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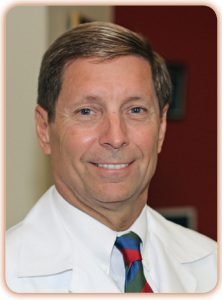

Kent W Small

Founder of Macula & Retina Institute, USA

North carolina macular dystrophy: Mutations found and new clinical findings

Purpose: We originally reported four mutations affecting PRDM13 in 11 families causing North Carolina Macular Dystrophy (NCMD/MCDR1). The purpose of this report is to present the analysis of an international cohort of an additional 10 families with NCMD.

Methods: We performed Sanger DNA sequencing of the DNASE 1 hypersensitivity binding site up stream of PRDM13 (chr6:100040800-100040950) in the family members with NCMD.

Results: Of the 10 new families studied with NCMD, six were found to have the same mutation as the original North Carolina family (Chr6: 100040906 G>T Het) and all families were in the USA. Four were found to have the "French mutation" (Chr6:100040987 G>C Het), three were European and one was American.

Conclusion: Additional families with the NCMD phenotype continue to support that these mutations are causative of MCDR1/NCMD.

Biography

Kent W Small is a board-certified Ophthalmologist with years of specialized training and over two decades of experience in retinal disorders, making him a leading expert in his field. He is a Professor and the Director of Macular Disease Center and Retinal Research Lab at the Jules Stein Eye Institute at UCLA.

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