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Peters anomaly and the clinical implications of rare genetic ocular diseases on the patient

Robert W Sault
Kelowna General Hospital, British Columbia

Peters anomaly is a very rare genetic autosomal recessive ocular malformation that can be associated with systemic abnormalities resulting in a significant reduction in the quality of life for patients. Rare genetic ocular diseases are repeatedly missed upon examination and subsequently underdiagnosed resulting in often preventable complications such as glaucoma, cardiac and functional impairments in patients. Behind the goal of academic research should be the question: "Why are we doing this?" With the intent of improving the base standard of clinical medicine through improved awareness, diagnostics, examination skills, and understanding of patho-physiological processes of rare ocular disease; our academic research in ocular medicine is about improving quality of life. We analyze the impact of rare genetic ocular diseases by examining specific genes; and coordinating emerging treatments in biomedicine to match the expectations of the patient, and the constantly evolving standard of clinical eye care.

rwsault@shaw.ca