Case Report

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Blalock-Taussig Shunt Thrombosis Prophylaxis in a Patient With Jacobsen Syndrome and Thrombocytopenia

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Abstract

Jacobsen syndrome (JS) is a rare chromosomal anomaly caused by deletions in the distal long arm of chromosome 11. Features of the syndrome include growth and developmental delays, a distinctive facial appearance, and a variety of physical problems including heart defects and bleeding disorders. Congenital heart defects occur in approximately 50% of children with JS. Hypoplastic left heart syndrome (HLHS) has been occasionally reported in association with JS. In such cases, the hematological abnormalities may influence the outcome from single-ventricle palliation through staged surgical reconstruction. Thrombotic obstruction or occlusion of the modified Blalock-Taussig (BT) shunt is a well-documented cause of interstage mortality following the Norwood operation. Although there is no consensus regarding the therapeutic value of antiplatelet therapy during the interstage period following the first stage of palliation, maintenance of shunt patency is critically important. For patients with JS undergoing single-ventricle palliation, decisions regarding antiplatelet therapy during the interstage period may be further complicated by the presence of thrombocytopenia and platelet dysfunction related to JS. We report the case of a patient with HLHS, JS, and thrombocytopenia who underwent the Norwood procedure, and we describe our strategy for prophylaxis against thrombosis of the BT shunt.

Keywords

Blalock-Taussig shunt, hypoplastic left heart syndrome, interstage mortality, Jacobsen syndrome

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Introduction

Jacobsen syndrome (JS) is a rare chromosomal condition caused by deletions in the distal region of the long arm of chromosome 11. These deletions, which may range from 7 to 20 Mb, involve bands distal to 11q23 and result in wide phenotypic variation. Features of JS include growth retardation, psychomotor retardation, and dysmorphic facies including skull malformations, hypertelorism, downslanting palpebral fissures, short nose, low-set, posteriorly rotated ears, and various ocular anomalies. Thrombocytopenia and platelet dysfunction are also common findings. In addition, immunological and hormonal abnormalities have been reported.[1]

Congenital heart defects occur in 56% of reported cases of JS. Ventricular septal defects, which occur in about 50% of the cases, are the most common congenital cardiac lesions reported.[2] The incidence of hypoplastic left heart syndrome (HLHS) has been reported as 5%. Other defects include double outlet right ventricle, transposition of the great arteries, truncus arteriosus, pulmonary atresia with intact ventricular septum, atrial septal defects, total anomalous pulmonary venous return, interrupted aortic arch, and complete atrioventricular canal.[3]

We report the case of a patient with HLHS, JS, and thrombocytopenia who underwent the Norwood procedure, and we describe our strategy for prophylaxis against thrombosis of the Blalock-Taussig (BT) shunt.

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Abbreviations and Acronyms

BT	Blalock-Taussig
HLHS	hypoplastic left heart syndrome
JS	Jacobsen syndrome
LPA	proximal left pulmonary artery
TEG	thromboelastogram

Case

A baby girl was born at term to a 25-year-old gravida-1 mother via C-section secondary to breech position. Birth weight was 5 pounds 13 ounces. Initial physical examination revealed trigonocephaly, upslanting palpebral fissures, wide nasal bridge, and low set, small, and posteriorly rotated ears. Apgar scores were 4, 6, and 8 at 1, 5, and 10 minutes, respectively. Positive pressure ventilation with eventual intubation of the trachea was required due to persistent respiratory distress. Worsening tachypnea developed following extubation, and a chest x-ray demonstrated cardiomegaly, increased pulmonary vascular markings, and vertebral anomalies. An echocardiogram performed on the third day of life revealed HLHS with severe aortic stenosis, mitral atresia, ventricular septal defect, hypoplastic aortic arch, total anomalous pulmonary venous return to the coronary sinus, and bilateral superior caval veins without the presence of a bridging vein. Prostaglandin E was administered intravenously and the patient was transferred to All Children's Hospital. Laboratory evaluation revealed thrombocytopenia. Bilateral pelvicaliectasis was seen on ultrasound examination of the abdomen. Chromosomal analysis and microarray showed a deletion of the long arm of chromosome 11 (11q) extending from band 11q24.2 to the terminal portion of the chromosome, adjacent to the critical region of 11q24.1 which is associated with JS. Given the constellation of physical features, congenital heart disease, thrombocytopenia, and chromosomal deletion, diagnosis of JS was established.

A Norwood operation with modified BT shunt was performed on the 15th day of life. Preoperative assays for platelet antibody and platelet glycoprotein expressions were negative. Preoperative coagulation studies, including prothrombin time, partial thromboplastin time, fibrinogen, and fibrin split products were all normal. Preoperative platelet count was 29 000/mm^3, and 4 units of platelets were transfused on the day of surgery to bring her platelet count to above 100 000/mm^3. Following surgery, a continuous infusion of heparin at 15 U/kg per h was administered as prophylaxis against shunt thrombosis. The infusion was continuously infused at this dose until postoperative day 15. Because of persistent thrombocytopenia and presumed platelet dysfunction, the patient was initially discharged without aspirin or other antiplatelet therapy. We did not consider using lovenox in this patient.

Respiratory distress and systemic arterial desaturations (measured by percutaneous pulse oximetry) prompted readmission to the hospital 4 weeks later. An echocardiogram showed possible narrowing of the distal BT shunt and the left pulmonary artery. Platelet transfusions were administered to bring the platelet count to above 100 000/mm^3. Cardiac catheterization was then performed, which demonstrated a patent BT shunt and proximal left pulmonary artery (LPA) stenosis. Saturations were improved following balloon angioplasty of the LPA. Platelet function was reinvestigated following catheterization. Platelet count was 84 000/mm^3. Platelet function was found to be functionally normal via thromboelastogram (TEG) study performed 10 days postcatheterization. Antiplatelet therapy was begun with baby aspirin 20.25 mg daily. A repeat TEG 9 days later showed evidence of adequate platelet inhibition.

Another cardiac catheterization was performed 6 weeks later, again prompted by desaturations. Angiography revealed narrowing of the distal third of the BT shunt without evidence of thrombus and poor interval growth of the branch pulmonary arteries. A 3.5-mm coronary artery stent was deployed within the lumen of the BT shunt to improve pulmonary blood flow. Saturations stabilized after placement of the stent. We hoped, in addition, that this would result in further growth of the pulmonary arteries prior to performance of the second-stage surgery, which would be a superior cavopulmonary anastomosis. Anticoagulation was maintained with baby aspirin 20.25 mg daily. Follow-up catheterization 8 weeks later showed continued patency of the stented BT shunt, good interval growth of the branch pulmonary arteries, and normal pulmonary vascular resistance.

At age 217 days, the patient underwent bilateral superior cavopulmonary anastomoses and surgical occlusion of the modified BT shunt. Baby aspirin was restarted at 20.25 mg daily on postoperative day 3. A repeat TEG scan on postoperative day 8 showed normal platelet function, and the dose was subsequently increased to 40.5 mg aspirin daily. The patient was discharged home 3 weeks after surgery and is currently doing well as she awaits a completion Fontan procedure.

Discussion

The Norwood procedure has afforded the opportunity for survival to infants affected by HLHS and HLHS-related malformations.[4-6] Refined operative techniques and improved perioperative management have resulted in improved survival after the Norwood procedure.[5-7] According to the STS Congenital Heart Surgery Database, aggregate survival to hospital discharge after the Norwood procedure was 80.7% in 2342 Norwood procedures performed at 69 North American Centers in the 5-year time interval of 2005-2009, inclusive.[6] More recent efforts have been directed toward identifying and reducing risk factors that adversely impact survival to stage II reconstruction.[8] Previously identified causes of interstage mortality include decreased coronary perfusion, catastrophic shunt thrombosis, systemic infection, arrhythmias, residual cardiac lesions, restrictive atrial septal defects, depressed cardiac function, elevated systemic vascular resistance, and growth failure.[7,9,10] The reported[7] incidence of systemic-to-pulmonary artery shunt thrombosis ranges from 8% to 16%.

Fenton and colleagues found BT shunt occlusion at autopsy to account for one third of cases of interstage mortality.[8] Therefore, maintenance of shunt patency is of major importance during the interstage period. A large multicenter study evaluating patients with systemic-to-pulmonary artery shunts demonstrated a decreased risk of shunt thrombosis for patients receiving aspirin therapy.[11]

Patients with JS often have evidence of platelet dysfunction in addition to thrombocytopenia. Low platelet counts in JS are related to an inherent deficiency in the platelet storage pool. Furthermore, hypoxia associated with congenital heart disease may further exacerbate thrombocytopenia. The theoretical risk of acute bleeding is increased upon initiation of long-term antiplatelet therapy.[12] However, this appears to be a heterogeneous patient population, and some patients with JS may have normal platelet function. Therefore, further assessment of platelet function may be warranted in certain clinical situations. Thromboelastography measures parameters of clot formation and lysis and historically has been used to monitor postoperative abnormalities of coagulation in the setting of liver transplantation and cardiac surgery.[13] Thromboelastography scans were performed in our patient prior to starting antiplatelet therapy and during longitudinal follow-up. Normal platelet function was determined by TEG analysis prior to starting aspirin therapy. Subsequent TEG analysis during platelet therapy demonstrated sufficient platelet inhibition and was used to guide antiplatelet dosing during the interstage period.

This case illustrates a special challenge in interstage management in a patient with JS and thrombocytopenia undergoing single-ventricle palliation. Despite the presence of thrombocytopenia, our patient had normal platelet function. Therefore, aspirin was eventually used as prophylaxis against thrombosis of the BT shunt. We suggest that in patients with JS, thrombocytopenia, and complex congenital heart disease, analysis of platelet function can be performed, and antiplatelet therapy should be considered.

Declaration of Conflicting Interests

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