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ORIGINAL ARTICLE

Ocular Manifestations Among Thalassemia Patients Presenting to Tertiary Care Hospital in Jammu Region

Sonika Syal, Rukhsana Kousar, Riya Gupta, Hans Raj Sharma, Manpreet Kaur

Abstract

Background: Thalassemia patients present with different types of ocular manifestations. These can be due to the disease process itself or due to iron chelation therapy. **Aims:** This study aims to find out the ocular manifestations in thalassemia patients at a tertiary care hospital in Jammu region. **Material and Methods:** It was a prospective, observational, cross-sectional, hospital-based study including 50 patients. A detailed history of the patients was taken. The ocular examination included the detailed slit lamp examination of the anterior segment, the colour vision, detailed fundus examination, refraction, and Schirmertest for dry eyes. **Results:** Out of 50 patients, 19 patients (38%) had ocular manifestations and these included dry eyes (30%), refractive errors (20%), colour vision defects (6%), disc hyperaemia (6%), AV tortuosity (4%), tessellated fundus (2%) and pseudoxanthoma elasticum (6%). **Conclusion:** In our study, we concluded that dry eyes were the most common ocular manifestations of thalassemia, followed by, refractive errors, colour vision defects, disc hyperaemia, AV tortuosity, tessellated fundus and pseudoxanthoma elasticum. Therefore, regular ophthalmological examination is essential to detect and manage these manifestations as soon as possible.

Key Words

Thalassemia, Ocular Abnormalties

Introduction

Thalassemia is a severe genetic blood disorder that is caused due to mutation in globin gene. Red blood cells are excessively destroyed due to abnormal globin chains. Patients with thalassemia major usually present to the hospital in the first two years of life. Lifelong blood transfusion is required for these patients to survive. Though mortality is prevented from repeated blood transfusion, iron that accumulates due to repeated blood transfusion can lead to organ failure.[1]

Ocular manifestations observed in thalassemia include decreased visual acuity, color vision defects, nyctalopia, cataract, retinopathy, visual field defects, optic neuropathy, pseudoxanthoma elasticum which includes angioid streaks, peau'd orange, pattern dystrophy like changes.[2]

The frequency of ocular manifestations in thalassemia ranges between 41.3% and 85% across different studies.[3,4,5,]

A few studies have been conducted in India on the ocular manifestations of Thalassemia.[6,7,8,9,10] In a tertiary care facility in GMC Jammu, patients diagnosed with thalassemia wereevaluated for ocular symptoms. The study intended to find out the different ocular

Department of Ophthalmology, Govt. Medical College Jammu, India.

Correspondence to: Dr Manpreet Kaur, Registrar, Department of Ophthalmology, Govt. Medical College Jammu. India.

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manifestations in patients with thalassemia.

Material and Methods

It was a prospective, cross-sectional, observational, hospital-based study conducted in the Department of Ophthalmology, GMC Hospital, Jammu over a period of 6 months from October 2023 to March 2024. The study was conducted after due approval from the Institutional Ethics Committee (IEC) of this institute (No: IEC/GMCJ/2012/1629, Dated: 08/12/2023). The study comprised of 50 patients of thalassemia including both thalassemia major and minor.

Study participants having diabetes, hypertension, other types of hemoglobinopathies, previous eye surgeries and those who were unwilling to participate in the study were excluded from the study.

Every participant provided the informed consent. A complete medical history was obtained. Detailed ocular examination was done which included visual acuity and refraction, slit lamp examination of the anterior segment, the colour vision, detailed fundus examination, and IOP measurements. Tear function tests included basal Schirmer's test.

Statistical Analysis

All the data was entered in an MS Excel spreadsheet. The quantitative data was represented as mean and standard deviation and qualitative data as number and percentage. Statistical analysis was done by using IBM SPSS statistics for windows version 25.0 (IBM Corp. Released 2017, Armonk, NY, USA). A *p*-value <0.05 was considered as statistically significant.

Results

The age and sex distribution of the study population was shown in Table 1. The mean age of the study population was 8.51 ± 2.52 years, ranging from 3-17 years. There were 27 (54%) males and 23 (46%) females. Male: Female ratio was 1.2:1. Family history of thalassemia was present in 12 (24%) patients.

Out of 50 patients, 37 (74%) patients had ocular manifestations and includeddry eyes (30%), refractive errors (20%), colour vision defects (6%), disc hyperaemia (6%), AV tortuosity (4%), tessellated fundus (2%) and pseudoxanthoma elasticum (6%) (Table 2). The slit lamp examination of the anterior segment of all the patients was normal. The mean IOP in right eye was 14.7 ± 1.14 mmHg and in the left eye was 15.1± 1.17 mmHg.

Table 1: Age and Sex Distribution of Patients

Age (Years)	No. of Patients		
	Male (%)	Female (%)	Total (%)
<4	3(6)	1(2)	4(8)
4-8	2(4)	2(4)	4(8)
9-13	12(24)	14(28)	26(52)
14-18	10(20)	6(12)	16(32)
Total	27	23	50(100)

Table 2: Distribution of Ocular Manifestations (n=50)

Ocular Manifestations	No. of Patients	Percentage (%)
Dry eyes	15	30
Refractive error	10	20
Color vision defects	3	6
Disc hyperemia	3	6
AV tortuosity	2	4
Tessellated fundus	1	2
Pseudoxanthoma elasticum	3	6

Discussion

Thalassemia is a group of inherited haematological disorders thatis caused due to defects in the synthesis of the haemoglobin chains. Ocular manifestations observed in thalassemia include decreased visual acuity, color vision defects, nyctalopia, cataract, retinopathy, visual field defects, optic neuropathy, pseudoxanthoma elasticum which includes angioid streaks, peau'd orange, pattern dystrophy like changes.[2] The frequency of ocular manifestations in thalassemia ranges between 41.3 % and 85 % across different studies.[3,4,5]

In our study, the mean age was 8.51 ± 2.52 years ranging from 3-17 ± 2.52 years. Kumble *etal*.[8] and Thakur *et al*.[6] in their studies reported the mean age of 9.73 years and 8.40 ± 2.61 years respectively. Thus, the results of our study were comparable to the above-mentioned studies.

In our study, 27 patients (54%) were males and 23 patients (46%) were females. The male-to-female ratio was found to be 1.2:1. Similar to our study, Ramakrishnan *et al*.[5] In theirstudy reported that 52% patients were males and 48% of patients were females whereas Baig *et al*.[2] in their study also reported that 53.7% of patients were males and 46.7% of patients were females.

In our study, 12 patients (24%) had family history of thalassemiawhich was similar to the study by Ramakrishnan e*t al*.[5], in which thalassemia was present in the family of 10 patients (20%).

In our study, ocular manifestations were seen in 74% of the patients. Similar to our study, Jafari *et al*.[4], Ramakrishnan *et al*.[5] and Abdel-Malak *et al*.[3] in their studies reported that ocular involvement was present in 68.5%, 74%, and 85% of the patients respectively. However, in contrast to the above studies, Gosai *et al*.[7], Baig *et al*.[2], Dewan *et al*.[11], and Taneja *et al*.[9] in their studies found that ocular manifestations were seen in 35.5%, 22.7 %, 36% and 58% of the patients respectively which was much lower than found in our study.

In our study, dry eyes were seen in 15 (30%) patients. Ramakrishnan *et al*.[5] and Prasad *et al*.[12] in their studies reported that dry eyes were seen in 38% and 33.3% of the patients respectively. However, in contrast to our study, Haghpanah *et al*.[13] in their study found that dry eyes were present in 5.1% of the patients only.

In our study, refractive errors were seen in 10 (20.0%) patients. Ramakrishnan *et al*.[5], Prasad *et al*.[12], Aksoy *et al*.[14], and Jafari *et al*.[4] in their studies found that refractive errors were present in 20%, 20.5%, 23.2%, and 21.2% of the patients respectively. Thus, the results of our study were similar to the above-mentioned studies. Colour vision defects were seen in 3 (6%) patients in our study. Similarly, Prasad *et al*.[12] and Jafari *et al*.[4] also reported colour vision defects in 5.1% and 3.7% of patients respectively.

In our study, disc hyperaemia was seen in 3 (6%) patients. Gosai *et al*.[7] in their study reported that disc hyperaemia was seen in 4.5% of the patients. Similarly, Baig *et a*l.[2] in their study also reported dischyperaemia in 1% of patients.

In our study, AV tortuosity was seen in 2(4%) patients. Gosai *et al*.[7] and Haghpanah *et al*.[13] in their studies reported tortuous blood vessels in 4.5% and 2.5% of the patients respectively. However, in contrast to our study, Ramakrishnan *et al*.[5], Kumble *et al*.[8], and Barteselli *et al*.[15] reported that in their studies, 14%, 17.3% and 16.9% of the patients had AV tortuosity respectively, which was much higher than in our study. Tessellated fundus was seen in 1 (2%) patient in our study. Similarly, Ramakrishnan *et al*.[5] in their study also reported tessellated fundus in 2% of patients.

In our study, pseudoxanthoma elasticum was seen in 3 (6%) patients. Similar to our study, Kumble *et al*.[8] in their study reported pseudoxanthoma elasticum in 2.7% of the patients. However, Barteselli *et al*.[15] in their study reported that 27.8% patients had fundus findings suggestive of pseudoxanthoma elasticum, which was much higher than in our study.

Conclusion

Children with thalassemia can present with different ocular manifestations. Therefore, regular ophthalmological examination is essential to detect these manifestations early so that management can be done early before irreversible loss of vision.

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Conflict of Interest : Nil

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