	Gene_Symbol	ID	Ref_Transcript	Exon	Nucleotide_Change	Amino_Acid_Change	Gene_Type	1000g2015aug_all	Pathogenic_Analysis	clinvar	MutRatio	Mutation_Type	dbsnp	PathSNP	MutInNorm	1000Genome	MutInDatal	1000g2015aug_all	ESP6500si	Inhouse	ExAC_ALL	ExAC_EAS	SIFT	SIFT_Predict	PolyPhen_2	PolyPhen_2_Predict	MutationTaster	MutationTaster_Predict	GERP++	GERP++_Predict	SPIDEX	REVEL_score	MCAP_score	MCAP_pred	InterVar	Highest-MAF	Mygeno_InterACMG	Pathogenic_Analysis
Family A	MTHFR	chr1-11855368	NM_005957	exon6	c.818A>G	p.K273R	het	-	Uncertain	-	0.52	SNV	-	#N/A	#N/A	#N/A	#N/A	-	-	-	-	-	0.059	Tolerated	0.038	Benign	1	Disease_causing	3.53	Conserved	-0.0865	0.368	0.071252519	P	Uncertain	-	PM2	Uncertain
	CYP1B1	chr2-38302213	NM_000104	exon2	c.319C>G	p.L107V	het	0.000399361	Uncertain	-	0.55	SNV	rs56339482	#N/A	#N/A	chr2-38302213	#N/A	0.000399361	-	0.0021739	0.0006	0.0097	-	-	1	Probably_damaging	0.997	Disease_causing	2.73	Conserved	0.0907	0.647	0.783094831	P	Uncertain	0.0097	PS1	Uncertain
	GDF6	chr8-97172666	NM_001001557	exon1	c.255G>T	p.P85P	het	0.00359425	Uncertain	-	0.52	SNV	rs1122968824	#N/A	#N/A	chr8-97172666	#N/A	0.00359425	0.0041	0.0057	0.0015	0.0051	-	-	-	-	-	-	-	-	-	-	-	-	Benign	0.0057	BP7	Uncertain
	EPG5	chr18-43496539	NM_020964	exon18	c.3248C>T	p.S1083L	het	0.014976	Uncertain	-	0.45	SNV	rs78339727	#N/A	#N/A	chr18-43496539	#N/A	0.014976	0.0208	0.0286	0.029	0.0021	0.019	Damaging	0.771	Possibly_damaging	0.999	Disease_causing	5.78	Conserved	-0.2802	0.184	-	-	Benign	0.029	BS1	Uncertain
	CRYBB2	chr22-25623876	NM_000496	exon4	c.230G>T	p.G77V	het	-	Uncertain	-	0.39	SNV	-	#N/A	#N/A	#N/A	#N/A	-	-	-	-	-	0	Damaging	0.999	Probably_damaging	1	Disease_causing	5.08	Conserved	0.5789	0.957	0.157318302	P	Uncertain	-	PM2;PP3	Likely pathogenic
Family B	POMGNT1	chr1-46655561	NM_001243766	exon20	c.1750G>A	p.D584N	het	-	Uncertain	-	0.53	SNV	-	#N/A	#N/A	#N/A	#N/A	-	-	-	-	-	0.389	Tolerated	0.079	Benign	1	Disease_causing	5.92	Conserved	-0.2323	0.077	0.015190102	B	Uncertain	-	PM2;BP1	Uncertain
	FOXE3	chr1-47882145	NM_012186	exon1	c.158C>T	p.P53L	het	0.00658946	Uncertain	-	0.6	SNV	rs534479543	#N/A	#N/A	0.00658946	#N/A	-	-	0.0114	-	-	0.159	Tolerated	0.013	Benign	1	Polymorphism	2.25	Conserved	-	0.113	-	-	Benign	0.0114	0	Uncertain
	PAX6	chr11-31811460 31811462	NM_001258464	-	c.821_*20delAA	-	-	-	Uncertain	-	0.5	unknown	rs759391101	#N/A	#N/A	#N/A	#N/A	-	-	-	0.0009	0.0008	-	-	-	-	-	-	-	-	-	-	-	-	Uncertain	0.0009	PM2	Uncertain
	CRYBB2	chr22-25623876	NM_000496	exon4	c.230G>A	p.G77D	het	-	Uncertain	-	0.45	SNV	-	#N/A	#N/A	#N/A	#N/A	-	-	-	-	-	0	Damaging	0.999	Probably_damaging	1	Disease_causing	5.08	Conserved	1.3267	0.968	0.219654258	P	Uncertain	-	PM2;PP3	Likely pathogenic
Family C	BFSP2	chr3-133119306	NM_003571	exon1	c.379C>G	p.Q127E	het	0.00139776	Uncertain	-	0.41	SNV	rs185816798	#N/A	#N/A	chr3-133119306	#N/A	0.00139776	-	0.0108696	0.0008	0.0115	0.235	Tolerated	0.021	Benign	0.989	Disease_causing	3.81	Conserved	-	0.227	-	-	Benign	0.0115	BS1	Uncertain
	BEST1	chr11-61719298	NM_004183	exon2	c.206C>A	p.S7Y	het	0.000199681	Likely pathogenic	-	0.57	SNV	rs199508634	#N/A	#N/A	chr11-61719298	#N/A	0.000199681	-	-	1.65E-05	0.0002	0.561	Tolerated	0.006	Benign	1	Disease_causing	-1.72	Nonconserved	0.1244	0.346	0.013177856	B	Uncertain	0.0002	PS1;PM2;PM5;PM5	Likely pathogenic
	MIP	chr12-56848079	NM_012064	exon1	c.319G>A	p.V107I	het	0.0219649	Uncertain	-	0.5	SNV	rs74641138	#N/A	#N/A	chr12-56848079	#N/A	0.0219649	0.019	0.0065217	0.0253	0.0395	0.543	Tolerated	0.012	Benign	1	Disease_causing	4.29	Conserved	-	0.233	-	-	Benign	0.0395	PM1;BS1	Uncertain
	CRYGD	chr2-208986446 208986447	NM_006891	exon3	c.475delG	p.A159Pfs*9	het	-	Likely pathogenic	-	0.55	deletion	-	#N/A	#N/A	#N/A	#N/A	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	Uncertain	-	PVS;PM2	Likely pathogenic