Decoding Syndromic Cone–Rod Dystrophies: The Complexities of Vision and Systemic Involvement

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Abstract:

Syndromic cone–rod dystrophies (CRDs) constitute a diverse spectrum of inherited retinal diseases marked by progressive loss of photoreceptor function. Because both cones and rods are affected, patients typically develop central vision decline, impaired color discrimination, and heightened photophobia. Beyond these ocular features, many individuals present with broader systemic involvement, ranging across skeletal, metabolic, neurological, renal, and cardiovascular abnormalities, adding further complexity to clinical recognition and treatment. The molecular basis is heterogeneous, with pathogenic variants in multiple genes contributing to syndromic presentations associated with disorders such as Bardet–Biedl, Usher, Alström, Jalili, Refsum disease, Senior–Løken, Cohen syndrome, Jeune, Sensenbrenner, and Joubert syndromes. Complementary investigations, particularly electroretinography, are essential for distinguishing syndromic CRDs from isolated cone dystrophies by documenting dysfunction in both cone- and rod-mediated pathways. This review seeks to outline the genetic diversity and defining clinical hallmarks of these conditions.

Keywords:

Syndromic Cone-Rod Dystrophies, Retinal Degeneration, Genetic Mutations, Photoreceptor Degeneration, Systemic Manifestations.