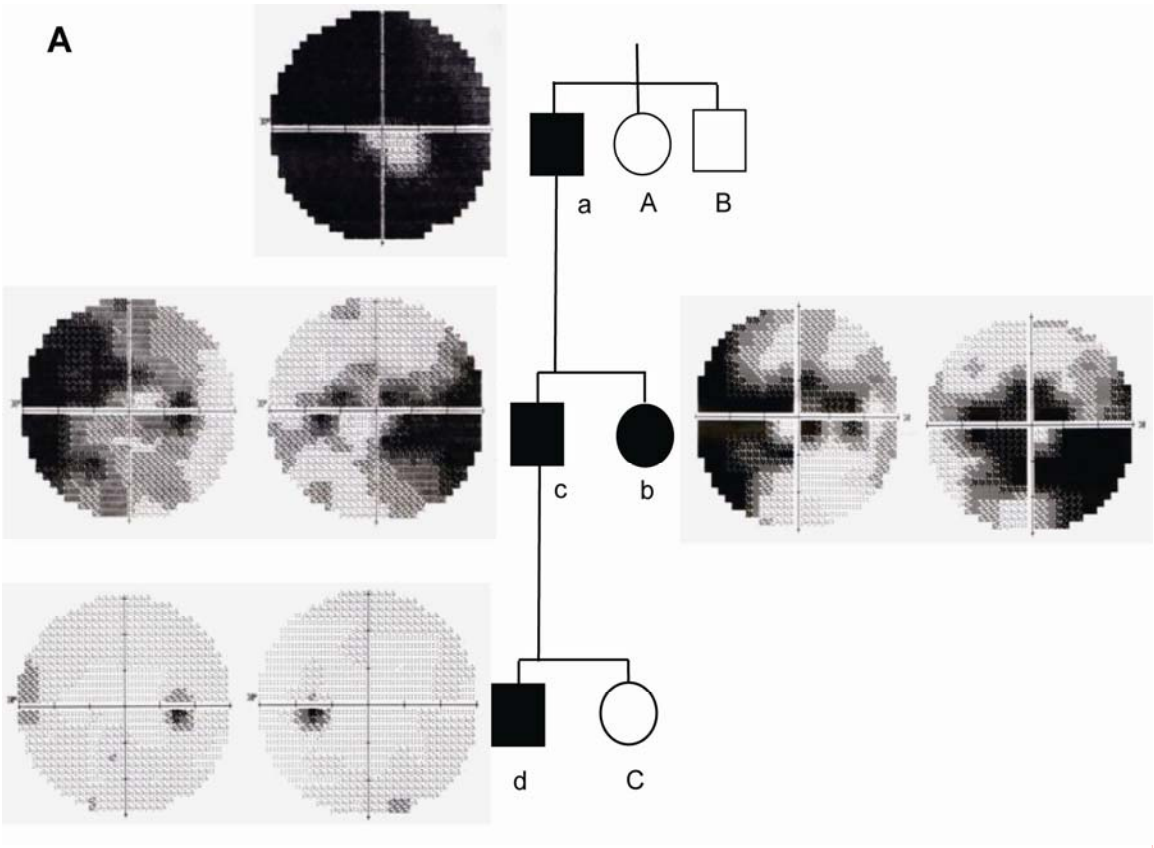
Supplement 1.

Pedigree of three generation NTG family with the *OPTN* E50K mutation. Three-generation Japanese NTG family with the E50K mutation was identified in Gifu Prefecture, Japan by Kawase and Yamamoto. A, Solid symbols (a, b, c, and d) and open symbols (A, B, and C) represent affected and unaffected disease status, respectively. Visual field defect (black) of Humphrey Field Analyzer (HFA, Carl Zeiss Medic, Dublin, CA) are depicted for each individual with carrying the *OPTN* E50K mutation in this pedigree. HFA represent severe (a), moderate (b and c), and mild/subnormal (d) visual field defect. B, Detection of the E50K mutation (+/-), age, IOP, and average visual field loss are listed. Patients (a, b, c) over the age of 40 yrs have severe visual field defects at normal IOP of 12 mmHg for both eyes, including large areas of visual field loss with mean deviations of between -16.43 to -26.50 dB. Patient d, positive for the E50K mutation, at normal IOP of 12 mmHg, experiences relatively minor damage to vision at the age of 17 yrs.

A



B

	OPTN E50K	Ref R/L	Age (yrs)	IOP (R/L mmHg)	Average Visual Field Loss MD (dB)
a	+	-3.0/-2.5	71	12/12	-26.50
b	+	-4.5/-5.5	47	12/12	-19.34
c	+	-5.5/-5.0	41	12/12	-16.43
d	+	-4.5/-5.0	17	16/16	-6.18
A	-	+2.5/+2.0	80	10/11	-3.51
B	-	+2.0/+2.0	72	12/11	+0.58
C	-	-9.5/-9.0	19	14/14	-1.61