Unique Phenotypic Expression of Pachydrusen in Two Bangladeshi-Americans with Pachychoroid and Central Serous Chorioretinopathy

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Purpose:
We present two Bangladeshi-Americans who had a phenotypic expression of pachydrusen that varied from previously reported cases of pachydrusen.

Methods:
Case Series

Results:
A 28 year old Bangladeshi-American woman who was monocular from a previous traumatic injury presented with longstanding decreased vision in her left eye. Vision in that eye was 20/80. Her anterior segment was normal. Funduscopic exam showed a shallow neurosensory macular detachment with multiple pachydrusen encircling the detachment. Fundus autofluorescence (FAF) showed these drusen to be autofluorescent. Spectral domain OCT (SD-OCT) showed a thicker choroid with pachyvessels and a neurosensory detachment without a "double-layer" sign. Fluorescein angiography showed a central window defect corresponding to the long standing macular detachment. Some pachydrusen exhibited staining while others showed blocked fluorescence. Neither leakage or evidence of dependent window defects characteristic of chronic Central Serous Chorioretinopathy (CSCR) were noted. The second patient, a 32 year-old Bangladeshi-American man, presented with long standing decreased vision in both eyes. His vision was 20/30 in the right eye and 20/50 in the left eye. Anterior segment was normal. The funduscopic exam showed bilateral neurosensory macular detachment with the similar pachydrusen encircling the macular detachments as well as symmetrical pachydrusen nasal to the optic nerve in both eyes. The FAF showed drusen to be autofluorescent. SD-OCT showed a thick choroid with pachyvessels and a neurosensory detachment without a "double-layer" sign. Fluorescein angiography findings were similar to the first patient with the exception that all of the pachydrusen in this patient were hypofluorescent.

Conclusions:
Pachychoroid disease is thought to be a continuum starting with pachychoroid pigment epitheliopathy that may lead to central serous chorioretinopathy (CSCR) which may then progress to pachychoroid neovasculopathy and/or polypoidal choroidal vasculopathy. Our patients may represent a unique phenotypic expression of CSCR. Typically pachydrusen occurring in CSCR are in patients that are much older than our patients (6th-7th decade). Also pachydrusen are not typically as numerous and don't have the strikingly similar configuration as seen in our patients. Given the similar ethnicity we hypothesize some common unknown genetic or environmental factor may be responsible for the phenotypic expression.