

Repair of Ventricular Septal Defect in a Patient with Hemophilia A: Case Report

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Introduction

Hemophilia is an X-linked recessive hemorrhagic disease that affects 1 in 10,000 males worldwide, in all ethnic groups; hemophilia A represents 80% of all cases. Congenital heart disease is the most commonly occurring birth defect; it is formed during the intrauterine period. The combination of these two disease is rather rare and requires the cardiologist's apprehensive attitude. Here, we present our experience in the surgical treatment of a boy of 1-year-old with Perimembranous Ventricular Septal Defect with Hemophilia A which was found in preoperative examination. He was successfully operated with cardiac pulmonary bypass on the background of factor VIII injection.

Keywords: congenital heart disease; perimembranous ventricular septal defect; hemophilia A

INTRODUCTION

Hemophilia A belongs to the group of hereditary coagulopathies characterized by deficiency of coagulation factor VIII. It is an X-linked recessive trait in which males are affected and the disorder is transmitted by females. The disease phenotype correlates with the activity of FVIII and can be classified as severe (<1%), moderate (1–5%), or mild (6–30%). Ventricular septal defects (VSDs) occur when any portion of the ventricular septum does not correctly form or if any of the components do not appropriately grow together. Perimembranous VSDs are caused by failure of the endocardial cushions, the conotruncal ridges, and the muscular septum to

fuse at a single point in space. They are located in the left ventricle outflow tract beneath the aortic valve and are the most common VSD subtype, occurring in 75–80% of cases. Here we present a case of Perimembranous VSD (1 cm) with Right ventricular outflow tract obstruction associated with Hemophilia A in a 1-year-old boy, who underwent a successful open heart surgery without any complication.

CASE PRESENTATION

A 1-year-old boy with a known case of Acyanotic Congenital Heart Disease presented to our outpatient department of cardiac surgery with a history of failure to thrive since birth with poor feeding and unable to attain his normal

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growth. On admission, his parents complained of perioral cyanosis during agitation and the appearance of dense subcutaneous masses with small ecchymoses on their surface. His parents also gave a history of bleeding from his gums and nose, now and then for the past 4 months. He was noted to have a heart murmur at birth. On admission to our unit, a physical examination showed a child whose weight and height were on the lower percentile. The precordium was hyperactive, and a thrill was palpable over the left sternal border. A harsh 5/6 holo systolic murmur was best heard at the lower left sternal border. The electrocardiogram showed a Subaortic VSD of 1 cm with moderate infundibular pulmonary stenosis with mild TR with mild PAH and a dilated LA and LV. His hematology consultation was done. Laboratory investigation revealed a factor VIII deficiency with only 4% factor VIII activity. The following diagnosis was made: Perimembranous VSD with moderate Hemophilia A. Taking into account indications for operation, and complete support with blood preparations according to hematologists recommendations, it was decided to operate the child under cardiopulmonary bypass.

The operation was performed 16 days later. The following values were obtained immediately before the procedure: factor VIII activity of 4%; partial thromboplastin time (PTT) 82 seconds (normal, 35-45 seconds); hemoglobin, 11.1 g/dl; hematocrit, 33.3%; fibrinogen, 215 mg/dl; platelets, 276000/cu mm; The required dose of factor VIII was calculated using body surface area. No fresh frozen plasma was administered in preparation for the operation. The chest was opened through a midline sternotomy. The defect of the ventricular septum was corrected using a PTFE graft and RV myomectomy was done on the background of factor VIII infusions at the rate of 250 UI every 8 hours as preoperative

preparation during 3 days and during 6 days after operation.

Surgical treatment was successful without significant bleeding. The postoperative course was unremarkable. The child was discharged from the hospital on the 10th day after the operation in satisfactory condition under the supervision of a cardiologist and hematologist.

DISCUSSION

Congenital heart disease is formed during the intrauterine period but, as a rule, they are not hereditary. According to medical statistics, the probability of congenital heart disease development as related to the total population is 1:100 healthy children. VSD is one of the most widespread acyanotic congenital heart diseases. Pathological manifestations vary because each of the anatomical signs can have different forms and intensities. Hemophilia A is a disease from the group of hereditary coagulopathies stipulated by deficiency of coagulation factor VIII and characterized by bleeding of hematoma-like type. Genes stipulating the development of hemophilia are X-linked; due to this, the disease runs in the family by recessive inheritance through female lineage. Hemophilia can manifest at whatever age. The disease manifests with bleeding of various localization and the appearance of painful intermuscular hematomas and hemarthroses. Replacement therapy is the main principle of hemophilia treatment. Nowadays it is performed with the help of intravenous infusions of concentrated preparations of factor VIII which allow for achieving the hemostatic factor level.

Calculation of the dose of concentrated factor VIII: Dose (IU) = body weight x (required activity – basal activity). Open heart surgery can be accomplished safely without any bleeding complications, even in a young child with moderate factor VIII deficiency. Our success

with this patient was based on meticulous preoperative planning and close cooperation among cardiac surgeons, hematologists, anesthesiologists, the blood bank, coagulation laboratory technicians, and perfusionists, as previously emphasized by others.

CONCLUSION

Surgical treatment has been and remains the

method of choice in large perimembranous VSD. The closest and long-term prognosis for patients operated with large perimembranous VSD is usually favorable. Timely and complete diagnosis of comorbidities and specific preparation for the intervention allow to perform surgery safely even in the case of Hemophilia A.

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