Commentary: Clinical and biometric characteristics of pediatric eyes with nanophthalmos

The development of the eye starts as early as three weeks of gestational age.[1] It involves the differentiation of specific cells from neuroepithelium and mesenchyme into various ocular tissues, which perform particular functions aiding in vision. Mutations in specific genes encoding for the development of the eye, such as *PAX6*, *MRFP*, *TMEM98*, *BEST1*, lead to developmental abnormalities in the eye.[2] These abnormalities can be as minimal as an isolated iris coloboma to gross defects like microphthalmos.

Microphthalmos is a decrease in the eye's size compared to age-matched population. Nanophthalmos is a clinical variant of microphthalmos in which the anterior and posterior segments of the eye stop developing without any other functional or structural abnormality.[3] Nanophthalmos is considered a rare/orphan disease by the Orphan Drug Act of 1983, which means the prevalence is considerably low in the general population to provide a minuscule financial incentive for the private sector to make and market new medications to treat or prevent it. The diagnosis and classification of this relatively rare disease is thus understudied. However, it is essential to study nanophthalmos as it provides us with information about the embryological development of the eye.

The article in the present issue of the journal details the characteristics of 40 patients with nanophthalmos, comparing them with the normal population.[4] It is crucial to understand that definition of nanophthalmos cannot be simplified to <20.5 mm, as used in the present article, for all age groups. Similarly, a blanket value of less than <16 years with no age range and an axial length cutoff value of 17 mm doesn't provide useful clinical information. There are varying classifications of nanophthalmos, but pediatric definitions are not clear.[5,6] Axial length less than two standard deviations for that age with other coexisting factors of high hyperopia (>+7 D), a small anterior chamber with a normal/thicker lens and steep, smaller corneas (<11 mm), as well as retinochoroidal thickening are essential in classifying a patient as nanophthalmic. Hence, it is necessary to measure the biometric parameters, including the retino-choroido-scleral thickness and the LT/ACD or LT/AL ratio when diagnosing nanophthalmos.[7] Identifying these factors is essential as almost half of those with higher ratios are at risk of developing glaucoma.[7] Clinical features of nanophthalmos play an essential role in diagnosing the disease and differentiating it from posterior microphthalmos with normal anterior segment dimensions (hence not at risk for glaucoma) but more retinal complications.[3,8] A combination of increased scleral thickness and abnormal collagen is hypothesized to impair vortex venous drainage and reduce the transscleral flow of proteins in these patients. Recent genetic studies underlie a strong familial basis of nanophthalmos, with five genes (*MFRP*, *TMEM98*, *PRSS56*, *BEST1*, and *CRB1*) being implicated.[3]

Nanophthalmos is an easily missed diagnosis due to the complexity of its evaluation and classification, making its diagnosis challenging. Historically speaking, the anatomy and histology of ocular structures in such cases were too complicated to evaluate with the available resources. With the advent of optical coherence tomography (OCT), it is easier to document and record findings in these patients. There is minimal source available on nanophthalmic pediatric patients. Pediatric ophthalmologists should also be well versed with this condition, which will help in the early diagnosis and treatment of associated complications like hyperopia (and the resulting amblyopia), angle-closure glaucoma, and uveal effusion syndrome.

Savleen Kaur, Jaspreet Sukhija, Vivekavardhan Chatla

Advanced Eye Centre, Post Graduate Institute of Medical Education and Research, Chandigarh, India

Correspondence to: Dr. Savleen Kaur, Assistant Professor, Advanced Eye Centre (Department of Ophthalmology), Post Graduate Institute of Medical Education and Research, Chandigarh, India.
E-mail: mailsavleen@gmail.com

References

1. Edward DP, Kaufman LM. Anatomy, development, and physiology of the visual system. Pediatr Clin North Am 2003;50:1-23.
2. Bosze B, Suarez-Navarro J, Soofi A, Lauderdale JD, Dressler GR, Brown NL. Multiple roles for Pax2 in the embryonic mouse eye. Dev Biol 2021;472:18-29.
3. Carricondo PC, Andrade T, Prasov L, Ayres BM, Moroi SE. Nanophthalmos: A review of the clinical spectrum and genetics. J Ophthalmol 2018;2018:2735465.
4. Rajendrababu S, Vaishali V, Senthilkumar VA, Ramesh S, Uduman MS. Comparison of clinical and biometric characteristics between nanophthalmic children and age-matched controls. Indian J Ophthalmol 2022;70:2448-5.
5. Wu W, Dawson DG, Sugar A, Elner SG, Meyer KA, McKey JB, *et al*. Cataract surgery in patients with nanophthalmos: Results and complications. J Cataract Refract Surg 2004;30:584-90.
6. Yalvac IS, Satana B, Ozkan G, Eksioglu U, Duman S. Management of glaucoma in patients with nanophthalmos. Eye (Lond) 2008;22:838-43.
7. Agarkar S, Koladiya N, Kumar M, Vijaya L, Raman R. Nanophthalmos in children: Morphometric and clinical characterization. J AAPOS 2020;24:27.e1-5.
8. Khan AO. Posterior microphthalmos versus nanophthalmos. Ophthalmic Genet 2008;29:189.

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_844_22

Cite this article as: Kaur S, Sukhija J, Chatla V. Commentary: Clinical and biometric characteristics of pediatric eyes with nanophthalmos. Indian J Ophthalmol 2022;70:2447-8.