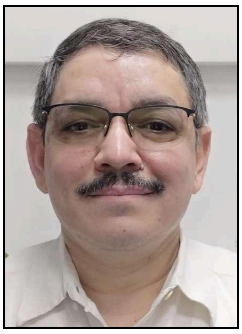
In vivo identification of angle dysgenesis and its relation to genetic markers associated with glaucoma using artificial intelligence



Dr. Dinesh Gupta*

Translational Bioinformatics Group, International Centre for Genetic Engineering and Biotechnology, New Delhi, India

Email: dinesh@icgeb.res.in

Purpose: To predict the presence of angle dysgenesis on anterior-segment optical coherence tomography (ADoA) by using deep learning (DL) and to correlate ADoA with mutations in known glaucoma genes.

Participants: In total, 800 high-definition anterior-segment optical coherence tomography (AS-OCT) images were included, of which 340 images were used to build the machine learning (ML) model. Images used to build the ML model included 170 scans of primary congenital glaucoma (16 patients), juvenile-onset open-angle glaucoma (62 patients), and adult-onset primary open-angle glaucoma eyes (37 patients); the rest were controls (n = 85). The genetic validation dataset consisted of another 393 images of patients with known mutations that were compared with 320 images of healthy controls.

Methods: ADoA was defined as the absence of Schlemm's canal, the presence of hyperreflectivity over the region of the trabecular meshwork, or a hyperreflective membrane. DL was used to classify a given AS-OCT image as either having angle dysgenesis or not. ADoA was then specifically looked for on AS-OCT images of patients with mutations in the known genes for glaucoma.

Results: The final prediction, which was a consensus-based outcome from the three optimized DL models, had an accuracy of >95%, a specificity of >97%, and a sensitivity of >96% in detecting ADoA in the internal test dataset. Among the patients with known gene mutations, (MYOC, CYP1B1, FOXC1, and LTBP2) ADoA was observed among all the patients in the majority of the images, compared to only 5% of the healthy controls.

Conclusion: ADoA can be objectively identified using models built with DL.