Congenital umbilical arterio-venous malformation: a word of caution

Horea Gozar*, Liliana Gozar, Catalin Constantin Badiu* and Horatiu Suciu*

INTRODUCTION

Blood flow through the umbilical arteries ceases after birth physiologically. The arteries and the umbilical vein narrow while the intimal and medial layers undergo aseptic necrosis. Within a few weeks, they are anatomically closed.

Umbilical arterio-venous fistulas are rare congenital malformations involving one, two or even more umbilical arteries and the umbilical vein. Clinically, they usually present as a pulsatile herniation. Due to high blood flow through the fistula, they may cause high-output heart failure in newborns.

PATIENT PRESENTATION

We report on a case of an 18-month old girl who was transferred to our unit with a pulsatile umbilical hernia, which was diagnosed initially as an uncomplicated umbilical bowel herniation. The girl presented in good clinical condition with normal age-related weight. Laboratory investigations showed only a slight elevation of serum aspartate aminotransferase (51 U/l). The umbilical herniation presented as an oval tumour (4.5 × 2 cm) fully covered with normally coloured skin and synchronously pulsating to her heart beat.

Echocardiography showed normal atrioventricular and ventricular connections with mildly dilated left cardiac chambers and a normal left ventricular function. The inferior caval vein was moderately dilated (1.12 cm). The Doppler ultrasound of the umbilical tumour revealed a large arterio-venous vascular malformation with a haemodynamically significant blood shunting [pulmonary (Qp) to systemic (Qs) blood flow ratio of 1.3:1].

As magnetic resonance imaging was not possible at that time at our institution, the performed CT-angiography revealed permeable umbilical veins and arteries, which were communicating in a dilated arterio-venous fistula (Fig. 1). Neighbouring major vessels including the abdominal aorta, the truncus coeliacus, the superior mesenteric artery and the renal arteries showed no abnormalities. The umbilical arteries, emerging from the hypogastric artery, ascend to the umbilicus and after a tortuous trajectory flow into the umbilical vein. The entire umbilical vein, with contrast in the arterial phase, met the left branch of the portal vein.

The case was evaluated in a multidisciplinary team and the patient was scheduled for operation. A semilunar incision (5 cm) was performed avoiding the umbilicus from the left side. The umbilical tumour was carefully dissected. On the caudal side of the incision we found three anatomic elements emerging from the tumour, two umbilical arteries and the urachus which had a large calibre and was patent sparing the junction of the vascular malformation. On the cranial side of the incision the umbilical vein was identified and dissected (Fig. 2). All vessels, as well as the urachus, were ligated and divided. The tumour was excised and the umbilical ring sutured, followed by a plasty of the umbilical skin. There were no operative complications. Postoperative recovery was uneventful and the patient was discharged 10 days after surgery.

Three months after the operation the girl presented in a normal clinical state and gained ~1 kg in weight. Blood samples showed normal transaminase activity. CT-angiography showed a normal aspect of the intra-abdominal vessels.

DISCUSSION

Umbilical hernia is a rare but well-known congenital malformation. The vast majority of these hernias are closing spontaneously prior to the fourth year of life with a very low incidence of
incarceration (0.2%). Therefore, it is safe to postpone or even avoid operation in this pathology. Nevertheless, a pulsatile hernia, as described in our patient, may represent a very rare vascular malformation, requiring further clinical investigations.

Several types of umbilical vessel malformations have been described in the literature: numerical aberrations of arteries or dilated umbilical veins [1, 2] or aberrant drainage of venous blood. Furthermore, umbilical arterio-venous fistulas with a direct connection between arterial and venous vessels leading to severe complications have been described [3, 4]. Nevertheless, in our patient both umbilical arteries and the umbilical vein were connected through a vascular tumour. Furthermore, the urachus was attached to the vascular tumour and still permeable but closed at the junction with the arterio-venous fistula. Significant complications might arise from significant blood shunting potentially leading to congestive heart failure. Moreover, haemorrhage from trauma or thrombus formation represents other potential complications [3, 4]. Thus, timely operative therapy should be taken into consideration.

Ultrasound is the diagnostic tool of choice for the detection of umbilical vessel malformations not only in the postnatal but also in the prenatal period [5]. In order to avoid severe complications, we advocate a close clinical and sonographic follow-up of the detected vascular malformation. Furthermore, if umbilical hernias are diagnosed after birth, even if not pulsatile, we advocate detailed ultrasound investigation, which is a simple and non-invasive method for exclusion of possible vascular tumours.

Conflict of interest: none declared.

REFERENCES