Usher syndrome

Category(ies): Inherited Eye Disease, Retina

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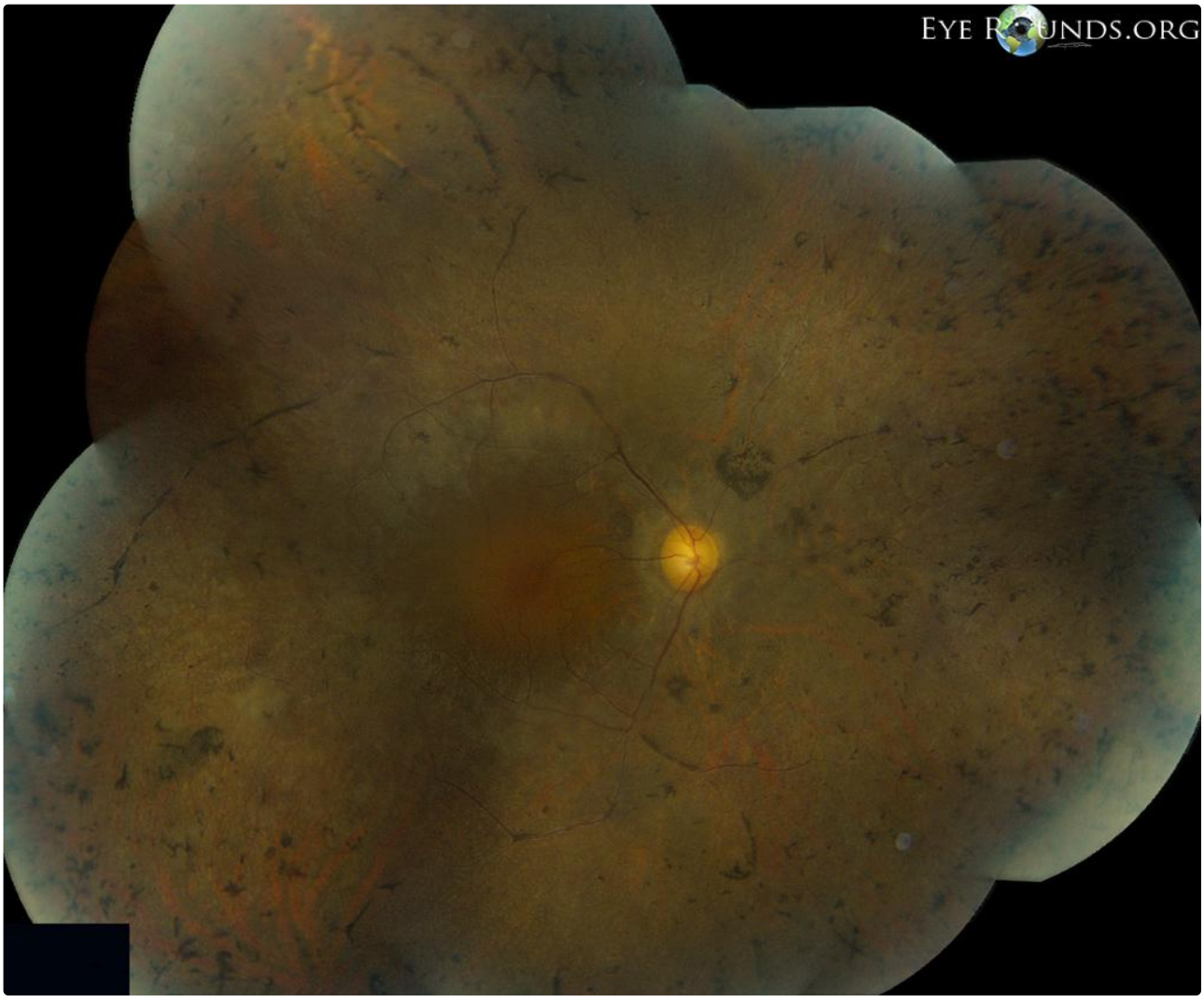
Usher syndrome is an autosomal recessive condition that results in retinitis pigmentosa with associated congenital hearing loss. There are three main types of Usher syndrome. Type 1 is associated with profound congenital sensorineural hearing loss and poor vestibular function, most commonly caused by mutation in the MYO7A gene. Type 2 results in mild congenital hearing loss with normal vestibular function and is most commonly caused by a mutation in the USH2A gene. Type 3 is rare and can have slowly progressive hearing loss. It is most commonly caused by a mutation in the USH3A gene. Similar to other types of retinitis pigmentosa, fundus features include peripheral retinal atrophy with bone-spicule-like pigmentation, vascular attenuation, and waxy-pallor of the optic disc.



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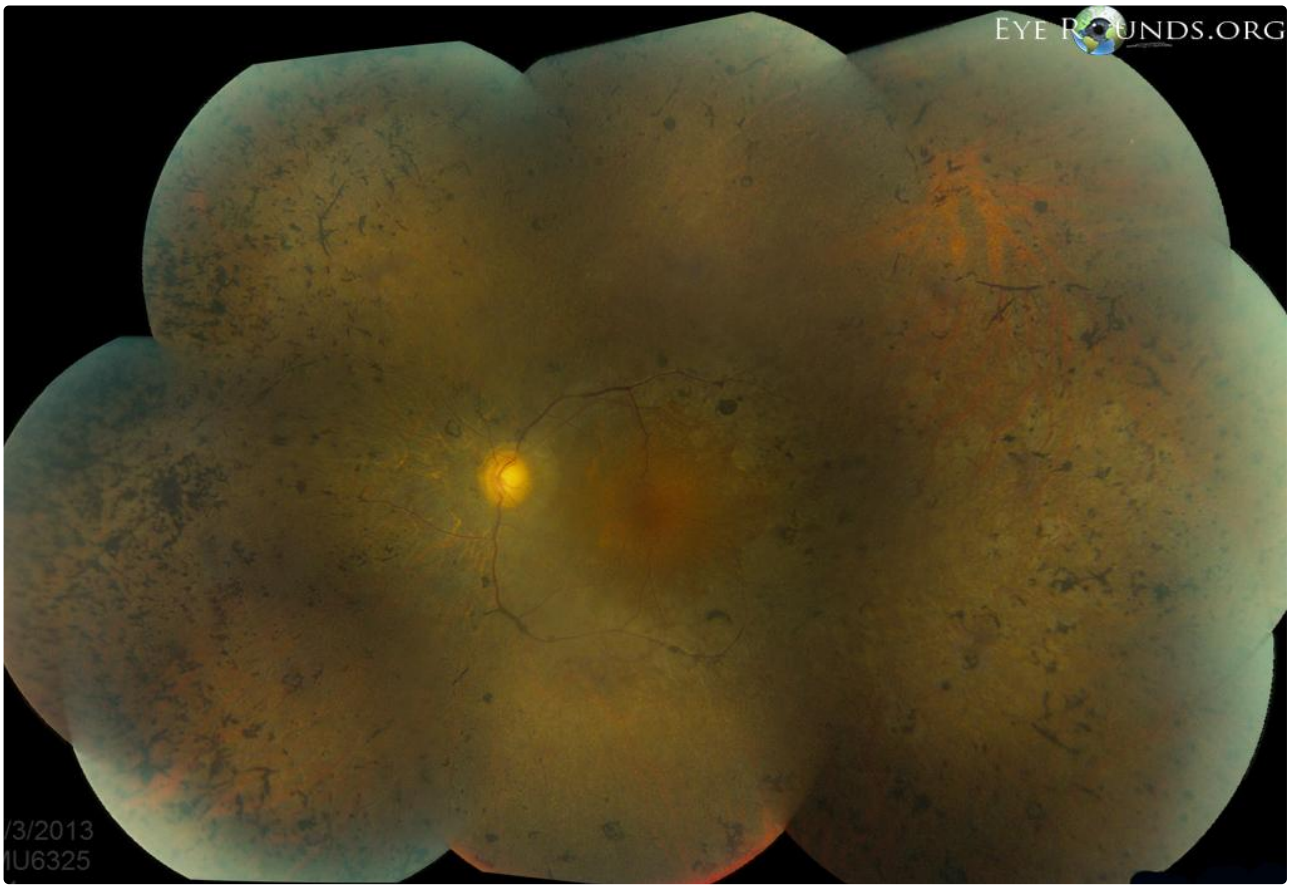

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