

Supplementary table 1. SNPs identified in *OPTN*

Genetic variation	Amino acid change	Minor allele frequency*	dbSNP / previous reports
c.102G>A	—	0.21	Tang <i>et al.</i> (2003)
c.147C>T	—	0.05	Tang <i>et al.</i> (2003)
c.293T>A	p.M98K	0.06	Rezaie <i>et al.</i> (2002)
c.811C>T**	p.R271C	0	novel
c.1634G>A	p.R545Q	0.04	Rezaie <i>et al.</i> (2002)
c.1713C>T	—	0	Alward <i>et al.</i> (2003)
IVS6-10G>A	—	0.14	rs11258210
IVS6-5T>C	—	0.25	rs2244380
IVS7+24G>A	—	0.17	rs11258211
IVS8+20G>A	—	0.01	Leung <i>et al.</i> (2003)
IVS9+19C>T	—	0	rs2277219
IVS11+28G>C	—	0	novel
IVS14+72G>A	—	0.03	novel
IVS15+101A>C	—	0.47	Yen <i>et al.</i> (2008)
IVS15-48C>A	—	0.47	rs10906310

* 94 Japanese controls were examined

** 940 Japanese controls were examined

Supplementary references

1. **Alward WL**, Kwon YH, Kawase K, Craig JE, Hayreh SS, Johnson AT, Khanna CL, Yamamoto T, Mackey DA, Roos BR, Affatigato LM, Sheffield VC, Stone EM. Evaluation of optineurin sequence variations in 1,048 patients with open-angle glaucoma. *Am J Ophthalmol* 2003; **136**: 904-10.
2. **Leung YF**, Fan BJ, Lam DS, Lee WS, Tam PO, Chua JK, Tham CC, Lai JS, Fan DS, Pang CP. Different optineurin mutation pattern in primary open-angle

glaucoma. *Invest Ophthalmol Vis Sci* 2003; **44**: 3880-84.

3. **Rezaie T**, Child A, Hitchings R, Brice G, Miller L, Coca-Prados M, Heon E, Krupin T, Ritch R, Kreutzer D, Crick RP, Sarfarazi M. Adult-onset primary open-angle glaucoma caused by mutations in optineurin. *Science* 2002; **295**: 1077-79.
4. **Tang S**, Toda Y, Kashiwagi K, Mabuchi F, Iijima H, Tsukahara S, Yamagata Z. The association between Japanese primary open-angle glaucoma and normal tension glaucoma patients and the optineurin gene. *Hum Genet* 2003; **113**: 276-79.
5. **Yen YC**, Yang JJ, Chou MC, Li SY. Absence of optineurin (OPTN) gene mutations in Taiwanese patients with juvenile-onset open-angle glaucoma. *Mol Vis* 2008; **14**: 487-94.