Widefield Retinal Imaging in Gyrate Atrophy: Correlation of Structural, Biochemical, and Functional Characteristics

Srikanta Kumar Padhy MD [1], Deepika C. Parameswarappa MS [2 3], Sumant Sharma MD [2], Tapas Ranjan Padhi MS [1], Subhadra Jalali MS [2], Brijesh Takkar MD [2], Raja Narayanan MS [2]

1- LV Prasad Eye Institute, Mithu Tulasi Chanrai Campus, Anant Bajaj Retina Institute, Bhubaneswar, India
2- LV Prasad Eye Institute, Kalam Anji Reddy Campus, Anant Bajaj Retina Institute, Srimati Kanuri Santhmma Centre for Vitreoretinal Diseases, Hyderabad, India
3- Ophthalmology and Vision Sciences, The Hospital for Sick Children, University of Toronto, Toronto, Ontario, Canada

Ophthalmology Retina
https://doi.org/10.1016/j.oret.2024.10.016Get rights and content

Objective

To profile a cohort of gyrate atrophy patients classified by widefield retinal imaging and correlate the structural, biochemical, and functional characteristics.

Design

Retrospective observational cohort study.

Participants

Sixty-five patients (129 eyes) with gyrate atrophy.

Methods

Data of participants with a diagnosis of gyrate atrophy were retrieved from their electronic medical records (January 2015 to December 2023). Retinal involvement was classified into 3 zones using widefield retinal images. Zone 3 had atrophic patches in the area anterior to the equator; zone 2 had involvement limited to the arcades but posterior to the equator; zone 1 had involvement within the vascular arcades and/or peripapillary region, with or without any other zone involvement. Macular assessment was performed using swept-source OCT (n = 104). Flash electroretinogram (ERG) was performed in 40 eyes. Serum ornithine levels (n = 35) were measured, and genetic analysis was conducted (n = 18).

Main Outcome Measures

Demography, patient profile, zone of retina involved, macular features, and serum ornithine levels.

Results

The average age at presentation was 26.4 (range, 5–67) years; the majority were male. Nyctalopia (n = 35, 53.8%) and blurred vision (n = 29, 44.6%) were the most common symptoms. Positive family history was reported in 32.3% of patients. Most eyes were myopic (69.8% <−3 diopters). Posterior subcapsular cataracts were documented in 36.4% of eyes. The highest frequency of retinal area

affected was zone 1 (57.14%), followed by zone 2 (33.33%) and zone 3 (9.52%), correlating with the age at presentation. Foveoschisis was observed in 57.7% of eyes, with a higher prevalence in eyes with zone 1 disease. Elevated serum ornithine levels (>163 µmol/L) were found in 77.14% of patients. The ERG showed nonrecordable (n = 32) or severely reduced (n = 8) responses in scotopic and photopic phases. Genetic analysis of 18 patients identified mutations in the *OAT* gene, including a novel missense variant (c.290T>C).

Conclusions

This large cohort of patients with gyrate atrophy revealed symmetrical involvement, predominantly in zone 1. Most patients presented between the first and third decades, experienced nyctalopia, vision reduction, early posterior subcapsular cataracts, and varying degrees of myopia. Zone 1 involvement was strongly associated with foveoschisis and visual compromise.