Genome-wide association analyses identify three new susceptibility loci for primary angle closure glaucoma

Vithana EN, Khor CC, Qiao C, Nongpiur ME, George R, Chen LJ, Do T, Abu-Amero K, Huang CK, Low S, Tajudin LS, Perera SA, Cheng CY, Xu L, Jia H, Ho CL, Sim KS, Wu RY, Tham CC, Chew PT, Su DH, Oen FT, Sarangapani S, Soumittra N, Osman EA, Wong HT, Tang G, Fan S, Meng H, Huong DT, Wang H, Feng B, Baskaran M, Shantha B, Ramprasad VL, Kumaramanickavel G, Iyengar SK, How AC, Lee KY, Sivakumaran TA, Yong VH, Ting SM, Li Y, Wang YX, Tay WT, Sim X, Lavanya R, Cornes BK, Zheng YF, Wong TT, Loon SC, Yong VK, Waseem N, Yaakub A, Chia KS, Allingham RR, Hauser MA, Lam DS, Hibberd ML, Bhattacharya SS, Zhang M, Teo YY, Tan DT, Jonas JB, Tai ES, Saw SM, Hon do N, Al-Obeidan SA, Liu J, Chau TN, Simmons CP, Bei JX, Zeng YX, Foster PJ, Vijaya L, Wong TY, Pang CP, Wang N, Aung T.

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Primary angle closure glaucoma (PACG) is a major cause of blindness worldwide. We conducted a genome-wide association study including 1,854 PACG cases and 9,608 controls across 5 sample collections in Asia.

Replication experiments were conducted in 1,917 PACG cases and 8,943 controls collected from a further 6 sample collections. We report significant associations at three new loci: rs11024102 in PLEKHA7 (per-allele odds ratio (OR) = 1.22; P = 5.33 × 10(-12)) , rs3753841 in COL11A1 (per-allele OR = 1.20; P = 9.22 × 10(-10)) and rs1015213 located between PCMTD1 and ST18 on chromosome 8q (per-allele OR = 1.50; P = 3.29 × 10(-9)) .

Our findings, accumulated across these independent worldwide collections, suggest possible mechanisms explaining the pathogenesis of PACG.

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