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INCIDENCE AND CLINICO-HAEMATOLOGICAL PROFILE OF HEMOPHILIA IN CHILDREN ATTENDING SKMCH, MUZAFFARPUR

Pediatrics

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ABSTRACT

Objective: To know the incidence and clinical presentation of Hemophilia in children & hematological profile to establish the cause of hemorrhage in children.

Method: study consists of 110 cases with hemorrhagic diathesis who attended pediatric department, sri krishna Medical College and Hospital, muzaffarpur in outdoor and indoor department during a period from 2018 to 2019. The detailed clinical history, physical examination and hematological and other related investigations were recorded in Performa and data were analyzed.

Result: During the study period 110 patient of hemorrhagic diathesis were studied. Majority of patient were of Hemophilia a (21.8%). Most common age group was 3-10 year (74.99%). there were total 28 cases of hemophilia of which , 24 cases of hemophilia A and 4 cases were of Hemophilia B . In majority of cases aPTT were abnormal. In all cases of Hemophilia most common presentation was haemarthrosis. Epistaxis was seen in 10 cases (41.66%) of hemophilia A and 2 cases (8.33%) of hemophilia A presented with intracranial bleed. Increased CT is found in 58.3% cases in our study and these were the cases in which there was severe deficiency of factor VIII. In our study severe deficiency of factor VIII is found in 58.3% cases, According to study moderate deficiency of factor IX is found in 100% cases.

KEYWORDS

Hemophilia, Factor assay, Coagulation disorder, Haemarthrosis

INTRODUCTION

The X-linked recessive inherited coagulation disorders, hemophilia A (Factor VIII deficiency) and hemophilia B (factor IX deficiency) are the commonest forms of hemophilia.(1,2) Incidence of hemophilia A is 1 per 5,000 male birth and hemophilia B is 1 per 25,000 male birth. Deficiency of associated factors hampers the process of hemostasis and predisposes hemophiliacs to spontaneous or post traumatic bleeding. Factor deficient individuals have severe, moderate and mild forms of disease, defined by plasma factor levels of <1%, 2-5%, 6-40%, respectively (3). It is sex linked recessive disease and gene located on xq28. Point mutation of the gene is the most prevalent type of genetic defect seen in 90-95% of cases. Male are sufferer and female are the carrier of the disease. Haemarthrosis is the most common, as well as most physically, psychologically and economically exhausting manifestation of hemophilia. CNS bleeding is the most serious complication of hemophilia which may occur following a trauma or spontaneously. Immediate factor replacement is a must in this case.(4) Activated partial thromboplastic time (APTT) is usually prolonged in patients with hemophilia (PwH). Factor VIII and factor IX assay are simple techniques, used for typing of severity of hemophilia and is done by two stage method, one stage method as well as micro-method. The one stage technique is used widely as it is simple to perform.(5) Bleeding episodes in hemophiliacs are treated with factor replacement therapy. The approximate least desired factor level required to control bleeding episodes is 30% (0.3 units/ml), 50 % (0.5 units/mi) and 80%-100% (0.8-1 unit/ml) in case of mild bleeding, major bleeding and major surgery respectively. Units of factor VIII to be administered are "required rise in % × weight in kg/2" and units of factor IX to be administered are "required rise in % × weight in kg".

MATERIALS AND METHODS

This Hospital based observational study was performed on all the 110 patients diagnosed as different types of hemorrhagic diathesis who attended pediatric department, Sri krishna Medical College and Hospital, muzaffarpur, in outdoor and indoor department during a period from 2018 to 2019.After taking written consent, all patients were subjected for clinical work up consist of detailed clinical history including relevant family history ,general as well as systemic examination & protocol wise laboratory test were done. Diagnosis of hemophilia was made on the basis of relevant history, physical examination ,and laboratory investigation such as prothrombin type (PT), Activated partial thromboplastic type (APTT) ,Bleeding time(BT),Clotting time(CT), correction studies and specific coagulation factor assay. Beside of this, other hematological investigation like complete blood counts including peripheral blood smear were also carried out to see blood cell morphology, platelet count. The physical examination and hematological and other related investigations finding were recorded in preformed Performa. Patients with other bleeding disorders were excluded from the study; Sample size was taken based on the convenience of the study.

RESULTS

This study was conducted in sri krishna medical college and hospital, muzaffarpur. During our study period a total no of 110 patients were studied, out of which 21.81 % patients were of hemophilia A and hemophilia B (3.6%). maximum patient of hemophilia A were between 3 to 10 year of age (74.99%). There were total 28 cases of hemophilia of which, 24 cases of hemophilia A and 4 cases were of Hemophilia B. the present study ratio of the hemophilia A to B is 6:1.

Table 1. Distribution Of Patients According To Type Of Hemophilia

Type of hemophilia	Number of Patients	Percentage
Congenital Coagulation factors defects		
i) Hemophilia A	24	21.81%
ii) Hemophilia B	04	3.6%
TOTAL	28	

Table 2. Age Wise Distribution Of Hemophiliac Patients

Age	No. given	% of cases
Upton 2 yrs.	04	16.66
3-5 yrs	10	41.66
6-10 yrs	08	33.33
10-18 yrs	02	8.33
Total	24	100

The commonest presentation in this study was Haemarthrosis.

Haemarthrosis were seen in 20 cases (83%) of hemophilia A and 1 (25%) cases of hemophilia B. Epistaxis was seen in 10 cases (41.66%) of hemophilia A and 2 cases (8.33%) of hemophilia A presented with intracranial bleed. Increased CT is found in 58.3% cases in our study and these were the cases in which there was severe deficiency of factor VIII. In our study severe deficiency of factor VIII is found in 58.3% cases, According to study moderate deficiency of factor IX is found in 100% cases. Family history found in 79.12 % cases.

HEMOPHILIA B SKIN and mucous membrane hemorrhage are present in maximum cases. Hemarthrosis present in only one case (25%).

Table – 3: Showing Pedigree History In Hemophilia A (n = 24)

Family history	n	%
No family history	05	20.83
In two generations family history present	16	66.66
In three generations family history present	03	12.55
In four generations family history present	Not Known	00
Total	**24**	100

Table – 4. Clinical Presentation Of Hemophilia A (n = 24)

Clinical presentation	No. of cases	%
Haemarthosis	20	83
Excessive bleeding and hematoma formation after mild trauma.	16	66.66
Spontaneous bruising with recurrent hematoma formation	12	50
Epistaxis	10	41.66
Intracranial hemorrhage	2	8.33

Table- 5 Distribution of Patients According to Severity of Haemophilia

Total no. of cases	Moderate deficiency of factor VIII		Severe deficiency of factor VIII	
	NO.	%	NO.	%
24	10	41.7	14	58.3

DISCUSSION

HEMOPHILIA:

In our study maximum patient of hemophilia A were between 3 to 10 year of age (74.99%) which correlate with the study of Vikas Payal et al[6], who found between the age group of 1 to 5 years (42.85%) cases, 5 to 10 year (32%) cases. Di Michele D et al. 1998 studied the incidence is 1 per 5,000 live births for hemophilia A and 1 per 30,000 live births for hemophilia B. These finding are similar to finding of our study. In the present study, there were total 28 cases of hemophilia of which, 24 cases of hemophilia A and 4 cases were of Hemophilia B. In the present study ratio of the hemophilia A to B is 6:1. Biggs and Mcfarlane[7] found the ratio to be 10:1 in England and 5:1 in USA .Finding of our study are similar to the finding of their study done in USA .In our study ratio of hemophilia A : B = 6:1. Study by Dr.Inaam Noure et al[13], it is 4.5:1. So our study correlates well with this study. In our study family history found in 79.12 % cases Strauss et al [8] also found positive family history in majority of the cases in their study of hemophilia. The commonest presentation in this study was Haemarthrosis. Haemarthrosis were seen in 20 cases (83%) of hemophilia A and 1 (25%) cases of hemophilia B. Epistaxis was seen in 10 cases (41.66%) of hemophilia A and 2 cases (8.33%) of hemophilia A presented with intracranial bleed. Study conducted by Vikas Payal et al, haemarthrosis found in 68% cases [6].Rajesh Kashyap et al[9], found in their study haemarthrosis in 74% cases. Our findings are similar to these workers. Increased CT is found in 58.3% cases in our study and these were the cases in which there was severe deficiency of factor VIII. Biggs and Macfarlane et al [7] also found that prolonged CT in only severe hemophilia. Abshireet al[10] clinical presentation findings were similar to present study. In our study severe deficiency of factor VIII is found in 58.3% cases, according to VikasPayal et al study severe deficiency found in 43-55.7%, so our study correlate well with the finding of Vikas Payal et al study. According to MA Karim[12] study 47.5% cases have severe deficiency of factor VIII. These finding are similar to finding of our study .But does not correlate well with finding of P Kannan et al [11] study where severe factor VIII deficiency found in 21% cases.According to study moderate deficiency of factor IX is found in 100% cases. According to study done by MA Karim mild deficiency (40%), moderate deficiency (50%), severe (10%). So our study does not correlate with these studies.

CONCLUSION

Hemophilias are distributed worldwide and have heterogeneous presentation depending upon the disease severity. Knowledge of the spectrum of presentation of hemophilia in the local population helps in early diagnosis and planning of management.Although ecchymoses and hemarthrosis were the leading clinical manifestations of hemophilia in children, in the present setting, posttraumatic bleeds and gum bleeds were the main features at the onset of presentation of these children. So, presence of these features in an otherwise normal child should be considered for the evaluation of hemophilia. More vigilance is to be needed for detection of hemophilia in children.

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