

VACTER syndrome with situs inversus totalis

Case report and a new syndrome

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Abstract

Introduction: The association of situs inversus totalis (SIT) and VACTERL syndrome an extremely rare coincidence.

Patients: The patient was first diagnosed as simple SIT with lumbosacral neoplasms according to the prenatal magnetic resonance imaging (MRI) examination; however, the local hospital ignored the important to physical examination so that missed anal atresia with fistula. The patient was presented to our hospital owing to constipation for 1 week. And then, she was diagnosed as VACTER syndrome with situs inversus totalis.

Results: Anorectoplasty was performed to treat constipation, one month later, we performed intramedullary tumor resection and pathological diagnosis of ependymal cyst. Postoperative recovery was uneventful and the baby was doing well at 5-months follow up.

Conclusion: It is extremely necessary for careful physical examination and detailed auxiliary examination to each system (including echocardiography, MRI, and so on) when diagnosing SIT. Also, recognizing and understanding the spectrum of situs anomalies is important, which aids in the diagnosis of disease and accordingly plan the therapeutic interventions.

Abbreviations: CT = computed tomography, MRI = magnetic resonance imaging, SIT = situs inversus totalis.

Keywords: ependymal cyst, situs inversus totalis, VACTERL syndrome

1. Introduction

VACTERL association is also known as VATER association or unusual syndrome, which refers to the non-random co-occurrence of congenital malformations, including vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula with esophageal atresia, and radial and renal dysplasia and limb defects,^[1,2] and the diagnosis requires the presence of at least 3 component features.

Situs inversus totalis (SIT) is an uncommon congenital condition, which is defined by a left-right transposition of the normally asymmetrical organs of the body as a mirror-image and its incidence accounts for 1/8000 to 1/25,000 live-born infants.^[3] This is characterized by dextrocardia with complete reversal of the heart chambers, the aorta turns to the right, and the left lung has 3 lobes and the right lung only 2. In the abdomen, the stomach, spleen, and pancreas are right sided of the body, the liver and gallbladder are left sided, and the colonic flexures

are reversed. The crucial structures associated with situs were evaluated by chest radiography, ultrasonography, computed tomography, or magnetic resonance imaging.^[4–6]

The association of SIT and VACTERL syndrome is an extremely rare coincidence. Till date there were no studies that reported from google scholar and PubMed on the subject.

2. Case report

Ethical approval was obtained from the Ethics Board of the Children's Hospital of Shanghai, Shanghai Jiao Tong University. Written informed consent was obtained from the parents on behalf of the child. The reasons of admission, the clinical and laboratory findings, the diagnostic and therapeutical approach

A 65-day-old babygirl (G2P2 5.5-kg with a healthy elder brother) was first misdiagnosed as simple SIT with lumbosacral neoplasms according to the prenatal MRI examination (Fig. 1). She was then admitted to the local hospital and suggested for observation of treatment. The reasons of admission: she was presented to the hospital owing to constipation for 1 week. The clinical and laboratory findings: physical examination revealed heart sound arising from the right, abdominal distention, and anal atresia with fistula (Fig. 2). Neurological examination was within normal limits. Parents denied of having any family history, no significant history of infection before the onset of disease, and no special history of drug use were present. Echocardiographic results reveal eddextrocardia and atrial septal defect (II) (Fig. 3). Chest plain film demonstrated dextrocardia (Fig. 4). Computed tomography (CT) revealed horseshoe kidney, peritoneal organ inversion (Fig. 5). Magnetic resonance imaging (MRI) showed lumbosacral neoplasms (cystic lesions), filum terminale fatty degeneration (Fig. 6). The diagnostic and therapeutical approach: this patient was diagnosed with VACTER syndrome (horseshoe kidney, anal atresia with fistula, atrial septal defect, ependymal cyst) and SIT. Anorectoplasty was performed to treat constipation. One month later, we performed intramedullary tumor

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The authors declare that they have no conflict of interest.

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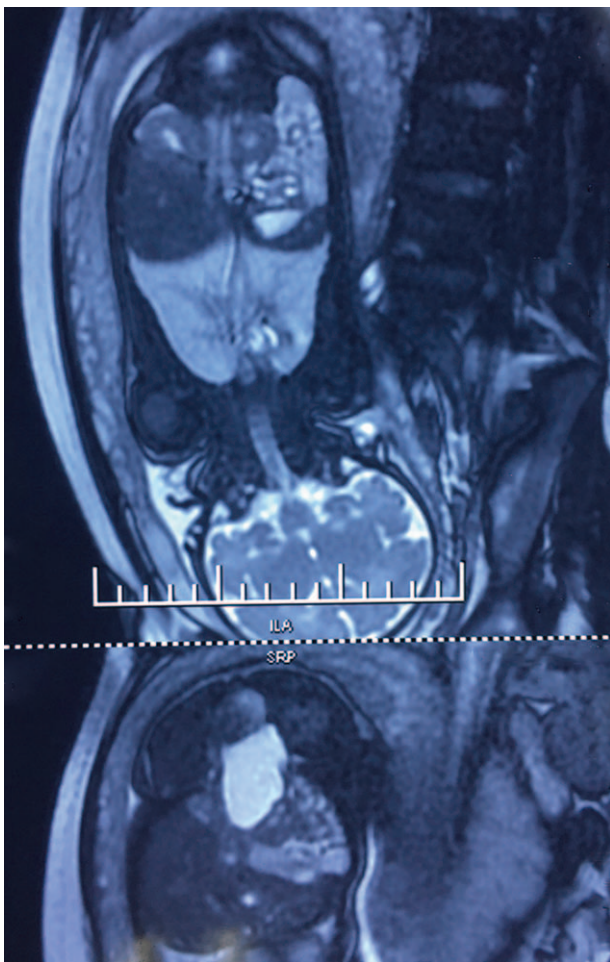


Figure 1. Prenatal MRI examination revealed situs inversus totalis, lumbosacral neoplasms. MRI = magnetic resonance imaging.

resection and pathological diagnosis of ependymal cyst (Fig. 7). Postoperative recovery was uneventful and the baby was doing well at 5-months follow up.

3. Discussion

VATER association was first named in the early 1970s. As initially described, the condition included of a group of congenital malformations: vertebral defects, anal atresia, tracheo-esophageal fistula (TEF) with esophageal atresia, and radial and renal dysplasia. And the diagnosis requires the presence of at least 3 component features. The incidence is estimated at 1/10,000 to 1/40,000 of the normal population throughout the world. However, we could not ignore the undeniable defect of diagnosis that the different hospitals have used differing diagnostic criteria, and there are no exist obvious genetic testing to make a definite diagnosis to this complex syndrome just like Down syndrome. And then some studies would likely overestimate or overlook the incidence of VACTERL association.^[1,2]

SIT is an uncommon entity that often occurs concomitantly with other abnormalities. The relationship between SIT and VACTERL syndrome still remains unclear and further studies are needed to identify the precise genetic and molecular reasons involved in the malformation of these patients. On account of the frequency of associated malformations of inverted organs and



Figure 2. Physical examination anal atresia with fistula.

vascular and nervous component variations that make it difficult and challenging for surgical management. Hence, more attentions should be paid during diagnosis and preoperative staging.

Few papers have reported gastrointestinal malformation or esophageal atresia with distal tracheoesophageal fistula or other malformations with SIT.^[5,7] Our case of SIT and VACTERL syndrome is quite rare, and this is the first time to report this new syndrome. Our patient was diagnosed with VACTER syndrome (horseshoe kidney, anal atresia with fistula, atrial septal defect, ependymal cyst) and SIT. Regarding the asymmetry and the

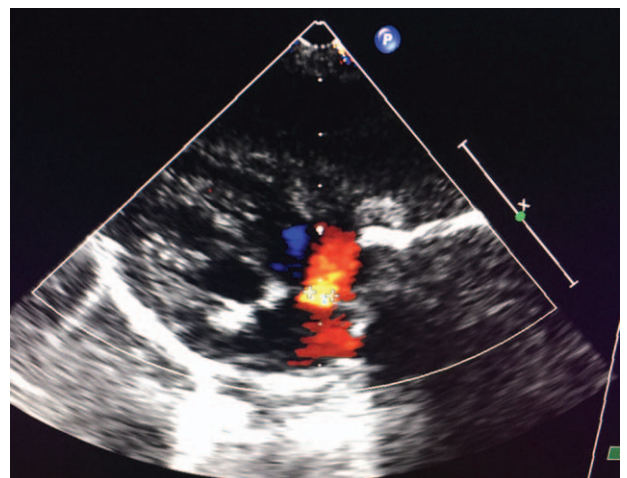


Figure 3. Echocardiography revealed dextrocardia and atrial septal defect (II).



Figure 4. Chest plain film demonstrated dextrocardia.



Figure 5. Computed tomography (CT) revealed horseshoe kidney, peritoneal organ inversion.

central nervous system, it is well known that the brain is an asymmetric organ and that cases of situs inversus can be involved in the deformation of the brain such as cerebellar hypoplasia,^[4,8] and fortunately, the patient's head MRI appeared normal. Two rare disease conditions occurred by coincidence, but the doctors lost their attentions to perform systematic physical examination. Auxiliary examination was conducted but missed the diseased anal area. Therefore, the possibility of malformation in other organs should always be considered early to avoid misdiagnosis and delayed treatment.

In summary, it is extremely necessary for careful physical examination and detailed auxiliary examination to each system (including echocardiography, magnetic resonance imaging, and so on) when diagnosing SIT. Also, recognizing and understanding the spectrum of situs anomalies is important, which aids in the diagnosis of disease and accordingly plan the therapeutic interventions.

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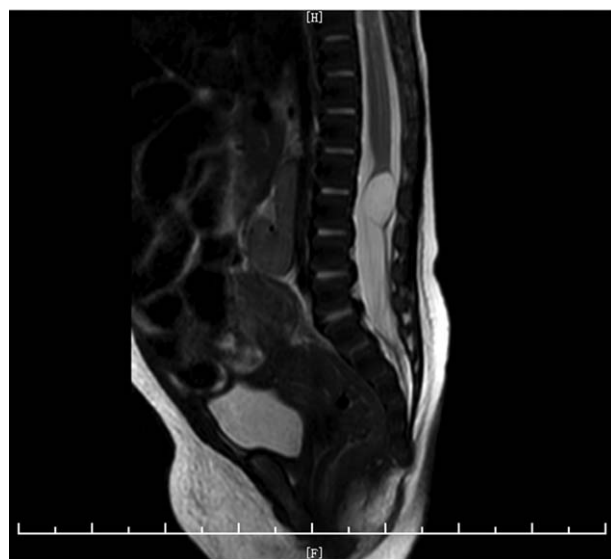


Figure 6. Magnetic resonance imaging (MRI) showed lumbosacral neoplasms (cystic lesions), filum terminale fatty degeneration.

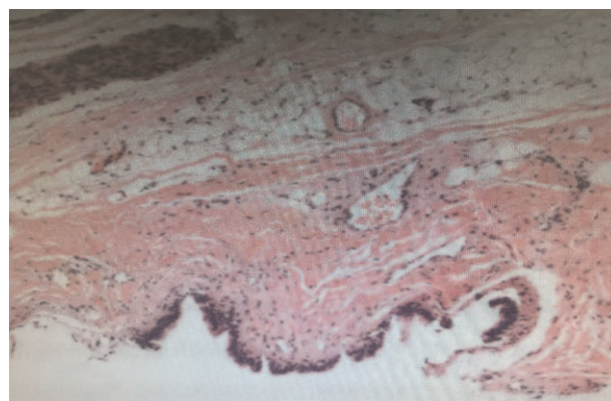


Figure 7. HE stain showed ependymal cyst.

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