Ching Yun Wang

Taichung Veterans General Hospital, Taichung
Taiwan

Instrumenting carotid sonography biomarkers and polygenic risk score as a novel screening approach for retinal detachment

Abstract:

Background: Retinal detachment (RD) is a blind-threatening disease without effective screening protocols. Thus, current medical practice deployed RD surveillance only after the first attack.

Aim: To foster a risk stratification framework attributing RD risk before disease onset, we instrumented the hemodynamic biomarkers of the carotid ultrasonography (CUS) and the RD polygenic risk score (PRSRD) from single nucleotide polymorphism (SNP) profiling.

Methods: For 21,441 Taiwan biobank participants, a backpropagation logistic regression model was built and visualized by nomogram to identify RD-associated CUS biomarkers. A PRSRD model was built by charting the expression of SNP functional genes from retina scRNA datasets. Last, a two-component RD prediction model (CUS and PRSRD) was assembled by logistic cumulative analysis.

Results: Hypertension (HTN) status was significantly associated with RD risk (OR=1.32). The CUS model (AUCHTN+=0.632, AUCHTN-=0.630) showed RD risk increased with the minimum flow of the right internal carotid artery and the timed average max velocity of the right common carotid artery (ICA-Qmin OR=1.04 and CCA-TAMAX OR=1.03 with p-value<0.05). The genome-wide association study (GWAS) identified three outstanding RD SNPs (IGFBPL1 rs117248428 OR=1.63, CELF2 rs56168975 OR=1.72, and PAX6 rs11825821 OR=1.61 with p-value<5.00x10-6) with coded genes highly-expressed in retinal pigment epithelium (RPE) and choroid. Notably, the two-component model achieved state-of-the-art prediction (AUCHTN+=0.95, AUCHTN-=0.93).

Conclusions: Through instrumenting CUS images and genetic PRSRD, we proposed a screening method for RD at-risk patients.