QUESTION 3

What Are the Age-Related Macular Degeneration Look-Alikes? Do I Treat Them Any Differently?

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The majority of clinical practice adheres to the classic medical aphorism, "When you hear hoof beats, think of horses not zebras." However, not every patient presenting with macular drusen, pigmentary changes, or areas of retinal pigment epithelium (RPE) atrophy carries the straightforward diagnosis of non-neovascular (or dry) age-related macular degeneration (AMD). Detecting zebras in the herd affords the clinician an opportunity to identify and treat systemic disease, neoplasia, or a genetic mutation with multigenerational implications.

Suspicion for an AMD look-alike should be spurred when typical signs of AMD are detected at an earlier age than expected or in association with specific clinical patterns. Variations in the typical signs (ie, drusen, pigmentary change, RPE atrophy) that should tip you off to an atypical case are listed in Table 3-1. When clinical suspicion for atypical AMD is high, increased attention should be paid to the clinical history, including age of presentation, initial fundus changes, the degree of visual acuity loss, and especially to subtle or pronounced nyctalopia. The review of systems should probe for problems with hearing, diabetes, renal disease, or skin problems. A history of current or prior steroid use should be elicited. The family history is also informative and will often identify relatives carrying a diagnosis of early onset AMD, unexplained visual loss and blindness, or occasionally nyctalopia. In some cases, examination of accompanying family members can be revealing. It is unusual for the diagnosis of an AMD mimic to be made solely with dilated fundus examination. Common ancillary tests including fundus photography, fundus autofluorescence (FAF), and optical coherence tomography (OCT) are complementary modalities used in developing the initial differential diagnosis. Ultimately, electroretinography (ERG) and/or electrooculograms (EOG) may be required in association with genetic testing to clarify the diagnosis.

Fekrat S, ed. *Curbside Consultation in Retina: 49 Clinical Questions, Second Edition.* (pp 9-17).