Original Article

Unveiling the Hidden Struggles of Hemophilia: A Comprehensive Cross-Sectional Study in South-India

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Abstract

Background: Hemophilia is a rare genetic bleeding disorder characterized by delayed blood clotting due to deficiencies in clotting factors. Despite advancements in treatment, hemophilia poses significant health and social challenges for patients.

Objective: This study aims to investigate the profile and consequences of hemophilia among patients in Mangalore, India, focusing on their awareness, treatment, and quality of life.

Methods: A cross-sectional study was conducted among hemophilic patients attending Government Wenlock District Hospital, KMC Hospital Attavar, and members of the Hemophilia Federation (India). Data was collected using a semi-structured questionnaire and analyzed using SPSS version 25.0.

Results: The study included 69 male participants aged 20-58 years. Most participants (81.2%) were diagnosed within the first year of life. Hemophilia A was more prevalent (81.2%) than Hemophilia B (18.8%). Bleeding episodes occurred once or twice a month for most participants (84%). Regular exercise was reported by 76.8% of participants, and 10.1% reported physical impairment due to hemophilia. Blood transfusions were common (26.1%), with 5.8% affected by Hepatitis C due to transfusions. Preventive prophylaxis was used by 60.9% of participants when required. Satisfaction with welfare centers was neutral to fairly satisfied for most participants.

Conclusion: Hemophilia significantly impacts the lives of patients, highlighting the need for improved awareness, treatment accessibility, and support systems to enhance their quality of life.

Keyword: Hemophilia, clotting factors, bleeding episodes

Introduction

Hemophilia is a rare, X-linked recessive genetic disorder resulting in delayed blood clotting due to deficiencies in clotting factors VIII (Hemophilia A)

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or IX (Hemophilia B)[1]. Hemophilia C, caused by a deficiency of factor XI, and parahemophilia, due to factor V deficiency, are less common variants[2]. This notoriety has brought hemophilia into public awareness, but it remains a misunderstood condition with significant health and social implications.

The incidence of hemophilia varies worldwide, with estimates suggesting that approximately 1 in 5,000 male births are affected by Hemophilia A, and 1 in 30,000 by Hemophilia B[3]. These figures underscore the rarity of the condition and the challenges in achieving widespread awareness and understanding. In India, the prevalence is believed to be lower, possibly due to underdiagnosis and lack of comprehensive data collection systems[4].

Hemophilia's clinical manifestations are characterized by spontaneous or prolonged bleeding, particularly into joints and muscles, leading to pain, swelling, and, if untreated, chronic joint disease and disability. The historical risk of blood-borne infections, such as HIV and Hepatitis C, from contaminated blood products has been a significant concern[5]. While modern treatments and stringent blood screening have mitigated these risks, they have not eliminated them entirely, especially in resource-limited settings.

Hemophilia's recognition as a disability under the 'Rights of Persons with Disabilities Act 2016' in India[6] marked a significant step toward improving the lives of affected individuals. This legislation provides legal protections and entitlements, including reservation in government jobs and access to disability benefits. However, challenges remain in ensuring that all patients benefit from these provisions, given the variability in healthcare access and quality across the country.

This study aims to examine the demographic profile, awareness, treatment practices, and social implications of hemophilia among patients in Mangalore, India. By understanding these factors, we hope to highlight areas for improvement in patient care and support, contributing to better health outcomes and quality of life for individuals with hemophilia.

Methods

A cross-sectional study was conducted among known hemophilic patients attending Government Wenlock District Hospital, KMC Hospital Attavar, and members of the Hemophilia Federation (India). The study included all patients who met the inclusion criteria: patients with hemophilia attending the specified hospitals and federation, aged 18 years or older, irrespective of their treatment status. Patients who were unwilling to participate were excluded from the study. A convenience sampling technique was used to select participants for this time-bound study.

After obtaining institutional ethics committee approval (IEC KMC MLR 04-19/202) and necessary hospital permissions, data collection commenced. A pretested semi-structured questionnaire, administered via Google Forms for federation members, was used to gather data. The questionnaire comprised of two parts: demographic details (Part A) and haemophilia-related questions (Part B). The data collected from the questionnaires were entered and analysed using SPSS version 25.0.

The questionnaire was designed to capture a comprehensive range of information, including demographic details (age, education, occupation), hemophilia diagnosis and type, frequency and severity of bleeding episodes, physical activity levels, complications arising from hemophilia (such as infections from blood transfusions), and awareness of treatment options and preventive measures. Participants were also asked about their satisfaction with the healthcare services they received and the impact of hemophilia on their social and economic lives.

Results

The results of this study are presented in a series of tables that summarize the demographic details, frequency of bleeding episodes, exercise habits, complications, awareness of hemophilia-related issues, and satisfaction with healthcare services among the participants.

The table no 1 shows that the majority of participants (76.8%) were aged between 20-40 years, with a significant proportion having attained higher education, primarily graduates (82.6%). This suggests a relatively young and educated hemophilic population in the study area.

The table no: 2 depicts, Hemophilia A is more in proportion (81.2%) than Hemophilia B (18.8%). Most participants (81.2%) were diagnosed within the first year of life, indicating early detection and diagnosis of the condition.The majority of participants (84%) experience bleeding episodes 1-2 times per month, highlighting the chronic and recurring nature of the condition.Most participants (91.3%) reported no immobilization days due to bleeding episodes, with a median of 0 days and an Inter quartile range (IQR) of 0 to 0 days, indicating effective management of the condition for the majority. A small percentage (8.6%) experienced immobilization for 2-7 days. More than half of the participants (55.1%) engaged in regular exercise for 2-5 hours per week, indicating a positive trend towards maintaining physical fitness despite their condition.Joint swelling is a common issue, with nearly half of the participants (49.3%) experiencing it 1-2 times per month, and a significant portion (44.9%) not experiencing any swelling.

The table no: 3 depicts that the high levels of awareness about carrier detection (91.3%) and prenatal diagnosis (91.3%) were observed. Prophylaxis utilization was noted in 60.9% of participants.

As shown in the table no 4, that the satisfaction with welfare centers had a median score of 3 (neutral) with an IQR of 3 to 4 (neutral to fairly satisfied), indicating general contentment but also room for improvement.

The table no 5 portrays that median impact on education and financial status was 0, with no significant financial impact reported. Social acceptance issues and employment denial had a notable presence, highlighting ongoing social challenges.

Table 1: Demographic Characteristics of Participants (n=69)

Characteristics	Frequency	Percentage (%)
Age (years)		
20-30	26	37.7
31-40	27	39.1
41-50	10	14.5
51-60	6	8.7
Education level		
Higher secondary	6	8.7
Graduate	57	82.6
Post graduate	6	8.7
Residence		
Urban	47	68.1
Rural	22	31.9

Table 2: Profile of hemophilia Type and diagnosis (n=69)

Items	Frequency	Percentage (%)
Haemophilia Type		
A	56	81.2
B	13	18.8
Diagnosing age		
Median (IQR)	0.5 (0.3-1.0)	
Within 1 year	56	81.2
After 1 year	13	18.8
Frequency of bleeding episodes per month		
1-2 times	58	84.0
3-4 times	10	14.5
≥5 times	1	1.4
Days of Immobilization		
None	63	91.3
2 to 3 days	3	4.3
4-7 days	3	4.3
Frequency of Joint Swelling per Month		
None	31	44.9
1-2 times	34	49.3
3-4 times	04	5.8

Table 3: Awareness of Hemophilia Management*

Awareness type	Frequency
Carrier detection	63
Pre natal diagnosis	63
Home remedies (RICE#)	59
Prophylaxis awareness	67
Prophylaxis utilization	42

*more than one option was selected

Rest, Ice, Compression and Elevation (RICE)

Table 4: Satisfaction with Welfare Centers (n=69)

Satisfaction level	Frequency	Percentage	Median (IQR)
Not satisfied	1	1.4	3 (3-4)
Insufficiently satisfied	6	8.7	
Neutral	39	56.5	
Fairly satisfied	20	29.0	
Strongly satisfied	3	4.3	

Table 5: Social and Educational Impact of Hemophilia

Impact type	Yes (n=69)	No (n=69)	Sometimes (n=69)
Physical impact	7	62	--
Social acceptance issues	7	44	18
Educational impact	2	67	--
Financial status impact	0	69	--
Employment denial	11	58	--

Discussion

The findings of this study provide a comprehensive understanding of the profile and consequences of hemophilia among patients in Mangalore, India, and highlight several key aspects when compared to previous studies.

Demographic Characteristics

The mean age of participants in this study was 34.2 years with a standard deviation of 9.1 years, indicating a relatively young population. This is consistent with global studies, such as the one by Stonebraker et al. (2010), which also reported a young demographic profile for hemophilia patients[7]even among the wealthiest of countries. The prevalence (per 100 000 males. The high level of education, with 82.6% of participants being graduates, is noteworthy and contrasts with some studies that suggest lower educational attainment due to chronic health conditions. This suggests that despite the challenges posed by hemophilia, many patients in this study area have managed to achieve significant educational milestones.

Hemophilia Type and Diagnosis

Hemophilia A was more prevalent (81.2%) than Hemophilia B (18.8%), aligning with global data, including the World Federation of Hemophilia's annual survey, which consistently shows a higher prevalence of Hemophilia A. The median age at diagnosis in this study was 0.5 years (IQR 0.3-1.0), indicating early diagnosis, which is crucial for effective management and aligns with the findings of Soucie et al.[8] in the United States, who also reported early diagnosis in most cases.

Frequency of Bleeding Episodes

Participants in this study reported a median of 2 bleeding episodes per month (IQR 1-2). This is comparable to the findings of Srivastava et al.[9], who reported similar frequencies, highlighting the chronic and recurring nature of the condition. The consistency in the frequency of bleeding episodes across different studies underscores the importance of regular and effective management strategies for hemophilia.

Physical Impairment Due to Bleeding Episodes

Most participants (91.3%) reported no immobilization days due to bleeding episodes, with a median of 0 days (IQR 0-0). This suggests effective management and contrasts with older studies(3), which reported higher rates of physical impairment. The improvement may be attributed to better prophylactic treatments and healthcare practices that have been implemented over the past few decades.

Exercise Habits

Regular exercise was reported by more than half of the participants, with a median of 5 hours per week

(IQR 3-6). This aligns with the recommendations by the World Federation of Hemophilia[10], which emphasize the benefits of physical activity in maintaining joint health and overall well-being. The fact that a significant portion of participants engage in regular exercise is a positive indicator of adherence to recommended health practices.

Awareness of Hemophilia Management

High levels of awareness were observed in this study, with 91.3% aware of carrier detection and prenatal diagnosis, and 97.1% aware of prophylaxis. This is in line with the findings of Padke[11] who reported high levels of awareness and knowledge about hemophilia management in urban settings. However, the actual utilization of prophylaxis at 60.9% indicates a gap between awareness and practice, similar to findings by Fischer et al. (2013), who noted that while awareness is high, consistent utilization of prophylactic treatment remains a challenge.

Satisfaction with Welfare Centers

The level of satisfaction with welfare centers was generally positive, with a median satisfaction score of 3 (neutral) and an IQR of 3 to 4 (neutral to fairly satisfied). This aligns with the findings of Brenda Riske[12]associated with adherence and better outcomes. However, satisfaction with US Hemophilia Treatment Centers (HTC, who reported varying levels of satisfaction with hemophilia care services, suggesting that while basic needs are met, there is room for improvement in service delivery and patient support.

Social and Educational Impact

The study found minimal impact on education and no significant financial status impact, with only 2.9% reporting educational impairment and none reporting financial impact. However, 10.1% faced social acceptance issues and 15.9% reported employment denial, indicating ongoing social challenges. These findings highlighted the social stigmas and employment challenges faced by individuals with hemophilia, despite advancements in medical care.

Conclusion

This study provides valuable insights into the current status of hemophilia patients in Mangalore, India. The findings highlight improvements in early diagnosis, education levels, and effective management of bleeding episodes. However, there are still gaps in the utilization of prophylactic treatments and social acceptance. Continued efforts are necessary to address these challenges and ensure comprehensive care and support for individuals with hemophilia. Further research with larger, more diverse populations is recommended to validate these findings and guide future healthcare policies and practices.

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