AB015. The relationship between copy number variations and high myopia in Chinese: a case-control study

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Abstract: As a complex disease, myopia is the most common eye disease worldwide. Many myopia susceptibility genes or variants have been successfully identified in the past years by genome-wide genetic association studies (GWAS), which focus mainly on the single-nucleotide polymorphisms. Little attention has been paid to examine the role of copy number variations (CNVs) in refractive error and myopia. This study adopted a systematic strategy to investigate the role of CNVs in high myopia. In the discovery phase, a pilot GWAS suggests putative CNVs for follow-up. Multiplex ligation-dependent probe amplification was then used to quantify the copy number of 89 CNV segments in 737 case-control samples in the second phase and then 24 top-ranking CNVs in a second group of 1,029 case-control samples in the final validation phase. This validation phase identified 22 significant CNVs. Further work is needed to examine the role of these few CNVs in myopia development.

Keywords: Myopia; copy number variations (CNVs); genetic susceptibility; case-control study; Chinese

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