

Age at assessment	Sex	Origins	Nucleotide change (NM 017890.3)	Amino acid change	First affected exon	Presence of cataract	Type of cataract	Reference
13	M	Iran	c.4028_4609del	p.(Glu1537Argfs*7)	32	No	N.A.	Alipour et al. 2019
8	M	Iran	c.11486dupG	p.(Leu3830Trpfs*13)	62	No	N.A.	Alipour et al. 2019
1	M	Iran	c.10360dupC	p.(Leu2454fs*7)	59	No	N.A.	Alipour et al. 2019
2	M	Italian	c.8697-9A>G ; c.10156dupA	? ; p.(Thr3386Asnfs*3)	51 ; 59	No	N.A.	Althasakis et al. 2012
11	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	No	N.A.	Douzou et al. 2011
12	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	No	N.A.	Douzou et al. 2011
13	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
15	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
21	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
26	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
28	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
29	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
37	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
37	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
42	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
53	M	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Nuclear sclerotic	Douzou et al. 2011
55	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Hypermetropia	Douzou et al. 2011
57	F	Greek	c.(580+1_581-1) (2333+1_2334-1)del	p.(Ala194Glyfs*9)	6	Yes	Hypermetropia	Douzou et al. 2011
15	M	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	Yes	N.D.	Falk et al. 2004
9	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
6	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
9	M	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
7	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
6	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
14	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	Yes	N.D.	Falk et al. 2004
13	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	No	N.A.	Falk et al. 2004
17	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thrfs*16)	54	No	N.A.	Horn et al. 2000
10	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thrfs*16)	54	No	N.A.	Horn et al. 2000
6	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thrfs*16)	54	No	N.A.	Horn et al. 2000
2	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
5	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
14	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
15	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	Cortical	Kivite-Kallio et al. 2000
19	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
20	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Cortical	Kivite-Kallio et al. 2000
20	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
21	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Posterior Subcapsular	Kivite-Kallio et al. 2000
26	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
27	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
34	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical	Kivite-Kallio et al. 2000
34	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	Nuclear sclerotic	Kivite-Kallio et al. 2000
34	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
39	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	No	N.A.	Kivite-Kallio et al. 2000
39	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical	Kivite-Kallio et al. 2000
40	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical / Posterior subcapsular	Kivite-Kallio et al. 2000
42	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical	Kivite-Kallio et al. 2000
46	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Posterior subcapsular	Kivite-Kallio et al. 2000
46	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical	Kivite-Kallio et al. 2000
46	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical	Kivite-Kallio et al. 2000
51	F	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical / Posterior subcapsular	Kivite-Kallio et al. 2000
51	M	Finnish	c.3348_3349delCT	p.(Cys1117Phefs*8)	22	Yes	Nuclear sclerotic / Cortical / Posterior subcapsular	Kivite-Kallio et al. 2000
19	F	Lebanese	c.9427G	p.(Glu2151)	2	No	N.A.	Kashir et al. 2015
53	M	Lebanese	c.9406-1G>C	p.(Tyr3136Thrfs*16)	54	Yes	Posterior Subcapsular	Megarbane et al. 2001 ; Megarbane et al. 2009
41	M	Lebanese	c.9406-1G>C	p.(Tyr3136Thrfs*16)	54	Yes	Posterior Subcapsular	Megarbane et al. 2001 ; Megarbane et al. 2009
10	F	Omani	c.7934G>A	p.(Glu2645Asp)	46	No	N.A.	Mochida et al. 2004
8	M	Omani	c.7934G>A	p.(Glu2645Asp)	46	No	N.A.	Mochida et al. 2004
5	M	Omani	c.7934G>A	p.(Glu2645Asp)	46	No	N.A.	Mochida et al. 2004
9	F	Saudi Arabian	c.1219C>T	p.(Gln407)	10	No	N.A.	Mochida et al. 2004
27	M	French	c.7051C>T ; c.11598delA	p.(Arg2351*) ; p.(Glu3867Tyrfs*11)	42 ; 64	No	N.A.	Mochida et al. 2004
24	F	French	c.7051C>T ; c.11598delA	p.(Arg2351*) ; p.(Glu3867Tyrfs*11)	42 ; 64	Yes	Posterior Polar	Mochida et al. 2004
24	F	French	c.7051C>T ; c.11598delA	p.(Arg2351*) ; p.(Glu3867Tyrfs*11)	42 ; 64	Yes	Posterior Polar	Mochida et al. 2004
9	F	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
6	F	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
2	F	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
<10	M	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
<10	M	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
9	M	Irish travellers	c.4471G>T	p.(Glu1491*)	32	No	N.A.	Murphy et al. 2007
9	F	German	c.2516-918 2824+2033dupC ; (8436+1_8437-1) (9258+1_9259-1)del	p.(p.Arg9313_Lys3086del) ; p.(Lys521fs*20) ; p.(Val2813_Lys3086del)	20 ; 46 ; 12 ; 46	Yes	Posterior Subcapsular	Nasser et al. 2019
19	M	Syrian	c.8112C>G	p.(Tyr2704*)	44	No	N.A.	Nasser et al. 2019
20	F	Syrian	c.6055_6056delGA	p.(Asn2019Glnfs*15)	35	No	N.A.	Nasser et al. 2019
14	F	Syrian	c.6055_6056delGA	p.(Asn2019Glnfs*15)	35	No	N.A.	Nasser et al. 2019
7	M	Lebanese	c.11327del	p.(Asn3776Thrfs*102)	54	No	N.A.	Prokudin et al. 2014
4	F	Lebanese	c.11327del	p.(Asn3776Thrfs*102)	54	No	N.A.	Prokudin et al. 2014
3	M	Arabian	c.3870-9884 3024+1867del ; c.5086C>T	p.(Glu2291Hisfs*30) ; p.(Ala1698T)	28 ; 35	No	N.A.	Rivera-Braquels et al. 2011
1	F	German	c.1207-2 2824+5250del ; c.11505delA	p.(Leu403Valfs*11) ; p.(Lys3835fs*43)	10 ; 60	No	N.A.	Rivera-Braquels et al. 2011
3	F	German/African	c.79752 2516-7756del ; c.3866C>G	p.(Thr1289Ser) ; p.(Leu1563Glnfs*15)	1 ; 27	No	N.A.	Rivera-Braquels et al. 2011
5	M	German/English	c.1563G>A ; c.13780delC	p.(Leu1563Glnfs*15) ; p.(Thr3927Lysfs*15)	12 ; 64	No	N.A.	Selferl et al. 2006
60	M	German	c.4923G>A ; c.4955C>G	p.(Tyr1641*) ; p.(Ser1652*)	34 ; 34	No	N.A.	Selferl et al. 2006
44	M	German	c.4923G>A ; c.4955C>G	p.(Tyr1641*) ; p.(Ser1652*)	34 ; 34	No	N.A.	Selferl et al. 2006
56	M	German	c.4923G>A ; c.4955C>G	p.(Tyr1641*) ; p.(Ser1652*)	34 ; 34	No	N.A.	Selferl et al. 2006
54	M	German	c.4923G>A ; c.4955C>G	p.(Tyr1641*) ; p.(Ser1652*)	34 ; 34	Yes	N.D.	Selferl et al. 2006
44	M	German	c.4923G>A ; c.4955C>G	p.(Tyr1641*) ; p.(Ser1652*)	34 ; 34	Yes	N.D.	Selferl et al. 2006
9	F	British	c.1504C>T ; c.6732-1G>A	p.(Arg502*) ; p.(Val2245fs*17)	12 ; 40	No	N.A.	Selferl et al. 2006
5	M	Turkish	c.11245G>T ; c.11564 11565delAT	p.(Glu3749*) ; p.(Tyr3855Valfs*30)	61 ; 63	No	N.A.	Selferl et al. 2006
17	M	Polish	c.65152 5295del ; c.5920C>T	p.(Glu1718_Glu1755del) ; p.(Arg1974*)	36 ; 37	No	N.A.	Selferl et al. 2006
17	M	Polish	c.6461 5462insC ; c.11507dupC	p.(Arg182Profs*18) ; p.(Ser390Glnfs*22)	37 ; 65	No	N.A.	Selferl et al. 2006
15	F	Belgian	c.10456 10457delAG	p.(Leu3487Profs*24)	59	No	N.A.	Selferl et al. 2006
7	F	French	c.2074C>T ; c.5426 5427dupAG	p.(Arg892*) ; p.(Gln1810Serfs*21)	16 ; 37	No	N.A.	Selferl et al. 2006
3	M	French	c.2074C>T ; c.5426 5427dupAG	p.(Arg892*) ; p.(Gln1810Serfs*21)	16 ; 37	No	N.A.	Selferl et al. 2006
16	F	French	c.7153G>T ; c.7322 7323+1delGAGATGGAGC	p.(Glu2885*) ; p.(Ser2641fs*13)	43 ; 44	No	N.A.	Selferl et al. 2006
11	M	German	c.2516 3083del ; c.2934+1 2934+2delGT	p.(Gly839_Thr1027del) ; p.(Glu242Alafs*14)	20 ; 23	No	N.A.	Selferl et al. 2006
28	M	Turkish	c.4878 4880dupATA	p.(Tyr1627*)	34	No	N.A.	Selferl et al. 2006
16	F	Turkish	c.8292C>A	p.(Cys2764*)	48	No	N.A.	Selferl et al. 2006
14	F	Turkish	c.8292C>A	p.(Cys2764*)	48	No	N.A.	Selferl et al. 2006
3	M	Polish	c.2516 4299del ; c.2727 2730dupGCTC	p.(Gly839Aspfs*37) ; p.(Asn911fs*3)	20 ; 21	No	N.A.	Selferl et al. 2006
3	M	Polish	c.2516 4299del ; c.2727 2730dupGCTC	p.(Gly839Aspfs*37) ; p.(Asn911fs*3)	20 ; 21	No	N.A.	Selferl et al. 2006
8	F	Turkish	c.8318C>T	p.(Ser2723Leu)	48	No	N.A.	Selferl et al. 2006
5	F	Turkish	c.8318C>T	p.(Ser2723Leu)	48	No	N.A.	Selferl et al. 2006
2	M	Turkish	c.8318C>T	p.(Ser2723Leu)	48	No	N.A.	Selferl et al. 2006
12	M	Palestinian	c.1225G>T	p.(Glu409*)	10	No	N.A.	Taban et al. 2007
11	M	Syrian	c.6538 6732del	p.(Ile2178Leufs*19)	40	No	N.A.	Taban et al. 2007
4	M	North American Caucasian	c.6733-2A>G	p.(Ile2178Leufs*19)	41	No	N.A.	Taban et al. 2007
26	F	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	Yes	Posterior Subcapsular	Taban et al. 2007
31	?	Amish	c.9258_9259insT	p.(Leu3087Phefs*20)	53	Yes	N.D.	Taban et al. 2007
11	M	French	c.1207-5364 2825-14554del ; c.11306 11307insTCCAGGTGCAATTGGTC	p.(Ile3770Profs*98)	10 ; 62	No	N.A.	This study and Duolomb et al. 2019
9	F	French	c.1207-5364 2825-14554del ; c.11306 11307insTCCAGGTGCAATTGGTC	? ; p.(Ile3770Profs*98)	10 ; 62	No	N.A.	This study and Duolomb et al. 2019
16	F	French	c.2911C>T ; c.3681T>A	p.(Arg971*) ; p.(Tyr1227*)	22 ; 25	No	N.A.	This study and Duolomb et al. 2019
20	M	French	c.3442C>T ; c.10805 10806insTGGCTGGAGCAGCTGTGGAC	p.(Arg134*) ; p.(Tyr3627Ile) ; p.(Tyr3627 His3633del)	23 ; 39	No	N.A.	This study and El Chehadeh et al. 2010
30	M	French	c.2074C>T ; c.5426 5427dupAG	p.(Arg892*) ; p.(Ile1810Serfs*21)	16 ; 37	No	N.A.	This study and El Chehadeh et al. 2010
26	M	French	c.2074C>T ; c.5426 5427dupAG	p.(Arg892*) ; p.(Ile1810Serfs*21)	16 ; 37	No	N.A.	This study and El Chehadeh et al. 2010
31	F	French	c.477 480del ; c.11859 11860insAA	p.(Leu160Asnfs*21) ; p.(Pro3954Asnfs*35)	6 ; 62	No	N.A.	This study and El Chehadeh et al. 2010

21	M	French	c.122046A ; c.7286delT	p.(Val408Leu) <sup>f</sup> *11 ; p.(Val2429Ala) <sup>f</sup> *2	10 ; 43	No	N.A.	This study and El Chehadeh et al. 2010
43	F	French	c.1206+5_1652-27dup ; c.10139_10143dupCGCCA	? ; p.(Glu3382Arg) <sup>f</sup> *15	10 ; 59	Yes	N.D.	This study and El Chehadeh et al. 2010, 2011
46	F	French	c.1206+5_1652-27dup ; c.10139_10143dupCGCCA	? ; p.(Glu3382Arg) <sup>f</sup> *15	10 ; 59	Yes	N.D.	This study and El Chehadeh et al. 2010, 2011
21	M	French	c.1733delT ; c.11620_11623delAGTG	p.(Ile679) <sup>f</sup> ; p.(Ser3876Arg) <sup>f</sup> *40	14 ; 64	No	N.A.	This study and El Chehadeh et al. 2013
11	F	French	c.7643_7644delinsAA ; arr Ra23 (100817911-100828887)x1	p.(Phe2548) <sup>f</sup> ; ?	45 ; 47	No	N.A.	This study and El Chehadeh et al. 2013
27	M	French	c.8515C>T	p.(Arg2839) <sup>f</sup>	49	No	N.A.	This study and El Chehadeh et al. 2013
37	F	French	c.5983+2dup ; c.11115+2T>C	? ; ?	38 ; 61	No	N.A.	This study and El Chehadeh et al. 2013
22	F	French	c.4633+2T>C ; c.9184-1G>T	? ; ?	29 ; 50	No	N.A.	This study and Gueneau et al. 2013
1	F	Irish/Hispanic	Large deletions	?	17 ; 26	No	N.A.	Urhah et al. 2018
5	M	Polish	c.2727_2730dupGGCTC	p.(Asp911Ala) <sup>f</sup> *3	21	No	N.A.	Hennies et al. 2004
3	M	Polish	c.2727_2730dupGGCTC	p.(Asp911Ala) <sup>f</sup> *3	21	No	N.A.	Hennies et al. 2004
13	F	Turkish	c.2911C>T	p.(Arg971) <sup>f</sup>	22	No	N.A.	Hennies et al. 2004
4	M	German	c.3618T>A ; c.7603C>T	p.(Cys1206) <sup>f</sup> ; p.(Arg2535) <sup>f</sup>	26 ; 45	No	N.A.	Hennies et al. 2004
30	M	German	c.4396dupA ; c.11216G>A	p.(Thr1466Asn) <sup>f</sup> *5 ; p.(Trp3739) <sup>f</sup>	32 ; 61	No	N.A.	Hennies et al. 2004
28	M	German	c.4396dupA ; c.11216G>A	p.(Thr1466Asn) <sup>f</sup> *5 ; p.(Trp3739) <sup>f</sup>	32 ; 61	No	N.A.	Hennies et al. 2004
25	M	German	c.5069T>A ; c.7610G>A	p.(Leu1690) <sup>f</sup> ; p.(Trp2537) <sup>f</sup>	35 ; 45	No	N.A.	Hennies et al. 2004
4	F	German	c.7027A>G ; c.13314C>T	p.(Tyr2341Cys) <sup>f</sup> ; p.(Gln3772) <sup>f</sup>	42 ; 62	No	N.A.	Hennies et al. 2004
7	F	Omani	c.7934G>A	p.(Gln2645Asp)	46	No	N.A.	Hennies et al. 2004
5	M	Omani	c.7934G>A	p.(Gln2645Asp)	46	No	N.A.	Hennies et al. 2004
2	M	Omani	c.7934G>A	p.(Gln2645Asp)	46	No	N.A.	Hennies et al. 2004
4	M	German	c.8611delA ; c.9731delA	p.(Thr2871His) <sup>f</sup> *16 ; p.(Thr3244Phe) <sup>f</sup> *2	50 ; 56	No	N.A.	Hennies et al. 2004
17	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thr) <sup>f</sup> *16	55	No	N.A.	Hennies et al. 2004
10	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thr) <sup>f</sup> *16	55	No	N.A.	Hennies et al. 2004
8	M	Lebanese	c.9406-1G>T	p.(Tyr3136Thr) <sup>f</sup> *16	55	No	N.A.	Hennies et al. 2004
6	M	Turkish	c.10888C>T	p.(Gln3630) <sup>f</sup>	59	No	N.A.	Hennies et al. 2004
30	F	?	c.5615dupA ; c.11169_11172dupGGAC	p.(Ser1873Gln) <sup>f</sup> *9 ; p.(Arg3725Gln) <sup>f</sup> *7	37 ; 61	Yes	Posterior Subcapsular	Khan et al. 2005
10	F	Italian	c.7504-1G>A ; c.11125delC	? ; p.(Leu3709Ser) <sup>f</sup> *61	45 ; 61	No	N.A.	Katzaki et al. 2007
17	M	Italian	c.11125delC ; c.11314C>T	p.(Leu3709Ser) <sup>f</sup> *61 ; p.(Gln3772) <sup>f</sup>	61 ; 62	No	N.A.	Katzaki et al. 2007
30	M	Italian	c.7603C>T ; c.10076_10077delCA	p.(Arg2535) <sup>f</sup> ; p.(Thr3359Ser) <sup>f</sup> *29	45 ; 58	No	N.A.	Katzaki et al. 2007
6	M	Italian	c.2047delC ; c.8114C>T	p.(Gln833Ser) <sup>f</sup> *51 ; p.(Arg2707) <sup>f</sup>	16 ; 47	No	N.A.	Katzaki et al. 2007
6	M	Italian	Large deletion ; c.11564delA	? ; p.(Tyr3855Leu) <sup>f</sup> *23	16 ; 60	No	N.A.	Katzaki et al. 2007