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Case Report

Paediatric Surgery Section

Acute Aortic Thrombosis with Massive Intestinal and Lower Limbs Ischaemia in a Girl with Down Syndrome

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ABSTRACT

Patients with Down syndrome may have an increased risk of venous and arterial thrombotic events. Despite these, some argued that there is no increased risk of prothrombotic conditions in trisomy 21 and the thrombosis is usually due to other associated risk factor. Here a report of a 3-year-old girl, known case of Down syndrome, with a past history of atrio-ventricular septal defect (AVSD) repair at the age of 4 months developed acute aortic thrombosis with massive intestinal and lower limbs ischemia without obvious cause.

Key Words: Acute aortic thrombosis, Down syndrome, Lower limbs ischemia. Massive intestinal ischemia

INTRODUCTION

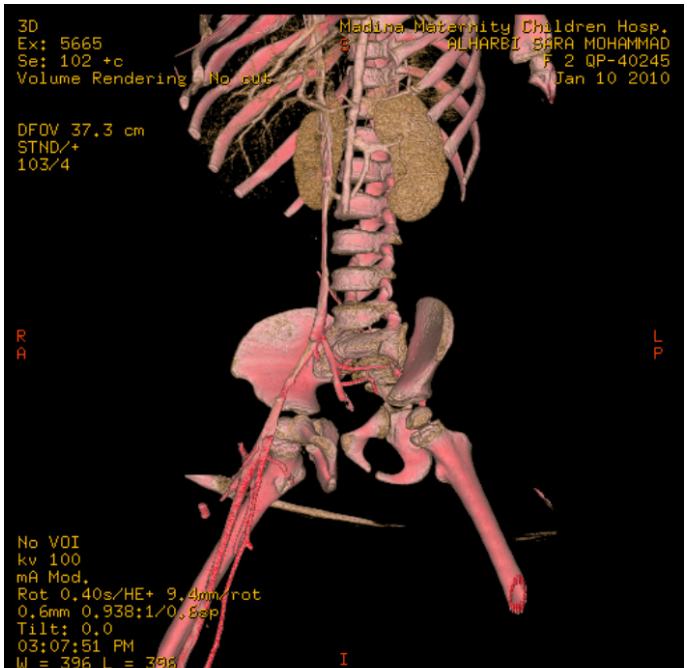
Thromboembolic events in children, especially after the neonatal period, are rare and most of these are venous [1]. Patients with Down syndrome may have an increased risk of venous and arterial thrombotic events [2-4]. Despite these, some argued that there is no increased risk of prothrombotic conditions in trisomy 21and the thrombosis is usually due to other associated risk factor [5]

Herein, the author presents a child with Down syndrome with complete aortic and superior mesenteric artery (SMA) occlusion with resultant mesenteric and bilateral lower limb ischaemia without evident cause. There is no such incidence reported in literature till now describing acute extensive multi-focal thrombotic events in a child or adolescent with Down syndrome.

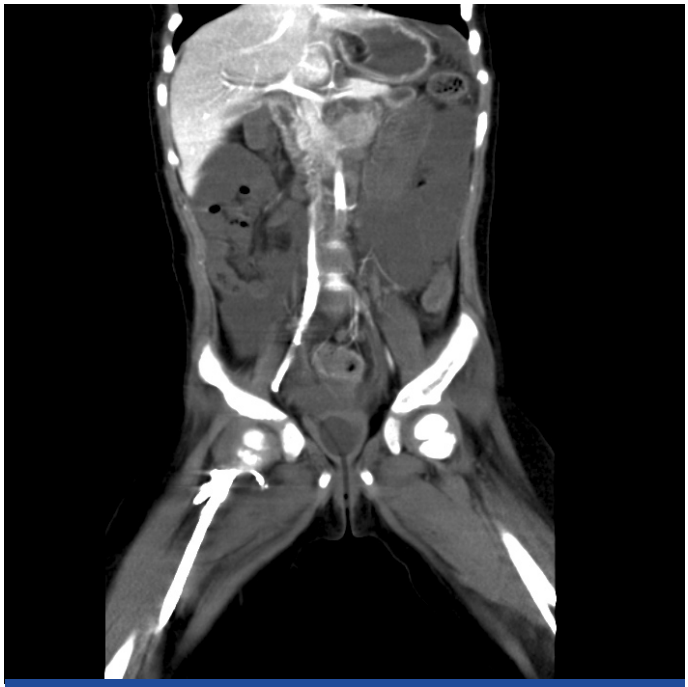
CASE REPORT

A 3-year-old girl, known case of Down syndrome, with a past history of atrio-ventricular septal defect (AVSD) repair at the age of 4 months was admitted with repeated non-bilious vomiting and diarrhea for 4 days. There were no other associated symptoms in the systemic review. She was not known to have any other medical illness, and was not on any medications. On examination, she was conscious, oriented, and afebrile. Systemic examination was unremarkable except for median sternotomy scar. Her initial electrolytes results demonstrated hyponatremia (125 mEq/L). She was diagnosed as a case of gastroenteritis with moderate dehydration and was treated with IV fluids. On 3rd day, she became hypotensive, irritable and developed a fever of 40° C, convulsions and bluish discoloration of her both lower limbs. She was shifted to the PICU with a diagnosis of meningitis. Lumbar puncture and brain CT results were normal. Following day, she developed abdominal pain and distension with bloody diarrhea. She looked sick, irritable, pale, tachypenic, hypotensive (65/35 mm Hg), and tachycardiac (pulse 160/min). She was intubated and started on inotropes. Abdomen became more distended, tense, tender guarding with absent intestinal sounds. Lower limbs became pale, cold and pulse less. Laboratorty study showed WBC (6.9 × 10^9 / L), Hb (8.6 gm/dl), PLT (57 × 10^9 / L), Glucose (6.3 mmol/L), and Alb (15 gm/L). Blood gas analysis showed metabolic acidosis. Factor V leiden , anti-thrombin III deficiency and anti-phosphlipid antibody were negative. Duplex scan of the abdomen and both lower limbs showed no signal in aorta and femoral arteries. Echocardiography demonstrated good cardiac function with no vegetation or thrombus in the heart or aortic arch. Contrast and volume enhanced CT angiography for the aorta demonstrated complete aortic and superior mesenteric artery occlusion and patent IVC [Table/Fig-1]. She was diagnosed to have bowel ischaemia [Table/Fig-2, 3], renal infarction [Table/Fig-4] and lower limb ischemia.

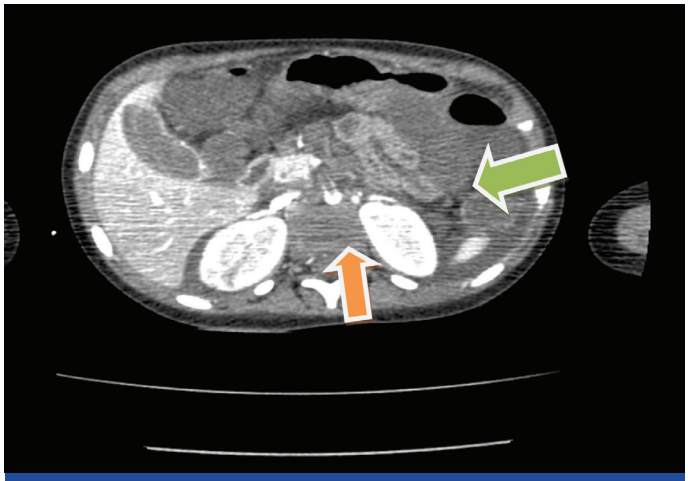
After adequate re-suscitation, the patient was subjected to laparotomy which revealed extensive small bowel necrosis, necessitating resection of the necrosed segment, followed by exteriorization of the proximal and distal ends. Stomach, Duodenum and colon



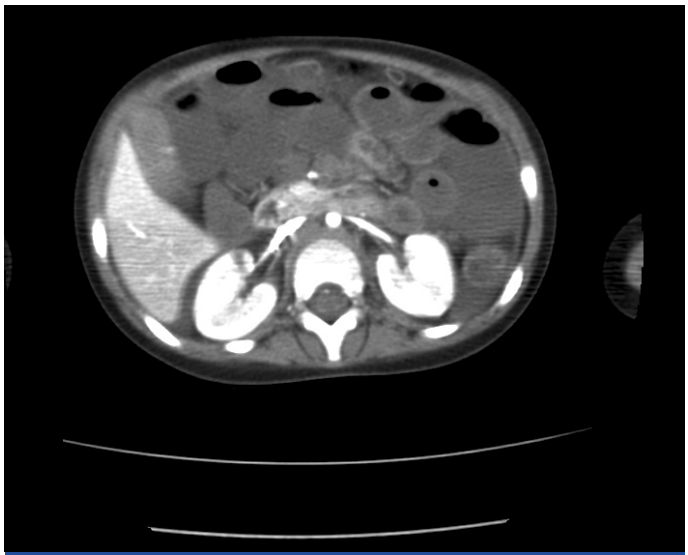
[Table/Fig-1]: Volume rendered CT abdomen reformated coronal veiw; showing complete occlusion of the aorta distal to the level of renal arteries



[Table/Fig-2]: Contrast enhanced CT abdomen coronal view showing no contrast distal to renal vessels and patchy enhancement of bowel loops



[Table/Fig-3]: Contrast enhanced CT abdomen showing complete occlusion of the aorta (orange arrow) with enhancing (viable) and non enhancing (necrotic) bowel loops; green arrow



[Table/Fig-4]: A hypoattenuatting wedge shaped area in right kidney indicating renal infarction

were normal. In the same setting, the vascular surgeon performed bilateral femoral artery exploration and retrograde aorto-iliac thrombectomy, resulting in good inflow and back flow. Fasciotomies for lower limbs were also done. 2.5 U of t-PA was infused into both superfacial femoral arteries. Good pulses reappeared in both lower limbs and heparin was continued post-operatively. After 48 hours, 2nd look laparotomy showed another ileal segament became gangrenous so it was resected, and both bowel ends are exteriorized and abdomen was closed. Central intravenous line was inserted for TPN purpose. On the 5th post-operative day, the exteriorized proximal segment of the bowel became black and the left leg revealed progressive ischaemia. She was taken for laparotomy which revealed that 15 cm of juejenum were ischaemic which was resected and both ends were exteriorized. At the same time, left above-knee amputation was done.

After 10 days inotropes were discontinoued, patient was extubated and vital signs were maintained. Her nutrition was maintained via TPN. One month later, she developed severe sepsis due to systemic candidiasis. The patient arrested and expired due to severe sepsis.

DISCUSSION

Thrombotic diseases are less frequent in children than in adults, but may result in severe morbidity and mortality. Thrombosis is commonly involve the venous system than the arterial one .The incidence of venous thromboembolism is 5.3 per 10.000 hospital admissions or 0.7 per 10.000 children and the mortality rate is 2.2% [6]. Complete aortic occlusion is even rarer but potentially catastrophic entity. Acute aortic occlusion bears an early mortality of 31–52% [7]. An acute thromboembolic event has not been reported in studies of post-operative repair in Down syndrome following repair of AVSD [8, 9], except one case of aortic thrombosis in a patient with Down syndrome has been reported recently [5]. This case was a 14-year-old boy with Down syndrome with repaired AVSD who presented with sudden onset of bilateral lower limb ischaemia. Transesophageal echocardiography detected a thrombus in the right atrium. The patient had complete occlusion of the infrarenal abdominal aorta with occlusion of superior mesenteric artery with extensive bowel necrosis and multiple areas of infarctions at the right kidney and bilateral lower limb ischaemia.

Thrombosis in a child warrants investigation of potential underlying prothrombotic conditions. These include abnormalities of the inherited anti-coagulant factors including protein C, protein S, anti-thrombin, and factor V Leiden, as well as acquired disorders such asantiphospholipid antibodies and prothrombotic mutations [10]. Despite the fact that congenital heart disease is considered as an acquired risk factor for thrombosis, a follow up of more than 100 patients after AVSD repair, with 24 (48%) of them having Down Syndrome failed to identify thrombosis or intra-cardiac thrombus as a cause of mortality, reported to be 10.7% [9]. Similar reports are recorded by various researchers [10-12]. Although, there are many reported cases of Down syndrome and thrombotic events; in most of them identifiable risk factor were present [13-15]. On the other hand some reports could not find thrombotic risk factor [16, 17]. This raises concern whether Down syndrome is a risk factor or not. In this case report, no cause was identified for the sudden arterial occlusion despite extensive investigations for possible causes. This case report highlights an unusual manifestation of Down syndrome in a child with previous uneventful repair of an AVSD. The likely cause for this extensive thrombosis in our patient

remains obscure. Is it Down syndrome itself? Is it the dehydration? Or it is a rare prothrombotic disorder.

Several proteins encoded on chromosome 21 are associated with an increased risk for vascular diseases. These include α-chains of collagen type VI, superoxide dismutase I, the interferon gamma receptor, and cystathionine β-synthase (2). In addition to these, a variety of haematologic abnormalities have been described in Down syndrome including an increased rate of laeukamoid reactions, neonatal polcythemia, thrombocytopenia and abnormal polymorphonuclear laeukocytic precursor cells, abnormal nailbed capillary morphology, high pulmonary vascular resistance, congenital heart disease, abnormalities of the retinal vessels, and primary intimal fibroplasia [3, 4]. In addition to that Down syndrome is associated with other vasculopathies. Autopsy finding in Down syndrome syndrome with cerebrovascular disorder revealed presence of fibrous hypertrophy of the arterial intimal walls, the tunica media was atrophic and internal elastic lamina was tortuous and duplicates [18]. Due to certain religious and legal issue, the autopsy of the deceased was not performed; although sometimes autopsy discloses certain unrevealed facts.

In conclusion, a case is reported with Down syndrome who developed extensive arterial thrombosis with no evidence of clear risk factor. This may indicate that Down syndrome itself may be a risk factor for thrombosis.

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