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AN UNUSUAL CASE OF ATYPICAL RETINITIS PIGMENTOSA WITH BILATERAL FULL THICKNESS MACULAR HOLES

Ophthalmology

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ABSTRACT

Retinitis pigmentosa (RP), an inherited retinal disorder can present with typical features of retinal pigment atrophy, bony spicules and arteriolar attenuation. It can also present in the less commonly observed atypical form whose features depend on the type of systemic association. Although it is associated with many posterior segment complications, macular holes can rarely be associated with this condition. A full thickness macular hole affecting both eyes is commonly seen in a typical case. Our patient with features of atypical RP presented with bilateral full thickness macular holes suggesting the rarity of this case.

KEYWORDS

Macular hole, Atypical retinitis pigmentosa, Bony spicules

INTRODUCTION

Retinitis pigmentosa (RP) is a term used for a group of disorders that are characterized by inherited, progressive dysfunction, cell loss, and atrophy of retinal tissue. RP is a slowly progressive disease which typically affects the rods before the cones. Initial involvement of photoreceptors lead to subsequent damage to the inner retinal cells.[1] Symptoms are common to both typical and atypical forms of RP and comprise of loss of peripheral and night vision.

The signs of typical RP are retinal pigment layer atrophy associated with mild arteriolar narrowing, intraretinal perivascular bone spicule pigmentary changes, tessellated fundus with gliotic waxy pallor of optic disc.[2] (Figure 1) The features of atypical retinitis pigmentosa depend on the pattern and the associated systemic conditions. They present with impairment of central vision where cones are affected earlier, absence of pigment accumulation, scattered whitish yellow spots and sectoral RP with involvement of inferior quadrants.[3] Our case with features of atypical retinitis pigmentosa presented with bilateral full thickness macular holes and myopia which are commonly seen in a typical case of RP. This is rarely encountered in clinical practice and such cases have to be evaluated thoroughly in order to establish the correct treatment protocol to prevent impending visual impairment.

CASE REPORT

A 60-year-old Indian female patient who works as an agriculturist presented to the outpatient department with painless diminution of vision in both eyes for 8 months. There was no history of trauma. The patient had no co morbidities and family history was insignificant.

Visual acuity in both eyes was 6/60. Anterior segment examination revealed senile immature cataract with nuclear sclerosis grade 2 with presence of vitreous bands. The Watzke Allen test showed a broken slit beam showing a macular hole. The fundus examination revealed bilateral central punched out lesions with a red base and yellow spotting with pigmented bony spicules in the inferonasal quadrant suggestive of atypical RP (Figure 2). Optical coherence tomography showed full thickness macular hole with posterior vitreous detachment in both eyes. The patient was treated with low visual aids which helped enhance residual vision by magnifying the image of the object at the retinal level.

DISCUSSION

The macular area is usually spared from photoreceptor degeneration until late stages of the disease in contrast to earlier stages where changes are observed. The commonly observed macular abnormalities are cystoid macular edema (CME) and macular holes (MHs).[4,5]

Although the mechanism of MH formation in RP cases is not completely understood, chronic blood–retinal barrier breakdown causes irregularities of the ILM in the inner surface of the retina.[6]

Macular cysts tend to coalesce and along with ILM wrinkling may sometimes lead to the formation of MH. MHs have been reported in 4%–8% of eyes with different shapes and stages.[7] Detection of these abnormalities is important because they can significantly reduce the central visual function in patients with RP whose peripheral visual function is already compromised. This case highlights the importance of investigating macular abnormalities in cases of RP. This is to detect pathologies which can be treated such as FTMH and CME which thereby helps maintain central vision in RP cases.

CONCLUSION

In the following case, the late onset symptomatology in the sixth decade was noticed but with severe structural impairment and peripheral visual field loss. Moreover, the patient presented with a FTMH in both eyes which is more commonly seen in typical cases of RP than in an atypical case. The long-term visual prognosis in such cases is often correlated with progression of macular degeneration, loss of photoreceptors at the fovea, evolution of cystoid macular edema and appearance of posterior subcapsular cataracts.

Furthermore, molecular genetic examinations are required to be performed in order to establish gene mutation and to refer the patient for genetic counselling.

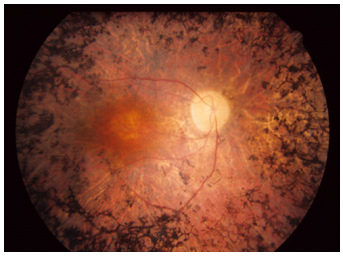

FIGURE 1: TYPICAL RETINITIS PIGMENTOSA

Showing the signs of typical RP are retinal pigment layer atrophy associated with mild arteriolar narrowing, intraretinal perivascular bone spicule pigmentary changes, tessellated fundus with gliotic waxy pallor of optic disc.

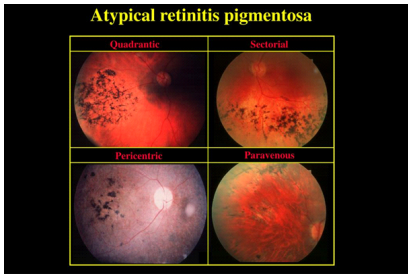

FIGURE 2: ATYPICAL RETINITIS PIGMENTOSA
Showing various forms of atypical retinitis pigmentosa

REFERENCES

1) Hisham, A. H., Mostafa, M. S., Sherif, S. K. & Hisham, F. K. (2010) Optical Coherence Tomography Study of Typical and Atypical Cases of Retinitis Pigmentosa: Journal of American Science, 6(10)
2) David, G. T. (2017) Retinitis Pigmentosa Clinical Presentation: Medscape
3) Howard, F., & Hansel, M.D. (1925) Atypical Retinitis Pigmentosa: American Journal of Ophthalmology, Volume 8, Issue 5, May 1925.Pages 374-376
4) Hagiwara, A., Yamamoto, S., Ogata, K., Sugawara, T., Hiramatsu, A., & Shibata, M. (2011) Macular abnormalities in patients with retinitis pigmentosa: Prevalence on OCT examination and outcomes of vitreoretinal surgery: Acta Ophthalmol, 89: e122-5
5) Fishman, G.A., Fishman, M., & Maggiano, J. (1977) Macular lesions associated with retinitis pigmentosa: Arch Ophthalmol,95:798-803
6) Takezawa, M., Tetsuka, S., & Kakehashi, A. (2011) Tangential vitreous traction: A possible mechanism of development of cystoid macular edema in retinitis pigmentosa. Clin Ophthalmol, 5:245-8.
7) Raja, M., Goldsmith, C., & Burton, B.J. (2011). Spontaneous resolution of full-thickness macular hole in retinitis pigmentosa: BMJ, Case Rep. pii: bcr0320113999.
8) https://binged.it/2OmjCRv eyesoncambodia.wordpress.com