Editorial

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http://journals.lww.com/TJOP DOI: 10.4103/tjo.TJO-D-23-00167 Advancing therapies for anterior segment developmental anomalies

"In the pursuit of understanding and treating these complex disorders of the anterior segment, we envision a world where everyone can enjoy the beauty of clear vision."

The development of the anterior segment of the eye is a highly intricate process that relies on sequential interactions between developing tissues and a multitude of genetic factors. Any disruptions in this process result in a spectrum of congenital anomalies, giving rise to anterior segment developmental anomalies (ASDA), including aniridia, Axenfeld-Rieger anomaly, congenital corneal opacities (CCO) (namely, primary and secondary CCO), and primary congenital glaucoma. Significant progress has been made in identifying the genetic factors responsible for ASDA, with a diagnostic success rate of about 50%. The success rate of specific phenotypes such as classical aniridia (PAX6) or classic Peters Plus syndrome (B3GALTL mutation) exceeds 90%. ASDA can be caused by pathogenic variants in genes with diverse functions, such as transcription factors, enzymes, structural proteins, extracellular matrix or cell adhesion proteins, signaling and membrane transport molecules, and genes with unknown functions. The involvement of numerous genes in overlapping phenotypes suggests their participation in a network of interconnected biological pathways crucial for anterior segment development.

In this theme-based issue, we will discuss the typical clinical presentations of various

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ASDA forms, the latest genetic data related to known factors and promising new candidate genes that might be involved in these phenotypes, and current and emerging treatments for these complex conditions. Further exploration of the affected genes and pathways in animal and cell culture models will facilitate the discovery of novel factors and empower the development of innovative therapeutic approaches by focusing on the most critical processes. Among these pioneering approaches, we highlight less invasive surgical interventions, such as optical iridectomy, rotational autokeratoplasty, selective endothelialectomy in Peters anomaly or selective endothelial removal, gene editing, and bioengineered corneas.

The path to conquering ASDA involves collaboration among clinicians, geneticists, researchers, and the ophthalmological community. This issue serves as a platform for disseminating knowledge and facilitating idea exchange, contributing to our shared vision of a world where clear vision is accessible to all, irrespective of genetic predisposition or developmental challenges. While challenges remain, the promise of a brighter future for individuals affected by developmental corneal abnormalities is evident. Ophthalmologists and researchers must collectively push the boundaries of knowledge and technology to deliver the precious gift of sight to those who entrust us with their vision.

Indeed, it is our great honor and privilege to serve as guest editors for the December

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Data availability statement

Data sharing not applicable to this article as no datasets were generated or analyzed during the current study.

Author contribution

Equally contributed: concept, design, & final revision as well.

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