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Schnyder corneal dystrophy- A window to insights on cholesterol, vitamin K and Parkinson's disease?

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Schnyder crystalline corneal dystrophy (SCD), first described in 1924, is an autosomal dominant inherited disease with corneal clouding resulting from abnormal cholesterol deposition in the front of the eye, the cornea. In the late 1980's, I discovered four large pedigrees of Swede Finn individuals with SCD. At that time, there were less than 100 articles on the disease in the peer reviewed literature. In 1992, I published the clinical findings in 18 affected members of these families I had examined which was the largest number of affected patients ever described in one publication at that time. One new finding was that although the disease name was Schnyder crystalline corneal dystrophy, only 50% of affected individuals actually had corneal crystals. Both affected and unaffected members of SCD pedigrees may have elevated blood cholesterol making it difficult to determine if high circulating cholesterol is associated with SCD. The eye disease progresses with age, independent of blood cholesterol value. From the beginning, even though a clinician, I continued recruitment of affected individuals in my hope that understanding this rare eye disease, could lead to better understanding of systemic cholesterol and lipid metabolism. One early collaborator, Howard Kruth MD, shared my vision. Later on, Mike Nickerson Ph.D. also agreed with the importance of finding the gene for this rare disease. I performed corneal transplantation surgery on patients visually disabled from the corneal clouding of SCD and sent the tissue to Dr. Kruth's laboratory for analysis which revealed that the abnormal lipid deposition was entirely HDL, not LDL suggesting a local abnormality of HDL metabolism. At the same time we found that the disease mapped to chromosome 1, but it took another decade before we and another group independently identified mutations in SCD patients in the UBIAD1 gene on chromosome 1p36. Recruitment efforts continued internationally. By 2007, I had followed 115 individuals who had SCD for up to 18 years. Although the disease had been thought to be visually benign, 54% of patients 50 and younger and 77% of patients 70 and younger had reported corneal transplant surgery. Many patients without corneal crystals eluded correct diagnosis because of the confusing nomenclature. For this reason, I founded the International Committee for Classification of Corneal Dystrophies (IC3D) which revised the entire corneal dystrophy nomenclature and also renamed this disease Schnyder corneal dystrophy. Most recently UBIAD1 has been found by others to be a human vitamin K2 (MK-4) biosynthetic enzyme and has been found to have links to bladder cancer and Parkinson's disease.

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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