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A COMPARATIVE STUDY OF HEMOPHILIA A VERSUS HEMOPHILIA B IN GOVERNMENT MEDICAL COLLEGE, KADAPA

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Abstract

Introduction: Hemophilia is a genetic bleeding disorder caused by deficiency of clotting factor VIII (Hemophilia A) or Factor IX (Christmas disease) or Factor XI (Hemophilia C). Hemophilia is a X linked recessive trait with defective F8 an F9 genes in long arm of X chromosome. Hemophilia can also be acquired due to development of antibodies directed against the clotting factors. **Aim:** To compare the prevalence of Hemophilia A and Hemophilia B. **Materials and Methods:** The present study is done over a period of two years i.e., from 2016 to 2018. During the above period 130 cases were studied. Among these 112 cases (86.1%) were Hemophilia A and 18 cases (13.8%) were Hemophilia B. **Requirement of materials:** Capillary tubes, Blotting paper, Blood samples, Sprit and cotton. **Results:** In the present study we registered 130 cases of Hemophilia. Among these 112 cases (86.1%) were Hemophilia A and 18 cases (13.8%) were Hemophilia B.

Keywords: Blood sample, Hemophilia A, Hemophilia B, Kadapa

INTRODUCTION

Haemophilia A and B are rare bleeding disorders caused by mutations in the genes encoding coagulation factor VIII (FVIII) and factor IX (FIX). [1] Hemophilia is a X linked recessive trait with defective F8 an F9 genes in long arm of X chromosome. Hemophilia can also be acquired due to development of antibodies directed against the clotting factors.

As per the world federation of hemophilia (WFH) 2016 global survey, India harbors the highest number (18,353) of patients with hemophilia (PwH).[2] However, this represents a significant under diagnosis, as with a population of 1.32 billion and a prevalence of 1/10000 male births, and the expected number of PwH in India should be approximately 1, 32, 000. This indicates that the proportion of patients actually diagnosed is <15%.[3]

PwH have life-long history of bleeding leading to musculoskeletal (MSK) complications. Asymptomatic patients or PwH with mild disease may experience bleeding only with trauma or during surgery. Lack of treatment with clotting factor concentrates (CFC) results in progressive disability, especially in severely affected PwH.[3]

At present, there is limited published data on demographic details and clinical status of the PwH from Andhra Pradesh. The aim of the study is to identify age, sex wise prevalence of Hemophilia A and Hemophilia B.

MATERIALS AND METHODS:

The present study is done over a period of two years in Department of Pathology, Government Medical College, Kadapa i.e., from 2016 to 2018. During the above period 130 cases were studied. Among these 112 cases (86.1%) were Hemophilia A and 18 cases (13.8%) were Hemophilia B.

RESULTS

Total registered Hemophilia cases -130.

Table 1: Age wise prevalence of Hemophilia A

Age	Number of cases
0-10	39
11-20	30
21-30	19
31-40	10
41-50	9
51-60	5
TOTAL	**112**

Table 2: Age wise prevalence of Hemophilia B

Age	Number of cases
0-10	06
11-20	01
21-30	06
31-40	05
TOTAL	**18**

Table 3: Sex wise prevalence of Hemophilia A & B

Sex	Hemophilia A	Hemophilia B
MALE	106	10
FEMALE	06	08
TOTAL	**112**	**18**

Table 4: Clinical Presentation in Hemophilia A

No of cases	Clinical signs / symptoms	BT	CT	aPTT	Hb g/dL
56	Haemarthrosis and Muscle hematoma following trauma	18 minutes	21 seconds	52 seconds	5.0
43	Epistaxis & Abdominal wall hematoma	14 minutes	19 seconds	47 seconds	7.0
13	Epistaxis, Haematuria and oro - pharyngeal bleeding	12 minutes	18 seconds	42 seconds	8.0

(BT-Bleeding time, CT- Clotting time, aPTT (activated partial thromboplastin time), Hb (Haemoglobin)

Table 5: Clinical Presentation in Hemophilia B

No of cases	Clinical signs / symptoms	BT	CT	aPTT	Hb g/dL
10	Epistaxis & Haemarthrosis	15 minutes	20 seconds	44 seconds	6.0
08	Epistaxis & hematoma	14 minutes	18 seconds	41 seconds	5.5

(BT-Bleeding time, CT- Clotting time, aPTT (activated partial thromboplastin time), Hb (Haemoglobin)

Among these 112 cases (86.1%) were Hemophilia A and 18 cases (13.8%) were Hemophilia B. The maximum number of cases was seen in first decade of life in both the conditions (Table 1 & 2). The male to Female ratio in Hemophilia A is 17:1.5, and in Hemophilia B is 8:1 with Male predominance in both the conditions (Table 3). The commonest clinical presentations were epistaxis, haemarthrosis and hematoma (Table 4 & 5).

DISCUSSION

This study describes the prevalence and baseline characteristics of PwH from Andhra Pradesh. The maximum numbers of cases were seen in first decade of life in both the conditions between 0-10 year age group. A study done by Joseph John et al reported that in Punjab, Hemophilia A & B was common in 19-44 year age group.[3]

The male to Female ratio in Hemophilia A is 17:1.5, and in Hemophilia B is 8:1 with Male predominance in both the conditions. Hemophilia B is inherited in an X-linked manner. The risk to sibs of a proband depends on the carrier status of the mother. Carrier females have a 50% chance of transmitting the F9 pathogenic variant in each pregnancy. Sons who inherit the pathogenic variant will be affected; daughters who inherit the pathogenic variant are carriers. Affected males transmit the pathogenic variant to all of their daughters and none of their sons. Carrier testing for family members at risk and prenatal testing for pregnancies at increased risk are possible if the *F9* pathogenic variant has been identified in a family member or if informative intragenic linked markers have been identified.[4]

The commonest clinical presentations were epistaxis, haemarthrosis and hematoma. A study done by Joseph John et al reported that in Punjab approximately 90% of bleeding episodes involve the MSK system, and in 80% of cases, the joints are particularly affected.[5] In this study, the prevalence of joint deformity (restricted ROM and clinical evidence of synovial thickening) was 83.5% in those with severe disease and much lesser in moderate and mild PwH. This is due to non affordability for prophylaxis and inaccessible CFC supply.

A study by Ljung et al reported that of the presenting symptoms, subcutaneous bleedings constituted 41% while joint and muscle bleedings were uncommon; 16% were bleedings in conjunction with puncture of vessels, injections or surgery. Fourteen percent had anaemia and received blood-transfusion at diagnosis; 9% were diagnosed post-neonatally but 20% had shown abnormal bleeding tendency already in the neonatal period; seven boys (5%) had intracranial haemorrhages, five of them neonatally.[6]

Despite significant advances in the treatment of haemophilia, including availability of recombinant coagulation factor replacement products and the use of prophylactic infusion regimens, the segment of haemophilic patients who develop inhibitory antibodies remain at higher risk for morbidity and mortality associated with recurrent or uncontrolled bleeding events. Bypassing agents represent the mainstay of treatment and prevention of bleeding. The most commonly used of the currently available therapeutic agents are a plasma-based therapy, factor eight inhibitor bypassing activity, vapour heated, and a recombinant therapy, NovoSeven (recombinant activated factor VIIa).[7]

CONCLUSION

In the present study, we registered 130 cases of Hemophilia. Among these 112 cases (86.1%) were Hemophilia A and 18 cases (13.8%) were Hemophilia B. The maximum numbers of cases were seen in first decade of life in both the conditions. The male to Female ratio in Hemophilia A is 17:1.5, and in Hemophilia B is 8:1 with Male predominance in both the conditions. The commonest clinical presentations were epistaxis, haemarthrosis and hematoma.

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