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A cross-sectional study of hemophilia in the Diyala government

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Abstract

Hemophilia is a group of X-linked recessive hereditary disorders due to a mutation in factor VIII and factor IX genes. Hemophilia A is a common hereditary hematological disease. Patients with hemophilia A have spontaneous and repeated bleeding, especially in the muscles and joints. Recurrent bleeding episodes result in disabilities by inducing persistent damage in these joints. Hemophilic arthropathy decreases the quality of life by restricting physical activity. In this study, we discussed the risk factors and prevalence of hemophilia in the Diyala governorate.

Methods: This is a cross-sectional descriptive study. We collected a sample of 50 hemophiliac patients who attended the specialized hematology center in Baquba teaching hospital in the period from July 2022 to December 2023.

Results: 50 patients were enrolled in this study, 40 males and 10 females. Their mean age was 18.7±15.04 years. Their mean age at diagnosis was 7.5±14.02 years. 54% of them suffered from anemia below 10 g/dL. There were significant associations between hemophilia and blood group, especially group O.

Conclusion: Hemophilia patients with the O + blood group have a high risk of severe bleeding.

Keywords: hemophilia, factor VII, Blood Group, patients.

Introduction

Hemophilia is a group of X-linked recessive hereditary disorders due to a mutation in factor VIII and factor IX genes. Males are mainly affected, while females remain asymptomatic. Hemophilia A is the most common (80% cases) with a prevalence of 1 in 5,000–10,000 male births and Hemophilia B with a prevalence of 1 in 20,000–34,000 male births.[1] Hemophilia lowers blood plasma clotting factors. Thus, when a blood vessel is injured, a temporary scab is formed but the missing coagulation factors prevent fibrin formation [1].

Hemophilia A is a common hereditary hematological disease. Patients with hemophilia A have spontaneous and repeated bleeding, especially in the muscles and joints. Severe hemophilia A accounts for 60% of patients with hemophilia and has highly frequent and serious joint bleeding and neoangiogenesis contributes to the development of hemophilic synovitis. At present, blood coagulation factors are used to measure and predict the bleeding risk for patients with hemophilia [2].

Spontaneous intra-articular hemorrhages may occur in 90% of the patients with severe hemophilia A. Recurrent bleeding episodes result in disabilities by inducing persistent damage in these joints. Hemophilic arthropathy decreases the quality of life by restricting the physical activity. Despite regular factor VIII (FVIII) concentrate replacements that are initiated at an early age, intra-articular hemorrhages (especially subclinical hemorrhages) have not yet been prevented completely and permanent joint damages continue to occur. It is not yet possible to determine the onset of intra-articular hemorrhage with clinical, laboratory, and radiologic imaging methods. Although only advanced findings of hemophilic arthropathy can be detected with conventional radiograms, MRI methods enable the detection of hemarthrosis findings in earlier phases [3,4].

The pathogenesis of hemophilic arthropathy is not well defined. Recent studies demonstrated that neo-angiogenesis plays a major role in the development of synovitis secondary to recurrent intra-articular hemorrhages in patients with hemophilia. It is shown that histologic abnormalities and neogenesis that occur within the synovia are similar to those in other joint diseases [5].

Aim of study

To discuss the nature, risk factors, and prevalence of hemophilia in the Diyala governorate and, relationships with blood groups.

Patients and methods

This is a cross-sectional descriptive study. We collected a sample of 50 hemophiliac patients who attended the specialized hematology center in Baquba teaching hospital in the period from July 2022 to December 2023. The patients were previously diagnosed with hemophilia and we excluded the newly diagnosed due to a lack of information about their symptoms. We collected information about their age, exact diagnosis, age at diagnosis, blood group, symptoms, etc. Through prepared written questionnaires and direct interviews with the patients and the parents.

Statistical package of social sciences (SPSS) version 25 was used to analyze the data. We expressed the qualitative and quantitative data frequencies by arithmetic mean and standard deviation. The chi-square test was used to identify the association between the variables when $P < 0.05$ was considered significant.

Results

50 patients were enrolled in this study, 40 males and 10 females. Their mean age was 18.7±15.04 years. Their mean age at diagnosis was 7.5±14.02 years. Their hemophilia type and symptoms are summarized in Tables 1 and 2.

Table 1. Diagnosis

Type	Frequency	Percent%
Hemophilia A	45	90.0
Hemophilia B	5	10.0
Total	50	100.0

Table 2. clinical features

Symptoms	Frequency	Percent%
Skin bleeding	22	44
Bleeding after trauma	25	50
Joint bleeding	21	42
Gum or tooth bleeding	6	12
Hematuria	3	6

Three were infected with hepatitis B and one with both types B and C.and 54% of them suffered from anemia below 10 g/dL as in table 3.

Table 3

Type	Normal	Anemia	Total
Hemophilia A	20	25	45
Hemophilia B	3	2	5
Total	23	27	50

Their blood group types are summarized in table 4.

Table 4. Blood group

ABO group	Frequency	Percent%
A+	15	30
A-	1	2
B+	11	22
B-	1	2
AB+	4	8
O+	16	32
O-	2	4
Total	50	100.0

There was a significant association between hemophilia and blood group as in table 5.

Table 5.

	Diagnosis			
Blood group	Hemophilia A	Hemophilia B	Total	Sig.
A+	11	4	15	P < 0.05
A-	1	0	1	
B+	11	0	11	
B-	1	0	1	
AB+	4	0	4	
O+	12	4	16	P < 0.05
O-	0	2	2	

Discussion

The best estimate of the prevalence of hemophilia, as calculated by comparative analyses of the most recent and most reliable registry data available from this study with other results in Canada, France, and the United Kingdom combined is 24.6 cases per 100 000 males for all severities of hemophilia A, 9.5 cases per 100 000 males for severe hemophilia A, 5.0 cases per 100 000 males for all severities of hemophilia B, and 1.5 cases per 100 000 males for severe hemophilia B. Applying a prevalence of 29.6 cases per 100 000 males for hemophilia A and B combined to the current world population estimate of 7.5 billion (3.8 billion males) [6].

Patients with blood group O were at increased risk of underdosing and had a higher rate of both overall bleeding and severe bleeding complications. and this agrees with the findings of Hazendonk et. Al., [7].

Differences in relation to tissue/organ targets, as indicated by the preponderance of epistaxis and hematomas as first symptoms in FVII deficiency and hemophilia B, respectively, support the presence of tissue and vascular bed-specific components interacting with the regulation of coagulation phases, eventually contributing to local hemostasis. Nevertheless, this analysis revealed a strong influence of the coagulation phenotype in determining the earliest and/or most severe bleeding phenotype in both diseases [8]. In our study, the most prevalent symptom was bleeding after simple injury (50%).

Our analysis indicates that symptoms occurred, on average, at an earlier age in hemophilia B compared with FVII deficiency, even when the same symptom is matched. Roughly half of the patients with hemophilia B have experienced an episode of hemarthrosis within the sixth year of age, whereas in FVII deficiency the

first hemarthrosis may occur later, even in teenagers. However, a small subset of patients with FVII deficiency was characterized by a very precocious and severe bleeding tendency, which shapes the bleeding-free survival curve in the first year of life, and could make it even steeper in FVII deficiency than in severe hemophilia B [9].

Conclusion

There was a strong association between blood group O and the risk of severe bleeding in hemophilia patients and increased in males infected with this disease.

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