ADITUM

Journal of Ophthalmology and Vision Care

Open Access Short Communication

Choroideremia Versus Retinitis Pigmentosa

Ali Nouraeinejad

Clinical Ophthalmology, Department of Clinical Ophthalmology, Institute of Ophthalmology, University College London (UCL), London, United Kingdom.

Article Info

Received: January 13, 2022
Accepted: February 26, 2022
Published: March 03, 2022

***Corresponding author:** Ali Nouraeinejad, Clinical Ophthalmology, Department of Clinical Ophthalmology, Institute of Ophthalmology, University College London (UCL), London, United Kingdom.

Citation Ali Nouraeinejad. (2022) "Choroideremia Versus Retinitis Pigmentosa", Ophthalmology and Vision Care, 2(1); DOI: http;//doi.org/02.2022/1.1021.

Retinal dystrophy is outlined as a heterogeneous group of hereditary disorders in which the loss of photoreceptor function results in progressive visual impairment [1]. The most common example is retinitis pigmentosa [1]. One of the retinal dystrophies that may be confused with retinitis pigmentosa is choroideremia [1]. Retinitis pigmentosa has largely attained the notice but little has been heard about choroideremia [1].

Choroideremia, an X-linked recessive inherited chorioretinal dystrophy/degeneration, is manifested as a progressive degenerative disorder of the photoreceptor layer, retinal pigment epithelium (RPE), and choroid [1-4].

Symptoms of patients with choroideremia often consist of gradual decrease in central visual acuity, nyctalopia, and constriction of the peripheral visual field (tunnel vision) [1]. Nyctalopia is usually the first symptom often with onset during childhood [1].

Given that choroideremia has an X-linked recessive pattern, male patients predominantly show the typical features of early nyctalopia for the period of the first decade of childhood that proceed into severe peripheral vision loss followed by legal blindness in late adulthood approximately the fifth to sixth decades [2-4]. On the other hand, female carriers remain asymptomatic or may experience nyctalopia and show evidence of pigmentary changes and chorioretinal degeneration in the fundus with subnormal visual sensitivity [2-4].

Since choroideremia and retinitis pigmentosa share diverse common clinical manifestations (e.g., the same family pedigree, nyctalopia, constriction of the visual field, gradually reduced visual acuity, and retinal degeneration), it makes difficulties in their diagnosis [1]. In this respect, about 6% of patients primarily diagnosed with retinitis pigmentosa were actually demonstrated to have choroideremia [5].

The author proposes for clinicians to reflect on the main classic triad of findings in retinitis pigmentosa that includes a bone spicule pigment migration pattern in the peripheral retina, optic disc pallor, and retinal vessel narrowing as well as another probable sign of epiretinal membrane formation [1,2,5]. As a clinical point, it is worth realizing that the scale of pigment migration into the retina that represents retinitis pigmentosa is not observed in patients with choroideremia [1]. In this respect, bilateral mid-peripheral intraretinal perivascular 'bone spicule' pigmentary changes and RPE atrophy related with arteriolar narrowing are noticed in retinitis pigmentosa [1].

Additionally, retinitis pigmentosa may occur as a sporadic (simplex) disorder or be inherited in an autosomal dominant (AD), autosomal recessive (AR) or X-linked recessive (XLR) mode [1]. On the other hand, the mode of inheritance is only X-linked recessive in patients with choroideremia [1]. The CHM gene is the only gene identified in patients with choroideremia so that it can be confirmed by genetic investigation in order to counsel the family [1].

Key Words: retinal dystrophy; choroideremia; retinitis pigmentosa; chorioretinal degeneration.

Compliance with ethical guidelines:

Review and original based materials have been appropriately cited and the ethical guidelines have been respected.

Funding:

This research did not receive any specific grant from funding agencies in the public, commercial, or nonprofit sectors.

Conflict of interest:

The author declares no conflict of interest.

Acknowledgements:

The author would like to express his honest gratitude and high respect for the lifetime support of his father, Mohammad Nouraeinejad.

References:

1. Nouraeinejad A. Differential Diagnosis in Optometry and Ophthalmology. Second Edition. Iran: Noruzi Publication; 2017.
2. Mitsios A, Dubis AM, Moosajee M. Choroideremia: from genetic and clinical phenotyping to gene therapy and future treatments. *Ther Adv Ophthalmol*. 2018;10:1-18.
3. Dong S, Tsao N, Hou Q, Bozkaya D, Leroy BP. US Health Resource Utilization and Cost Burden Associated with Choroideremia. *Clin Ophthalmol*. 2021;15:3459-3465.
4. Moosajee M, Ramsden SC, Black GC, Seabra MC, Webster AR. Clinical utility gene card for: choroideremia. *Eur J Hum Genet*. 2014;22(4):e1-e4.
5. MacDonald IM, Sereda C, McTaggart K, et al. Choroideremia gene testing. *Expert Rev Mol Diagn.* 2004; 4: 478–484.