160038797	T>C	rs116418256	0.0081	0.008	0.004	2.3	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160039359	G>A	rs192835895	0.012	0.0028	<0.001	9.9	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160039373	GACACACA C>G	rs56656397	0.14	0.16	<0.001	2	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160039373	GAC>G	rs56656397	0.13	0.14	<0.001	2.2	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160039373	GACACAC> G	rs56656397	0.06	0.081	<0.001	1.7	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160041721	C>T	rs3795339	0.0012	0.00037	0.03	7.6	MODERATE	missense variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160042003	T>C	rs145947380	0.0032	0.00063	<0.001	12	MODERATE	missense variant	Conflicting interpretations of pathogenicity
KCNJ10	chr1:160042480	C>T	rs115466046	0.015	0.017	0.002	2	MODERATE	missense variant	Conflicting interpretations of pathogenicity
MYH9	chr22:36292033	G>A	rs727503286	0.00081	7.3e-05	0.02	25	MODERATE	missense variant	Conflicting interpretations of pathogenicity
PALB2	chr16:23635002	T>C	rs515726072	0.0016	0.00018	<0.001	21	MODERATE	missense variant	Conflicting interpretations of pathogenicity
PALB2	chr16:23637964	A>T	rs774949203	0.00041	0	0.04	N/A	LOW	splice polypyrimidine tract variant,intron variant	Conflicting interpretations of pathogenicity
PALB2	chr16:23641145	G>A	rs377085677	0.00081	8.8e-05	0.02	21	MODERATE	missense variant	Conflicting interpretations of pathogenicity
PALB2	chr16:23641315	C>G	rs138200248	0.019	0.0073	<0.001	5.9	MODIFIER	upstream gene variant	Conflicting interpretations of pathogenicity
PLCB1	chr20:8132322	C>T	rs532302075	0.021	0.0098	<0.001	4.8	MODIFIER	5 prime UTR variant	Conflicting interpretations of pathogenicity
PLCB1	chr20:8739374	G>C	rs768572485	0.00081	4.4e-05	0.008	42	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
PLCB1	chr20:8788453	T>C	rs75820839	0.00081	1E-04	0.03	18	MODERATE	missense variant	Conflicting interpretations of pathogenicity
POT1	chr7:124851922	T>C	rs573222502	0.00041	0	0.034	N/A	MODERATE	missense variant	Conflicting interpretations of pathogenicity
PTEN	chr10:87958028	T>C	rs1060503839	0.00041	0	0.034	N/A	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
PTEN	chr10:87965754	G>A	rs576872432	0.0012	5.9e-05	<0.001	47	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
PTEN	chr10:87966971	C>T	rs180953647	0.027	0.0043	<0.001	14	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
RET	chr10:43106469	G>A	rs377767388	0.0016	7.4e-05	<0.001	51	MODERATE	missense variant	Conflicting interpretations of pathogenicity
RET	chr10:43111500	C>T	rs377130948	0.0041	0.00024	<0.001	40	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
RET	chr10:43114546	C>T	rs148935214	0.0045	0.00076	<0.001	13	MODERATE	missense variant	Conflicting interpretations of pathogenicity
RET	chr10:43114671	G>A	rs1799939	0.2	0.18	<0.001	2.6	MODERATE	missense variant	Conflicting interpretations of pathogenicity
RET	chr10:43118460	A>T	rs77724903	0.011	0.0018	<0.001	14	MODERATE	missense variant	Conflicting interpretations of pathogenicity
RET	chr10:43124887	C>T	rs17158558	0.024	0.016	<0.001	3.5	MODERATE	missense variant	Conflicting interpretations of pathogenicity

RET	chr10:43126647	A>G	rs201740483	0.0081	0.00084	<0.001	22	MODERATE	missense variant	Conflicting interpretations of pathogenicity
RET	chr10:43129699	A>G	rs775114955	0.0012	4E-04	0.03	7	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
SEC23B	chr20:18524971	C>T	rs201418257	0.00081	0	0.001	N/A	HIGH	stop gained	Pathogenic
SEC23B	chr20:18525868	C>T	rs146917730	0.022	0.016	<0.001	3.2	MODERATE	missense variant	Conflicting interpretations of pathogenicity
SERPINA1	chr14:94388565	A>G	rs11558258	0.051	0.039	<0.001	3	MODIFIER	5 prime UTR variant	Conflicting interpretations of pathogenicity
SLC26A4	chr7:107674187	A>C	CM033462	0.00041	0	0.04	N/A	MODERATE	missense variant	Likely pathogenic
SLC26A4	chr7:107683277	G>A	rs727505080	0.00081	0.00016	0.04	11	MODERATE	missense variant	Conflicting interpretations of pathogenicity
SLC26A4	chr7:107683349	AT>A	COSV105848651	0.00041	0	0.04	N/A	HIGH	frameshift variant	Likely pathogenic
SLC26A4	chr7:107689054	T>C	rs111033212	0.0028	0.0011	0.001	5.9	MODERATE	missense variant,splice region variant	Likely pathogenic
SLC26A4	chr7:107690220	A>C	rs28939086	0.0016	0.00032	0.003	11	MODERATE	missense variant	Conflicting interpretations of pathogenicity
SLC26A4	chr7:107694718	T>G	rs397516418	0.00041	0	0.04	N/A	HIGH	splice donor variant	Pathogenic/Likely pathogenic
SLC26A4	chr7:107698071	C>T	rs765197819	0.00041	0	0.04	N/A	MODERATE	missense variant	Likely pathogenic
SLC26A4	chr7:107715796	C>T	rs17154362	0.0061	9E-04	<0.001	16	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
SMAD3	chr15:67138026	C>A	rs958007552	0.00081	1E-04	0.03	18	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
SMAD3	chr15:67191325	A>G	rs191679355	0.00081	1E-04	0.03	18	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
STK11	chr19:1206797	C>CT	rs927999961	0.0012	0.00022	0.009	13	MODIFIER	5 prime UTR variant	Conflicting interpretations of pathogenicity
STK11	chr19:1219421	C>T	rs863224669	0.00081	3E-05	0.005	63	LOW	splice region variant,intron variant	Conflicting interpretations of pathogenicity
STK11	chr19:1221319	C>A	rs377208033	0.00081	1.5e-05	0.003	130	MODERATE	missense variant	Conflicting interpretations of pathogenicity
STK11	chr19:1226556	C>T	rs200078204	0.0016	0.00087	0.04	4.3	MODERATE	missense variant	Conflicting interpretations of pathogenicity
STK11	chr19:1226655	C>T	rs587782259	0.0041	0.00038	<0.001	24	MODIFIER	3 prime UTR variant	Conflicting interpretations of pathogenicity
TERT	chr5:1278812	C>T	rs146768484	0.0089	0.00094	<0.001	22	LOW	splice polypyrimidine tract variant,intron variant	Conflicting interpretations of pathogenicity
TERT	chr5:1293652	G>A	rs34094720	0.0057	0.0049	0.005	2.7	MODERATE	missense variant	Conflicting interpretations of pathogenicity
TOE1	chr1:45340381	G>A	rs3219466	0.039	0.034	<0.001	2.6	MODIFIER	intron variant	Conflicting interpretations of pathogenicity
WRN	chr8:31058542	A>G	rs34477820	0.01	0.0054	<0.001	4.3	MODERATE	missense variant,splice region variant	Conflicting interpretations of pathogenicity
WRN	chr8:31064438	G>C	rs145764920	0.0061	0.0027	<0.001	5.2	LOW	splice donor region variant,intron variant	Conflicting interpretations of pathogenicity
WRN	chr8:31081176	G>T	rs4987238	0.0045	0.0018	<0.001	5.7	MODERATE	missense variant	Conflicting interpretations of pathogenicity
WRN	chr8:31141445	G>A	rs140768346	0.0045	0.0025	<0.001	4.1	MODERATE	missense variant	Conflicting interpretations of pathogenicity

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