

Gene	ensembl_gene_id	ensembl_transcript_id	protein_coding	protein_change	Genotype	Chromosome	Start	End
ASXL1	G000001711	000003060	c.3874G>A	p.Val1292Ile	HET	20	32436601	32436601
BLM	G000001977	000003551	c.1847A>G	p.Asn616Ser	HET	15	90761220	90761220
CD48	G000001177	000006137	c.324C>G	p.Ile108Met	HET	1	1.61E+08	1.61E+08
CR2	G000001177	000003670	c.1676G>A	p.Gly559Glu	HET	1	2.07E+08	2.07E+08
FANCA	G000001877	000003893	c.797C>T	p.Thr266Met	HET	16	89799634	89799634
FANCM	G000001877	000002674	c.5440G>A	p.Glu1814Lys	HET	14	45196271	45196271
FPR1	G000001711	000003047	c.205G>A	p.Val69Met	HET	19	51746790	51746790
IL12RB1	G000000967	000005939	c.842C>T	p.Thr281Ile	HET	19	18072291	18072291
KLF1	G000001057	000002648	c.493G>A	p.Ala165Thr	HET	19	12885737	12885737
LRBA	G000001987	000003571	c.5031T>A	p.Asn1677Lys	HET	4	1.51E+08	1.51E+08
LRBA	G000001987	000003571	c.2209G>A	p.Val737Ile	HET	4	1.51E+08	1.51E+08
MASP2	G000000097	000004008	c.352C>T	p.Arg118Cys	HET	1	11046616	11046616
MEFV	G000001037	000000219	c.2082G>A	p.Met694Ile	HET	16	3243405	3243405
MEFV	G000001037	000000219	c.2040G>C	p.Met680Ile	HET	16	3243447	3243447
MSH6	G000001167	000002344	c.2561A>T	p.Lys854Met	HET	2	47800544	47800544
NEIL3	G000001097	000002645	c.1524T>A	p.Ser508Arg	HET	4	1.77E+08	1.77E+08
NLRP1	G000000917	000005722	c.760C>T	p.Pro254Ser	HET	17	5559936	5559936
POLE	G000001777	000003205	c.1320C>G	p.Asp440Glu	HET	12	1.33E+08	1.33E+08
RET	G000001657	000000340	c.200G>A	p.Arg67His	HET	10	43100585	43100585
RTEL1	G000002587	000003700	c.2278G>T	p.Ala760Ser	HET	20	63690306	63690306
STAT2	G000001707	000003141	c.2191C>T	p.Pro731Ser	HET	12	56344047	56344047
TINF2	G000000927	000002674	c.734C>A	p.Ser245Tyr	HET	14	24240746	24240746

Ref_Allele	Alternative_Allele	rsid	Manually curated ACMG classification	SIFT_score	SIFT_pred
G	A	rs14013726	Likely benign	0.023	Deleterious
A	G	.	Likely benign	0.059	Tolerate
G	C	rs13919821	Likely benign	0.025	Deleterious
G	A	rs14361433	Likely benign	0.025	Deleterious
G	A	rs75279944	Likely benign	0.024	Deleterious
G	A	rs13907468	VUS	0.135	Tolerate
C	T	rs19953414	Likely benign	0.013	Deleterious
G	A	.	Likely benign	.	.
C	T	rs104096320	Likely benign	0.631	Tolerate
A	T	.	VUS	0.002	Deleterious
C	T	rs15121344	Likely benign	0.292	Tolerate
G	A	rs14727078	Likely benign	0.017	Deleterious
C	T	rs28940578	Pathogenic	0.113	Tolerate
C	G	rs28940580	Pathogenic	0.359	Tolerate
A	T	rs34374438	VUS	0.002	Deleterious
T	A	.	Likely benign	0.079	Tolerate
G	A	rs37466280	Likely benign	0.074	Tolerate
G	C	rs76335022	VUS	0.017	Deleterious
G	A	rs19248901	Benign	0.611	Tolerate
G	T	rs77665799	Likely benign	0.71	Tolerate
G	A	rs95503031	Likely benign	0.711	Tolerate
G	T	rs14277786	Benign	0.003	Deleterious

hen2_HDIV	hen2_HDIV	hen2_HVAR	hen2_HVAR	tionTaster_	MutationTaster_pred	ionAssessor
0.501	sibly_Dama	0.058	Benign	1	Polymorphism	1.735
0.023	Benign	0.004	Benign	1	Polymorphism	1.745
0.181	Benign	0.774	sibly_Dama	1	Polymorphism	2.085
1	pably_Dama	0.992	pably_Dama	0.991	Disease_Causing	2.91
0.389	Benign	0.057	Benign	1	Polymorphism	0
0.236	Benign	0.031	Benign	1	Polymorphism	1.63
0.8	sibly_Dama	0.327	Benign	1	Polymorphism	1.145
0.999	pably_Dama	0.974	pably_Dama	1	Polymorphism	2.135
0.001	Benign	0.001	Benign	1	Polymorphism	0.345
0.999	pably_Dama	0.994	pably_Dama	1	Disease_Causing	3.2
0.992	pably_Dama	0.989	pably_Dama	1	Disease_Causing	1.485
0.202	Benign	0.029	Benign	1	Disease_Causing	3.4
0.786	sibly_Dama	0.337	Benign	0	Polymorphism Automatic	0.965
0	Benign	0.001	Benign	1	Polymorphism	-0.605
1	pably_Dama	0.997	pably_Dama	1	Disease_Causing	3.14
0.035	Benign	0.038	Benign	1	Polymorphism	1.46
0.005	Benign	0.001	Benign	1	Polymorphism	-0.345
0.957	pably_Dama	0.781	sibly_Dama	1	Disease_Causing	2.405
0.002	Benign	0.001	Benign	0.926	Polymorphism	0.905
0.529	sibly_Dama	0.167	Benign	1	Polymorphism	1.245
0.002	Benign	0.002	Benign	1	Polymorphism	1.845
0.907	sibly_Dama	0.264	Benign	0	Polymorphism Automatic	0.69

ionAssessor	CADD_raw	ADD_phred	Var_autom	AD_genom
Low	1.449	13.05	ertain signific	0.0005
Low	-0.838	0.035	likely benign	.
Medium	1.868	15.39	.	0.0006
Medium	4.809	24.8	ertain signific	0.0002
Neutral	2.148	17.17	likely benign	.
Low	1.087	11.15	ertain signific	0.0001
Low	1.202	11.76	ertain signific	6.46E-05
Medium	1.732	14.59	ertain signific	3.23E-05
Neutral	1.841	15.23	.	9.70E-05
Medium	4.133	23.8	ertain signific	.
Low	3.656	23.2	ertain signific	0.0007
Medium	5.129	25.4	ertain signific	0.0005
Low	-0.499	0.221	ertain signific	3.23E-05
Neutral	-1.836	0.002	ertain signific	.
Medium	3.028	22.3	ertain signific	0.0003
Low	2.121	16.99	ertain signific	.
Neutral	-1.638	0.002	ertain signific	3.23E-05
Medium	4.324	24	ertain signific	.
Low	1.507	13.35	likely benign	0.0008
Low	-0.451	0.29	ertain signific	.
Low	0.317	5.859	.	3.23E-05
Neutral	3.206	22.7	likely benign	0.0001

<b>CLNALLEID</b>
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267279
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CLNDN
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Common_variable_immunodeficiency_7 not_specified
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not_specified
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Familial_Mediterranean_fever Familial_mediterranean_fever, Hereditary_nonpolyposis_colon_cancer Hereditary_cancer-pr
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Colorectal_cancer,_susceptibility_to,_12
Pheochromocytoma Multiple_endocrine_neoplasia Hereditar
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Dyskeratosis_congenita Revesz_syndrome Dyskeratosis_conge

CLNDISDB
MedGen:CN169374
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MedGen:C3542922,OMIM:614699   MedGen:CN169374
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MedGen:CN169374
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MedGen:C0031069,OMIM:249100,Orphanet:ORPHA342,SI
MedGen:C0009405,Orphanet:ORPHA443090   MedGen:C00
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MedGen:C3554460,OMIM:615083
Human_Phenotype_Ontology:HP:0002666,MedGen:C0031
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MedGen:C0265965,Orphanet:ORPHA1775,SNOMED_CT:74

CLNREVSTAT
no_assertion_provided
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criteria_provided,_conflicting_interpretations
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criteria_provided,_single_submitter
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criteria_provided,_multiple_submitters,_no_conflicts
reviewed_by_expert_panel
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criteria_provided,_single_submitter
criteria_provided,_conflicting_interpretations
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criteria_provided,_conflicting_interpretations

CLNSIG
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Conflicting_interpretations_of_pathogenicity
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Uncertain_significance
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Pathogenic
Uncertain_significance
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Uncertain_significance
Conflicting_interpretations_of_pathogenicity
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Conflicting_interpretations_of_pathogenicity