International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Nanophthalmos with Associated Retinitis Pigmentosa

Totli Kuruba Mayuri[1], Malleswari Medikonda[2]

[1, 2]Department of Glaucoma, Sankara Eye Hospital, Guntur, Andhra Pradesh, India

Abstract: ***Nanophthalmos is a rare genetic disorder characterized by abnormally small eyes due to compromised growth, often resulting in high hyperopia and various ocular complications. This article presents two cases of nanophthalmos, highlighting its clinical manifestations and associated conditions. Case 1 describes a 41 - year - old female with chronic angle - closure glaucoma, high hypermetropia, amblyopia, and retinitis pigmentosa. Case 2 involves a 35 - year - old female with nystagmus, esotropia, and retinitis pigmentosa. The discussion explores the relationship between nanophthalmos and retinal abnormalities, proposing two potential theories for pigmentary retinal changes. Uveal effusion and non - rhegmatogenous retinal detachment are also discussed as complications of nanophthalmos. These cases underscore the importance of recognizing the diverse clinical presentations of nanophthalmos, including its impact on visual function and the potential for pigmentary retinal degenerations like retinitis pigmentosa.***

Keywords: Nanophthalmos, high hyperopia, amblyopia, glaucoma, retinitis pigmentosa

1. Introduction

Nanophthalmos is a rare genetic disease with a small eye secondary to compromised growth and is characterized by ([1)] Shortened axial length (generally 19.6mm or less; at least two standard deviations below age - matched controls), High lens/eye volume ratio with narrow iridocorneal angle, High hyperopia (ranging from +8.00 to +25.00 DS) and Scleral thickening.

It can be associated with [(2, 3, 4)] Angle - closure glaucoma, Axial high hypermetropia, amblyopia, corneal steepening and irregular astigmatism, Absent or rudimentary foveal avascular zone, Optic disc drusen, Retinitis pigmentosa, Retinoschisis and foveoschisis, Crowded optic disk, chorioretinal folds, and retinal cysts, Central retinal vein occlusion and increased subfoveal choroidal thickness

Case 1

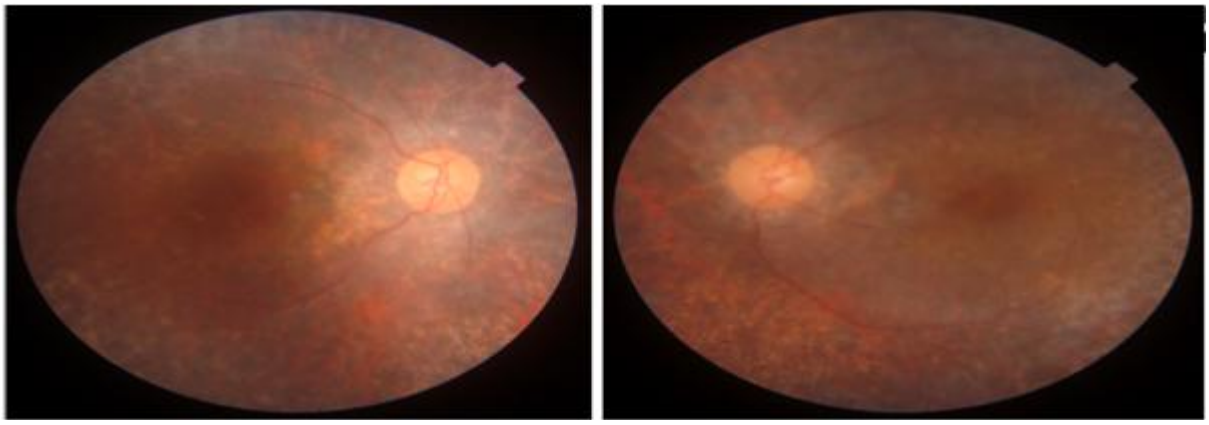
A 41yrs old female presented to us with complaints of redness and pain both eyes occasionally from 1year. Has a history of night blindness from the age of 20yrs and has never used glasses since childhood.

On examination her vision was 2/60 in right eye (RE) and hand movements in left eye (LE) with refractive error of +14.0DS RE and +13.0DS LE. Slit lamp examination showed RE clear cornea with shallow anterior chamber, sluggishly reacting pupil and early lenticular changes. LE showed edematous cornea, shallow anterior chamber, mid dilated and not reacting pupil with early lenticular changes. IOP - 12mm Hg RE, 68mm Hg LE, Gonioscopy closed angles BE. YAG peripheral iridotomy was done BE after controlling IOP. During one week follow up - Vision 2/60 RE, 3/60 LE with patent PI both eyes and IOP - 14 mm Hg RE and 20mm Hg LE.

Fundus showed Pale disc BE with arteriolar attenuation and pigmentary degeneration of retina, macula was normal with dull FR. (Fig 1)

Axial length - 15.45mm RE and 15.71 mm LE.

This patient had BE nanophthalmos with chronic angle closure with high hypermetropia, amblyopia and retinitis pigmentosa



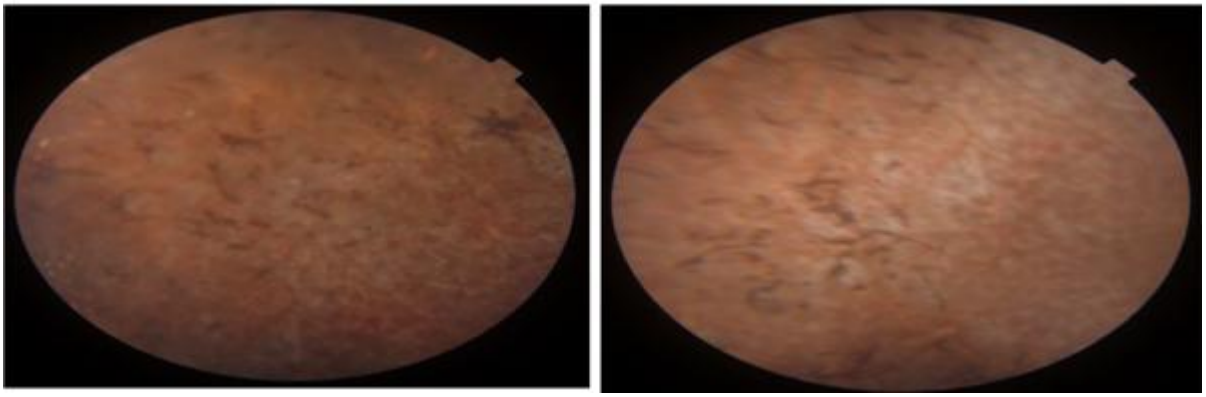


Figure 1: Image showing fundus of both eyes with pale disc, attenuated arterioles and pigmentary degeneration of retina

Case 2:
A 35yrs old female presented to us with complaints of diminution of vision and abnormal eye movements BE since childhood. On examination, BE had Nystagmus, esotropia with vision of PL+, clear cornea, Sluggish pupils and pseudophakia with IOP 16mm Hg and open angles BE.

Axial length was 19.12 mm RE and 20.45 mm LE.

Fundus showed pale disc, attenuation of arteries with pigmentary degeneration of retina suggestive of retinitis pigmentosa with consecutive optic atrophy BE and RE nanophthalmos.

2. Discussion

Nanophthalmos is a rare clinical spectrum of disorders with a phenotypically small but morphologically normal eyes. [(5)] Maldevelopment of the retina, retinitis pigmentosa, and other retinal anamolies have also been reported in nanophthalmos. [(6)] It can be explained with two theories:

1) In underdeveloped eye of nanophthalmos, lack of full development of the retina in the later part of fetal life is responsible for pigmentary retinal dystrophic changes.
2) Possibility of the presence of a prior uveal effusion causing a secondary pseudo - RP after retinal detachment. [(7)] Uveal effusion and non rhegmatogenous retinal detachment may occur spontaneously or as a complication of ocular surgery in nanophthalmos. [(8)] due to impaired venous drainage because of thickend sclera [(9)]

3. Conclusion

Nanophthalmos can be unilateral and the cause for visual decline in nanophthalmos is not only the high hyperopia, amblyopia, glaucoma, uveal effusion but also pigmentary retinal degenerations like RP.

References

[1] Sundin OH, Dharmaraj S, Bhutto IA, et al. Developmental basis of nanophthalmos: MFRP is required for both postnatal ocular growth and postnatal emmetropization. Ophthalmic Genet.2008; 19 (1): 1 - 9.
[2] M. K. Walsh and M. F. Goldberg, “Abnormal foveal avascular zone in nanophthalmos, ” American Journal of Ophthalmol_x0002_ogy, vol.143, no.6, pp.1067 - 1068, 2007.
[3] J. Cresp´ı, J. A. Buil, F. Bassaganyas et al., “A novel mutation confirms MFRP as the gene causing the syndrome of nanophthalmos–renititis pigmentosa–foveoschisis–optic disk drusen, ” American Journal of Ophthalmology, vol.146, no.2, pp.323–328, 2008.
[4] L. C. Zacharias, R. Susanna, O. Sundin, S. Finzi, B. N. Susanna, and W. Y. Takahashi, “Efficacy of topical dorzolamide therapy for cystoid macular edema in a patient with MFRP - related nanophthalmos–retinitis pigmentosa–foveoschisis–optic disk drusen syndrome, ” Retinal Cases and Brief Reports, vol.9, no.1, pp.61–63, 2015.
[5] Singh OS, Simmons RJ, Brockhurst RJ, Trempe CL. Nanophthalmos: a perspective on identification and therapy. Ophthalmology.1982; 89: 1006–12.
[6] Waardenburg PJ. In: Waardenburg PJ, Franceschetti A, KleinD, eds. Genetics and ophthalmology. Assen: Royal VanGorcum, 1961; 1: 764.
[7] Chang CJ, Lai WW, Edward DP, Tso MOM. Apoptotic pho_x0002_toreceptor cell death after traumatic retinal detachment in humans. Arch Ophthalmol 1995; 113: 880–6.
[8] Ryan EA, Zwaan J, Chylack LT Jr. Nanophthalmos with uveal effusion. Clinical and embryologic considerations. Ophthalmology 1982; 89: 1013–7.
[9] Shaffer RN. Discussion. Trans Am Ophthalmol Soc 1975; 73: 119–20. Comment on: Trans Am Ophthalmol Soc 1975; 73: 97–118.