

Supplemental Table 1. ABCA4 patient cohort with pathogenic ABCA4 changes and the c.5603A>T (p.Asn1868Ile) status. Nucleotide positions and protein translation correspond to CCDS747.1 and NP_000341.2, respectively. Nucleotide numbering uses the A of the ATG translation initiation start site as nucleotide 1.

PATIENT #	segregation of variants performed	Age of Onset	Foveal Sparing	ABCA4 disease alleles	c.5603A>T (p.Asn1868Ile) genotype	ABCA4 locus seq	ABCA4 pathogenic variant	ABCA4 pathogenic variant
3007	x			2	++		c.2588G>C (p.Gly863Ala)	c.5461-10T>C (p.?)
3807	x	51	N	2	++		c.2588G>C (p.Gly863Ala)	c.5461-10T>C (p.?)
3306				2	++		c.2588G>C (p.Gly863Ala)	c.5461-10T>C (p.?)
3465		10	N	2	++		c.2588G>C (p.Gly863Ala)	c.5461-10T>C (p.?)
3334				2	++		c.2588G>C (p.Gly863Ala)	c.5461-10T>C (p.?)
3467		33		2	++		c.6191C>T (p.Ala2064Val)	c.6191C>T (p.Ala2064Val)
4247				2	++		c.4594G>A (p.Asp1532Asn)	c.4594G>A (p.Asp1532Asn)
3356		41	N	2	++		c.2588G>C (p.Gly863Ala)	c.2588G>C (p.Gly863Ala)
3122	x	32	Y	2	++		c.2588G>C (p.Gly863Ala)	c.2588G>C (p.Gly863Ala)
3341		35	Y	2	++		c.2588G>C (p.Gly863Ala)	c.5177C>A (p.Thr1726Asn)
3611	x			2	+/-		c.[1715G>A;2588G>C] (p.[Arg572Gln;Gly863Ala])	c.5898+1G>A (p.?)
3597		21	N	2	+/-		c.[2549A>G;5882G>A] (p.[Tyr850Cys;Gly1961Glu])	c.5461-10T>C (p.?)
3408	x	18	N	2	+/-	x	c.5882G>A (p.Gly1961Glu)	c.2160+584A>G (p.?)
3269		60	Y	2	+/-	x	c.2588G>C (p.Gly863Ala)	c.4539+2028C>T (p.?)
3179	x			2	+/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5018+5G>A (p.?)
3358				2	+/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5461-10T>C (p.?)
3084	x			2	+/-		c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])	c.5461-10T>C (p.?)
3427		16	N	2	+/-		c.1592A>G (p.Glu531Gly)	c.5461-10T>C (p.?)
3336	x			2	+/-		c.161G>A (p.Cys54Tyr)	c.5461-10T>C (p.?)
3837	x	11	N	2	+/-		c.1648G>A (p.Gly550Arg)	c.5461-10T>C (p.?)
3250				2	+/-		c.3292C>T (p.Arg1098Cys)	c.5461-10T>C (p.?)
3347				2	+/-		c.3322C>T (p.Arg1108Cys)	c.5461-10T>C (p.?)
3546				2	+/-		c.4139C>T (p.Pro1380Leu)	c.5461-10T>C (p.?)
3839	x	9	Y	2	+/-		c.4139C>T (p.Pro1380Leu)	c.5461-10T>C (p.?)
7565				2	+/-		c.4139C>T (p.Pro1380Leu)	c.5461-10T>C (p.?)
3186				2	+/-		c.4139C>T (p.Pro1380Leu)	c.5461-10T>C (p.?)
3303	x			2	+/-		c.5882G>A (p.Gly1961Glu)	c.5461-10T>C (p.?)
3284	x	10	N	2	+/-		c.5461-10T>C (p.?)	c.5714+5G>A (p.?)
3342	x			2	+/-		c.5461-10T>C (p.?)	c.5714+5G>A (p.?)
3004	x			2	+/-		c.4469G>A (p.Cys1490Tyr)	c.5714+5G>A (p.?)
5020	x			2	+/-		c.770T>G (p.Leu257Arg)	c.5714+5G>A (p.?)
3857				2	+/-		c.2588G>C (p.Gly863Ala)	c.5898+1G>A (p.?)
4581		45	N	2	+/-		c.5461-10T>C (p.?)	c.6006-609T>A (p.?)
3063	x			2	+/-		c.[1715G>A;2588G>C] (p.[Arg572Gln;Gly863Ala])	c.[3322C>T;6320G>A] (p.[Arg1108Cys;Arg2107His])
3595	x		Y	2	+/-		c.1805G>A (p.Arg602Gln)	c.4469G>A (p.Cys1490Tyr)

3129			Y	2	+/-		c.3113C>T (p.Ala1038Val)	c.4469G>A (p.Cys1490Tyr)
3003				2	+/-		c.2588G>C (p.Gly863Ala)	c.4506C>A (p.Cys1502*)
3738		25	N	2	+/-		c.3050+5G>A (p.?)	c.4594G>A (p.Asp1532Asn)
3204		18	N	2	+/-		c.4139C>T (p.Pro1380Leu)	c.4594G>A (p.Asp1532Asn)
3077				2	+/-		c.4139C>T (p.Pro1380Leu)	c.4594G>A (p.Asp1532Asn)
Y703				2	+/-		c.454C>T (p.Arg152*)	c.4594G>A (p.Asp1532Asn)
3553				2	+/-		c.2588G>C (p.Gly863Ala)	c.6286G>A (p.Glu2096Lys)
3104				2	+/-		c.5461-10T>C (p.?)	c.5882G>A (p.Gly1961Glu)
Y647				2	+/-		c.5461-10T>C (p.?)	c.5882G>A (p.Gly1961Glu)
3332		23	N	2	+/-		c.5461-10T>C (p.?)	c.5882G>A (p.Gly1961Glu)
3747		17	N	2	+/-		c.5461-10T>C (p.?)	c.5882G>A (p.Gly1961Glu)
3021				2	+/-		c.3322C>T (p.Arg1108Cys)	c.5882G>A (p.Gly1961Glu)
3458		13	N	2	+/-		c.4216C>T (p.His1406Tyr)	c.5882G>A (p.Gly1961Glu)
3946		42	Y	2	+/-	x	c.4216C>T (p.His1406Tyr)	c.5882G>A (p.Gly1961Glu)
3635		17	Y	2	+/-		c.4469G>A (p.Cys1490Tyr)	c.5882G>A (p.Gly1961Glu)
3473	x		Y	2	+/-		c.4469G>A (p.Cys1490Tyr)	c.5882G>A (p.Gly1961Glu)
2020	x	55	Y	2	+/-		c.4594G>A (p.Asp1532Asn)	c.5882G>A (p.Gly1961Glu)
3724		58	Y	2	+/-		c.4594G>A (p.Asp1532Asn)	c.5882G>A (p.Gly1961Glu)
4351	x	12	Y	2	+/-	x	c.5318C>T (p.Ala1773Val)	c.5882G>A (p.Gly1961Glu)
3508		38	Y	2	+/-		c.67-2A>G (p.?)	c.2588G>C (p.Gly863Ala)
3009				2	+/-		c.768G>T (p.?)	c.2588G>C (p.Gly863Ala)
3125				2	+/-		c.1015T>G (p.Trp339Gly)	c.2588G>C (p.Gly863Ala)
3345				2	+/-		c.1574T>C (p.Phe525Ser)	c.2588G>C (p.Gly863Ala)
9026	x	22	N	2	+/-	x	c.1819G>A (p.Gly607Arg)	c.2588G>C (p.Gly863Ala)
3691			N	2	+/-		c.1849C>T (p.Gln617*)	c.2588G>C (p.Gly863Ala)
3787		21	Y	2	+/-		c.1921T>G (p.Cys641Gly)	c.2588G>C (p.Gly863Ala)
3454				2	+/-		c.1964T>G (p.Phe655Cys)	c.2588G>C (p.Gly863Ala)
3264				2	+/-		c.318T>G (p.Tyr106*)	c.2588G>C (p.Gly863Ala)
3366	x	5		2	+/-		c.5572T>A (p.Tyr1858Asn)	c.5822A>C (p.His1941Pro)
3255		18	N	2	+/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5537T>C (p.Ile1846Thr)
3101				2	+/-		c.5461-10T>C (p.?)	c.6079C>T (p.Leu2027Phe)
3298	x	11	N	2	+/-		c.2552G>A (p.Gly851Asp)	c.6079C>T (p.Leu2027Phe)
3155			Y	2	+/-		c.1253T>C (p.Phe418Ser)	c.4326C>A (p.Asn1442Lys)
3800		18	N	2	+/-		c.2588G>C (p.Gly863Ala)	c.3085C>T (p.Gln1029*)
3216	x			2	+/-		c.2588G>C (p.Gly863Ala)	c.4234C>T (p.Gln1412*)
3292			N	2	+/-		c.2894A>G (p.Asn965Ser)	c.5138A>G (p.Gln1713Arg)
5007	x	5	N	2	+/-		c.4537del (p.Gln1513fs)	c.319C>T (p.Arg107*)
4563	x	48	N	2	+/-		c.2588G>C (p.Gly863Ala)	c.3322C>T (p.Arg1108Cys)
3237	x	9	N	2	+/-		c.1253T>C (p.Phe418Ser)	c.3322C>T (p.Arg1108Cys)
3070				2	+/-		c.2588G>C (p.Gly863Ala)	c.4919G>A (p.Arg1640Gln)
3965	x	12	N	2	+/-	x	c.2564G>A (p.Trp855*)	c.5114G>A (p.Arg1705Gln)

3648	x			2	+/-		c.2588G>C (p.Gly863Ala)		c.5778_5779del (p.Arg1927fs)
3865	x		Y	2	+/-		c.4469G>A (p.Cys1490Tyr)		c.6089G>A (p.Arg2030Gln)
4514		7	N	2	+/-		c.[2041C>T;2588G>C] (p.[Arg681*;Gly863Ala])		c.6088C>T (p.Arg2030*)
3981				2	+/-		c.2588G>C (p.Gly863Ala)		c.6088C>T (p.Arg2030*)
3400				2	+/-		c.5461-10T>C (p.?)		c.6119G>A (p.Arg2040Gln)
3902		50	Y	2	+/-		c.1957C>T (p.Arg653Cys)		c.1957C>T (p.Arg653Cys)
9018		45	N	2	+/-		c.2966T>C (p.Val989Ala)		c.3210_3211dup (p.Ser1071fs)
3257				2	+/-		c.2588G>C (p.Gly863Ala)		c.3335C>A (p.Thr1112Asn)
3600		8	N	2	+/-		c.3322C>T (p.Arg1108Cys)		c.4577C>T (p.Thr1526Met)
3188				2	+/-		c.2919-2A>G (p.?)		c.6718A>G (p.Thr2240Ala)
3049	x			2	+/-		c.2588G>C (p.Gly863Ala)		c.2947A>G (p.Thr983Ala)
3074	x	36	Y	2	+/-		c.5461-10T>C (p.?)		c.1843G>T (p.Val615Phe)
3349				2	+/-		c.5461-10T>C (p.?)		c.122G>A (p.Trp41*)
3509	x			2	+/-		c.2588G>C (p.Gly863Ala)		c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])
3477	x			2	+/-		c.2588G>C (p.Gly863Ala)		c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])
3348	x			2	-/-	x	c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])		c.1938-619A>G (p.?)
3449	x			2	-/-		c.571-1G>T (p.?)		c.2160+1G>C (p.?)
5225	x			2	-/-		c.161G>A (p.Cys54Tyr)		c.2160+1G>C (p.?)
3193				2	-/-		c.3322C>T (p.Arg1108Cys)		c.3522+5del (p.?)
3061				2	-/-		c.3113C>T (p.Ala1038Val)		c.3813+1G>T (p.?)
3326			N	2	-/-		c.2971G>C (p.Gly991Arg)		c.4253+4C>T (p.?)
3241				2	-/-		c.2588G>C (p.Gly863Ala)		c.4253+5G>T (p.?)
4542	x			2	-/-	x	c.2966T>C (p.Val989Ala)		c.4253+5G>T (p.?)
3031				2	-/-		c.3386G>A (p.Arg1129His)		c.4352+1G>A (p.?)
3525	x			2	-/-	x	c.768G>T (p.?)		c.4539+2001G>A (p.?)
3134		5	N	2	-/-	x	c.2564G>A (p.Trp855*)		c.4539+2001G>A (p.?)
3786		14	N	2	-/-	x	c.1022A>G (p.Glu341Gly)		c.4539+2028C>T (p.?)
3513	x			2	-/-	x	c.161G>A (p.Cys54Tyr)		c.4539+2028C>T (p.?)
3291				2	-/-	x	c.4577C>T (p.Thr1526Met)		c.4539+2028C>T (p.?)
3798		6	N	2	-/-	x	c.5113C>T (p.Arg1705Trp)		c.4539+2028C>T (p.?)
3142	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])		c.4540-2A>G (p.?)
3221	x			2	-/-		c.4328G>A (p.Arg1443His)		c.4667+1G>A (p.?)
9004				2	-/-	x	c.161G>A (p.Cys54Tyr)		c.4773+3A>G (p.?)
3399	x			2	-/-		c.5018+2T>C (p.?)		c.5018+2T>C (p.?)
3642		19	N	2	-/-		c.1726G>C (p.Asp576His)		c.5196+1056A>G (p.?)
3556				2	-/-		c.3413T>A (p.Leu1138His)		c.5196+1056A>G (p.?)
4271	x			2	-/-	x	c.5882G>A (p.Gly1961Glu)		c.5196+1056A>G (p.?)
3330				2	-/-	x	c.5882G>A (p.Gly1961Glu)		c.5196+1056A>G (p.?)
3430	x			2	-/-	x	c.161G>A (p.Cys54Tyr)		c.5196+1137G>A (p.?)
3827	x			2	-/-	x	c.161G>A (p.Cys54Tyr)		c.5196+1137G>A (p.?)
3151				2	-/-	x	c.666_678del (p.Lys223_Arg226delfs)		c.5196+1137G>A (p.?)

3788	x	32	Y	2	-/-	x	c.666_678del (p.Lys223_Arg226delfs)	c.5196+1137G>A (p.?)
3097				2	-/-		c.571-2_c.575del (p.?)	c.5313-2_c.5316del (p.?)
3143				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5460+1G>A (p.?)
3295				2	-/-	x	c.2971G>C (p.Gly991Arg)	c.570+1798A>G (p.?)
Y417				2	-/-	x	c.52C>T (p.Arg18Trp)	c.570+1798A>G (p.?)
3954	x			2	-/-	x	c.571-1G>T (p.?)	c.571-1G>T (p.?)
3192				2	-/-		c.3522+5del (p.?)	c.5714+5G>A (p.?)
9025		42	Y	2	-/-	x	c.5714+5G>A (p.?)	c.5714+5G>A (p.?)
3353	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5714+5G>A (p.?)
3730		13	N	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5714+5G>A (p.?)
3225				2	-/-		c.1622T>C (p.Leu541Pro)	c.5714+5G>A (p.?)
3570		24	N	2	-/-		c.1648G>A (p.Gly550Arg)	c.5714+5G>A (p.?)
3226				2	-/-	x	c.2092T>C (p.Cys698Arg)	c.5714+5G>A (p.?)
3725				2	-/-		c.2915C>A (p.Thr972Asn)	c.5714+5G>A (p.?)
9001	x	17	Y	2	-/-	x	c.3352C>T (p.His1118Tyr)	c.5714+5G>A (p.?)
3709	x	19	N	2	-/-		c.4139C>T (p.Pro1380Leu)	c.5714+5G>A (p.?)
3859	x	8	N	2	-/-		c.4139C>T (p.Pro1380Leu)	c.5714+5G>A (p.?)
3888	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.5714+5G>A (p.?)
3071				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5714+5G>A (p.?)
3387		43	Y	2	-/-		c.4139C>T (p.Pro1380Leu)	c.5714+5G>A (p.?)
3059	x			2	-/-		c.106del (p.Leu37fs)	c.5714+5G>A (p.?)
3378				2	-/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5836-3C>A (p.?)
3391	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.6005+1G>T (p.?)
3124		42	Y	2	-/-	x	c.3814-2A>G (p.?)	c.6006-609T>A (p.?)
3023	x			2	-/-	x	c.5312+2T>G (p.?)	c.6006-609T>A (p.?)
9029		34	Y	2	-/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6006-609T>A (p.?)
3782	x			2	-/-	x	c.161G>A (p.Cys54Tyr)	c.6006-609T>A (p.?)
3001				2	-/-	x	c.1988G>A (p.Trp663*)	c.6006-609T>A (p.?)
3638	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.6148-698_c.6670del (p.?)
3971	x			2	-/-	x	c.4577C>T (p.Thr1526Met)	c.6386+2C>G (p.?)
3398				2	-/-		c.5882G>A (p.Gly1961Glu)	c.6387-1G>T (p.?)
3809	x	17	N	2	-/-		c.5882G>A (p.Gly1961Glu)	c.6479+1G>A (p.?)
Y416				2	-/-		c.71G>A (p.Arg24His)	c.6479+1G>C (p.?)
3925		31		2	-/-	x	c.5882G>A (p.Gly1961Glu)	c.67-1845_c.160+345del (p.?)
3539	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.6729+5_6729+19del (p.?)
3928				2	-/-		c.5882G>A (p.Gly1961Glu)	c.6729+5_6729+19del (p.?)
3572				2	-/-		c.859-9T>C (p.?)	c.859-9T>C (p.?)
3106	x			2	-/-		c.859-9T>C (p.?)	c.859-9T>C (p.?)
3392				2	-/-		c.5318C>T (p.Ala1773Val)	c.4773G>C (p.?)
3035				2	-/-	x	c.5882G>A (p.Gly1961Glu)	c.6342G>A (p.?)
3516				2	-/-		c.2894A>G (p.Asn965Ser)	c.[3416A>G;6119G>A] (p.[Tyr1139Cys;Arg2040Gln])

3263				2	-/-		c.194G>A (p.Gly65Glu)	c.3113C>T (p.Ala1038Val)
3384	x			2	-/-		c.1988G>A (p.Trp663*)	c.3113C>T (p.Ala1038Val)
9084	x			2	-/-		c.247_250dup (p.Ser84fs)	c.3113C>T (p.Ala1038Val)
3511	x			2	-/-		c.4457C>T (p.Pro1486Leu)	c.4793C>A (p.Ala1598Asp)
3198	x			2	-/-		c.4793C>A (p.Ala1598Asp)	c.4793C>A (p.Ala1598Asp)
3385	x			2	-/-		c.4793C>A (p.Ala1598Asp)	c.4793C>A (p.Ala1598Asp)
4515	x	12	N	2	-/-		c.3386G>T (p.Arg1129Leu)	c.5318C>T (p.Ala1773Val)
3785		11	N	2	-/-		c.5380G>C (p.Ala1794Pro)	c.5380G>C (p.Ala1794Pro)
3052				2	-/-	x	c.3287C>T (p.Ser1096Leu)	c.6108_6130dup (p.Ala2044ValinsProGlyPheGluValTyrGln)
3483		10	N	2	-/-		c.2453G>A (p.Gly818Glu)	c.4462T>C (p.Cys1488Arg)
3818	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.6448T>C (p.Cys2150Arg)
3416	x			2	-/-		c.[2560G>A;3113C>T] (p.[Ala854Thr;Ala1038Val])	c.6449G>A (p.Cys2150Tyr)
3498	x			2	-/-		c.2461T>A (p.Trp821Arg)	c.6449G>A (p.Cys2150Tyr)
3561				2	-/-		c.5882G>A (p.Gly1961Glu)	c.6449G>A (p.Cys2150Tyr)
4185		12	N	2	-/-		c.5714+5G>A (p.?)	c.161G>A (p.Cys54Tyr)
3231				2	-/-		c.247_250dup (p.Ser84fs)	c.323A>T (p.Asp108Val)
3075				2	-/-		c.52C>T (p.Arg18Trp)	c.3606T>A (p.Asp1202Glu)
3748	x	3	N	2	-/-		c.834del (p.Asp279fs)	c.834delT (p.Asp279fs)
3732		32	Y	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.1726G>C (p.Asp576His)
							c.[1622T>C;1933G>A;3113C>T]	
3423		12	N	2	-/-		(p.[Leu541Pro;Asp645Asn;Ala1038Val])	c.1933G>A (p.Asp645Asn)
3821	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.3259G>A (p.Glu1087Lys)
3960	x	17	N	2	-/-	x	c.2401G>A (p.Ala801Thr)	c.3259G>A (p.Glu1087Lys)
9064	x			2	-/-		c.3988G>T (p.Glu1330*)	c.3988G>T (p.Glu1330*)
3113				2	-/-		c.4854G>T (p.Trp1618Cys)	c.6694G>A (p.Glu2232Lys)
4333	x			2	-/-		c.1222C>T (p.Arg408*)	c.4249_4251del (p.Phe1417del)
3211				2	-/-		c.3323G>A (p.Arg1108His)	c.4318T>G (p.Phe1440Val)
3478				2	-/-		c.71G>A (p.Arg24His)	c.751_753del (p.Phe251del)
3220	x			2	-/-		c.101_106del (p.Ser34_Leu35delfs)	c.3547G>T (p.Gly1183Cys)
3578	x			2	-/-		c.3113C>T (p.Ala1038Val)	c.4322G>A (p.Gly1441Asp)
3494	x			2	-/-		c.[1622T>C;4328G>A] (p.[Leu541Pro;Arg1443His])	c.5882G>A (p.Gly1961Glu)
3262	x	32	Y	2	-/-		c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])	c.5882G>A (p.Gly1961Glu)
3586	x			2	-/-		c.[1A>G;6089G>A] (p.[?;Arg2030Gln])	c.5882G>A (p.Gly1961Glu)
3401	x			2	-/-		c.[3608G>A;4537del] (p.[Gly1203Glu;Gln1513fs])	c.5882G>A (p.Gly1961Glu)
3501	x			2	-/-		c.1100-1G>A (p.?)	c.5882G>A (p.Gly1961Glu)
3418		19	N	2	-/-		c.160+2T>C (p.?)	c.5882G>A (p.Gly1961Glu)
3267				2	-/-		c.160+5G>C (p.?)	c.5882G>A (p.Gly1961Glu)
3039				2	-/-		c.2382+1G>A (p.?)	c.5882G>A (p.Gly1961Glu)
3343	x			2	-/-		c.3050+5G>A (p.?)	c.5882G>A (p.Gly1961Glu)
3594	x			2	-/-		c.3050+5G>A (p.?)	c.5882G>A (p.Gly1961Glu)
3708		13		2	-/-		c.3050+5G>A (p.?)	c.5882G>A (p.Gly1961Glu)

4385				2	-/-		c.3050+5G>A (p.?)	c.5882G>A (p.Gly1961Glu)
3309				2	-/-		c.4539+1G>T (p.?)	c.5882G>A (p.Gly1961Glu)
3533				2	-/-		c.4540-2A>G (p.?)	c.5882G>A (p.Gly1961Glu)
3453				2	-/-		c.5018+2T>C (p.?)	c.5882G>A (p.Gly1961Glu)
3260	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3214	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
4547				2	-/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3248				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3324				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3964		11		2	-/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3167		10	Y	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3249				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3322				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3373				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
3091		33	Y	2	-/-		c.1208A>T (p.Asp403Val)	c.5882G>A (p.Gly1961Glu)
3066				2	-/-		c.161G>A (p.Cys54Tyr)	c.5882G>A (p.Gly1961Glu)
3289	x			2	-/-		c.1622T>C (p.Leu541Pro)	c.5882G>A (p.Gly1961Glu)
3371				2	-/-		c.1804C>T (p.Arg602Trp)	c.5882G>A (p.Gly1961Glu)
3842				2	-/-	x	c.1819G>T (p.Gly607Trp)	c.5882G>A (p.Gly1961Glu)
3000				2	-/-		c.1908G>T (p.Gln636His)	c.5882G>A (p.Gly1961Glu)
3852				2	-/-		c.1957C>T (p.Arg653Cys)	c.5882G>A (p.Gly1961Glu)
3109				2	-/-		c.1964T>G (p.Phe655Cys)	c.5882G>A (p.Gly1961Glu)
3409	x			2	-/-		c.1988G>A (p.Trp663*)	c.5882G>A (p.Gly1961Glu)
3016	x			2	-/-		c.1A>G (p.?)	c.5882G>A (p.Gly1961Glu)
3175				2	-/-		c.2453G>T (p.Gly818Val)	c.5882G>A (p.Gly1961Glu)
3824	x			2	-/-		c.286A>G (p.Asn96Asp)	c.5882G>A (p.Gly1961Glu)
3792				2	-/-		c.3007C>T (p.Gln1003*)	c.5882G>A (p.Gly1961Glu)
3130		36	Y	2	-/-		c.3322C>T (p.Arg1108Cys)	c.5882G>A (p.Gly1961Glu)
3208				2	-/-		c.3364G>A (p.Glu1122Lys)	c.5882G>A (p.Gly1961Glu)
3493				2	-/-		c.3385C>T (p.Arg1129Cys)	c.5882G>A (p.Gly1961Glu)
3475				2	-/-		c.4070C>T (p.Ala1357Val)	c.5882G>A (p.Gly1961Glu)
3435	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
3451	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
3543	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
3565	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
7076				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
Y391				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
4361				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5882G>A (p.Gly1961Glu)
Y395				2	-/-		c.4217A>G (p.His1406Arg)	c.5882G>A (p.Gly1961Glu)
3233				2	-/-		c.4234C>T (p.Gln1412*)	c.5882G>A (p.Gly1961Glu)
3382				2	-/-		c.4234C>T (p.Gln1412*)	c.5882G>A (p.Gly1961Glu)

3784		27	N	2	-/-		c.4462T>C (p.Cys1488Arg)	c.5882G>A (p.Gly1961Glu)
3323				2	-/-		c.454C>T (p.Arg152*)	c.5882G>A (p.Gly1961Glu)
3254	x			2	-/-		c.45G>A (p.Trp15*)	c.5882G>A (p.Gly1961Glu)
3067				2	-/-		c.5087G>A (p.Ser1696Asn)	c.5882G>A (p.Gly1961Glu)
3196				2	-/-		c.5114G>T (p.Arg1705Leu)	c.5882G>A (p.Gly1961Glu)
3062				2	-/-		c.5316G>A (p.Trp1772*)	c.5882G>A (p.Gly1961Glu)
3469				2	-/-	x	c.5318C>T (p.Ala1773Val)	c.5882G>A (p.Gly1961Glu)
6046	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
3329	x			2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
3190				2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
3235				2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
4085	x			2	-/-		c.634C>T (p.Arg212Cys)	c.5882G>A (p.Gly1961Glu)
3432	x			2	-/-		c.634C>T (p.Arg212Cys)	c.5882G>A (p.Gly1961Glu)
3222				2	-/-		c.564del (p.Glu189fs)	c.5882G>A (p.Gly1961Glu)
3804				2	-/-		c.2191_2199del (p.Phe731_Leu733del)	c.5882G>A (p.Gly1961Glu)
3266				2	-/-		c.4734del (p.Leu1580*)	c.5882G>A (p.Gly1961Glu)
3275		13	Y	2	-/-		c.4537dup (p.Gln1513fs)	c.5882G>A (p.Gly1961Glu)
9028		16	Y	2	-/-		c.5196+1G>A (p.?)	c.5882G>A (p.Gly1961Glu)
4602	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
9090	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5882G>A (p.Gly1961Glu)
9056				2	-/-		c.3407G>T (p.Gly1136Val)	c.5882G>A (p.Gly1961Glu)
3993				2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
3998		49	Y	2	-/-		c.5882G>A (p.Gly1961Glu)	c.5882G>A (p.Gly1961Glu)
9068	x			2	-/-		c.4947del (p.Glu1650fs)	c.5882G>A (p.Gly1961Glu)
3480				2	-/-		c.2947A>G (p.Thr983Ala)	c.5882G>A (p.Gly1961Glu)
3174				2	-/-		c.286A>G (p.Asn96Asp)	c.[5959G>T;c.5961_5964del] (p.Gly1987*)
9019				2	-/-		c.2588G>C (p.Gly863Ala)	c.2897G>A (p.Gly966Glu)
3044				2	-/-		c.2971G>C (p.Gly991Arg)	c.4875T>A (p.His1625Gln)
3904		20	Y	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5051T>A (p.Ile1684Asn)
4060	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.3543del (p.Lys1182fs)
3688		42	Y	2	-/-		c.2925C>G (p.Ile975Met)	c.5932A>G (p.Lys1978Glu)
3137		36	Y	2	-/-	x	c.2971G>C (p.Gly991Arg)	c.3377T>C (p.Leu1126Pro)
3470	x			2	-/-	x	c.2971G>C (p.Gly991Arg)	c.3413T>C (p.Leu1138Pro)
3015				2	-/-		c.3279C>A (p.Asp1093Glu)	c.3819dup (p.Leu1274fs)
3956		14	Y	2	-/-	x	c.2813T>C (p.Phe938Ser)	c.5186T>C (p.Leu1729Pro)
3502				2	-/-		c.2947A>G (p.Thr983Ala)	c.5186T>C (p.Leu1729Pro)
3968				2	-/-		c.5059del (p.Ile1687fs)	c.5351T>G (p.Leu1784Arg)
3173				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5819T>A (p.Leu1940Gln)
9012		22	Y	2	-/-	x	c.[71G>A;5882G>A] (p.[Arg24His;Gly1961Glu])	c.6079C>T (p.Leu2027Phe)
3313				2	-/-		c.4253+5G>T (p.?)	c.6079C>T (p.Leu2027Phe)
3010	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6079C>T (p.Leu2027Phe)

3778	x			2	-/-			c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6079C>T (p.Leu2027Phe)
3466				2	-/-			c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6079C>T (p.Leu2027Phe)
3604	x			2	-/-			c.2915C>A (p.Thr972Asn)	c.6079C>T (p.Leu2027Phe)
3252	x			2	-/-			c.3323G>A (p.Arg1108His)	c.6079C>T (p.Leu2027Phe)
3209				2	-/-			c.4139C>T (p.Pro1380Leu)	c.6079C>T (p.Leu2027Phe)
3765				2	-/-			c.4457C>T (p.Pro1486Leu)	c.6079C>T (p.Leu2027Phe)
3011				2	-/-			c.4577C>T (p.Thr1526Met)	c.6079C>T (p.Leu2027Phe)
3962		44	Y	2	-/-		x	c.5882G>A (p.Gly1961Glu)	c.6079C>T (p.Leu2027Phe)
3242				2	-/-			c.91T>C (p.Trp31Arg)	c.6079C>T (p.Leu2027Phe)
3817		12	Y	2	-/-			c.5882G>A (p.Gly1961Glu)	c.6098T>A (p.Leu2033His)
3160				2	-/-			c.3292C>T (p.Arg1098Cys)	c.6179T>G (p.Leu2060Arg)
3403	x			2	-/-			c.4437G>A (p.Trp1479*)	c.6419T>A (p.Leu2140Gln)
3164		47	Y	2	-/-			c.6543_6578del (p.Leu2182_Phe2193del)	c.6543_6578del (p.Leu2182_Phe2193del)
3395				2	-/-		x	c.859-9T>C (p.?)	c.885del (p.Leu296fs)
3178				2	-/-			c.1622T>C (p.Leu541Pro)	c.1622T>C (p.Leu541Pro)
3412				2	-/-			c.1804C>T (p.Arg602Trp)	c.5646G>A (p.Met1882Ile)
3411				2	-/-			c.2023G>A (p.Val675Ile)	c.5646G>A (p.Met1882Ile)
3390				2	-/-			c.634C>T (p.Arg212Cys)	c.4326C>A (p.Asn1442Lys)
3686		5	N	2	-/-			c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.2894A>G (p.Asn965Ser)
3811		6	N	2	-/-			c.2461T>A (p.Trp821Arg)	c.2894A>G (p.Asn965Ser)
3364	x			2	-/-			c.2894A>G (p.Asn965Ser)	c.2894A>G (p.Asn965Ser)
3136				2	-/-		x	c.2041C>T (p.Arg681*)	c.294C>G (p.Asn98Lys)
3942		13	N	2	-/-		x	c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])	c.4139C>T (p.Pro1380Leu)
3887				2	-/-			c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])	c.4139C>T (p.Pro1380Leu)
3413				2	-/-			c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])	c.4139C>T (p.Pro1380Leu)
3236				2	-/-			c.[1A>G;6089G>A] (p.[?;Arg2030Gln])	c.4139C>T (p.Pro1380Leu)
5180	x	6	N	2	-/-			c.3050+5G>A (p.?)	c.4139C>T (p.Pro1380Leu)
3419				2	-/-		x	c.859-9T>C (p.?)	c.4139C>T (p.Pro1380Leu)
3548				2	-/-			c.3322C>T (p.Arg1108Cys)	c.4139C>T (p.Pro1380Leu)
3243				2	-/-			c.3322C>T (p.Arg1108Cys)	c.4139C>T (p.Pro1380Leu)
3801				2	-/-			c.3322C>T (p.Arg1108Cys)	c.4139C>T (p.Pro1380Leu)
3858				2	-/-			c.3322C>T (p.Arg1108Cys)	c.4139C>T (p.Pro1380Leu)
3126	x			2	-/-			c.3323G>A (p.Arg1108His)	c.4139C>T (p.Pro1380Leu)
Y423				2	-/-			c.3323G>A (p.Arg1108His)	c.4139C>T (p.Pro1380Leu)
4428		10	N	2	-/-			c.3911T>A (p.Leu1304*)	c.4139C>T (p.Pro1380Leu)
3072	x			2	-/-			c.4139C>T (p.Pro1380Leu)	c.4139C>T (p.Pro1380Leu)
3402				2	-/-			c.4139C>T (p.Pro1380Leu)	c.4139C>T (p.Pro1380Leu)
Y485				2	-/-			c.4139C>T (p.Pro1380Leu)	c.4139C>T (p.Pro1380Leu)
3486				2	-/-			c.4139C>T (p.Pro1380Leu)	c.4139C>T (p.Pro1380Leu)
3953				2	-/-		x	c.4139C>T (p.Pro1380Leu)	c.4139C>T (p.Pro1380Leu)
3006				2	-/-			c.5882G>A (p.Gly1961Glu)	c.4139C>T (p.Pro1380Leu)

3624		5	N	2	-/-		c.768G>T (p.?)	c.4139C>T (p.Pro1380Leu)
3926	x	11	N	2	-/-		c.1025_1038del (p.Asp342_Lys346delfs)	c.4139C>T (p.Pro1380Leu)
3279	x	31	Y	2	-/-		c.2893A>T (p.Asn965Tyr)	c.4457C>T (p.Pro1486Leu)
3805				2	-/-		c.3470T>G (p.Leu1157*)	c.3876G>C (p.Gln1292His)
3154				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.4234C>T (p.Gln1412*)
4286	x			2	-/-		c.3322C>T (p.Arg1108Cys)	c.4234C>T (p.Gln1412*)
3656	x			2	-/-		c.66G>A (p.?)	c.6658C>T (p.Gln2220*)
3128		32	Y	2	-/-		c.[2627A>C;3322C>T] (p.[Gln876Pro;Arg1108Cys])	c.2627A>C (p.Gln876Pro)
9015				2	-/-		c.1819G>A (p.Gly607Arg)	c.3292C>T (p.Arg1098Cys)
3771		8	N	2	-/-		c.2617T>C (p.Phe873Leu)	c.3292C>T (p.Arg1098Cys)
9060		22	N	2	-/-		c.3322C>T (p.Arg1108Cys)	c.3322C>T (p.Arg1108Cys)
3121				2	-/-		c.2947A>G (p.Thr983Ala)	c.3322C>T (p.Arg1108Cys)
3969	x			2	-/-	x	c.3287C>T (p.Ser1096Leu)	c.3322C>T (p.Arg1108Cys)
3873	x			2	-/-		c.478G>T (p.Glu160*)	c.3322C>T (p.Arg1108Cys)
3234				2	-/-		c.634C>T (p.Arg212Cys)	c.3323G>T (p.Arg1108Leu)
3102				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.3385C>T (p.Arg1129Cys)
3288				2	-/-		c.2894A>G (p.Asn965Ser)	c.3386G>T (p.Arg1129Leu)
3899	x	9	N	2	-/-		c.1025_1038del (p.Asp342_Lys346delfs)	c.3386G>T (p.Arg1129Leu)
3912	x			2	-/-		c.2813T>C (p.Phe938Ser)	c.3898C>T (p.Arg1300*)
3060	x			2	-/-		c.1804C>T (p.Arg602Trp)	c.3898C>T (p.Arg1300*)
3447				2	-/-		c.4667G>A (p.Arg1556Lys)	c.4667G>A (p.Arg1556Lys)
3081				2	-/-		c.3056C>T (p.Thr1019Met)	c.4919G>A (p.Arg1640Gln)
3036	x			2	-/-		c.3322C>T (p.Arg1108Cys)	c.4919G>A (p.Arg1640Gln)
3365				2	-/-		c.4139C>T (p.Pro1380Leu)	c.4919G>A (p.Arg1640Gln)
3187				2	-/-		c.5196+1G>A (p.?)	c.6089G>A (p.Arg2030Gln)
3183		33	Y	2	-/-		c.1648G>A (p.Gly550Arg)	c.6089G>A (p.Arg2030Gln)
3407	x			2	-/-		c.4139C>T (p.Pro1380Leu)	c.6089G>A (p.Arg2030Gln)
4273	x			2	-/-		c.4639A>T (p.Lys1547*)	c.6089G>A (p.Arg2030Gln)
4405				2	-/-		c.6079C>T (p.L2027Phe)	c.6089G>A (p.Arg2030Gln)
3529		33	Y	2	-/-		c.716G>A (p.Trp239*)	c.6089G>A (p.Arg2030Gln)
3870	x			2	-/-		c.5312+1G>A (p.?)	c.6088C>T (p.Arg2030*)
3978		10	N	2	-/-	x	c.5312+2T>G (p.?)	c.6088C>T (p.Arg2030*)
3650				2	-/-		c.6088C>T (p.Arg2030*)	c.6088C>T (p.Arg2030*)
5010				2	-/-		c.1374del (p.Thr459fs)	c.6088C>T (p.Arg2030*)
9057	x	11	Y	2	-/-		c.4577C>T (p.Thr1526Met)	c.6119G>A (p.Arg2040Gln)
3328		7	N	2	-/-		c.1819G>A (p.Gly607Arg)	c.6119G>A (p.Arg2040Gln)
3670		9	N	2	-/-		c.5882G>A (p.Gly1961Glu)	c.6118C>T (p.Arg2040*)
3338	x			2	-/-		c.688T>A (p.Cys230Ser)	c.6229C>G (p.Arg2077Gly)
3723		10	N	2	-/-		c.3385C>T (p.Arg1129Cys)	c.6229C>T (p.Arg2077Trp)
3259				2	-/-		c.5882G>A (p.Gly1961Glu)	c.6229C>T (p.Arg2077Trp)
3583	x			2	-/-		c.6079C>T (p.L2027Phe)	c.6229C>T (p.Arg2077Trp)

3990	x	16	Y	2	-/-		c.4253+5G>A (p.?)	c.6316C>T (p.Arg2106Cys)
3258				2	-/-		c.1100-6T>A (p.?)	c.6316C>T (p.Arg2106Cys)
3598				2	-/-	x	c.3898C>T (p.Arg1300*)	c.6316C>T (p.Arg2106Cys)
3934				2	-/-		c.4538A>G (p.Gln1513Arg)	c.6316C>T (p.Arg2106Cys)
3812				2	-/-		c.3554_3555CC>GA (p.Ser1185*)	c.6317G>A (p.Arg2106His)
Y265				2	-/-		c.1805G>A (p.Arg602Gln)	c.6317_6323del (p.Arg2107_Met2108delfs)
3986				2	-/-		c.1804C>T (p.Arg602Trp)	c.6320G>A (p.Arg2107His)
3775		21	N	2	-/-		c.302+1G>A (p.?)	c.6320G>A (p.Arg2107His)
3523	x			2	-/-		c.3050+5G>A (p.?)	c.6320G>A (p.Arg2107His)
3261				2	-/-		c.3523-1G>A (p.?)	c.6320G>A (p.Arg2107His)
3517	x			2	-/-		c.4540-2A>G (p.?)	c.6320G>A (p.Arg2107His)
3299				2	-/-		c.768G>T (p.?)	c.6320G>A (p.Arg2107His)
3380				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6320G>A (p.Arg2107His)
3559				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.6320G>A (p.Arg2107His)
3664				2	-/-		c.1015T>G (p.Trp339Gly)	c.6320G>A (p.Arg2107His)
3894	x			2	-/-		c.1577A>C (p.Glu526Ala)	c.6320G>A (p.Arg2107His)
3920				2	-/-		c.1577A>C (p.Glu526Ala)	c.6320G>A (p.Arg2107His)
3337		19	N	2	-/-		c.1609C>T (p.Arg537Cys)	c.6320G>A (p.Arg2107His)
3321		44		2	-/-		c.2966T>C (p.Val989Ala)	c.6320G>A (p.Arg2107His)
4556				2	-/-	x	c.2971G>C (p.Gly991Arg)	c.6320G>A (p.Arg2107His)
3092		25	Y	2	-/-		c.4223G>T (p.Trp1408Leu)	c.6320G>A (p.Arg2107His)
3940	x	7		2	-/-	x	c.428C>T (p.Pro143Leu)	c.6320G>A (p.Arg2107His)
3952		43	Y	2	-/-	x	c.6079C>T (p.L2027Phe)	c.6320G>A (p.Arg2107His)
3191				2	-/-		c.6229C>G (p.Arg2077Gly)	c.6320G>A (p.Arg2107His)
Y415				2	-/-		c.6316C>T (p.Arg2106Cys)	c.6320G>A (p.Arg2107His)
3794		60	Y	2	-/-		c.6320G>A (p.Arg2107His)	c.6320G>A (p.Arg2107His)
3082		50	Y	2	-/-		c.658C>T (p.Arg220Cys)	c.6320G>A (p.Arg2107His)
4360				2	-/-		c.247_250dup (p.Ser84fs)	c.6320G>A (p.Arg2107His)
3760		8	N	2	-/-		c.634C>T (p.Arg212Cys)	c.634C>T (p.Arg212Cys)
3376				2	-/-		c.454C>T (p.Arg152*)	c.6445C>T (p.Arg2149*)
3562	x			2	-/-		c.885del (p.Leu296fs)	c.1609C>T (p.Arg537Cys)
3170				2	-/-		c.885del (p.Leu296fs)	c.1609C>T (p.Arg537Cys)
3397				2	-/-		c.1805G>A (p.Arg602Gln)	c.1805G>A (p.Arg602Gln)
3099				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5087G>A (p.Ser1696Asn)
3580	x			2	-/-		c.5196+1G>A (p.?)	c.6703T>C (p.Ser2235Pro)
3182	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.2385C>G (p.Ser795Arg)
4472	x	7	N	2	-/-		c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])	c.3056C>T (p.Thr1019Met)
3589				2	-/-		c.3056C>T (p.Thr1019Met)	c.3056C>T (p.Thr1019Met)
3331				2	-/-		c.1037A>C (p.Lys346Thr)	c.3350C>T (p.Thr1117Ile)
3489				2	-/-		c.4036_4037del (p.Thr1346fs)	c.4036_4037del (p.Thr1346fs)
3424	x			2	-/-		c.2564G>A (p.Trp855*)	c.4577C>T (p.Thr1526Met)

3863	x	10	N	2	-/-		c.4577C>T (p.Thr1526Met)	c.4577C>T (p.Thr1526Met)
3240	x			2	-/-		c.1222C>T (p.Arg408*)	c.5935del (p.Thr1979fs)
3351				2	-/-		c.4139C>T (p.Pro1380Leu)	c.5056G>A (p.Val1686Met)
3474	x			2	-/-		c.4537dup (p.Gln1513fs)	c.5056G>A (p.Val1686Met)
3176				2	-/-		c.2966T>C (p.Val989Ala)	c.5289delT (p.Val1764fs)
3945				2	-/-	x	c.2966T>C (p.Val989Ala)	c.5289delT (p.Val1764fs)
3228				2	-/-		c.5714+5G>A (p.?)	c.5917del (p.Val1973*)
3692	x			2	-/-		c.859-9T>C (p.?)	c.5917del (p.Val1973*)
3227				2	-/-		c.4524_4534del (p.Leu1509_Pro1511delfs)	c.5917del (p.Val1973*)
3107	x			2	-/-		c.5714+5G>A (p.?)	c.5917del (p.Val1973*)
3117				2	-/-		c.5882G>A (p.Gly1961Glu)	c.5917del (p.Val1973*)
9081	x			2	-/-		c.2966T>C (p.Val989Ala)	c.5917del (p.Val1973*)
3957	x	11		2	-/-	x	c.5882G>A (p.Gly1961Glu)	c.5917del (p.Val1973*)
9032				2	-/-	x	c.71G>A (p.Arg24His)	c.1561del (p.Val521Serfs)
3002				2	-/-		c.1957C>T (p.Arg653Cys)	c.2966T>C (p.Val989Ala)
3977	x	10	N	2	-/-	x	c.1995C>A (p.Tyr665*)	c.2966T>C (p.Val989Ala)
3510		39	Y	2	-/-		c.2966T>C (p.Val989Ala)	c.2966T>C (p.Val989Ala)
4338				2	-/-		c.52C>T (p.Arg18Trp)	c.2966T>C (p.Val989Ala)
3504				2	-/-		c.1317G>A (p.Trp439*)	c.4347G>T (p.Trp1449Cys)
3446		9	N	2	-/-	x	c.4139C>T (p.Pro1380Leu)	c.4437G>A (p.Trp1479*)
4225		12	N	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.5316G>A (p.Trp1772*)
3769		8	N	2	-/-		c.91T>C (p.Trp31Arg)	c.91T>C (p.Trp31Arg)
3790	x			2	-/-		c.93G>A (p.Trp31*)	c.93G>A (p.Trp31*)
3687		13	N	2	-/-		c.5018+2T>C (p.?)	c.6107A>G (p.Tyr2036Cys)
9099	x	8	N	2	-/-		c.[179C>T;3830C>T] (p.[Ala60Val;Thr1277Met])	c.[179C>T;3830C>T] (p.[Ala60Val;Thr1277Met])
9040				2	-/-		c.[2549A>G;5882G>A] (p.[Tyr850Cys;Gly1961Glu])	c.[2549A>G;5882G>A] (p.[Tyr850Cys;Gly1961Glu])
3375	x			2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
4574	x	5	N	2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3557				2	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
4580		7	N	2	-/-		c.1529T>C (p.Leu510Pro)	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3876	x			2	-/-		c.3352C>T (p.His1118Tyr)	c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])
3335				2	-/-		c.4139C>T (p.Pro1380Leu)	c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])
3085	x			2	-/-		c.71G>A (p.Arg24His)	c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])
3080		30	Y	2	-/-		c.[872C>T;1531C>T] (p.[Pro291Leu;Arg511Cys])	c.[872C>T;1531C>T] (p.[Pro291Leu;Arg511Cys])
3542		43	Y	1	++	x	c.1027_1028del (p.Asn343*)	
3496		35	Y	1	++	x	c.3754G>T (p.Glu1252*)	
9097	x	32	Y	1	++		c.5461-10T>C (p.?)	
9013		35	Y	1	++		c.4469G>A (p.Cys1490Tyr)	
Y522				1	++	x	c.5461-10T>C (p.?)	
3057				1	++		c.5461-10T>C (p.?)	
3087		30	Y	1	++	x	c.5461-10T>C (p.?)	

3452		46	Y	1	++	x	c.5461-10T>C (p.?)
3464		24	Y	1	++	x	c.2069del (p.Gly690fs)
3195				1	++	x	c.5461-10T>C (p.?)
3051	x			1	+/-	x	c.4918C>T (p.Arg1640Trp)
3426	x	32	Y	1	+/-	x	c.5395A>G (p.Asn1799Asp)
3094	x			1	+/-	x	c.2588G>C (p.Gly863Ala)
3286	x	42	Y	1	+/-	x	c.4670A>G (p.Tyr1557Cys)
3521		57	Y	1	+/-	x	c.5461-10T>C (p.?)
3064				1	+/-		c.5461-10T>C (p.?)
3584		30	Y	1	+/-	x	c.655A>T (p.Arg219*)
3601	x	22	Y	1	+/-	x	c.[3481C>A;5714+5G>A] (p.[Arg1161Ser;?])
3495	x	10	N	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3531		43	N	1	+/-	x	c.4249_4251del (p.Phe1417del)
7203	x	52	Y	1	+/-	x	c.247_250dup (p.Ser84fs)
3100	x		Y	1	+/-	x	c.161G>A (p.Cys54Tyr)
3488		47	Y	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3671	x		Y	1	+/-	x	c.571-1G>T (p.?)
3767	x	50	Y	1	+/-	x	c.6229C>T (p.Arg2077Trp)
3892	x	20	Y	1	+/-	x	c.1804C>T (p.Arg602Trp)
3938	x		Y	1	+/-	x	c.5196+1G>A (p.?)
3381	x	45	Y	1	+/-	x	c.2894A>G (p.Asn965Ser)
3405	x	30	Y	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
9007			Y	1	+/-	x	c.4234C>T (p.Gln1412*)
9011		45	Y	1	+/-	x	c.1957C>T (p.Arg653Cys)
9045				1	+/-		c.5461-10T>C (p.?)
9047				1	+/-		c.6386+1G>A (p.?)
9051			Y	1	+/-		c.52C>T (p.Arg18Trp)
9095		26	N	1	+/-		c.262G>A (p.Gly88Arg)
4546		10		1	+/-	x	c.2588G>C (p.Gly863Ala)
3422				1	+/-	x	c.3328+1G>A (p.?)
4512			Y	1	+/-		c.4234C>T (p.Gln1412*)
3357		48	Y	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3293				1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3028		13	N	1	+/-		c.2894A>G (p.Asn965Ser)
3704		50	Y	1	+/-	x	c.4139C>T (p.Pro1380Leu)
3766		31	Y	1	+/-	x	c.1921T>C (p.Cys641Arg)
3429		27	Y	1	+/-	x	c.4918C>T (p.Arg1640Trp)
3361		28	Y	1	+/-	x	c.6166A>T (p.Lys2056*)
3127		52	Y	1	+/-	x	c.3846del (p.Gly1283fs)
3046				1	+/-		c.6386+2C>G (p.?)
3527		46		1	+/-	x	c.161G>A (p.Cys54Tyr)

4115		44		1	+/-	x	c.1222C>T (p.Arg408*)
3813		40	Y	1	+/-	x	c.868C>T (p.Arg290Trp)
3110		36		1	+/-	x	c.6543_6578del (p.Leu2182_Phe2193del)
4594		35	Y	1	+/-		c.4139C>T (p.Pro1380Leu)
3245		22	N	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3862		71	Y	1	+/-	x	c.1903C>T (p.Gln635*)
3803		67	Y	1	+/-	x	c.3261A>C (p.Glu1087Asp)
3098		52	Y	1	+/-	x	c.161-1G>A (p.?)
3485		48	Y	1	+/-	x	c.5923G>C (p.Gly1975Arg)
3456		40	Y	1	+/-	x	c.3259G>A (p.Glu1087Lys)
3089		40	Y	1	+/-	x	c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3903		32	Y	1	+/-	x	c.45G>A (p.Trp15*)
3123		31	Y	1	+/-	x	c.3812A>G (p.Glu1271Gly)
3492		29	Y	1	+/-	x	c.6229C>T (p.Arg2077Trp)
3162		21	Y	1	+/-	x	c.1906C>T (p.Gln636*)
3159		18	Y	1	+/-	x	c.5461-10T>C (p.?)
3450				1	+/-	x	c.[4222T>C;4918C>T] (p.[Trp1408Arg;Arg1640Trp])
3045				1	+/-	x	c.247_250dup (p.Ser84fs)
3020				1	+/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
3008	x			1	-/-		c.3148G>A (p.Gly1050Ser)
3032	x			1	-/-	x	c.[735T>G;2300T>A] (p.[Tyr245*;Val767Asp])
3073	x			1	-/-	x	c.3148G>A (p.Gly1050Ser)
3037	x			1	-/-	x	c.[5882G>A;6445C>T] (p.[Gly1961Glu;Arg2149*])
3468	x			1	-/-	x	c.5714+5G>A (p.?)
3268		42	Y	1	-/-	x	c.4248C>A (p.Phe1416Leu)
3520		59	Y	1	-/-	x	c.5714+5G>A (p.?)
3316				1	-/-	x	c.247_250dup (p.Ser84fs)
3532		57	Y	1	-/-	x	c.3043T>C (p.Phe1015Leu)
3603				1	-/-	x	c.5714+5G>A (p.?)
4367				1	-/-	x	c.2971G>C (p.Gly991Arg)
3609	x			1	-/-	x	c.6320G>A (p.Arg2107His)
3487				1	-/-	x	c.4918C>T (p.Arg1640Trp)
3706		16	N	1	-/-	x	c.2966T>C (p.Val989Ala)
3733	x			1	-/-	x	c.4577C>T (p.Thr1526Met)
3832	x		N	1	-/-	x	c.5882G>A (p.Gly1961Glu)
4565	x		N	1	-/-	x	c.733T>C (p.Tyr245His)
3982	x			1	-/-		c.4793C>A (p.Ala1598Asp)
3999	x			1	-/-		c.5882G>A (p.Gly1961Glu)
3150	x	22	Y	1	-/-	x	c.3322C>T (p.Arg1108Cys)
4638	x			1	-/-		c.5761G>A (p.Val1921Met)
9066	x			1	-/-		c.3292C>T (p.Arg1098Cys)

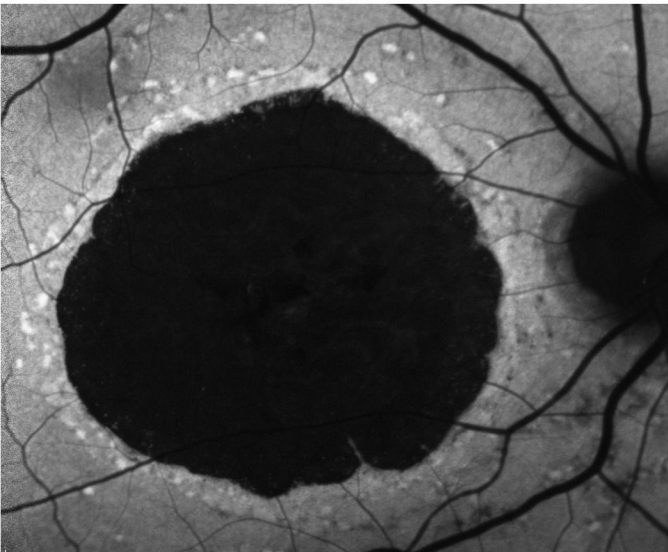
9096	x			1	-/-		c.5882G>A (p.Gly1961Glu)
9017				1	-/-	x	c.4353-4T>C (p.?)
Y256				1	-/-	x	c.634C>T (p.Arg212Cys)
5360				1	-/-	x	c.5882G>A (p.Gly1961Glu)
4720			N	1	-/-		c.6089G>A (p.Arg2030Gln)
3297				1	-/-	x	c.5882G>A (p.Gly1961Glu)
3943		28	Y	1	-/-	x	c.2616del (p.Leu274fs)
3363				1	-/-	x	c.161G>A (p.Cys54Tyr)
3056				1	-/-		c.3113C>T(p.Ala1038Val)
3118		44	Y	1	-/-	x	c.6320G>A (p.Arg2107His)
3133		41	Y	1	-/-	x	c.5312+1G>A (p.?)
3901		10	Y	1	-/-	x	c.3113C>T(p.Ala1038Val)
5222			N	1	-/-	x	c.2966T>C (p.Val989Ala)
4632			N	1	-/-		c.[1622T>C;3113C>T] (p.[Leu541Pro;Ala1038Val])
4391				1	-/-	x	c.6229C>T (p.Arg2077Trp)
3944				1	-/-	x	c.2041C>T (p.Arg681*)
3579				1	-/-	x	c.6320G>A (p.Arg2107His)
4120				1	-/-	x	c.5881G>A (p.Gly1961Arg)
3278				1	-/-	x	c.6694G>A (p.Glu2232Lys)
4203				1	-/-	x	c.634C>T (p.Arg212Cys)
3898				1	-/-	x	c.1034A>G (p.Tyr345Cys)
3315				1	-/-	x	c.6088C>T (p.Arg2030*)
4541				1	-/-	x	c.1699G>A (p.Val567Met)
3103				1	-/-	x	c.184C>T (p.Pro62Ser)
3327				1	-/-	x	c.[3758C>T;5882G>A] (p.[Thr1253Met;Gly1961Glu])
3740				1	-/-	x	c.4577C>T (p.Thr1526Met)
5056				1	-/-		c.6320G>A (p.Arg2107His)
3018				1	-/-		c.4139C>T (p.Pro1380Leu)
3636		37		0	++		
3212	x			0	+/-		
3119	x	47	Y	0	+/-	x	
3834	x	24	Y	0	+/-		
3939	x	13	N	0	+/-	x	
9093		22	N	0	+/-		
Y382				0	+/-		
Y394				0	+/-		
Y458				0	+/-		
Y533				0	+/-		
3992		10	N	0	+/-		
3314		50	Y	0	+/-		
3280		14	Y	0	+/-		

3120			Y	0	+/-	
3161		44	Y	0	+/-	
3967		3	N	0	+/-	x
3163			N	0	+/-	
3325				0	+/-	
3194				0	-/-	
3224				0	-/-	
3247	x			0	-/-	
3414	x			0	-/-	
3459	x			0	-/-	
3497			Y	0	-/-	
3530	x		Y	0	-/-	
3746	x		Y	0	-/-	
3768	x		Y	0	-/-	
4629	x			0	-/-	
7124				0	-/-	
7728				0	-/-	
9003			N	0	-/-	x
9052			N	0	-/-	
4618			N	0	-/-	
4270				0	-/-	
4184				0	-/-	
4205				0	-/-	
4201				0	-/-	
3294				0	-/-	
3307				0	-/-	
3239				0	-/-	
3795			N	0	-/-	
3796				0	-/-	
3797				0	-/-	
3362				0	-/-	
3189				0	-/-	
3132			N	0	-/-	
3643			N	0	-/-	
3948			N	0	-/-	x
3949			N	0	-/-	x
3088			Y	0	-/-	x
3281			Y	0	-/-	
3659				0	-/-	
3726				0	-/-	
3053				0	-/-	
4172				0	-/-	

3396	0	-/-	x
3029	0	-/-	
3034	0	-/-	
3041	0	-/-	
3069	0	-/-	
3111	0	-/-	
3131	0	-/-	
3135	0	-/-	
3144	0	-/-	
3145	0	-/-	
3157	0	-/-	
3165	0	-/-	
3166	0	-/-	
3205	0	-/-	
3206	0	-/-	
3223	0	-/-	
3251	0	-/-	
3282	0	-/-	
3296	0	-/-	
3311	0	-/-	
3369	0	-/-	
3370	0	-/-	
3372	0	-/-	
3374	0	-/-	
3377	0	-/-	
3383	0	-/-	
3393	0	-/-	
3404	0	-/-	
3448	0	-/-	
3554	0	-/-	
3555	0	-/-	
3558	0	-/-	
3560	0	-/-	
3552	0	-/-	

Unifocal Atrophy with Lesion-Centric Flecks

c.5603A>T (p.Asn1868Ile)



c.5882G>A (p.Gly1961Glu)

