

Inherited Bleeding Disorder: A Prospective Study

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Abstract

Introduction: Inherited bleeding disorders are relatively rare genetic disorders characterized by increased or prolonged bleeding due to abnormal coagulation (the ability of the blood to clot).

Aim: To study the various inherited bleeding disorders and to study about the clinical features among these disorders.

Materials and Methods: In this prospective study, patients who are admitted in Tirunelveli Medical College Hospital in hemophilia society were included. Clinical features of all these patients were undertaken to study their prevalence among these disorders.

Results: Out of these 70 patients with inherited bleeding disorders, 87% have Factor VIII deficiency, 8% have Factor VIII deficiency with inhibitor, 4% have Factor IX deficiency, 7% have von willebrand factor deficiency, and 5% have others such as Factor V deficiency, and Factor XIII deficiency.

Conclusion: Management of patients with inherited bleeding disorders should reflect knowledge of the specific disorder to be treated plus careful consideration of the clinical circumstance for which therapy is proposed. In all cases, once a decision to treat has been made, the safest efficacious therapy should be given.

Key words: Bleeding, Coagulation factors, Hemophilia

INTRODUCTION

Inherited bleeding disorders are relatively rare genetic disorders characterized by increased or prolonged bleeding due to abnormal coagulation (the ability of the blood to clot). The cause is a decrease in amount or function of one of the 11 proteins in the blood, called clotting factors, that work together to make the blood clot.¹ Inherited bleeding disorders lead to a lifelong bleeding tendency. Diagnosis, frequently made in childhood, is based on clinical presentation of bleeding and family history together with the laboratory tests. Hemophilia A and B are the most frequent of these disorders. Together with von willebrand's disease (vWD), these X-linked disorders comprise 95%-97% of all inherited bleeding disorders.² The remaining defects,

generally transmitted as autosomal recessive traits in both sexes are rare with low prevalence rate. However, in countries where consanguineous marriages are relatively common, these autosomal recessive disorders occur more frequently in homozygosity.³ These inherited bleeding disorders occur in mild, moderate, and severe forms depending on the plasma factor levels of 6-30%, 1-5%, and <1%, respectively. While some patients may only have mild bruising or bleeding following trauma, others with severe deficiency may exhibit intracranial hemorrhages and hemarthroses. With the exception of vWD, these disorders produce similar signs and symptoms, regardless of the particular factor that is lacking.^{4,5}

Aim

The aim of the study was to study the various inherited bleeding disorders and to study about the clinical features among these disorders.

MATERIALS AND METHODS

In this prospective study, patients who are admitted in Tirunelveli Medical College Hospital in hemophilia society were included.

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Exclusion Criteria

Patients with other acquired causes of bleeding disorders, patients with bleeding disorders associated with fever and liver diseases. Detailed medical history and physical examination of the patients were taken. Clinical features of all these patients were undertaken to study their prevalence among these disorders.

RESULTS

In this study, detailed data collection of 70 patients from hemophilia society was taken into account and following conclusions are made. 37% patients were from 11 to 20 years age group followed by 21 to 30 years 26% (Figure 1). 93% of patients were male, 7% were female. (Figure 2). Out of these, 70 patients with inherited bleeding disorders, 87% have Factor VIII deficiency, 8% have Factor VIII deficiency with inhibitor, 4% have Factor IX deficiency, 7% have von willebrand factor deficiency, and 5% have others such as Factor V deficiency, and Factor XIII deficiency (Figure 3). Out of these, 70 patients with these inherited bleeding disorders, 50% have knee joint hemarthrosis, 25% have elbow joint hemarthrosis, 4% have subdural hematoma,

13% have psoas hematoma, 4% have gum bleeding, and 4% have hematuria (Figure 4).

DISCUSSION

Hemophilia can be referred to as a disorder that causes joint damage leading to limitation in conducting daily activities and changes in social functioning. In developed countries, hemophiliacs have a quality of life very similar to that seen in general population due to the provision of safety factor concentrates and a multidisciplinary comprehensive care approach.^{6,7} In the opinion of the author, in developing countries like Pakistan, hemophiliacs are not treated with safe products and appropriate quantities of the products because of cost related issues. Hence, the lack of adequate treatment can result in pain, arthropathy, and disability. It is estimated that only 25% of all hemophiliacs around the world receive adequate treatment and most of them die before the age of 20. Despite financial limitations, some developing countries such as Chile, Iran, Venezuela, and Vietnam are optimizing their resources for hemophilia care.⁸⁻¹⁰

Estimations based on the World Federation of Hemophilia annual global surveys indicate that the number of people with hemophilia in the world is approximately 400,000.

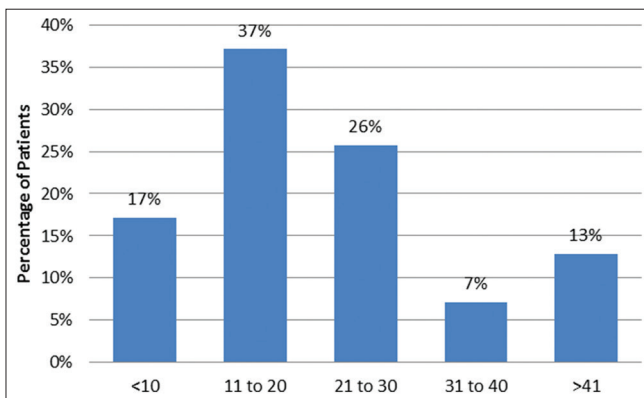


Figure 1: Age group distribution

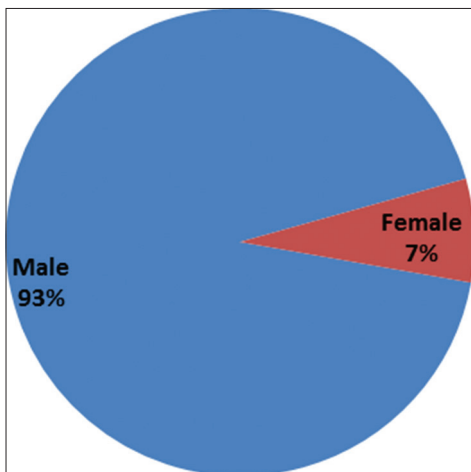


Figure 2: Gender distribution

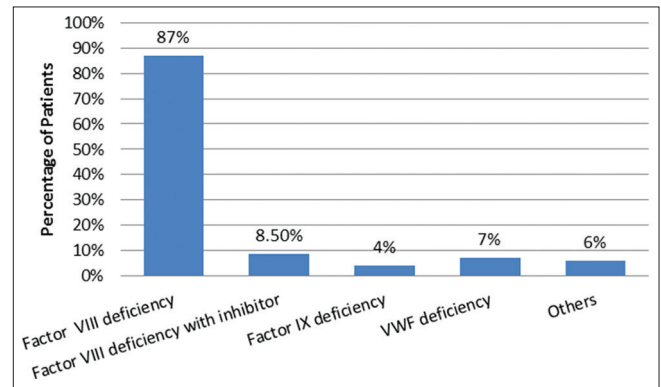


Figure 3: Distribution of bleeding disorder

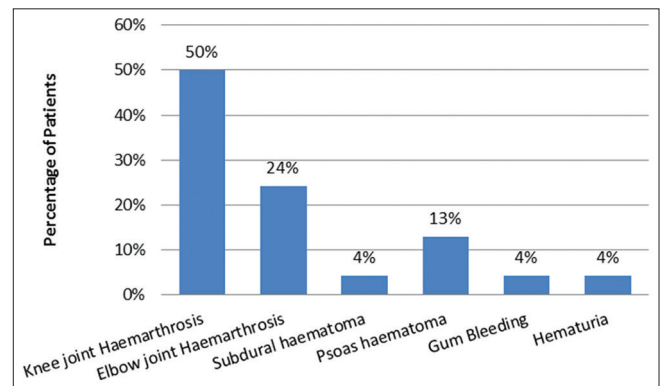


Figure 4: Distribution of clinical features

Hemophilia A is more common than Hemophilia B, representing 80-85% of the total hemophilia population. Hemophilia generally affects males on the maternal side. However, both F8 and F9 genes are prone to new mutations, and as many as 1/3 of all cases are the result of spontaneous mutation where there is no prior family history.¹¹

CONCLUSION

For each phase of hemostasis, screening tests which help in distinguishing a platelet disorder from a coagulation defect are available. The approach to a patient with a bleeding disorder needs a comprehensive detailed history and thorough physical examination. There must be a logical systematic approach and a discriminate use of laboratory investigations to reach the diagnosis and assess severity.

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