EJVES Extra **13**, 9–12 (2007)
doi:10.1016/j.ejvsextra.2006.07.005, available online at http://www.sciencedirect.com on ScienceDirect

SHORT REPORT

Spontaneous Non-traumatic Rupture of a Non-aneurysmatic Infrarenal Abdominal Aorta in a 10-Year Old Girl without Histological Evidence of Connective Tissue or Autoimmune Disease: A Case Report

R.A. Pol,[1] H.A.J.M. Kurvers,[2] J.B.G.M. Verheij,[4] G. Pals,[5] A.M. van der Wal,[6] J. Boers[3] and P.J.G. Jörning[1*]

[1]*Department of Surgery, Isala Klinieken, Zwolle, The Netherlands*
[2]*Department of Surgery, University Medical Center, St Radboud, Nijmegen, The Netherlands*
[3]*Department of Pathology, Isala Klinieken, Zwolle, The Netherlands*
[4]*Department of Genetics, University Medical Center Groningen, The Netherlands*
[5]*Department of Clinical Genetics, VU Medical Center Amsterdam, The Netherlands*
[6]*Department of Pathology, University Medical Center, Amsterdam, The Netherlands*

Spontaneous non-traumatic rupture of a non-aneurysmatic infrarenal abdominal aorta is an extremely rare condition, which to our knowledge, has only been described in adults. Rupture is usually associated with some form of connective tissue or autoimmune disease, which causes weakness of the aortic tunica media. This report is the first to present a case of a non-traumatic rupture of a non-aneurysmatic abdominal aorta in a 10-year old girl. Histological investigation of microscopic sections of the ruptured aorta, molecular biological, and DNA mutation analyses, did not demonstrate any abnormalities.

Keywords: Abdominal aorta; Spontaneous rupture; Connective tissue disease; Autoimmune disease; Child.

Introduction

Spontaneous non-traumatic rupture (SNR) of a non-aneurysmatic infrarenal abdominal aorta is a rare condition, which, to our knowledge, has only been described in adults.[1] In these cases, rupture may result from well-defined causes such as (hereditary) structural weakness of the aortic tunica media caused by some form of connective tissue or autoimmune[2] disease. The nature of this life-threatening condition necessitates early identification and immediate surgical repair.

In most cases presented in the literature, the cause of SNR of a non-aneurysmatic infrarenal abdominal aorta was shown histologically to be associated with some kind of connective tissue or autoimmune[1] disease as indicated by structural weakness of the aortic tunica media.

This report is the first to present a case of a spontaneous rupture of a non-aneurysmatic abdominal aorta in a 10-year old girl in the absence of histological abnormalities usually seen in connective tissue or autoimmune disease.

Medical History

Patient A, a 10-year old Caucasian girl, was referred to the emergency room by a general practitioner because of a sudden onset of acute abdominal pain and nausea. Significant past medical problems were denied. There was no history of trauma. On physical examination, the patient was agitated, screaming, and diaphoretic. Her blood pressure was 114/75 mm Hg and her pulse rate 146 beats/minute. Palpation of the abdomen was diffusely painful. No pulsatile mass, guarding, or rebound tenderness was found. Laboratory

*Corresponding author. P.J.G. Jörning, Department of surgery, Isala Clinics, Dr. van Heesweg 2, 8025 AB Zwolle, The Netherlands.
E-mail address: p.j.g.jorning@isala.nl

tests, consisting of full blood count and differential, urea and electrolytes, only revealed a mild leucocytosis (14.6×10^3) and a normale erythrocyte sedimentation rate (ESR). Abdominal ultrasonography revealed no signs of appendicitis, volvulus, intussusception, or free fluid. A plain abdominal X-ray suggested a volvulus of the sigmoid.

Because of the probability of a volvulus of the sigmoid, a colonoscopy was performed by the pediatrician, which revealed no abnormalities. Following colonoscopy, the patient had less pain and discomfort and she was admitted to the pediatric ward for further observation.

Eleven hours after admission the patient became unresponsive and hypotensive, her blood pressure decreased to 70/30 mm Hg and her pulse rate increased to 120 beats/minute. Laboratory tests revealed a hemoglobin level of 2.1 mmol/L. These findings suggested hemorrhagic shock of unknown origin. The abdominal ultrasound was repeated and revealed a large retroperitoneal hematoma. Following successful resuscitation, the patient was rushed to the operating room where an emergency midline laparotomy was performed. This revealed a large midline retroperitoneal hematoma originating from a tear in the infrarenal aorta. The tear was about 1.5 cm in length and originated 1–2 centimeters below the renal arteries. The aortic wall appeared normal, aneurysmatic dilation was not observed. Following clamping of the aorta proximal and distal to the rupture, primary repair of the aortic tear was attempted using 6/0 Prolene (Ethicon inc., Johnson&Johnson, Somersville, NJ, USA). The aortic tissue, however, was found to be extremely fragile as a result of which multiple attempts to achieve primary repair failed. This prompted us to place one vascular clamp directly below the renal arteries and another just distal to the initially most distally placed clamp. Removal of the initially placed vascular clamps demonstrated severe laceration at the site of clamping. We therefore opted to replace the damaged aortic segment by a 10 mm PTFE tube graft (GORE-TEX® Vascular grafts, Gore Medical Products, Flagstaff, Arizona, USA) that was anastomosed in an end-to-end way with 5/0 prolene. Following careful removal of the proximal vascular clamp, the aorta was severed at the site of clamping with partial avulsion of the origin of the left renal artery. In order to obtain more proximal control, the cardiothoracic surgeon was consulted who performed a thoracotomy. To this end, the incision of the laparotomy was extended over the fifth left intercostal space. To enhance exposure of the aorta, the diaphragm was divided in a circumferential fashion. Following identification and exposure of the descending thoracic aorta a vessel loop was placed around the aorta. Mild traction on the vessel loop already caused immediate and complete transection of the thoracic aorta. Aortic retraction resulted in uncontrollable hemorrhage, cardiac arrest and death.

During autopsy, a large retroperitoneal and intramesenteric haematoma was observed. No malformations were observed suggestive of an inherited connective tissue disorder. There were no pre-existing macroscopic abnormalities of the aorta (Fig. 1).

Two samples of iliac, renal and coeliac arteries each as well as 5 samples (normal and ruptured areas) of the abdominal aorta were submitted for histologic examination. HE, PAS, EvG, Gomorri Silver and Alcean Blue stain did not reveal any pathology, as judged by a local pathologist (JEB) and by the Academic consultant (AvdW). Mucoid degeneration, fragmentation of elastic fibers or any inflammation was absent. (Fig. 2). A skin biopsy was taken for fibroblast culture. Immunohistochemical stains were judged to be of very limited value and were not performed. Tissue was not specifically submitted for electron microscopy (EM) at the time of autopsy; tissue taken from paraffin blocks was judged to be less informative being of insufficient quality for high power EM examination of collagen fibers. At autopsy, no abnormalities were found other than ruptured vessels.

The genes involved in Marfan syndrome (*FBN1* and *TGFBR2*), and the arterial form of Ehlers-Danlos syndrome (*COL3A1*) were analyzed. Collagen protein analysis was performed in cultured fibroblasts and showed no abnormalities. The electrophoresis patterns of metabolically labelled collagens type I, III and V were normal and the relative amount of excreted collagen type III was 15% (normal range 5–18%). No mutations were detected in the *COL3A1* gene, by sequencing of the complete coding sequence on cDNA from mRNA. Bi-allelic expression of

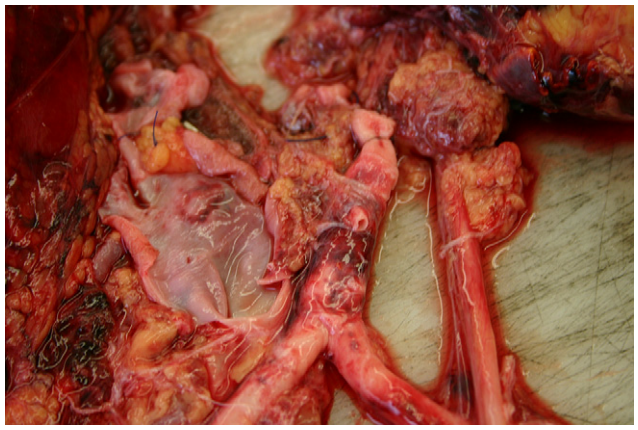

Fig. 1. Macroscopic view of the aorta during autopsy. No abnormalities (aneurysmatic dilation, dissection) are seen.