Marfan Syndrome with Spontaneous Rupture of Aneurysm of Common Iliac Artery

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Received: November 2, 2012; Initial review: November 26, 2012; Accepted: December 06, 2012.

Marfan syndrome rarely presents with peripheral artery and pulmonary artery aneurysms. We are presenting a case of a fifteen-year-old boy who presented to us with progressive lower abdominal pulsatile swelling with bruit in the right inguinal region for one month. He had typical marfanoid habitus, though there was no history of similar illness in family. CT angiogram revealed the presence having giant right common iliac aneurysm along with interlobar pulmonary artery aneurysm. He had spontaneous rupture of aneurysm in right common iliac artery.

Key words: *Aneurysm; Iliac artery; Marfan syndrome; Pulmonary artery.*

Marfan syndrome is an autosomal dominant, multisystem disease characterized by skeletal deformity like long bone overgrowth and other structural abnormalities, such as dislocation of the ocular lens, pneumothorax, mitral valve prolapse, and aortic arch dilatation [1]. Isolated iliac artery aneurysms are rare in the general population (0.03%) and represent 2% of all abdominal aortic aneurysms [2]. This association with Marfan syndrome is even rare. Pulmonary artery dilation is a minor cardiovascular criterion in the diagnosis of Marfan syndrome and very rarely seen [1]. We are reporting this case who had aneurysm of both pulmonary (right inter lobar) and right common iliac artery; along with sudden and spontaneous rupture of right common iliac artery.

CASE REPORT

A 15-years-old boy presented to us with progressive swelling of lower abdomen for last one month. There was no history of fever, dysurea, hematuria, and dyspnea or chest pain. Examination revealed marfanoid habitus with reduced upper and lower segment ratio or arm span to height ratio greater than 1.05, presence of wrist and thumb sign, arachnodactyly, clinodactyly, pectus carinatum, scoliosis and high arched palate. Ocular examination didn't reveal presence of ectopia lentis or other ocular disorder. Cardiovascular, respiratory and central nervous system examination was within normal limit. Per abdominal examination revealed presence of pulsatile mass in the pelvic area along with bruit in the right inguinal region. Peripheral pulses were equally palpable in the both the limbs. Routine blood investigation and chest x-ray was within normal limit. Echocardiography was also found to be normal. CT angiogram revealed the presence of giant aneurysm of right common iliac artery (***Fig.*** **1**) and peripheral pulmonary artery aneurysm of right inter lobar artery. Though he was posted for endovascular repair with stenting but suddenly he developed spontaneous rupture of common iliac artery with disappearance of abdominal swelling and increasing pallor. Finally he succumbed to his illness.

DISCUSSION

The most common cardiovascular abnormalities in Marfan syndrome include aortic and mitral valvular disease along with aneurysm and dissection of ascending aorta. Aneurysm in other location is uncommon in Marfan syndrome [4]. Though there are several case reports of patient of Marfan syndrome having common iliac artery aneurysm [4-6] in the literature but presence of pulmonary artery aneurysm is very rare with approximately eight cases per 100,000 autopsies [1]. However, there is no

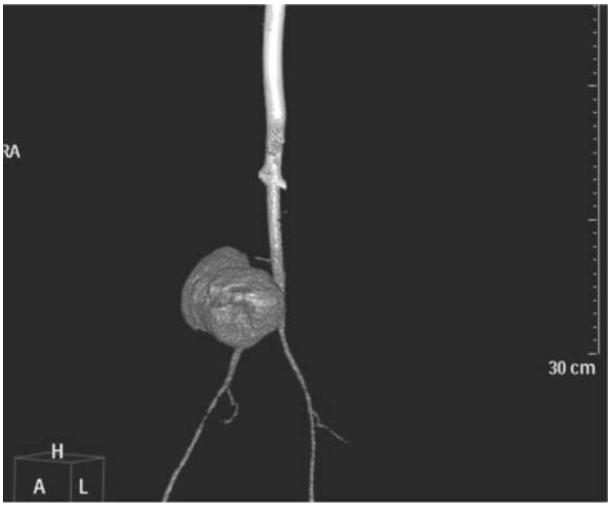

FIG. 1 *CT peripheral angiogram showed giant aneurysm of right common iliac artery.*

reported case so far which is showing presence of aneurysm in both peripheral and pulmonary artery. In our patient we found that he had aneurysm in both right common iliac artery along with right inter lober pulmonary artery and he was a previously undiagnosed sporadic case without any positive family history of Marfan syndrome. There is a reported case of spontaneous rupture of dissecting aneurysm of left common iliac artery during playing in a forty years old male patient [4]. But in our case it was sudden and spontaneous rupture had occurred without any physical exertion. While lying down, he developed acute onset pain followed by sudden collapse with severe pallor and disappearance of swelling in the abdomen. The occurrence of such an aneurysm suggests that the inherent mural weakness in Marfan syndrome is more widespread in the arterial tree than is generally appreciated [3]. It is secondary to cystic medial necrosis in the aorta and in multiple visceral arteries with extensive mucoid degeneration of the media of arterial wall [4]. So, patients with Marfan syndrome should be followed for signs of weakness of the peripheral arterial system and sometimes even with minor straining aneurysmal arterial wall can rupture. The peculiarity in our case was that even during rest the aneurysm got ruptured and he died almost immediately before resuscitation. When we diagnosed this case, we had planned early endovascular treatment with covered stenting of right common iliac artery as per recent advancement [7], but due to financial constraint it was delayed. Even in the acute condition, if we could have diagnosed rupture of the aneurysm earlier, he could have been saved by immediate closure of that artery by balloon dilatation just before the ruptured segment followed by immediate surgery with simple excision and end-to-end repair.

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Acute Lymphoblastic Leukemia with Treatment–Naïve Fanconi Anemia

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Received: April 25, 2012; Initial review: May 25, 2011; Accepted: December 14, 2012.

Fanconi anemia is known to have a predisposition to cancer, mostly associated with acute myeloid leukemia. We report an eight-year-old girl with treatment and naïve FA who developed acute-lymphoblastic-leukemia (ALL). She was initiated on chemotherapy but she failed to respond to treatment and died during induction phase of chemotherapy. While this association may be coincidental but possibility of transition of Fanconi anemia to ALL should be considered in view of high predisposition to cancer in this disorder.

Key words: *Acute lymphoblastic leukemia; Aplastic anemia; Fanconi anemia; Malignancy.*

Fanconi anemia is a genomic instability syndrome characterized by a wide array of congenital malformations, bone marrow failure, and a predisposition to cancer [1]. These patients usually present with reduced numbers of progenitor cells, including myeloid, erythroid, and multipotent progenitors [2]. In addition, almost 25% of patients develop a neoplastic disorder including acute