

Intrathoracic kidney in a case of trisomy 18

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SUMMARY: Chu I-W, Yeh S-J, Lin Y-C. Intrathoracic kidney in a case of trisomy 18. *Turk J Pediatr* 2008; 50: 176-178.

We report a case of trisomy 18 (47, XX+18) with intrathoracic kidney. It was discovered incidentally as an opacity at the right lower hemithorax on the chest radiograph, later confirmed by magnetic resonance imaging. Ultrasound can be used as the first-line imaging modality for intrathoracic kidney, even in the prenatal period.

Key words: trisomy 18, intrathoracic kidney, chest radiograph, MRI, double outlet right ventricle, ventricular septal defect, pulmonary stenosis, patent ductus arteriosus.

Trisomy 18 syndrome is the second most common chromosomal aberration syndrome caused by an extra number 18 chromosome. It was first described in 1960 by Edwards et al.¹; therefore, it is also named as Edward's syndrome. Its incidence is given as 0.3 per 1,000 live births². A variety of anatomic abnormalities involving almost all organ systems have been noted during obstetric ultrasonography and at the time of autopsy³. Congenital kidney abnormalities occur in 33 to 70% of the cases^{4,5}. Horseshoe kidney, polycystic kidney, hydronephrosis and hydroureter are the more common findings, while ectopic kidney is less seen, with an incidence of about 10%. We present a case of intrathoracic kidney in a baby with trisomy 18 (47, XX+18). To our knowledge, this is the first such case ever reported in the literature.

Case Report

This 33-day-old female infant was referred to our department from a local district hospital due to congestive heart failure. She was the third child born to Chinese, non-consanguineous, healthy parents. Her mother was 38 years old and her father was 43 years old at the time of conception. She was delivered by cesarean section, with a birth weight of 1.6 kg. On admission, the ultrasonography study confirmed multiple congenital heart defects, including double outlet right ventricle, ventricular septal defect, pulmonary stenosis and patent ductus arteriosus. Her congenital

heart disease was managed with digoxin and diuretics. She also had other congenital anomalies, including clenched hands, rocker bottom feet and talipes calcaneovalgus.

Chromosome analysis of the peripheral blood confirmed trisomy of chromosome 18 (47, XX+18). Her chest radiograph showed an opacity in the posterior aspect of the right lower hemithorax (Fig. 1A). Magnetic resonance imaging (MRI) of the thorax revealed an intrathoracic eventration of the right kidney (Fig. 1B) and confirmed the diagnosis of intrathoracic kidney. Her renal function was normal.

Discussion

The majority of ectopic kidneys lie inferior to the normal position such as in the lower lumbar or pelvic region because of ascending failure⁶. Intrathoracic kidney is very rare and is the least frequent of all ectopic kidneys, with a prevalence of less than 1 per 10,000⁷. In our review of the literature, two cases of trisomy 21 with thoracic kidney have been reported by Navarro et al.⁸ and Stein et al.⁹. However, there has been no case of intrathoracic kidney reported in a baby with trisomy 18. The majority of intrathoracic kidney cases occur on the left side and occur almost twice as frequently in male as in female patients¹⁰. In this case, the ectopic kidney was located in the right instead of the left chest cavity. Interestingly, the intrathoracic kidneys in the previously reported two cases of trisomy 21 were also located in the right chest cavity.

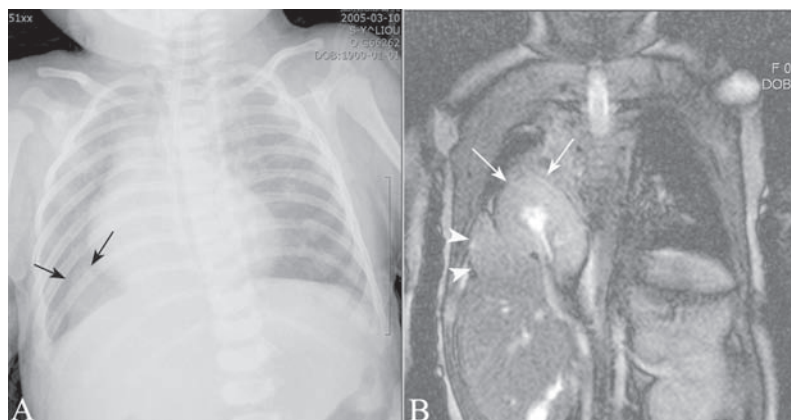


Fig. 1. (A) The posteroanterior chest X-ray reveals an opacity in the posterior aspect of the right lower hemithorax (arrows). (B) The coronal section of T2-weighted magnetic resonance thoracic imaging shows an intrathoracic eventration of the right kidney (arrows) and a portion of the liver (arrowheads). Intrathoracic kidney was diagnosed.

Intrathoracic kidneys have been classified into four subtypes: 1) true thoracic ectopia with a normally developed closed diaphragm; 2) eventration of the diaphragm; 3) diaphragmatic hernia (Bochdalek), either congenital or acquired; and 4) traumatic rupture of the diaphragm with renal ectopia⁷. Unlike the low ectopic kidneys, which are the frequent sites for stone formation, ureteropelvic junction obstruction and infection, most intrathoracic kidneys function normally. The thoracic kidney often is deformed in size and shape but remains fully functional⁷. Most cases of intrathoracic kidneys are asymptomatic and often present as incidental findings of a round, well-defined opacity on the chest radiographs. Usually, the thoracic kidney requires no medical or surgical therapy.

In this case, the renal function was normal and no symptom was related to this anomaly. An opacity at the posterior aspect of the right hemithorax on the chest radiograph was the first finding. Benign or malignant pulmonary or pleural mass, pneumonia and diaphragmatic eventration over the right lower lung were considered. An MRI confirmed the diagnosis of intrathoracic kidney. As we mentioned earlier, most cases with intrathoracic kidneys are incidentally found on the chest radiograph. We consider that chest radiographs can be used as a screening tool for intrathoracic kidney. However, such diagnosis cannot be made based on the chest radiography alone. Other imaging modalities should be performed to confirm the diagnosis of intrathoracic kidney. Prior to the advent of crossed-section tomography, the diagnosis of intrathoracic kidney was confirmed,

in most patients, by intravenous pyelography (IVP)⁶. Although IVP can confirm the diagnosis of intrathoracic kidney, it does not provide any information regarding associated anomalies or the possibility of malignancy. Since the 1980s, other imaging modalities, including ultrasonography, computed tomography (CT) and MRI have been reported as useful diagnostic tools^{7,8}. Ultrasonography is a safe and useful tool for the diagnosis of intrathoracic kidney even in the prenatal period¹¹ and can be considered as the first-line imaging modality when intrathoracic kidney is suspected.

The incidence of malformations of the kidney and urinary tract is much higher in many forms of chromosome aberrations than in the general population. Therefore, an intrathoracic kidney should be considered when there is an opacity in the lower lung field on chest radiograph, especially when the patient has chromosomal aberrations.

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