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Relevance of Molecular Diagnosis of Corneal Dystrophies

Preeti Paliwal and Arundhati Sharma

Laboratory of Cyto-Molecular Genetics, Department of Anatomy, All India Institute of Medical Sciences, New Delhi, India

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ABSTRACT Corneal dystrophies are defined as a group of inherited corneal disorders characterized by opacification of the cornea. Initial classification of corneal dystrophies was based on the layer of cornea involved but with the advent of better technology, a larger picture has evolved that includes both phenotypic and genotypic variants. With the evolving knowledge, a revised classification has been proposed by the International Committee for Classification of Corneal Dystrophies (IC3D). This classification has taken into account the clinical, histologic and genetic basis of the disease, integrating them into one. Our understanding of corneal dystrophies has reached new heights with mutations identified in at least 14 genes. Even though there has been a vast addition of information to the database, we still come across new variants which may seem enigmatic phenotypically. With the additional new information refining the molecular basis, there is a need to explore further the underlying molecular pathology so as to enable the possibility of better treatment modalities to the affected patients and their families. In view of the recent advances, we hereby review the dystrophies with an aim to provide updated information on the clinical and molecular aspects of corneal dystrophies which will aid in their differential diagnosis and management.