Current Ocular Therapy for Cystinosis

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Cystinosis is a rare genetic disorder characterized by the accumulation of cystine crystals in various tissues throughout the body. Among its ocular manifestations, corneal involvement is particularly notable, progressing from the periphery to affect all layers of the cornea. This progression can lead to epithelial erosions and ocular surface disease. In vivo confocal microscopy (IVCM) offers a valuable tool for visualizing these changes, providing detailed images of the cornea and conjunctiva in a layer-by-layer fashion. Furthermore, IVCM aids in distinguishing cystinosis-related corneal crystal accumulation from other conditions such as infectious crystalline keratopathy and corneal dystrophies. Topical cysteamine treatment has shown promise in managing corneal cystinosis, preventing irreversible damage in the absence of proper intervention.

Beyond the cornea, cystinosis affects the retina, where cystine crystals irreversibly accumulate, leading to pigmentary retinopathy and photoreceptor degeneration. This retinal involvement manifests as decreased visual acuity, color vision, night vision, and visual field deficits. Current pharmacologic therapies aim to mitigate these retinal complications, although further clinical investigational initiatives are underway to explore additional treatment modalities.

Understanding the ocular manifestations of cystinosis, particularly its effects on the cornea and retina, is essential for timely diagnosis and management. Advances in imaging techniques like IVCM and Keratography provide insights into the pathogenesis of cystinosis-related ocular changes and aid in distinguishing them from other similar conditions. With ongoing research and therapeutic developments, there is hope for improved outcomes and quality of life for individuals living with cystinosis.