Macular abnormalities in patients with Retinitis Pigmentosa.

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ABSTRACT:

Objective: To assess the frequency of macular abnormalities identified through OCT in individuals with retinitis pigmentosa.

Methodology: This quantitative cross-sectional research was conducted in the Department of Ophthalmology at Civil Hospital Karachi. A total of 50 eyes from patients aged 18 to 65 years, diagnosed with retinitis pigmentosa (RP), were examined. The diagnosis of retinal pigmentosa was based on symptoms such as poor night vision or visual field loss, along with an ophthalmological assessment revealing mid-peripheral pigmentary changes, arteriolar narrowing, and a waxy pallor of the optic disc. To ensure high-quality retinal optical coherence tomography (OCT) images, eyes with significant media opacities that obstructed the view of the fundus were excluded from the study.

Results: Average The mean age of the patients was 41.2 ±3.24 years. Majority of the patients were between 36-45 years, 17 (68%) were females and 08 (32%) were males. 31(62%) patients had normal or mild impairment. The lens status examination revealed that 7(14%) eyes had cataract and 5(10%) eyes had aphakia. 12 (24%) patients showed up having normal macula and 3 (6%) had Cystoid macular oedema (CME).

Conclusion: Macular finding of OCT revealed that macular finding in patients with retinitis pigmentosa (RP) manifest in various forms. Therefore, genetic testing is essential for individuals with visual impairments.

Keywords: Inherited retinal dystrophy, macular atrophy, macular edema, optical coherence tomography, retinitis pigmentosa

Introduction:

Approximately 1 in every 4000 individuals globally is affected by retinitis pigmentosa (RP), with over 100 genes identified as associated with this condition.[1,2] Night blindness frequently serves as the initial symptom of RP, followed by a progressive loss of peripheral vision.[3] The clinical manifestations of RP follow a typical pattern, and in the advanced stages, only a small central portion of the visual field may remain, resulting in severely limited vision, often referred to as 'tunnel vision.' [4] However, it is not uncommon for patients with RP to exhibit macular abnormalities during fundus examinations, which can lead to additional visual complications such as vitreomacular interface disorders, choroidal neovascularization, macular atrophy, and cystoid macular edema (CME). The occurrence of macular holes (MH) as a consequence of RP is rare, with reported rates ranging from 0.5% to 4.5%.[5,6] Previous studies have indicated a possible pathophysiological link between RP and MH.[7,8] The use of high-definition spectral-domain optical coherence tomography (SD-OCT) significantly improves our understanding of the clinical features of these eyes, which remain largely unexplored. The age at which retinitis pigmentosa (RP) manifests can vary from infancy to late adulthood.[9] The emergence of clinical symptoms is linked to the mode of inheritance, which may be X-linked, autosomal recessive, or autosomal dominant. Numerous investigations have employed optical coherence tomography (OCT) to assess the overall retinal structure in RP patients, and the latest advancements in high-resolution OCT technology facilitate the observation of finer retinal details. With the introduction of spectral-domain SD-OCT, it has become feasible to obtain structural information regarding retinal anatomical irregularities in individuals with RP.[10] Consequently, given the progress in technology that allows for the visualization of more intricate retinal structures, we undertook this study to assess the prevalence of macular abnormalities identified by OCT in patients diagnosed with retinitis pigmentosa.

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Objective:

To assess the frequency of macular abnormalities identified through OCT in individuals with retinitis pigmentosa.

Methodology:

This quantitative cross-sectional research was conducted, between July2022 till December 2022, in the Department of Ophthalmology at Civil Hospital Karachi. A total of 50 eyes from patients aged 18 to 65 years, diagnosed with retinitis pigmentosa (RP), were examined after taking approval from ethical board of Dow University of Health Sciences through Institute of Research Board (IRB); IRB-2276/DUHS/Approval/2021/678. The diagnosis of retinal pigmentosa was based on symptoms such as poor night vision or visual field loss, along with an ophthalmological assessment revealing mid-peripheral pigmentary changes, arteriolar narrowing, and a waxy pallor of the optic disc. To ensure high-quality retinal optical coherence tomography (OCT) images, eyes with significant media opacities that

obstructed the view of the fundus were excluded from the study. After taking both verbal and written consent, data was collected using a meticulously designed performa. The performa was translated into local languages when necessary. Diagnoses of RP was established based on both the patient history and ophthalmic examinations. Patients exhibiting poor night vision or compromised visual fields, along with slit lamp examinations revealing mid-peripheral intra-retinal perivascular 'bone-spicule', arteriolar attenuation, and waxy disc pallor, were diagnosed with retinal pigmentosa. To obtain high-quality retinal optical coherence tomography (OCT) images, the eyes with significant media opacities that obstructed a thorough visualization of the fundus were excluded. Additionally, patients with a history of concurrent ocular conditions or treatments (such as intra -vitreal antivascular endothelial growth factor injections, diabetic macular oedema, and laser therapy) were also excluded. Visual acuity was classified according to WHO guidelines: [11]

- Mild visual impairment: <6/12 but 6/18 or better;
- Moderate visual impairment: <6/18 but 6/60 or better;
- Severe visual impairment: <6/60 but 3/60 or better;
- Blindness: <6/60 or <10° in central radius field.

The statistical analysis performed using SPSS® 26. Frequencies and percentage were used for descriptive variables. Visual acuity and central macular thickness (CMT) were correlated using Pearson's Chi-square; a p-value of less than 0.05 was regarded as statistically significant.

Results:

A total of 50 patients participated in the study. The average age was 41.2 ±3.24 years. Most of the patients were between 36-45 years followed next in frequency 46-55 years. Range patients succeeded next. While both the extreme of age groups presented less in number as depicted in table 1.

Table No 1: Distribution of patients according to age groups.

Age (Years)	n	(%)
18-25	4	8
26-25	12	24
36-45	17	34
46-55	13	26
56-65	4	8
Total	50	100
Mean age	42.1 ±3.24 years	

There were 17 (68%) females and 08 (32%) males in the study. The table 2 depicts frequency and percentage of visual acuity and status of the lens. There were 31(62%) patients with normal or mild impairment, 11(22%) patients had moderate and 8(16%) patients had severe visual impairment and blindness. The lens status examination revealed that 7(14%) eyes had cataract, 5(10%) eyes had aphakia while 11(22%) had pseudo-phakia.

Amongst all 12 (24%) patients had normal macula and 3 (6%) had CME. The distribution of macular morphology based on optic coherence tomography is detailed in table 3.

Table No 2: Distribution of patients according to visual acuity and status of lens

Distribution	Number of eyes	Eyes (%)
Visual acuity		
Normal or Mild visual impairment	31	62
Moderate visual impairment	11	22
Severe visual impairment and blindness	8	16
Total	50	100
Status of Lens		
Cataract	7	14
Aphakia	5	10
Pseudophakia	11	22
Clear	21	42
Early opacity	6	12
Total	50	100

Table No 3: Macular abnormalities.

Macular Morphology	Number	Frequency (%)
Normal macula	12	24
Cystoid macular oedema (CME)	3	6
ERM/VMA Epiretinal Membrane/ Vitreomacular Adhesion	2	4
Atrophic	20	40
Atrophic/VMA	5	10
ERM/atrophic	8	16
Total	50	100

Discussion:

In this study, one fifth of the eyes exhibited moderate-to-profound vision loss. Lens abnormalities, such as pseudophakia, aphakia, and well-formed cataracts were observed in more than two third of eyes. However, no statistically significant correlation (p >0.05). was found between macular morphology, CMT, and best corrected visual acuity (BCVA).

Impaired vision and diminished night vision (22%) were identified as the most prevalent symptoms, followed by normal and mild impairments that prompted RP patients to seek ophthalmological evaluations. A prior hospital-based study conducted by Eballe AO et al.[12] in a Cameroonian hospital reported a visual impairment rate of 57.5%, which exceeds the findings of our study. Even higher rates were observed in research by Ahmed J et al.[13] in Pakistani hospitals (80.3%), as well as by Onakpoya OH[14] and Ukponmwan CU15 in Nigeria, where visual impairment rates of 69.8% and 76.7% were documented, respectively.

During current study, a notable prevalence of cataracts was observed in 14% of the examined eyes. This rate aligns with findings previously documented in Cameroon[12]

and Nigeria.[14] However, it is lower than the incidences reported by Auffarth GU et al[16] in Germany and Pruett RC et al[17] in the United States. Cataracts can significantly contribute to central vision impairment. Surgical procedures for cataract removal in eyes affected by RP may improve vision, although the extent of visual enhancement is contingent upon the preoperative status of the ellipsoid zone.[18-21] Through the use of Optical Coherence Tomography (OCT) for examination, we found that macular atrophy was the most common macular abnormality, present in 40% of participants, which is lower than the 53.8% reported by Ibrahim W et al.[22] in a longitudinal study conducted in Egypt. Nevertheless, the incidence of atrophy in our research was higher than that observed in previous studies from Pakistan[13] and Nigeria,[14] which relied on clinical examination for the identification of macular atrophy. This reliance may have resulted in underreporting, as OCT is known to have a superior detection rate compared to clinical evaluations. According to our findings, cystoid macular edema (CME) was present in only 6% of cases. Therefore, OCT demonstrates greater efficacy than clinical examination in detecting macular oedema in eyes affected by Retinitis Pigmentosa (RP).[23] CME significantly contributes to vision loss in patients with RP and can be treated with topical administration of carbonic anhydrase inhibitors, which may also be given in oral form.

In the present study, the small sample size may be the primary reason for the inability to identify rare macular abnormalities, including macular holes and epiretinal membranes. Subsequent research should encompass a larger cohort and multi-centre investigations to facilitate a more thorough examination of macular abnormalities in individuals with retinitis pigmentosa.

Conclusion:

Results of current study obtained through OCT, reveal that macular abnormalities in patients with retinitis pigmentosa (RP) manifest in various forms. Therefore, genetic testing is essential for individuals with visual impairments.

Conflict of Interest:

The authors hold no conflict of interest.

Source of Funding:

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