Medical Retina

ABCA4-RELATED RETINITIS PIGMENTOSA FICTION OR REALITY

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Since the genetic locus of Stargardt disease was mapped and ABCA4 (formerly ABCR) gene was cloned in the 1990s, a clinical spectrum of ABCA4-related disease has been described. Currently Online Mendelian Inheritance in Man (OMIM) describes 4 phenotypes associated with ABCA4 mutations: Stargardt disease/fundus flavimaculatus/early-onset severe retinal dystrophy (STGD1; MIM#248200), cone-rod dystrophy (MIM#604116), retinitis pigmentosa 19 (MIM#601718) and disease susceptibility loci in age-related macular degeneration (AMD; MIM#153800). Literature review and author`s clinical experience indicate that ABCA4 disease is associated with macular only, cone or cone-rod disease (also refered to as inverse retinitis pigmentosa) and the association with rod-cone disease (retinitis pigmentosa) is a misconception perpetuated in current biomedical literature.