Original Research Article

Hemophilia- A profile study including ocular complications at a tertiary care centre in Chennai

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Abstract

Aim: a profile study of hemophilia, including ocular complications in 110 patients with hemophilia at a tertiary care centre in Chennai. **Materials and Methods:** A retrospective study was carried out on 110 patients with hemophilia over a span of one year at the Government Royapettah Hospital-a tertiary care centre in Chennai. 110 hemophiliac patients in the age group 6-63 were included in this study. The diagnosis of hemophilia was made on the basis of screening tests- complete blood count, Activated Partial Thromboplastin Time, Prothrombin time and Fibrinogen test and clotting factor assays which diagnose a bleeding disorder. Coagulation profile study including clotting factor assay was performed for all patients. A general examination was done to assess the site of bleed followed by ocular examination in detail, which included both anterior and posterior segment examination. **Results and Conclusion:** non-ocular bleeding complications were more common. The commonest site of ocular bleed was the bleed under the skin around the eye-periorbital bleeding or ecchymoses along with subconjunctival haemorrhage. Retinopathy in hemophilia is rare.
Key Words: bleeding disorder, clotting factors, hemophilia, haemorrhage, replacement therapy.

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INTRODUCTION

Hemophilia is an inherited bleeding disorder that causes abnormal or exaggerated bleeding and poor blood clotting due to deficient clotting factors in the blood, which impair the body's ability to form blood clots. People with hemophilia bleed easily, and the blood takes a longer time to clot. The three forms of hemophilia are hemophilia A, B, and C. Hemophilia A is the most common type of hemophilia, and it is caused by a deficiency in factor VIII. Hemophilia B, also called Christmas disease, is caused by a deficiency of factor IX. Clotting factor levels in normal persons who do not have hemophilia is between 50 and 100 percent. Mild hemophilia is indicated by a clotting factor level in the plasma that is between 5 and 40 percent. Moderate hemophilia is indicated by a clotting factor level in the plasma that is between 1 and 5 percent. Severe hemophilia is indicated by a clotting factor level in the plasma of less than 1 percent. Hemophilia varies in its severity among affected individuals. Symptoms include excessive bleeding from any site in the body. Though bleeding manifestations are mostly non ocular, ocular complications like subconjunctival hemorrhage, retinal or orbital hemorrhage and postsurgical hemorrhage can occur. Treatment involves coagulation factor replacement therapy.

MATERIALS AND METHODS

This retrospective study was carried out on 110 patients with hemophilia over a span of 1 year at Government Royapettah Hospital- a tertiary care centre in Chennai. 110 hemophiliac patients in the age group 6-63 were included in this study. The diagnosis of hemophilia was made on the basis of screening tests- complete blood count(CBC), Activated Partial Thromboplastin Time(APTT), Prothrombin time(PT) and Fibrinogen test

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and specific clotting factor assays which diagnose a bleeding disorder and determines the type of hemophilia as well as the severity of the disease. These clotting factor assays are mandatory for management of hemophilia. Coagulation profile study was performed for all patients.

Exclusion criteria: Hemophiliacs with co morbidities like diabetes mellitus, systemic hypertension, ischemic heart disease and other systemic illnesses like systemic lupus erythematosus and rheumatoid arthritis were excluded from the study.

Clinical evaluation: Detailed history was obtained regarding blood group, severity of disease, factor levels at the time of presentation, co morbidities including diabetes mellitus and hypertension, recent site of bleed ,whether the bleed was spontaneous or post traumatic, complaints pertaining to the eye in terms of bleeding in the eye or around the eye, any defective vision or loss of vision, usage of glasses, any surgical intervention in the eye and if so,any major operative or post operative bleed.

Investigations done

Blood: Blood group confirmation, complete blood count, Activated Partial Thromboplastin Time, Prothrombin time and Fibrinogen test, blood sugar, bleeding and clotting time

Clotting factor assays: factor VIII /factor IX levels

Blood pressure recording

Examination: in detail was carried out in all cases. A general examination was done to assess the site of bleed followed by ocular examination, which included both anterior and posterior segment examination. Vision assessment –uncorrected and best corrected visual acuity testing was done for all patients. Intra ocular pressure recording was done with applanation tonometer. A thorough anterior segment examination using the slit lamp was carried out. Posterior segment examination was done after dilatation of the pupil using slit lamp biomicroscopy with 90 D lens or indirect ophthalmoscopy. Observations regarding media clarity, disc and vessels, macula and any abnormalities were recorded with a fundus camera.

OBSERVATION AND ANALYSIS

Based on patient's history and our examination, a profile of hemophilia patients was made and the results were tabulated and analysed.

Table 1: Age Wise Distribution Of Patients

Age group in years	No. of persons and %
Less than 10	8 (7.27%)
11-20	37 (33.63%)
21-30	30(27.27%)
31-40	19 (17.27%)
41-50	9 (8.18%)
51-60	5 (4.54%)
61-70	2 (1.81%)

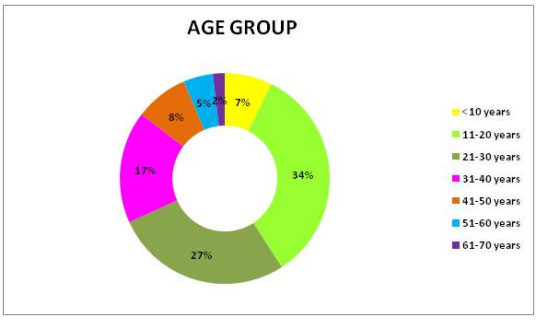


Table 2: Type Of Factor Deficiency

Factor type	No. Of patients and %
Factor 8	97 (88.18%)
Factor 9	9 (8.18%)
Factor 8 inhibitor	4 (3.63%)

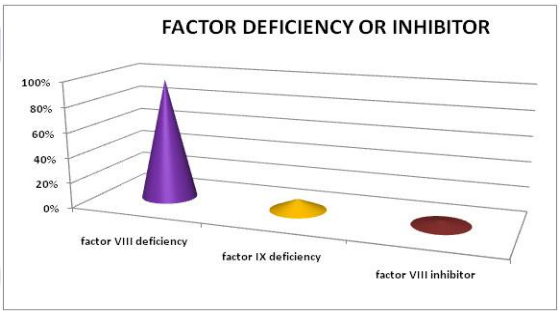


Table 3: Blood Group

Blood group	No of patients and %
A positive	24 (21.81%)
B positive	34 (30.90%)
AB positive	3 (2.72%)
O positive	41 (37.27%)
A negative	2 (1.81%)
B negative	1 (0.9%)
AB negative	1 (0.9%)
O negative	4 (3.63%)

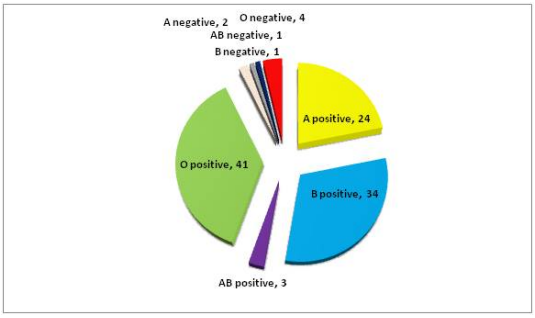
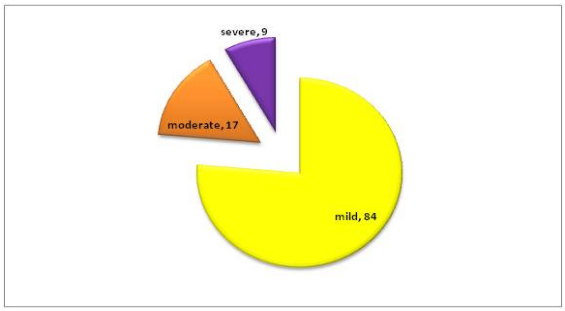


Table 4: Severity Of Disease

Severity of disease	No. Of patients and %
Mild	84 (76.36%)
Moderate	17 (15.45%)
Severe	9 (8.18%)



76% of hemophilia patients had mild disease i.e clotting factor level between 5-40% while 15% fell in the moderate level category and around 8% had severe hemophilia.

Table 5: Site Of Last Bleed

Site of bleed	No of patients and %
Joint or intramuscular	67 (60.90%)
Gum	16 (14.54%)
Eye	14 (12.72%)
Nasal	2 (1.81%)
Anal	2 (1.81%)
Others	9 (8.18%)

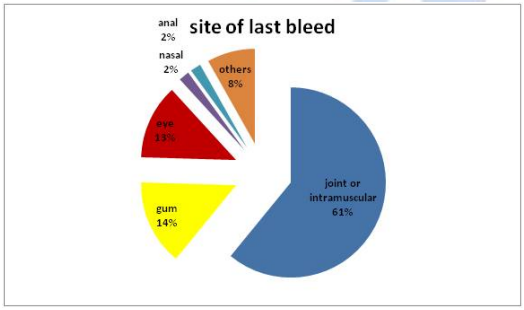
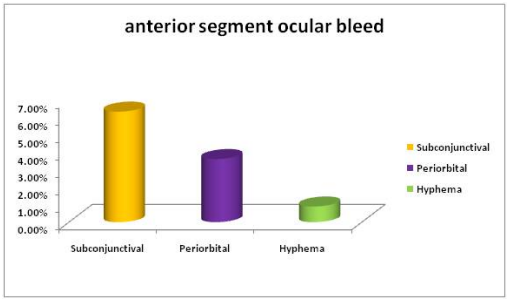


Table 6: Ocular Bleed – Anterior Segment

Type of haemorrhage	No of patients and %
ANTERIOR SEGMENT	
Subconjunctival	7 (6.36%)
Periorbital (Ecchymoses)	4 (3.63%)
Hyphema	1 (0.9%)
POSTERIOR SEGMENT	2 (1.81%)



Non ocular bleeding complications were more common than ocular. Joint bleeds and intramuscular bleeds accounted for the majority of bleeding complications. Other causes of non ocular bleeding were the dental and nasal bleeding, followed by anal bleeding. The less common causes of bleeding were hematuria and malena. Anterior segment examination using slit lamp did not reveal any abnormalities in 96 patients. One7 year old had bilateral mild congenital ptosis. Ocular bleeding (haemorrhages in and around the eye) accounted for 13% of all bleeds. 14 patients had ocular bleeding which is substantially lesser than that observed in a study by Robert A Rubenstein *et al*[2], Archives of Ophthalmology. comprising 125 hemophiliac patients. This could probably be attributed to the increased awareness and treatment options available nowadays. The commonest site of ocular bleed was the bleed under the skin around the eye-periorbital bleeding or ecchymoses along with subconjunctival haemorrhage. Sub conjunctival hemorrhage was seen in seven patients. Periorbital ecchymoses was seen in four patients while one had hyphema which settled with factor replacement therapy. Posterior segment examination revealed simple tortuosity of retinal vessels in 6 patients. Retinopathy due to hemophilia per se, were not seen in any of our patients. Retinopathy in hemophilia is very rare[8], which is consistent with studies by Kataria *et al* in their study on fundus in blood dyscrasias[8]. It is of prime importance to directly measure factor levels in a patient with ocular haemorrhages, as Partial Thromboplastin Time can be normal under conditions of stress[9]. Posterior segment bleed was not seen in any of our patients, except two patients who lost their vision ultimately. Intra orbital hemorrhage leading to loss of vision occurred in these two patients in our study. One was 45 year old male with no co morbidities with moderate hemophilia, whose only initial presenting symptom was sudden onset unilateral undiagnosed headache which was later attributed to intraorbital hemorrhage which eventually led to loss of vision in the affected eye. The other patient also lost vision in one eye following a spontaneous bleed in one eye which was attributed to probable choroidal hemorrhage.

Such intraocular or retrobulbar bleeding leading to loss of vision is rare as reports in literature are very few – Orbital hematoma following retrobulbar block for cataract surgery by White *et al*[6], Case report of a case of delayed spontaneous bleeding in the blind eye of a hemophilia patient by Gi Sung Son *et al*[7].

DISCUSSION AND RESULTS

- Patients in the age group 11-20(34%) were the most affected seeking factor replacement therapy,followed by those in the age group 21-30(27%).
- Factor VIII deficiency was the most common - seen in 97 persons (88%), followed by factor IX deficiency in 9 persons (8.18%).
- Blood group O was the most prevalent blood group seen in 45 persons(41%) ,followed by blood group B(32%) and A(24%).
- Severity of the disease was mild in 84 persons(76%), moderate in 17 persons(15%) and severe in 9(8%).
- Joint and intramuscular bleeding was the most common non ocular complication followed by gum bleeds.
- Of the ocular complications, subconjunctival and periorbital bleeding were the common anterior segment complications.
- Posterior segment bleed was seen in only 2 persons (1.81%), who however lost vision in that eye.
- A high index of suspicion of bleed due to hemophilia, in cases of undiagnosed refractory headache is needed.
- Precautionary measures in patients with hemophilia have a definite role not only in life saving but also in sight saving.
- Clotting factor assays are mandatory for management of hemophilia. An early diagnosis and appropriate treatment with replacement coagulation factors go a long way in preventing vision loss.

CONCLUSION

Non ocular complications were more common than ocular. Retinopathy in hemophilia is rare. Of the ocular complications, anterior segment haemorrhages were significantly higher than posterior segment haemorrhages, which however caused potential sight threatening complications and vision loss.

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