Choroideremia is a generalized choroidal dystrophy with X-linked inheritance and involvement of the mid – peripheral receptors, mainly rods, similar to Retinitis Pigmentosa. These patients present with complaints of progressive night blindness and loss of peripheral vision. Being an X-linked disease, the carrier state is seen in females, and has a fundus appearance ranging between normal to very rarely a very advanced form of disease as seen in a male Choroideremia patient.

We report an eighteen year old girl who had presented to our General Ophthalmology clinic with asthenopic symptoms and referred to the retina department for opinion regarding abnormal looking fundus. Her vision unaided was 20/20 with normal near vision in both eyes. Fundus examination showed diffuse pigmentary mottling (Fig 1&2) with normal discs and vessels.

The differential diagnoses under consideration were congenital syphilis, congenital rubella or a variant of Retinitis Pigmentosa. But the vessels and discs were normal and there was no history of night blindness. Also there were no other clinical signs to support the diagnosis of congenital rubella or congenital syphilis. On further enquiry, she gave the history of her father having decreased vision since few years and was hence asked to report the next day with her father.

On examination of the father, fundus showed diffuse chorio-retinal atrophy with an island of intact chorio-retinal tissue (Fig3&4) in the foveal area. He was suffering from night blindness and also gave similar history in his maternal uncle. The diagnosis in father was chorioderemia and the daughter was diagnosed to have the carrier state of the disease.

Choroideremia is easy to diagnose in the full - blown stage in a male patient, when the clinical picture is characterized by history of night blindness and the retinal examination findings of generalized chorio- retinal atrophy sparing the macular area with normal discs and vessels. But it can cause diagnostic difficulty in a carrier, if the relatives are not available for examination and in the absence of the history of similar eye disease in the male members of the family.

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A carrier of chorioderemia is often asymptomatic and can have fundus with normal appearance or with diffuse pigmentary mottling.[1] The vessels are of normal caliber and discs do not show any pallor. Electrophysiological studies are normal in asymptomatic carriers and is not useful in diagnosing the carrier state.[2] Focal area of choroidal atrophy may appear later in life and can progress. Rarely, this can lead to generalized chorioretinal atrophy causing visual dysfunction and fundus appearance similar to the male counterpart were the affected female could be the offspring of an affected father and a carrier mother.[3,4]

In conclusion, Choroideremia carrier state is a diagnosis which is to be made with the help of examination of family members and proper elicitation of family history.

References:


