Hemophilia as a hereditary disease

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Introduction

Hemophilia is a serious hereditary genetic blood disease which is caused by quantitative and qualitative insufficiency of coagulation factors. There are four types of hemophilia: hemophilia A (clotting factor VII deficiency), -hemophilia B (factor IX deficiency– Christmas),

hemophilia C (factor XI deficiency– Rosenthal), hemophilia D (factor XII deficiency – Hageman).

Aim

The aim is to characterize types of hemophilia, to examine symptoms and causes, to study methods of treatment and prevention.

Materials and methods

Hemophilia is a sex-linked recessive disorder and is more likely to occur in males than females. Children of patients with hemophilia are always healthy but daughters are carriers of the pathological gene which is located in one of X-chromosomes. Since a male receives his single X-chromosome from his mother, the son of a healthy female silently carrying the deficient gene will have a 50% chance of inheriting that gene from her and with it the disease.

Results

Hemophilia is a treatable disease. It is characterized by impaired blood clotting and is manifested by frequent bleeding into joints, muscles and visceral organs. Hemorrhage in vital organs can lead to the patient's death and frequent hemorrhages in joints lead to their irreversible destruction and early disability of the patient. Without treatment it leads to permanent disability and premature death. At the beginning of the 20th century the life expectancy of hemophiliacs did not exceed 15 years. Today due to supportive replacement therapy the life expectancy of hemophiliacs and healthy people is equal.

Conclusion

Despite the severity of the disease patients have reasons for optimism. Scientists carry out research directed on a healthy gene selection, and introduction of it into the DNA of the patient to normalize the synthesis of missing factors. These studies show that hemophilia can be cured completely instead of simply dealing with its manifestations.