

**Table 1: Clinical and imaging data of the 27 included patients carrying the *SSBP1* c.113G>A variant: from asymptomatic carriers to rod cone dystrophy.**

Patient, Gender	Age of onset	VA RE/LE Age	Pallor	Initial RNFL TSNI RE/LE Age	Final RNFL TSNI RE/LE Age	Initial ETDRS Thickness RE-LE Foveal Central 1mm Nasal 3mm Sup 3mm Temp 3mm Inf 3mm Age	Final ETDRS Thickness RE-LE Foveal Central 1mm Nasal 3mm Sup 3mm Temp 3mm Inf 3mm Age	Phenotypes
IV:6, M	12	20/50-20/50 at 12 years  20/400-20/400 at 83 years.	T	No SD-OCT at baseline	T 33/39 S 60/110 N 52/88 I 76/53 At 83 years	No SD-OCT at baseline	F208-209 C241-235 N266-254 S257-257 T274-256 I262-253 At 83 years	DOA AMD drusen
V:13, M	6	20/40-20/40 at 30 years  20/60 -20/50 at 46 years	T	No SD-OCT at baseline	T 32/NA S 33/NA N 48/NA I 69/NA At 46 years	No SD-OCT at baseline	F203-215 C240-267 N260-263 S233-259 T266-273 I279-292 At 46 years	DOA
V:28, F	30	20/25-20/32 at 33 years  20/50-20/40 at 45 years	T	No SD-OCT at baseline	T 49/53 S 106/90 N 58/68 I 100/119 At 45 years	No SD-OCT at baseline	F141-135 C185-179 N257-251 S255-244 T249-246 I254-271 At 45 years	DOA
V:41, F	6	20/200-20/200 at 15 years  20/4000-20/1000 At 67 years	T	No SD-OCT at baseline	T 17/4 S 57/51 N 41/36 I 52/47 At 67 years	No SD-OCT at baseline	F137-128 C191-189 N246-228 S233-229 T224-221 I237-227 At 67 years	DOA

VI:8, F	36	20/20-20/20 at 36 years	T	T 34/37 S 77/84 N 51/50 I 91-83 At 36 years	No follow-up	F185-196 C239-247 N285-289 S286-291 T281-285 I280-283 At 36 years	No follow-up	DOA
VI:19, M	12	20/16-20/16 at 12 years	T	T 34/36 S 123/111 N 79/85 I 130/97 At 12 years	No follow-up	F217-216 C249-247 N280-279 SNR-NR T280-277 UD-UD At 12 years	No follow-up	DOA
VI:20, M	6	20/63-20/63 at 6 years  20/80-20/100 at 10 years	T	No SD-OCT at baseline	T 25/24 S 96/90 N 52/47 I 81/79 At 10 years	No SD-OCT at baseline	F217-212 C245-243 N287-286 S286-286 T280-276 I287-286 At 10 years	DOA
VI:34, F	6	20/125-20/125 at 45 years	T	No SD-OCT at baseline	T 23/24 S 72/68 N 66/64 I 81/68 At 45 years	No SD-OCT at baseline	F153-152 C187-187 N263-260 S259-256 T253-250 I255-253 At 45 years	DOA
VI:35, F	6	20/600-20/600 at 10 years  20/600-20/600 at 28 years	T	No SD-OCT at baseline	T 33/33 S 44/28 N 27/22 I 39/46 At 28 years	No SD-OCT at baseline	F155-146 UD At 28 years	DOA

VII:3, F	10	20/20-20/20 at 10 years  20/20-20/20 at 13 years	T	T 58/49 S 121/121 N 76/72 I 127/130 At 10 years	T 53/40 S 114/122 N 55/61 I 130/132 At 13 years	F 200-200 Cube not done At 10 years	F197-196 C255-253 N320-320 S328-327 T323-321 I324-323 At 13 years	DOA
V:3, F	8	20/50-20/50 at 65 years  20/50-20/50 at 68 years	T	No SD-OCT at baseline	T 25/26 S 67/68 N 52/49 I 70/64 At 68 years	No SD-OCT at baseline	F152-147 C214-209 N273-270 S271-267 T264-261 I267-263 At 68 years	DOA+F
V:11, M	6	20/400-20/400 at 46 years  20/800-20/800 at 50 years  20/800-20/800 at 53 years	T	T 10/16 S 59/59 N 40/40 I 46/48 At 46 years	T 10/21 S 59/59 N 40/40 I 46/48 At 53 years	F 200-201 C 231-235 N 256-261 S 249-250 T 242-244 I 248-251 At 46 years	F203-215 C230-238 N255-259 S246-251 T241-245 I247-251 At 53 years	DOA+F
V:39, F	37	20/160-20/160 at 56 years  20/160-20/160 at 72 years	T	No SD-OCT at baseline	T 12/10 S 52/60 N 41/49 I 58/61 At 72 years	No SD-OCT at baseline	F161-175 C192-198 N247-247 UD-UD T235-231 UD-UD At 72 years	DOA+F

V:47, F	16	20/32-20/32 At 16 years  20/32-20/32 At 60 years	T	No SD-OCT at baseline	T 52/46 S 106/115 N 87/89 I 116/110 At 60 years	No SD-OCT at baseline	F186-181 C235-230 N315-313 S305-308 T303-303 I309-308 At 60 years	DOA+F
VI:4, M	9	20/32-20/32 at 11 years  20/63- 20/40 at 48 years	T	T 27/26 S 71/70 N 43/43 I 91/76 At 45 years	T 26/27 S 72/72 N 42/43 I 90/79 At 48 years	F198-207 C240-248 N287-291 S288-292 T286-286 I283-286 At 45 years	F204-214 C246-251 N290-293 S290-294 T286-287 I284-285 At 48 years	DOA+F
VI:17, M	3	20/40-20/40 at 22 years  20/40-20/50 at 25 years	T	T 26/26 S 65/87 N 50/44 I 110/113 At 22 years	T 28/26 S 64/88 N 49/42 I 113/110 At 25 years	F176-177 C218-223 N285-285 S271-272 T267-269 I276-275 At 22 years	F173-184 C219-225 N287-286 S273-273 T268-272 I277-278 At 25 years	DOA+F
VI:21, M	24	20/25-20/25 At 24 years  20/32-20/32 At 30 years	T	No SD-OCT at baseline	T 36/38 S 90/105 N 68/65 I 86/93 At 30 years	No SD-OCT at baseline	F175-174 C208-205 N261-264 S258-264 T254-256 I260-258 At 30 years	DOA+F
VI:24, M	4	20/32-20/32 at 4 years  20/32-20/32 at 11 years	T	No SD-OCT at baseline	T 30/27 S 113/110 N 62/73 I 107/108 At 11 years	No SD-OCT at baseline	F164-16 C205-203 N257-255 S256-259 T255-255 I256-255 At 11 years	DOA+F

VI:25, F	5	20/25-20/25 At 5 years  20/25-20/25 At 14 years	T	No SD-OCT at baseline	T 40/39 S 97/110 N 74/70 I 113/108 At 14 years	No SD-OCT at baseline	F169-170 C202-199 N269-269 UD-UD T262-259 UD-UD At 14 years	DOA+F
VI:38, M	17	20/32-20/32 at 17 years  20/63-20/50 at 28 years	T	No SD-OCT at baseline	T 24/26 S 86/82 N 62/47 I 77/86 At 28 years	No SD-OCT at baseline	F183-179 C234-232 N293-294 S282-287 T288-293 I282-287 At 28 years	DOA+F
VII:1, M	5	20/50-20/50 at 6 years  20/50-20/50 at 8 years	T	T 40/44 S 107/104 N 76/74 I 102/125 At 6 years	ND at 8 years	ND at 6 years	F179-182 C216-208 N274-265 S269-266 T269-263 I277-269 At 8 years	DOA+F
VII:8, F	6	20/40-20/50 at 6 years  20/125-20/100 at 11 years	T	No SD-OCT at baseline	RNFL unrealizable	No SD-OCT at baseline	F 169-175 UD	DOA+F
IV:17, F	5	20/400-20/400 at 70 years	T	NA OCT-3	Dead	NA OCT-3	dead	DOA+RCD
V:42, M	6	VA < 20/40 at the age of 18.  20/400-20/400 at 69 years	T	No SD-OCT	T 15/UD S 40/UD N 5/UD I 66/UD At 69 years	No SD-OCT	F136-156 C223-197 N285-249 S263-231 T247-225 I256-229 At 69 years	DOA+RCD

V:9, M		20/20-20/20 At 51 years	No	T 69/71 S 131/140 N 54/54 I 122/115 At 51 years	No follow-up	F195-197 C246-246 N303-300 S301-302 T289-286 I296-295 At 51 years	No follow-up	Asymptomatic
VI:14, M		20/20-20/20 At 15 years	No	T 79/73 S 134/142 N 77/89 I 149/140 At 15 years	No follow-up	F254-232 No macular cube	No follow-up	Asymptomatic
VI:15, M		20/20-20/20 At 11 years	No	T 79/74 S 140/141 N 104/96 I 146/151 At 11 years	No follow-up	F237-233 C285-282 N355-355 S349-346 T341-341 I355-356 At 11 years	No follow-up	Asymptomatic

DOA: dominant optic atrophy. DOA+F: dominant optic atrophy and combined foveopathy. DOA+RCD: dominant optic atrophy and combined rod cone dystrophy. LE: Left eye. ND: not done. RE: right eye. RNFL: retinal nerve fiber layer. UD: unreliable data.