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Clinical Profile of Haemophilia Patients at Govt. Royapettah Hospital

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Abstract:
Background *Haemophilia group of related bleeding disorders that are inherited. Inherited bleeding disorders include abnormalities of coagulation factors and platelet function.Our country has the second highest burden of haemophilia patients in the world.*
Methods: *This was an observational cross-sectional study .This study was done at Government Royapettah Hospital, Chennai for a period of six months. The study was performed after procuring informed written consent from all the participants involved.*
Results: *The mean age of the study population was 25.28yrs.All the patients under the study were male.Of the study population of 50, only 8% knew the carrier status of their mother. The rest of the 92% were not aware of their maternal carrier status. In the group of 50 haemophiliacs, 30% had no family history. 20% were not aware of their grandfather's or uncle's disease status. 42% had a positive family history either in their maternal grandfather or maternal uncle.Of the study population 80% were Haemophilia A and 64% - Severe Haemophilia A, 16% had significant joint deformities. The rest of the 84% had normal range of movements.aPTT analysis showed an abnormal value (>40seconds) in about 48% of the study population..Analysis showed that the weight bearing joints – knees and ankles were most commonly affected. Right knee bleed contributed to 22% of the total bleeds in Haemophilia patients. Left knee contributed to 24%*
Conclusion: *The severe type of haemophilia A was more common than mild and moderate types. The study suggest clinical profile of haemophiliac patients in our Government Royapettah hospital/KMCH*
Keywords: *Clinical profile, haemophilia,*

I. Introduction

Haemophilia a group of related bleeding disorders that are inherited. Inherited bleeding disorders include abnormalities of coagulation factors and platelet function; the most common of which is von Willebrand disease. However, when the term "haemophilia" is used, it most often refers to Haemophilia A and Haemophilia B. Our country has the second highest burden of haemophilia patients in the world. The pathophysiology of Haemophilia A and Haemophilia B is based on the insufficient generation of thrombin by the factorIXa/factor VIIIa complex through the intrinsic pathway of the coagulation cascade1. Bleeding may occur anywhere in patients with haemophilia. The most common sites are into joints and muscles and from the gastrointestinal tract. Approximately 80 percent of haemorrhage occurs in the joints; the ankles are most commonly affected in children, and the knees, elbows, and ankles in adolescents and adult. Spontaneous heamarthrosis are characteristic of severe disease. Haemophilia has an incidence of about 1 in 10000 in our country. Patients present with spontaneous bleeds or post traumatic bleeds depending on the severity of the disease. This group of patients are prone to develop chronic disability if under treated or untreated. Incidence of premature death is also high in this group of patients.

Discovery of the molecular structure of both factor VIII and factor IX has recently allowed the development of genetically engineered products, prepared using recombinant DNA technology. In the long term, Preparations presently prepared form human plasma may be superseded by recombinant clotting factor concentrates (a DNA). Nearly one third of cases of haemophilia occur with no preceding family history, possibly from new genetic mutation. When recorded family history is available, efforts should be made to identify female carriers. Identification depends on family history, measurement of clotting profile and DNA analysis.

Aims And Objectives: To study the clinical profile of haemophilia patients in Govt. Royapettah hospital/kmch age wise, sex wise, severity, mean age, joint involvement, family history

II. Materials And Methods

This study was done at Government Royapettah Hospital, Chennai for a period of six months. The study was performed after procuring informed written consent from all the participants involved.

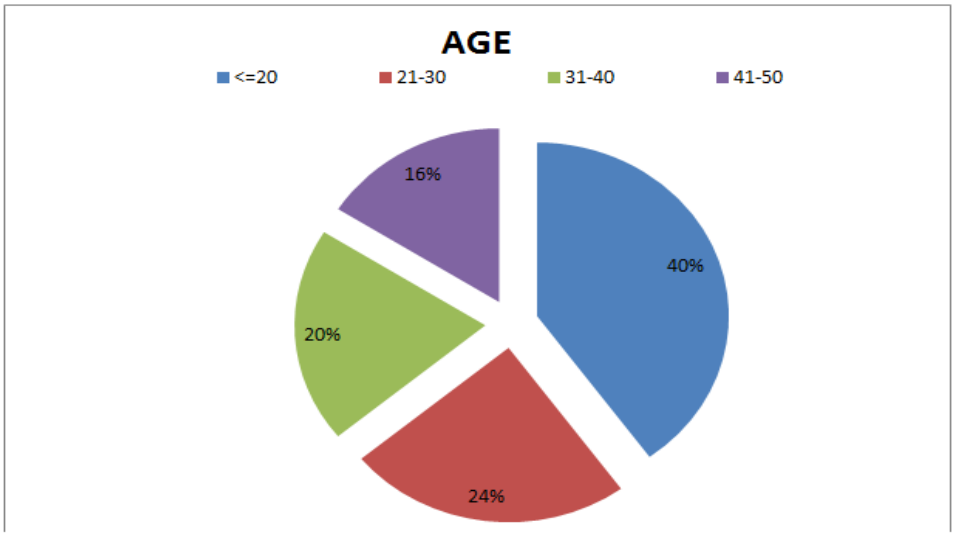
Study design:
The study design is a cross sectional study.
Population:
The study population included 50 patients who attended the Haemophilia OP at Government Royapettah Hospital and in-patients in the same hospital.
Inclusion criteria:
Patients diagnosed as Haemophilia and attending the Haemophilia OP.
Newly diagnosed in- patients in Government Royapettah Hospital.
Exclusion criteria:
The patients having factor deficiency other than factor VIII/IX and VWF were excluded from the study
Methodology:
All patients, diagnosed and registered in the Haemophilia clinic were taken as the study population. The sample size was set to be 50. A detailed history regarding the onset and progression of the disease, family history, maternal carrier status, treatment history and the presence of complications were taken.
After obtaining consent blood was drawn for investigations.
The following investigations were done:
- APTT
- BT
- CBC

III. Results

The data obtained was analysed using the SSPS software.

Age Distribution

Valid	Frequency	Percent	Valid Percent	Valid Percent
<= 20	20	40.0	40.0	40.0
21-30	12	24.0	24.0	64.0
31-40	10	20.0	20.0	84.0
41-50	8	16.0	16.0	100.0
Total	50	100.0	100.0	



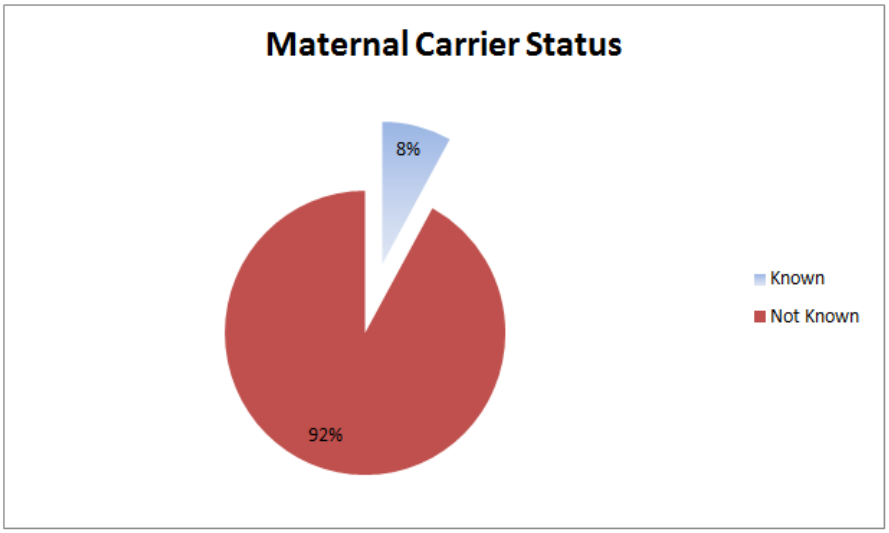
From the analysis, 40% of the patients were less than 20 yrs of age contributing to the highest percentage. 16% of the patients were between the age group of 41 to 50 yrs of age. The mean age of the study population was 25.28yrs.

Sex distribution:

All the patients under the study were male. Female haemophiliacs were not identified in the study population.

Carrier status distribution:

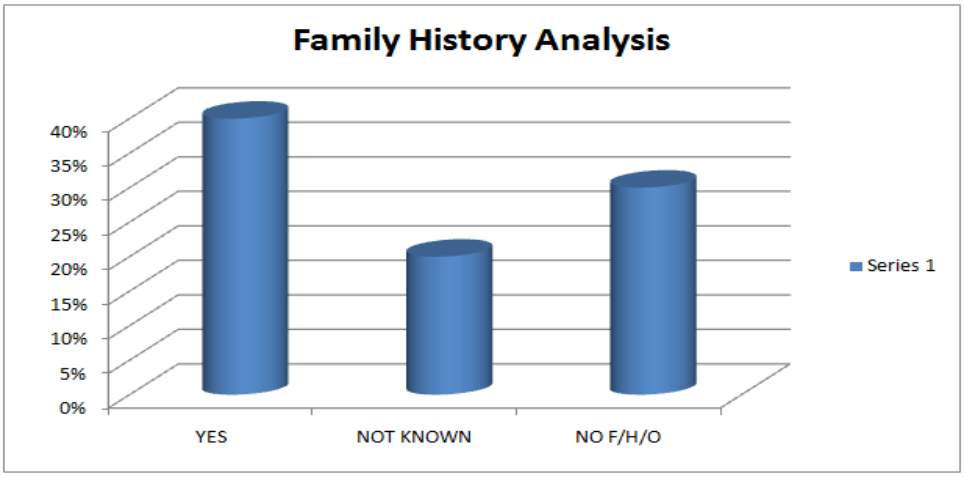
	Frequency	Percent	Valid Percent	Cumulative Percent
Valid Not Known	46	92.0	92.0	92.0
Carrier Positive	4	8.0	8.0	100.0
Total	50	100.0	100.0	



Of the study population of 50, only 8% knew the carrier status of their mother. The rest of the 92% were not aware of their maternal carrier status.

Family history analysis:

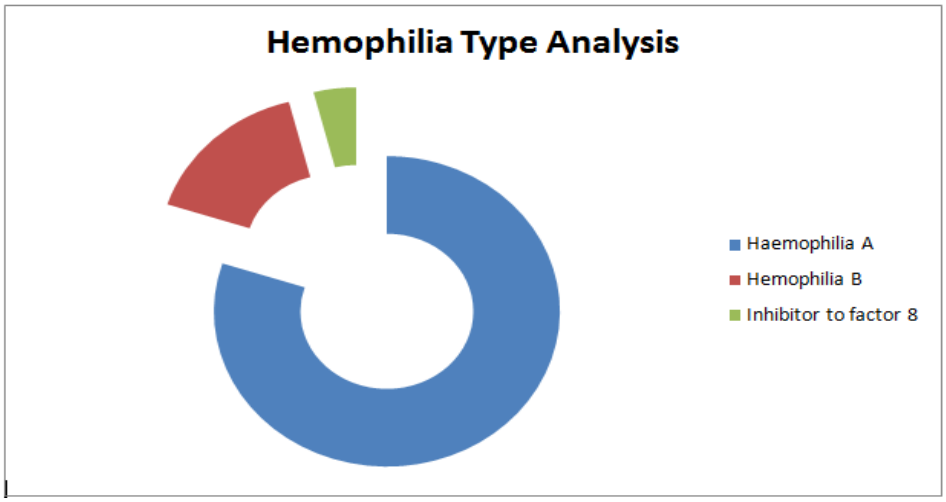
	Frequency	Percent	Valid Percent	Cumulative Perecent
Valid No	19.0	38.0	38.0	38.0
Not Known	10.0	20.0	20.0	58.0
Yes	21.0	42.0	42.0	100.0
Total	50.0	100.0	100.0	



In the group of 50 haemophiliacs, 30% had no family history. 20% were not aware of their grandfather's or uncle's disease status. 42% had a positive family history either in their maternal grandfather or maternal uncle.

Haemophilia type analysis:

		Frequency	Percent	Valid Percent	Cumulative Percent
Valid	A	40	80.0	80.0	80.0
	B	6	12.0	12.0	92.0
Inhibitor to Factor 8		4	8.0	8.0	100.0
Total		50	100.0	100.0	



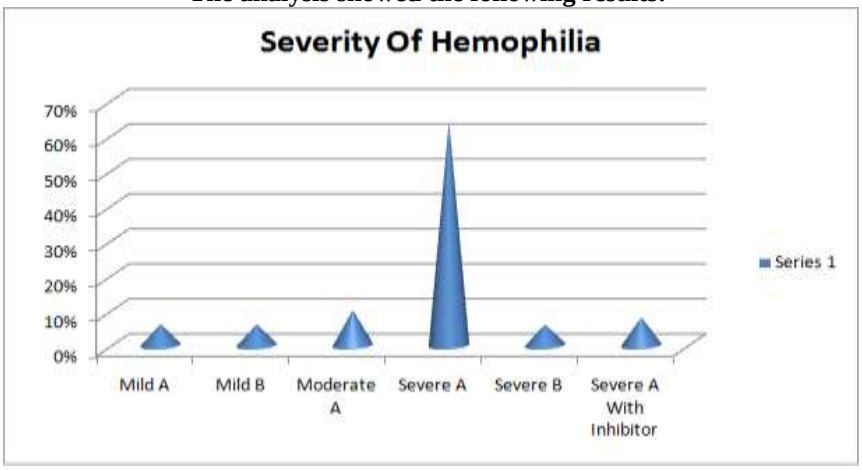
Of the study population

- 80% were Haemophilia A
- 16% were Haemophilia B
- 4% hadpositive inhibitors to Factor VIII and were severe haemophiliacs

Severity of Haemophilia:

		Frequency	Percent	Valid Percent	Cumulative Percent
Valid	Mild	3	6.0	6.0	6.0
	Mild B	3	6.0	6.0	12.0
	Moderate	5	10.0	10.0	22.0
	Severe	32	64.0	64.0	86.0
	Severe B	3	6.0	6.0	92.0
	Severe with Inhibitor	4	8.0	8.0	100.0
	Total	50	100.0	100.0	

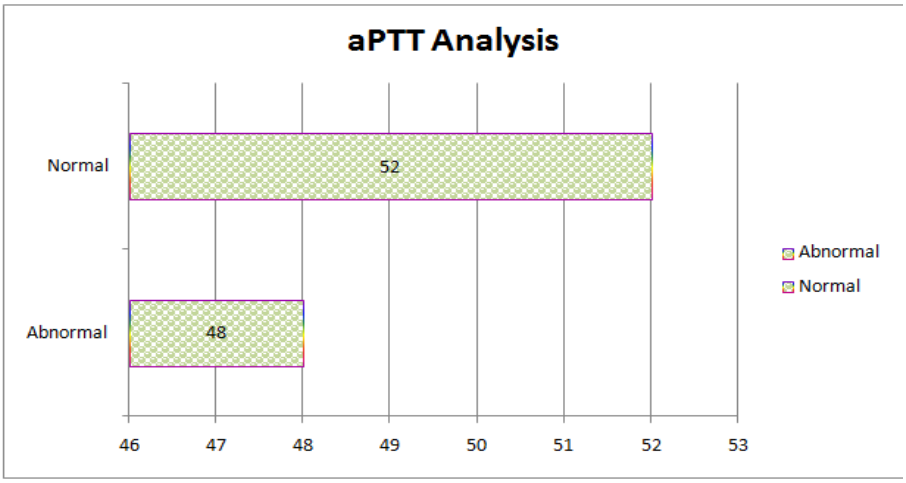
The analysis showed the following results:



- 6% Mild Haemophilia A
- 10% Moderate Haemophilia A
- 64% Severe Haemophilia A
- 8% Severe Haemophilia A with positive inhibitors
- 6% Mild Haemophilia B
- 6% Severe Haemophilia B

Ananlysis of aptt:

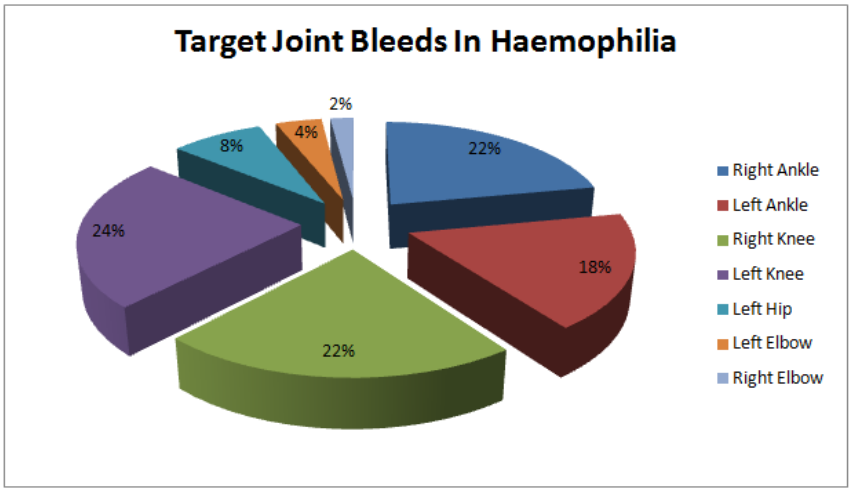
		Frequency	Percent	Valid Percent	Cumulative Percent
Valid	Normal	26	52.0	52.0	52.0
	Abnormal	24	48.0	48.0	100.0
	Total	50	100.0	100.0	



APTT analysis showed an abnormal value (>40seconds) in about 48% of the study population. The rest of the 52% had normal aPTT values. The prolonged aPTT signifies a clotting factor defect especially in the intrinsic and the common pathway.
The average aPTT is 54.20 seconds

Target joints:

Analysis showed that the weight bearing joints – knees and ankles weremost commonly affected. Right knee bleed contributed to 22% of the total bleeds in Haemophilia patients. Left knee contributed to 24% of the total joint bleeds in Haemophilia patients. Right ankle bleeds contributed to 22% and left ankle bleeds to 18% of the bleeding manifestations. This shows that weight bearing joints are more commonly affected. The other joints had a minor percentage contribution.



IV. Discussion

Prevelance And Global Stand

The analysis showed that the age group that formed the major group of patients was less than 20 yrs of age. This is probably due to the better diagnostic facilities available and better awareness among the general public and the carrier parents. Patients between 41 and 50 yrs of age contributed a less proportion of the total study population. Indian data from 2011 showed that there were 14,718 patients with bleeding disorder and of that 11,586 patients were HaemophiliaA. India reports the second largest number in patients with bleeding disorders and the third largest in those with HaemophiliaA

Estimated Number Of Patients

The prevalence of Haemophilia A in India is around 0.9 per 1,00,000 people. This low number of prevalence might reflect the under diagnosis, under reporting and early mortality of cases. When compared with the developed world the rate of case detection in India is 5 times lesser than that in the developed world.
Using the population data for 2011 from the Census of India15 and a prevalence of haemophilia A of 4 per 1,00,000, the estimated number of haemophilia patients in India would be around 48,4076 Haemophilia B has a lower prevalence rate of 0.1 per 1,00,000 population. This shows the need for better surveillance and case reporting in India. The estimated patients with HaemophiliaB would cross 20,000 in number. With this study incidence and prevalence of the study could not be estimated as its a cross sectional descriptive study.

Case Detection

India has reported the second maximum newly diagnosed cases in the world, in the last one year. However, studies for predicting the future trends in haemophilia in India is absent. With better laboratory services and better awareness among both the patients and the doctors, the number of newly diagnosed cases might increase. Observed estimated and real prevalence of Haemophilia patients in the various states of India and the union territories have been represented in the following pictorial representation.

Orthopedic Complications

In this study arthropathy was seen in 16% of the study population. However, in a study by Kar et al, prevalence of the disability has been estimated. According to this study, of 148 patients with severe Haemophilia A only 9 were free of disability. The orthopaedic complications were seen more in those who belonged to the lower socio economic group. Kar et al found the increased incidence of fractures in patients with severe Haemophilia on follow up. In this study follow up was not done and the socio economic status was not analysed. In this study only 16% had arthropathy but all the 16% were severe Haemophilia patients.

Inhibitors

Development of inhibitors to factor VIII concentrates is the most dreaded of complications. Factor VIII concentrate is neutralized by the inhibitors. These inhibitors are alloantibodies with high affinity. They belong to the IgG subclass. In this study, 8% were positive for inhibitors and all were severe Haemophilia with Factor VIII levels less than 1%. Studies in India showed that the prevalence of inhibitors were found to be 8.3 to 12% The treatment for inhibitors is Immune tolerance induction. In view of the expense involved in ITI treatment, initiating treatment plans in India is still under question.

Type Of Treatment

There are two types of treatment in Haemophilia– On demand and Prophylactic treatment. Though prophylactic treatment has a decreased incidence of joint complications, the cost of treatment per patient is high. Though India is the second largest in the number of Haemophilia patients our per capita usage of factor products was only 0.032 when compared to USA which had a per capita usage of 5.17.In view of the cost and the inaccessibility to Haemophilia treatment centres, Indian patients are still under on demand treatment. Prophylactic treatment is still under study in our population.

Joint Involvement

The weight bearing joints are more commonly affected in Haemophilia. This has been shown in the study by the greater percentage contribution of knee and ankle bleeds. The frequent joint bleeds cause damage and result in arthropathy that can severely affect the quality of patient's life. Most people tend to discontinue their job in view of their arthropathy thereby increasing the economic burden of the family.

V. Conclusion

Haemophilia is an inherited bleeding disorder with X linked inheritance. Majority of the Haemophilia are factor VIII deficient and have factor levels less than 1%. The genetic make up and the profile of the patients vary in the ethnic groups of India. Prevention of Haemophilic birth is limited in our country due to the absence of prenatal diagnostic facilities. Treatment facilities in our country need to be improved to make the factors available for the patients belonging to the lower socio economic group.