ISSN: 2581 – 3633
PubMed - National Library of Medicine - ID: 101745081

International Journal of Medical Science and Advanced Clinical Research (IJMACR)

Available Online at:www.ijmacr.com

Volume – 8, Issue – 4, August - 2025, Page No.: 200 – 206

Spectrum of Hemophilia in Children: A Cross-Sectional Study from A Tertiary Care Unit in Western India

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How to citation this article: Dr. Satyawan S. More, "Spectrum of Hemophilia in Children: A Cross-Sectional Study from A Tertiary Care Unit in Western India", IJMACR- August - 2025, Volume – 8, Issue - 4, P. No. 200 – 206.

Type of Publication: Original Research Article

Conflicts of Interest: Nil

Abstract

Background: Hemophilia is a rare, X-linked bleeding disorder, predominantly affecting males, caused by deficiencies of clotting factors. The disease imposes significant morbidity, especially in resource-limited settings. Diagnosis and management challenges, especially in rural areas, contribute to delayed care and adverse outcomes in Indian children.

Objectives: To describe the clinico-epidemiological profile, bleeding patterns, complications, and challenges in management of children with hemophilia admitted to a tertiary care hospital in Western India, aiming to inform improved strategies for care.

Methodology: This cross-sectional hospital-based study was conducted at CPR Hospital, Kolhapur, from June 2024 to June 2025. All children below 12 years, diagnosed with hemophilia and admitted for bleeding episodes or complications, were included following informed consent. Demographic, clinical, and laboratory data regarding disease severity, age at diagnosis, family history, bleeding patterns, joint involvement, complications, and inhibitor status were collected and analyzed descriptively using SPSS.

Results: A total of 45 children were included; all had Hemophilia A. Severe disease predominated (64.4%). Most were aged 5–10 years (51.1%) at admission. Rural residence (64.4%) was common, and family history was present in 46.7%. At first diagnosis, bruising (37.8%) and gum bleed (22.2%) were common presenting symptoms, whereas hemarthrosis (51.1%) predominated at admission. Bleeding episodes were more often traumatic (60%). The major joints affected were the knees (62.2%), elbows (46.7%), and ankles (42.2%). Complications included intracranial hemorrhage (15.6%) and joint movement limitation (6.7%). Inhibitor testing showed 31.3% positivity, complicating management. Most children had 1–5 bleeding episodes annually; the majority had no bleeding after vaccination.

Conclusion: Hemophilia A in Western Indian children shows predominance of severe disease, frequent joint bleeds, diagnostic delays, rural-urban disparities, and high inhibitor prevalence. Early diagnosis, enhanced

rural care, regular inhibitor screening, and improved factor availability are essential for optimizing outcomes.

Keywords: Hemophilia A, children, epidemiology, inhibitors, joint bleeds

Introduction

Hemophilia is a rare, X-linked recessive disorder characterized by deficiencies in clotting factors, leading to a lifelong propensity for bleeding. Hemophilia A (factor VIII deficiency) and Hemophilia B (factor IX deficiency) account for the majority of cases, with Hemophilia A being approximately five times more common. [1] Global estimates suggest that over 800,000 people live with hemophilia, yet a significant number, particularly in developing nations, remain undiagnosed due to inadequate screening and healthcare infrastructure. [2] In India, the Hemophilia Federation estimates a patient population of nearly 70,000, but only about 21,800 are registered, highlighting a massive gap in identification and management. [3]

The disease imposes a severe physical, psychological, and financial burden on patients and their families. Management primarily relies on replacement therapy with clotting factor concentrates (CFCs), which is costly and often inaccessible.[4] A major complication of treatment is the development of neutralizing antibodies (inhibitors), which renders standard factor replacement ineffective and complicates management.[5] Prophylactic therapy, aimed at preventing bleeding episodes, is standard of care in developed nations and has been shown to preserve joint function and improve quality of life. However, in India, most patients still receive on-demand therapy due to economic constraints and limited availability of factors. [6]

Early diagnosis and a comprehensive care approach are crucial for improving outcomes. Unfortunately, a lack of awareness among primary care physicians and the community often leads to delayed diagnosis, frequently after a significant bleeding event.[7] This study was undertaken to describe the clinical and epidemiological profile of children with hemophilia admitted to a tertiary care hospital in Western India, to better understand the local challenges and inform strategies for improved care.

Material and Methods

This hospital-based cross-sectional study was conducted in the Department of Pediatrics at CPR Hospital, Kolhapur, A tertiary care center in Western India. The study duration was from June 2024 to June 2025. All children aged below 12 years, either previously diagnosed with hemophilia or newly diagnosed during the study period, who were admitted to the pediatric ward for management of a bleeding episode or related complications, were included in the study. Children whose parents did not provide informed consent were excluded.

After obtaining ethical clearance from the Institutional Ethics Committee and written informed consent from parents, detailed history and clinical examination were performed. Data regarding demographic details, age at diagnosis, family history, Severity of hemophilia, presenting bleeding manifestations, number of bleeding episodes per year, joints involved, complications, and inhibitor status were recorded on a pre-designed proforma. The severity of hemophilia was classified based on factor VIII levels: severe (<1 IU/dL), moderate (1-5 IU/dL), and mild (>5 IU/dL).[8] Data were compiled in Microsoft Excel and analyzed using SPSS software version 26. Descriptive statistics were used to represent the data.

Results

A total of 45 children with hemophilia were included in the study. All patients (100%) were diagnosed with Hemophilia A. The majority of children (64.4%) had severe hemophilia.

Table 1: Demographic and Baseline Characteristics of the Study Population (N=45)

Demographic and Baseline Characteristics		n	(%)
Severity of Hemophilia	Mild (>5 IU/dL)	8	17.8
	Moderate (1-5 IU/dL)	8	17.8
	Severe (<1 IU/dL)	29	64.4
Age at Admission (years)	<1	2	4.4
	1-4	7	15.6
	5-10	23	51.1
	>10	13	28.9
Age at Diagnosis	<6 months	14	31.1
	6 months - 1 year	10	22.2
	1-4 years	18	40.0
	5-10 years	3	6.7
Residence	Urban	16	35.6
	Rural	29	64.4
Family History	Negative	24	53.3
	Positive	21	46.7
If Positive:	Sibling	6	28.6
	Maternal Uncle	5	23.8
	Grandfather	5	23.8
	Cousin	3	14.3
	Grandfather + Sibling	2	9.5
Total		45	100.0

There was predominance of severe Hemophilia A, affecting nearly two-thirds of the patients (64.4%), while mild and moderate types were less common (each 17.8%). The age distribution at admission shows that the study primarily from age group 5-10 years old (51.1%), followed by those over 10 years (28.9%). This suggests that the condition leads to multiple hospitalizations throughout childhood. The age at first diagnosis is a critical finding: while 31.1% were diagnosed very early (before 6 months), a significant 40.0% were not diagnosed until they were between 1-4 years old, indicating potential delays in recognition.

A majority of patients (64.4%) came from rural areas, highlighting potential disparities in healthcare access. A positive family history was a strong feature, present in 46.7% of cases. Among these, the most frequently affected relatives were siblings and maternal uncles

(each 23.8%), which is consistent with the X-linked inheritance pattern.

Table 2: Clinical Presentation and Bleeding Patterns (N=45):

Clinical Presentation		n	(%)
Bleeding Type at Admission	Hemarthrosis	23	51.1
	Bruises/Ecchymosis	14	31.1
	Intracranial Hemorrhage	4	8.9
	Gum Bleed	2	4.4
	Hematuria	1	2.2
	Melena	1	2.2
Bleeding Type at 1st Diagnosis	Bruises/Ecchymosis	17	37.8
	Gum Bleed	10	22.2
	Hemarthrosis	6	13.3
	Scalp Hematoma	4	8.9
	Tongue Bite	3	6.7
	Intracranial Hemorrhage	3	6.7
	Epistaxis	2	4.4
Onset of Bleeding	Traumatic	27	60.0
	Spontaneous	18	40.0
Bleeding Episodes/Year	1 to 5	31	68.9
	5 to 10	9	20.0
	>10	3	6.7
	<1	2	4.4
Bleeding during Vaccination	Yes	10	22.2
	No	35	77.8
Total		45	100.0

The primary reason for hospital admission was hemarthrosis (51.1%), underscoring the debilitating impact of joint bleeds on quality of life and the need for acute care. This is contrasted with the initial symptoms that led to diagnosis; the first sign of a problem was most often bruises/ecchymosis (37.8%) or gum bleeds (22.2%), which are less specific and may be missed. Only 13.3% presented with hemarthrosis at first diagnosis. The trigger for bleeding episodes was more often traumatic (60.0%) than spontaneous, though a substantial 40.0% experienced spontaneous bleeds, a hallmark of severe disease. The annual bleeding rate was manageable for most, with 68.9% experiencing a relatively low frequency of 1 to 5 episodes per year. Reassuringly, the vast majority (77.8%) did not experience bleeding after vaccination, which is crucial information for reassuring parents and ensuring immunization compliance.

Table 3: Joint Involvement and Complications (N=45):

Joint Involvement and Complications		Frequency	Percent
Joints Involved	Knee	28	62.2
	Elbow	21	46.7
	Ankle	19	42.2
	Shoulder	6	13.3
	Hip	5	11.1
	Wrist	1	2.2
Complications	Intracranial Hemorrhage	7	15.6
	Limitation of Joint Movement	3	6.7
	Compartment Syndrome	1	2.2
Inhibitors (Tested n=16)	Positive	5	31.3*
	Negative	11	68.7*
Total		45	100.0

The knee was overwhelmingly the most common target joint, affected in 62.2% of patients, followed by the elbow (46.7%) and ankle (42.2%). This pattern aligns with known literature and points to the weight-bearing joints being most vulnerable. The most severe complication was intracranial hemorrhage (ICH), which occurred in a concerning 15.6% of the children, a life-threatening event that represents a major cause of morbidity and mortality.

A notably high proportion of those tested for inhibitors (31.3%) were positive. This is a critical finding as the development of inhibitors is one of the most challenging complications in hemophilia care, rendering standard clotting factor treatments ineffective and significantly increasing treatment complexity and cost.

Discussion

This cross-sectional study provides a detailed overview of the clinico epidemiological profile of 45 children with hemophilia A from a tertiary care center in Western India. Our findings highlight several critical aspects of the disease burden in this region, some of which align with national data while others present distinct challenges.

The overwhelming predominance of severe hemophilia (64.4%) in our cohort is consistent with several Indian studies, which typically report a higher proportion of severe cases in hospital-based settings. A study by John et al. from Kerala reported that 63% of their cohort had severe hemophilia, nearly identical to our finding[8]. Similarly, Singh et al. documented that 70% of their patients in a North Indian tertiary center presented with the severe form of the disease [6]. This is often attributed to a referral bias, as children with severe disease experience more frequent and serious bleeding episodes, necessitating hospitalization. In contrast, a study from Eluru, Andhra Pradesh, by Pusapati LCV et al, reported a much lower proportion of severe cases (12%) with a majority of mild cases (49%), suggesting possible regional variations in disease severity or, more likely, significant under-diagnosis of mild cases in the community, which do not present to tertiary hospitals [9].

The mean age at diagnosis in our study was 11 months, with 40% of children diagnosed between 1-4 years of age, indicating a considerable diagnostic delay. This finding is corroborated by a study from Bangladesh by Islam et al., where the mean age at diagnosis was 3 years, and a large number of patients were diagnosed after the age of 2[10]. John et al. also reported a mean presentation age of 5 years[8]. This delay can be attributed to a lack of awareness among parents and primary care physicians, who may dismiss early signs like bruising (37.8% at first diagnosis in our study) as normal childhood trauma. The higher proportion of early diagnosis (<6 months) in our study (31.1%) compared to others is likely due to a few cases presenting with catastrophic bleeding like intracranial hemorrhage (ICH) in infancy, which prompts immediate investigation.

A significant finding was the high burden of patients from rural areas (64.4%), underscoring the healthcare accessibility issues in India. The Hemophilia Federation India (HFI) annual report consistently notes that over 80% of registered patients face challenges in accessing adequate care, a figure that is even more pronounced for those in rural and remote locations [3]. The high rate of inhibitor development (31.3% among those tested) is another alarming finding. This is substantially higher than the often-cited global average of 25-30% for severe hemophilia A patient but aligns with a multi-center Indian study by Pinto et al., which reported an overall inhibitor prevalence of 6% in Hemophilia A, with a notably higher incidence of 13% in Southern India [14]. This elevated risk in the Indian population could be linked to genetic predispositions and, critically, to intermittent, on-demand treatment patterns due to factor non-availability, which is a known risk factor for inhibitor development.

The pattern of joint involvement, with the knee (62.2%), elbow (46.7%), and ankle (42.2%) being the most common target joints, is universally reported and matches findings from study by Razaq M et al and Payal V et al in Uttar Pradesh and other parts of India [12,13]. However, the disturbingly high rate of intracranial hemorrhage (15.6%) as a complication in our study is a serious concern. While a systematic review by Davies et al. estimates the rate of ICH in neonates with hemophilia to be between 2-4%, the lifetime risk is higher[15]. Our finding suggests that the lack of prophylactic therapy and delayed treatment might be contributing to a higher incidence of this life-threatening complication in our population.

Conclusion

This study concludes that hemophilia A in Western Indian children is predominantly severe, with frequent joint bleeds and a high incidence of intracranial hemorrhage. Significant diagnostic delays and a rural-urban care disparity are evident. The substantial prevalence of inhibitors further complicates management.

Therefore, strategies must prioritize early diagnosis through heightened awareness, improved access to clotting factors and prophylactic therapy in rural areas, and systematic inhibitor screening to optimize outcomes and reduce lifelong disability. Implement newborn screening for high-risk families, enhance rural access to clotting factors and prophylaxis, and mandate routine inhibitor screening to improve comprehensive hemophilia care.

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