Case Report

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Von willebrand disease and gastrointestinal bleeding: case presentation

Abstract

Introduction: Von Willebrand diseases is a hereditary bleeding disorder caused by a quantitative or qualitative deficiency of von Willebrand factor, characterized by light mucocutaneous bleeding, although other bleeding such as gastrointestinal and joint bleeding may occur in severely affected patients. It treatment is fundamentally based on replacement therapy and the use of desmoprecin.

Objective: Describe the characteristics and clinical evolution of a patient who presented upper gastrointestinal bleeding as a form of presentation of von Willebrand disease.

Conclusions: von Willebrand diseases hould be suspected in patients with abnormal bleeding symptoms without apparent causes.

Keywords: von Willebrand disease, digestive bleeding

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Introduction

Von Willebrand disease is a hereditary bleeding disorder caused by a quantitative or qualitative deficiency of von Willebrand factor, a large protein synthesized in the vascular endothelium and megakaryocytes of the bone marrow.[1]

It is characterized by mucocutaneous bleeding such as ecchymoses, hematomas, epistaxis, gingivorrhagia, and menorrhagia. However, in severely affected patients, gastrointestinal and joint bleeding may also occur.[2] The diagnosis is based on family history, clinical manifestations, and screening tests, although the determination of the von Willebrand factor: antigenic and the ristocetin cofactor are basic tests for the diagnosis of the disease.[3] Treatment includes desmopressin, replacement therapy that can be done through cryoprecipotation or concentrates of factor VIII-von Willebrand factor and antifibrinolytics.[1,4] A clinical case of a two-year-old female transitional with upper digestive bleeding as the presentation of von Willebrand disease is presented. The objective is to describe the characteristics and clinical evolution of this patient. The girl's legal guardians were informed, informed consent was requested and, respecting ethical principles, the data of the patient and her relatives will not be published.

Case presentation

Two-year-old female transitional product of a high-risk obstetric pregnancy due to intrauterine growth retardation, normal delivery at 38 weeks of gestation with a birth weight of 2650 grams, adequate apgar. She presented a history of hematomas in the lower limbs that were initially related to trauma. She was referred from her health area because she began to have dark pasty stools in the morning and in the evening she had a very abundant stool like coffee grounds and vomited blood. The history of treatment with oral amoxicillin is recorded 72 hours earlier due to the diagnosis of acute otitis media. She was examined in the guard room and had dry and hypocolored skin and mucous membranes, slightly sunken eyes, distal coldness, bruises on the lower limbs, heart rate: 155 per minute, blood pressure: 90/60 mm/Hg, respiratory rate: 27 per minute. Rectal examination: presence of blood on the glove. In addition, the following complementary activities were carried out:

Hb: 56 g/l

Bleeding time: 7 minutes

Activated partial thromboplastin time (TPTK): 53 seconds (normal 24-36 seconds)

Rest of the coagulogram: Normal

She was admitted to intensive care unit and she was transfused with red blood cells, and appropriate measures were taken for digestive bleeding. A collective discussion was held with hematology, suggesting von Willebrand Disease, so cryoprecipitate every 12 hours and tranexamic acid every 8 hours intravenously were added to the treatment. After two doses of cryoprecipitate, clinical and complementary improvement began and after 48 hours of treatment the TPTK had normalized and there was no evidence of active bleeding. After discharge, she continued with follow-up in the hematology clinic where, in coordination with the Institute of Hematology and Immunology, specific studies were performed that showed a decreased ristocetin cofactor (less than 18%), which confirmed the diagnosis of von Willebrand desease.

Discussion

Von Willebrand disease is characterized by deficiency or dysfunction of von Willebrand factor, a plasma glycoprotein that mediates platelet adhesion and protects circulating FVIII from proteolysis, contributing to primary and secondary hemostasis, initially described by Erik von Willebrand in 1924.[4,5] It is considered the most common hereditary cause of bleeding, with an incidence that can reach up to 1% of the general population and an estimated prevalence in patients with bleeding symptoms of 1:10,000.[1,2] It is associated with mutations in chromosome 12 (p13.2), which codes for von Willebrand factor (although a type of acquired von Willebrand disease secondary to several diseases, such as neoplasms, autoimmune diseases, cardiovascular diseases, among others) has

been described.[6,7] According to the guidelines of the World Federation of Hemophilia, there are 3 types of the disease: 1, 2A, 2B, 2M, 2N, and 3. Types 1 and 3 are quantitative defects of the factor at the plasma level; type 3 being the most serious. Type 2 is divided into 4 subtypes and is classified as a qualitative factor defect.[6] It is characterized by mucocutaneous bleeding, such as ecchymoses, hematomas, epistaxis, and menorrhagia, although more serious bleeding such as gastrointestinal and joint bleeding may occur, reported in only 14% and 8% respectively.[2,5]

Von Willebrand disease and angiodysplasia (as a cause of digestive bleeding) have been associated in up to 20% of patients, especially in types 2A and 3, where it is associated with the loss of high molecular weight multimers, with a prevalence of only 6% in the general population.[8,9] In this case the girl had a history of small hematomas in the lower limbs and despite not being a very common symptom, she presented upper digestive bleeding that required admission and compromised her life. It was not possible to associate it with angiodysplasia, the studies carried out by the digestive tract were negative after this he has continued to present spontaneously appearing ecchymosis and hematomas with minimal trauma. The diagnosis is based on the personal history of bleeding, family history of von Willebrand disease, and evaluation of the results of complementary tests.[10] The first phase of complementary tests constitutes global tests that include complete blood count, platelet count, bleeding time and activated partial thromboplastin time (PTT K), the second phase is specific diagnostic tests where the coagulant activity of FVIII, the von Willebrand factor antigen, the activity of the ristocetin cofactor and the study of the multimers of the von Willebrand factor and the third phase that allows the classification of the disease, the binding capacity of the factor is measured von Willebrand factor to collagen, the binding of von Willebrand factor to FVIII and platelet agglutination induced by ristocetin.[1]

Due to the girl's symptoms and the alteration in bleeding time and TPTK, the disease was suspected, which was subsequently confirmed by determining a decrease in the ristocetin cofactor by less than 18%. There is no family history of abnormal bleeding. Treatment is based fundamentally on bleeding symptoms; essential therapeutic options include the use of desmopressin, which induces the release of von Willebrand factor, FVIII, and tissue plasminogen activator of endothelial cells, in addition to replacement treatment with concentrates of plasma-derived or recombinant FVIII-von Willebrand factor and cryoprecipitate. In addition to adjuvant therapies such as antifibrinolytics (tranexamic acid and epsilon amino caproic acid).[1,4] Given the suspected diagnosis in this case, we used cryoprecipitate every 12 hours as a source of von Willebrand factor in addition to intravenous tranexamic acid every 8 hours, achieving clinical and complementary improvement. Samples from the girl were later sent to the Institute of Hematology and Immunology where the diagnosis was confirmed. It can be concluded that von Willebrand disease should be suspected in patients with symptoms of abnormal bleeding without an apparent cause, in order to provide timely treatment.

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Conflicts of interest

The authors declare that there is no conflicts of interest.

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