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+Clinical Profile and Treatment Outcomes of Children Diagnosed with Retinoblastoma

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Abstract

Background: *Retinoblastoma is the most common primary intraocular malignancy in children and remains a major cause of ocular morbidity and mortality when diagnosis and treatment are delayed. Understanding the clinical profile and treatment patterns is essential for improving early detection and outcomes.*

Methods: *This hospital-based descriptive study was conducted in the Department of Ophthalmology, National Institute of Ophthalmology and Hospital, Dhaka, from April 2011 to March 2013. A total of 46 children clinically suspected of having retinoblastoma, including referred cases, were enrolled. Patients with other ocular diseases or incomplete records were excluded. Detailed demographic data, clinical presentation, stage at diagnosis and treatment modalities were recorded.*

Results: *Most patients (56.5%) presented between 1 and 3 years of age, with a slight male predominance (56.5%). Leukocoria was the most common presenting feature, observed in 69.6% of cases, followed by leukocoria with proptosis (15.2%), pseudohypopyon (10.9%) and hyphema (4.3%). At presentation, the majority of patients were diagnosed at Stage II (60.9%), while 19.6% each were at Stage I and Stage III; no Stage IV cases were observed. The most common treatment modality was enucleation combined with radiotherapy (26.1%), followed by enucleation alone (21.7%) and chemotherapy (19.6%). A small proportion of patients (6.5%) were lost to follow-up.*

Conclusion: *Children with retinoblastoma commonly presented at an early age but often with intermediate-stage disease, resulting in a high rate of enucleation-based treatment. Early diagnosis and timely referral are crucial to enable globe-sparing management and improve overall outcomes.*

Keywords: *Retinoblastoma, Leukocoria, Enucleation, Pediatric ocular tumors, Clinical profile.*

1. INTRODUCTION

Retinoblastoma is the most common primary intraocular malignancy of childhood and represents a significant cause of morbidity and mortality if not diagnosed and treated promptly [1]. The disease arises from immature retinal cells and typically affects children under five years of age [2]. Although retinoblastoma is highly curable when detected early, delayed presentation remains a major challenge in many low- and middle-income settings, leading to advanced disease at diagnosis and poor visual and survival outcomes [3].

Clinically, retinoblastoma presents with a wide spectrum of signs and symptoms. Leukocoria is the most frequent presenting feature, followed by strabismus, redness, pain, watering, proptosis, hyphema and visual impairment [4]. The pattern of presentation often reflects the stage of the tumor at diagnosis, which plays a

critical role in determining treatment options and prognosis [5]. Advanced intraocular or extraocular disease frequently necessitates aggressive interventions, including enucleation and adjuvant therapy, to ensure patient survival [6].

The management of retinoblastoma has evolved considerably over time, with increasing emphasis on globe salvage and visual preservation in early-stage disease [7]. Treatment modalities include enucleation, systemic chemotherapy, focal therapies such as cryotherapy and laser photocoagulation and radiotherapy in selected cases [8]. The choice of treatment depends on tumor stage, laterality, extent of ocular involvement and availability of specialized facilities. In resource-limited settings, however, late presentation and limited access to advanced treatment modalities often restrict management options, resulting in higher rates of enucleation [9].

Understanding the clinical profile and treatment patterns of children diagnosed with retinoblastoma is essential for identifying gaps in early detection, referral pathways and management strategies [10]. Hospital-based data provide valuable insights into real-world presentation, disease severity at diagnosis and therapeutic approaches, which can inform public health interventions and clinical practice [11]. Moreover, evaluating treatment outcomes helps assess the effectiveness of existing management protocols and highlights areas requiring improvement [12].

Despite the clinical importance of retinoblastoma, data on its presentation and management in many developing regions remain limited. This study was undertaken to describe the clinical characteristics, stage at presentation, treatment modalities and outcomes of children diagnosed with retinoblastoma in a tertiary eye care setting. By analyzing these parameters, the study aimed to contribute to a better understanding of disease patterns and support efforts toward earlier diagnosis, optimized treatment and improved survival outcomes for affected children.

2. Methodology & Materials

This hospital-based descriptive study was conducted in the Department of Ophthalmology, National Institute of Ophthalmology and Hospital, Dhaka, from April 2011 to March 2013. A total of 46 children clinically suspected of having retinoblastoma, including those referred from other hospitals, were enrolled in the study. Children with ocular conditions other than retinoblastoma and those with incomplete medical records were excluded.

A detailed clinical history was obtained from the parents or guardians, focusing on presenting symptoms such as leukocoria (white pupillary reflex), watering, pain, redness, proptosis, squint, hyphema and defective vision. Information regarding laterality of the disease, duration of symptoms and disease progression was recorded. Family history was documented, including parental consanguinity and the presence of similar ocular conditions among siblings or other relatives.

All patients underwent comprehensive ocular examination, including assessment of visual acuity, pupillary reaction, anterior and posterior segment evaluation, measurement of intraocular pressure and corneal diameter. Relevant investigations included X-ray of the orbit and skull, computed tomography (CT) scan of the orbit and brain, B-scan ultrasonography and estimation of aqueous lactate dehydrogenase levels. Enucleated eyes were subjected to histopathological examination.

Treatment modalities were selected based on tumor stage and severity and included enucleation, radiotherapy (administered in collaboration with the Department of Radiology), cryotherapy and chemotherapy. All patients were followed up regularly, with careful examination of the anophthalmic socket and the contralateral eye at each visit to detect recurrence or involvement.

Data were entered and analyzed using Microsoft Excel. Descriptive statistics, including frequency, percentage, mean and standard deviation, were used to summarize demographic characteristics, clinical features, tumor stage, laterality and treatment modalities.

3. Results

Table1. *Age and Sex Distribution of Patients (n = 46)*

Age	Males	Females	Total	Percentage (%)
<1 year	5	3	8	17.4

1–3 years	14	12	26	56.5
3–5 years	4	3	7	15.2
>5 years	3	2	5	10.9
Total	26	20	46	100

Table 1 shows the age and sex distribution of the 46 patients included in the study. The majority of cases (56.5%) presented between 1 and 3 years of age, followed by 15.2% between 3 and 5 years, 17.4% under 1 year and 10.9% over 5 years. Male patients (26/46, 56.5%) were slightly more than female patients (20/46, 43.5%) across all age groups.

Table2. *Clinical Presentation of Retinoblastoma (n = 46)*

Presentation	Number of Cases	Percentage (%)
White reflex in the pupil	32	69.6
White reflex with proptosis	7	15.2
Pseudohypopyon	5	10.9
Hyphema	2	4.3
Total	46	100

Table 2 presents the clinical presentations of retinoblastoma among the 46 patients. The most common presenting feature was white reflex in the pupil (leukocoria), observed in 32 patients (69.6%), followed by white reflex with proptosis in 7 patients (15.2%). Pseudohypopyon was seen in 5 patients (10.9%) and hyphema was the least common, occurring in 2 patients (4.3%).

Table3. *Distribution of Retinoblastoma Stage at Presentation (n = 46)*

Stage	Number of Cases	Percentage (%)
I	9	19.6
II	28	60.9
III	9	19.6
Total	46	100

Table 3 illustrates the distribution of retinoblastoma stages at presentation among the 46 patients. The majority of cases, 28 patients (60.9%), presented at Stage II, followed by 9 patients (19.6%) at Stage I and 9 patients (19.6%) at Stage III. No patients were observed in Stage IV.

Table4. *Treatment Modalities Applied (n = 46)*

Mode of Treatment	Number of Cases	Percentage (%)
Enucleation	10	21.7
Enucleation + Radiotherapy	12	26.1
Radiotherapy alone	8	17.4
Enucleation + Cryotherapy	4	8.7
Chemotherapy	9	19.6
Lost to Follow-up	3	6.5
Total	46	100

Table 4 shows the treatment modalities applied to the 46 patients with retinoblastoma. The most commonly used treatment was enucleation combined with radiotherapy (12 patients, 26.1%), followed by enucleation alone in 10 patients (21.7%) and chemotherapy in 9 patients (19.6%). Radiotherapy alone was administered to 8 patients (17.4%), while enucleation with cryotherapy was used in 4 patients (8.7%). Three patients (6.5%) were lost to follow-up.

4. DISCUSSION

Retinoblastoma remains the most common intraocular malignancy of childhood and continues to pose significant diagnostic and therapeutic challenges, particularly in settings

where delayed presentation is frequent. In the present study, the majority of patients (56.5%) were diagnosed between 1 and 3 years of age, which is consistent with the natural history of retinoblastoma as a disease of early childhood. Similar age distributions have been reported by Lavaju et al. and Nabie et al., who observed peak presentation within the first three years of life in tertiary care settings [13,14]. These findings reinforce the importance of vigilant screening during infancy and early childhood.

A slight male predominance (56.5%) was observed in this study, which aligns with reports from Nepal, Iran and Malaysia, where male children were more frequently affected [13,14,15]. Although retinoblastoma is not known to have a strong gender predisposition, this pattern may reflect sociocultural factors influencing healthcare-seeking behavior rather than true biological differences, as suggested by Dimaras et al. and Ali et al [16, 17].

Leukocoria was the most common presenting feature in our cohort, occurring in 69.6% of patients, either alone or in association with proptosis. This finding is consistent with previous studies from South and Southeast Asia, where leukocoria has been reported as the predominant symptom in 60–75% of cases [15,18]]. However, a considerable proportion of children in our study presented with advanced signs such as proptosis (15.2%), pseudohypopyon (10.9%) and hyphema (4.3%), indicating delayed presentation. Similar advanced presentations have been documented in studies from the Democratic Republic of Congo and other resource-limited regions, where late diagnosis remains a major concern [19].

Stage at presentation is a critical determinant of treatment choice and prognosis. In this study, the majority of patients (60.9%) presented with Stage II disease, while 19.6% each presented with Stage I and Stage III disease. The absence of Stage IV disease in our cohort may reflect referral patterns or early mortality before presentation. Comparable distributions with a predominance of intermediate to advanced intraocular stages have been reported by Lavaju et al. and Bonanomi et al., underscoring the persistent challenge of early detection [13, 20]. According to Dimaras et al. and Houston et al., late-stage presentation significantly reduces the likelihood of globe salvage and necessitates more aggressive treatment [21, 22].

Treatment modalities in this study largely reflected disease severity at diagnosis. Enucleation combined with radiotherapy was the most common treatment (26.1%), followed by enucleation alone (21.7%) and chemotherapy (19.6%). These findings are consistent with treatment patterns described by Meel et al. and Lin and O'Brien, where enucleation remains a cornerstone of management for advanced intraocular disease [23, 24]. The relatively high rate of enucleation in our study highlights the limited scope for conservative, globe-sparing therapies in late-presenting cases. Radiotherapy alone (17.4%) and enucleation with cryotherapy (8.7%) were used selectively, depending on tumor extent and response.

Loss to follow-up was observed in 6.5% of patients, a finding comparable to reports from other low-resource settings where treatment abandonment remains a challenge [25]. This emphasizes the need for improved counseling, social support and follow-up systems to ensure continuity of care.

5. Limitations of the Study

This study has several limitations that should be acknowledged. The hospital-based descriptive design and relatively small sample size limit the generalizability of the findings to the broader population. In addition, detailed long-term outcomes such as survival rates, globe salvage and visual acuity could not be systematically analyzed due to variable follow-up and loss to follow-up in a small proportion of patients.

6. Conclusion

Retinoblastoma in children commonly presented at an early age, with leukocoria as the most frequent clinical feature and a majority of patients diagnosed at intermediate stages. Treatment was largely influenced by disease stage at presentation, resulting in a high rate of enucleation and combined modality therapy. Early detection, timely referral and improved awareness are essential to facilitate globe-preserving treatments and improve overall outcomes in affected children.

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