



Subnormal visual acuity after compliant amblyopia therapy: residual/refractory amblyopia or co-existing pathology? - a retrospective analysis

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Strabismus, 2024 Feb 4:1-12.

- <https://doi.org/10.1080/09273972.2023.2294997>

Purpose:

To assess the prevalence of alternate etiology/co-existing pathology among patients with amblyopia, and to characterize factors contributing to over-diagnosis of amblyopia.

Methods:

We retrospectively reviewed records of children (from 1 January 2016 to 31 December 2019) who were initially diagnosed as “amblyopia” but later an alternate diagnosis for subnormal vision was established. Patients who had a best corrected visual acuity (BCVA) of $\leq 20/32$ (0.2 logMAR) after compliant amblyopia therapy were divided into 2 groups: those with refractory amblyopia (BCVA improvement from baseline < 1 logMAR line) and residual amblyopia (BCVA improvement from baseline > 1 logMAR line). Data was collected for presence/absence of amblyogenic risk factors, history, ocular examination, and investigations leading to the final alternate diagnosis. We analyzed the factors that contributed to the initial over-diagnosis of amblyopia using the diagnostic error evaluation and research (DEER) taxonomy tool.

Results:

During the study period, 508 children with an initial diagnosis of amblyopia met the study criteria. Among these 508 children, 466 were diagnosed to have amblyopia alone, while 26 children (5.1%, median age: 7 years, 17 boys: 9 girls) were revised to have an alternate diagnosis/co-existing pathology. These 26 patients comprised of 2 groups: children referred to us as amblyopia but re diagnosed to have an alternate diagnosis; and a second subset, initially diagnosed by us to have amblyopia, but later found to have alternate diagnosis/co-existing pathology. Subclinical optic neuritis (50%, 13 children), and occult macular dystrophy (OMD) (38.4%, 10 children) were the most frequent alternative diagnoses. Children with ametropic amblyopia (8/26, 30.7%) were most frequently misdiagnosed. Risk factors that led to an initial diagnosis of amblyopia were: high refractive error and heterotropia in 7 patients each (26.9%), anisometropia in 12 (46.1%), and prior pediatric cataract surgery in 4(15.3%). No improvement in BCVA in 21/26 (80.7%) children led to suspicion of co-existing etiology. Other clues were optic disc pallor (11), subnormal color vision (7), history of parental consanguinity in 7, and preceding febrile illness/rhinitis in 1 child. The DEER taxonomy tool suggested that the most common reasons for diagnostic errors were over-emphasis on amblyopia.

Conclusion:

Our study suggests that 5% of children diagnosed with amblyopia might have co-existing/alternate etiology. Most common co-existing etiologies were subclinical optic neuropathy, and OMD. No improvement in BCVA, subtle history and examination findings prompted further workup. Not considering co-existing etiologies was the most common reason for an initial overdiagnosis of amblyopia.

KEYWORDS:

- [Amblyopia](#)
- [co-existing](#)
- [macular dystrophy](#)
- [optic neuritis](#)
- [subclinical](#)

Acknowledgements

We acknowledge the support of the following:

Dr. Rajat Kapoor and Dr. Mohan Kannam (ex-faculty, LV Prasad Eye Institute, in the diagnosis and management of the patients, and contributing the patients).

Mr. Mahalakshmi Naidu, (photography and audio-visual departments), GMRV campus, L V Prasad Eye Institute for their assistance in documentation of the clinical photographs.

We also thank our diagnostic team with the help in the documentation and management of these patients.

English writing: We thank Dr. Akhila Kekunnaya for help with the English writing.

Funding

Financial support and sponsorship: Hyderabad Eye Research Foundation (HERF), Telangana, India