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# STUDY OF LABORATORY PARAMETERS IN HEMOPHILIA PATIENTS IN A TERTIARY CARE CENTER : STUDY OF 86 CASES

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**ABSTRACT** **Background:** Hemophilia is an X-linked congenital bleeding disorder caused by deficiency of factor VIII (hemophilia A) or factor IX (hemophilia B). India carries a high burden of hemophilia. Bleeding into soft tissues, muscles and joints is the clinical hallmark of hemophilia. laboratory evaluation of is crucial for confirmation and grading of severity **Methods:** This cross-sectional observational study was conducted at a tertiary care hemophilia center from September 2018 to September 2020. A total of 86 patients were evaluated. Coagulation tests included prothrombin time (PT), activated partial thromboplastin time (aPTT), and factor VIIIand IX assays. **Results:** Of 86 cases, 75 (87.2%) had hemophilia A and 11 (12.8%) had hemophilia B. Almost all patients were male. aPTT was prolonged in 100% of patients tested in both groups, whereas PT remained within normal limits in all tested cases. According to Factor VIII and factor IX assays, hemophilia classify into mild, moderate and severe categories. **Conclusion:** Hemophilia A was more frequent than hemophilia B. Prolonged aPTT with normal PT was a consistent screening pattern in all patients, aPTT as a sensitive screening test. FactorVIII and IX assays are mandatory for definitive diagnosis and accurate grading of disease severity.

**KEYWORDS :** Hemophilia A,Hemophilia B,PT,APTT

## INTRODUCTION

Hemophilia is a congenital X- linked bleeding disorder. It is a coagulation disorder occurring either because of deficiency of factor VIII (Hemophilia-A) or deficiency of factor IX (Hemophilia-B)[1] affecting 1 in 5000 – 10,000 males for Hemophilia A and 1 in 25,000 – 30,000 males in hemophilia B[2]. India has the second highest burden of hemophilia patients in the world[3].

As factor VIII and IX genes are both located on the X chromosome, hemophilia primarily affecting males. and females carrying a hemophilia mutation on one of their two X chromosomes and act as carriers. About one third of hemophilia A cases arise from spontaneous mutations[4] Bleeding manifestations of hemophilia A and B, are hemarthrosis; soft tissue hematomas;easy bruising; excessive bleeding with surgery, trauma, dental extraction; bleeding in the gastrointestinal or genitourinary tract; and uncommonly umbilical stump bleeding.

Both the disorders (A & B) are indistinguishable clinically from each other as the signs and symptoms are same. Laboratory evaluation of patients with such a bleeding history should include an aPTT and PT. Diagnosis is confirmed by a FVIII or FIX assay. Replacement of the deficient factor is the mainstay of treatment; it may be "on demand" or "prophylactic" to prevent hemarthrosis in severe deficiency.

Accurate diagnosis is important and essential for effective management. It is dependent on laboratory following strict protocols and procedures which requires knowledge and expertise in coagulation laboratory testing and use of correct equipment and reagents[5]. Thus purpose of this study is to study various applicable laboratory parameters, effectiveness of various screening and confirmatory tests for diagnosis of hemophilia

## MATERIALS & METHODS

This was a hospital-based cross-sectional observational study carried out at hemophilia center of a tertiary care hospital from September 2018 to September 2020. It was The study was approved by the local ethical committee. Total 86 study cases were enrolled in this study.Written informed consent was obtained from adult patients and from parents or guardians in pediatric cases.

**Data collection:** All 86 samples collected in Tri sodium citrate 3.2 g% vacutainer (1:9) for coagulation studies (i.e PT, aPTT, Factor VIII, Factor IX) at hemophilia center. clinical details (age,sex,clinical features) were recorded.

PT and aPTT were performed using commercially available reagents (Diagnostica Stago®) on an ARX-CLOT semi-automated coagulometer. whileFactor VIII and IX assays were performed using one-stage clotting assays with calibrator plasma and internal quality controls at two levels (normal and pathological) run daily

**Reference range for PT: 11–14.5 seconds and aPTT: 28–38 seconds**
Severity of hemophilia is classified on the basis of plasma factor level deficit into 3 types –'[6]Mild (5-40%), Moderate (1-5%) and Severe (< 1%)

**Statistical Analysis:** Results were summarized using descriptive statistics and are presented as frequencies, percentages and mean values.

## RESULT:

A total of 86 patients with hemophilia were studied over the two-year period. Hemophilia – A75 (87.2%) is much more common than hemophilia – B. 11 (12.8%)(figure1)

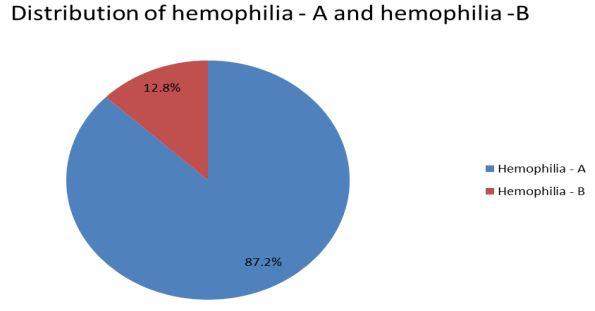


**Figure 1**
Among hemophilia A cases, 74 of 75 (98.67%) were male and A single female patient with hemophilia A was identified.

**Table 1:Age wise distribution in Hemophilia A and Hemophilia B**

| Age Group | Hemophilia A | Hemophilia B | Total cases | Percentage total |
|---|---|---|---|---|
| < 1 | 05 (6.6%) | 00 (0%) | 05 | 5.81% |
| 1 – 5 | 09 (12%) | 02 (18.2%) | 11 | 12.79% |
| 5 – 10 | 10 (13.4%) | 04 (36.4%) | 14 | 16.28% |
| 10 – 15 | 16 (21.3%) | 02 (18.2%) | 18 | 20.94% |
| 15 – 20 | 09 (12%) | 00 (0%) | 09 | 10.46% |
| 20 – 30 | 13 (17.3%) | 01 (9.1%) | 14 | 16.27% |
| 30 – 40 | 03 (4%) | 01 (9.1%) | 04 | 4.65% |
| > 40 | 10 (13.4%) | 01 (9.1%) | 11 | 12.8% |
| Total | 75 (100%) | 11 (100%) | 86 | 100% |

All 11 hemophilia B patients (100%) were male. In our study Age distribution was from 7 months to 66 years. (table 1)

Hemophilia A peaked in the 10–15 years age group, while Hemophilia B was most frequent in the 5–10 years group.

The study found a consistent correlation between hemophilia and elevated aPTT, while PT remained consistently normal. (table2)

**Table 2: APTT Distribution across Hemophilia**

| APTT Range (Seconds) | Hemophilia A Cases | Hemophilia B Cases |
|---|---|---|
| 40 – 60 | 28.3% | 18.2% |
| 60 – 80 | 26.6% | 36.4% |
| 80 – 100 | 28.4% | 36.3% |
| 100 | 11.7% | 9.1% |

According to Factor VIII and factor IX assays, confirmed the diagnosis of hemophilia A or B and allowed classification into mild, moderate and severe categories.

**Table 3: Severity Distribution in Hemophilia A and B**

| Factor level | Severity | Hemophilia A | Hemophilia B |
|---|---|---|---|
| 5 – 40% | Mild | 29 (38.66%) | 4 (36.37%) |
| 1 – 5% | Moderate | 14 (18.67%) | 2 (18.18%) |
| <1% | Severe | 32 (42.67%) | 5 (45.45%) |

## DISCUSSION

In this study, hemophilia A was considerably more frequent than hemophilia B,which is correlate with other similar studies, [6,7,8] in keeping with global and Indian data. Male predominance reflects the X-linked pattern of inheritance compatible with other studies[9,10]the occurrence of a single female patient with hemophilia A is unusual. Although genetic confirmation was not feasible, this have been attributed to skewed lyonization or homozygous mutations, underscoring the importance of genetic testing in atypical presentations[11]

The age distribution with clustering in childhood and adolescence is similar to other studies ([12,13] A relatively high proportion of patients in the severe category was observed in both hemophilia A and B, which may reflect referral bias to a tertiary care center and under-recognition of mild cases in the community.[6,12,14]

All patients tested in our series had prolonged aPTT with normal PT, In intrinsic pathway defects. aPTT as a highly sensitive screening test ,this has been compatible with other studies[7,12,13,14]. Normal PT helps to exclude combined intrinsic and extrinsic pathway defects and, when interpreted together with clinical features, strongly suggests hemophilia.

Specific factor VIII and IX assays remain the gold standard for diagnosis, allowing distinction between hemophilia A and B and enabling grading of severity. Early identification of the severity category is essential for planning prophylactic factor replacement and preventing joint damage and long-term disability. The clinical utility of this cohort lies in establishing region-specific laboratory patterns that can guide early diagnosis, appropriate severity stratification, and judicious use of factor concentrates. Although correlation with bleeding scores and imaging findings was not performed, the laboratory data provide a strong foundation for clinical decision-making in resource-limited settings.

## LIMITATIONS OF THE STUDY :

Single-center design and the absence of detailed correlation with clinical bleeding scores and imaging findings.

## CONCLUSION

Hemophilia is a lifelong congenital X-linked bleeding disorder predominantly affecting males. In this study hemophilia A was more common than hemophilia B. Prolonged aPTT with normal PT was a universal finding and represents a simple and effective screening pattern for hemophilia. Definitive diagnosis and severity assessment require factor VIII and IX assays, which guide appropriate replacement therapy. the study reinforces the pivotal role of basic coagulation tests in the diagnostic approach to suspected hemophilia in resource-limited settings and contributes valuable regional data that can support patient management strategies and inform broader public health planning for hemophilia. Strengthening coagulation laboratory services, improving awareness among clinicians, general public and ensuring timely referral to hemophilia centers are essential steps to reduce morbidity, disability and improve quality of life in patients with hemophilia

**Declaration by Authors**
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