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Schnyder corneal dystrophy- A case study in translational research

Schnyder Corneal Dystrophy (SCD) has been an exceptional model for questions and answers that result from the bedside to bench approach of translational research. A medline search indicates the term translational research appeared as early as 1993, a few years after I examined the originalitial four large pedigrees of affected patients with Schnyder dystrophy. Initial investigation led to my simple clinical observation that only 50% of affected patients with Schnyder crystalline corneal dystrophy had corneal crystals. Patients without crystals were typically misdiagnosed. The nomenclature challenges later led to creation of the International Committee for Classification of Corneal Dystrophies and revision of the dystrophy nomenclature in 2008 to be based on genetic, histopathologic and clinical findings rather than just phenotype. Histopathologic changes of affected SCD corneas demonstrated abnormal collection of intra and extracellular cholesterol reminiscent of atherosclerosis. The assumption that that understanding pathogenesis of SCD might increase our understanding of lipid metabolism, led to the search for the affected chromosome, chromosome 1 and then the isolation of the abnormal gene, UBIAD1. Although arcus, a peripheral corneal ring composed of LDL is the most common type of cholesterol deposition in humans, the central corneal deposit of SCD was found to be HDL. Specific dog species often have deposition of cholesterol in the central cornea, while peripheral arcus occurs less commonly. Interestingly, the predominant blood cholesterol component in humans is LDL while in dogs is HDL. Can comparisons between human and dog cholesterol HDL and LDL distribution provide further understanding about SCD and lipid metabolism in the cornea and systemically?

Biography

Jayne S. Weiss is Chair of the LSU Department of Ophthalmology, Herbert E Kaufman MD endowed Chair, Professor of Ophthalmology, Pharmacology and Pathology and Director of LSU Eye Center in New Orleans, Louisiana. She began her research on Schnyder corneal dystrophy 25 years ago recruiting patients internationally leading to the gene discovery. The misleading disease nomenclature led her to establish the International Committee for Classification of Corneal Dystrophies whose new nomenclature system including genetic information is now used for the dystrophies. She is a member of the National Advisory Eye Council, a consultant to the FDA Ophthalmic Devices Panel and Assistant Editor of the journal, Cornea.

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