

Horseshoe Kidney And Spinal Cord Lipoma In An Infant With Vater Association

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Citation

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Abstract

VATER association is basically a combination of vertebral, anal, tracheoesophageal and radial limb anomalies that for the diagnosis at least three of these anomalies should be present. Various renal anomalies and distal spinal cord pathologies are among the spectrum of this association. In this case report we present an infant with two rare anomalies, i.e. horseshoe kidney and spinal cord lipoma.

INTRODUCTION

The VATER association is a collection of certain congenital morphological malformations typically including vertebral, anal, tracheoesophageal, radial and renal anomalies (₁). Many renal and spinal cord anomalies have been described previously (_{2, 3}). In this case report, we describe a two-month-old male infant with horseshoe kidney and spinal cord lipoma, an association which has not been previously published.

CASE HISTORY

A two-month-old male infant referred to our Pediatric department due to deformity in his arms. He was born via normal spontaneous vaginal delivery in the 8th gestational month. His weight was 2000g, length 47 cm, head circumference 34 cm. His mother was a smoker and had a history of gentamicin injection in the first trimester. There was no consanguinity between the parents and they were healthy. Physical examination revealed hypoplasia of both thumbs and radial deviation of both hands. Neurological examination was normal. Cardiac examination revealed a systolic murmur of 2/6 grade. He also had bilateral inguinal hernia. The results of laboratory examinations were within normal limits. Upper extremity roentgenograms showed absence of radius bilaterally and bilateral club hand (Figure 1).

Figure 1

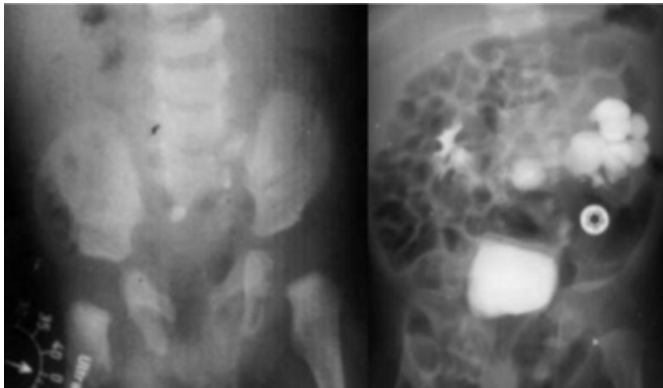
Figure 1: Extremity roentgenogram showing absence of radius and radial deviation of the hand.



Echocardiography showed an atrial septal defect. Abdominal ultrasound revealed a mild echogenicity increase and ectasia of pelvicaliceal system in the left kidney and a hypoechoic connecting band-like structure between lower poles of kidneys. The lengths of the left and right kidneys were 6 and 5 cm respectively. The parenchymal thicknesses of left and right kidneys were 7 and 6 mm respectively. Radiographs taken as part of an excretory urogram showed partial agenesis of sacrum, dilatation of the left collecting system and a midline dilated structure with contrast enhancement that was connected to the left kidney (Figure 2A, B).

Figure 2

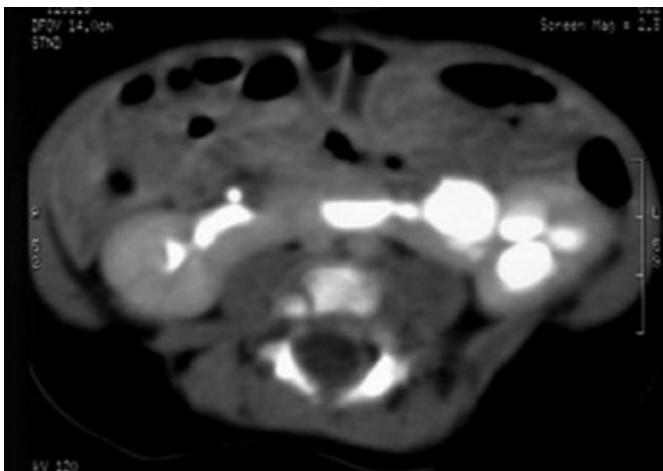
Figure 2: Excretory urogram showing partial agenesis of sacrum (A) and dilatation of the left collecting system and a midline dilated contrast-enhancing structure (B).



Abdominal CT showed dilatation of the left kidney, fibrous connection between lower poles of both kidneys and a midline dilated structure with contrast pooling in the fibrous band (Figure 3).

Figure 3

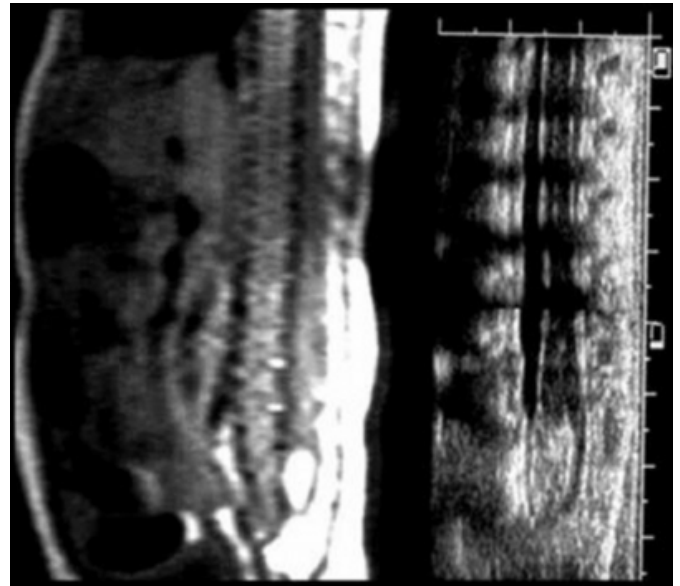
Figure 3: Abdominal CT slice demonstrating dilatation of the left kidney, fibrous connection between lower poles of both kidneys and a midline dilated structure with contrast pooling in the fibrous band.



Cranial CT was normal. Due to partial sacral agenesis, spinal ultrasound and MRI examinations were performed that revealed spinal cord lipoma at the level of sacrum (Figure 4).

Figure 4

Figure 4: Composite spinal ultrasonogram and sagittal T1 weighted MR scan showing spinal cord lipoma 11x7 mm in diameters at the level of sacrum.



Our patient was diagnosed as having horseshoe kidney with VATER association.

Cystoscopy was attempted in order to perform retrograde pyelography and, if necessary, place a ureteral stent. We were unable to pass an 11 F cystoscope through the urethra and the intervention was terminated by placing a 8F nelaton catheter. One month later the patient was operated in order to relieve hydronephrosis by relieving the ureteropelvic region, and to correct malrotation. In the operation, the isthmus, which was in the midline with many aberrant vessels, was resected. The kidneys were mobilized and than suspended to psoas muscles and lateral abdominal wall. Control excretory urogram, performed on the second month after operation, revealed persistence of the dilatation and absence of midline structure. Ultrasound examination on ten months after the operation demonstrated normal kidneys without evidence of hydronephrosis.

DISCUSSION

The VATER syndrome is first described by a Turkish physician, Prof. Burhan Say, in 1968 (4). It is a combination of certain congenital anomalies including vertebral, anal, tracheoesophageal, radial and renal defects (2, 3). A wide range of congenital anomalies attributed to this syndrome thus in 1973 the VATER association term was first used as an acronym for a non-random association of anomalies (5). It is diagnosed as having at least three of the following five VATER anomalies: vertebral anomalies, anal atresia,

tracheoesophageal fistula and/or esophageal atresia, radial-ray limb anomalies, renal anomalies (3). Although many studies require three or more of the defects for the diagnosis, cases with two features may be viewed as belonging to the same continuum. The limits of the VATER association and the extent of the overlap with other defects or associations are as yet unclear (6). Since our case has radial, vertebral and renal anomalies, he was diagnosed as having VATER association.

The aetiology and pathogenesis of the VATER association are unknown. No teratogens, chromosomal aberrations or familial occurrences have been implicated. It has been postulated that associations are defects of blastogenesis, originating from the midline developmental field of the early embryo (7). Defects in mesodermal development during embryogenesis were suggested as a possible etiology. Since the critical period of organogenesis is at or before the sixth or seventh week of gestation, faulty mesodermal development during the first five weeks of gestation is thought to be the possible cause of this syndrome, but it is generally accepted as a multifocal developmental disorder (1,2,3,4,5). It was reported that the presence of associated anomalies, such as those encountered in humans with the VATER (or VACTERL) or CHARGE associations, may have a common pathogenesis, but at this stage that remains largely unknown. Several observations point to the possible involvement of the neural crest in the origin of some components of these associations (8). Apoptosis, programmed cell death, which is distinct from cell necrosis, plays an important role during embryologic morphogenesis besides cell proliferation and differentiation (8). Therefore apoptosis could also have important consequences in VATER association.

In the literature there are reports of various genitourinary abnormalities in this syndrome like duplications of the bladder, agenesis of ipsilateral kidney, anomalous insertion of the native ureter into the ejaculatory duct, congenital uterovesical fistula, single vaginal ectopic ureter, urolithiasis, ureteropelvic junction obstruction, crossed fused ectopia and severe reflux. Also there are other anomalies like radial artery hypoplasia, tracheal agenesis with associated laryngoesophageal cleft and, tetralogy of Fallot, single umbilical artery, imperforate anus with cloaca, hydrocephalus and aqueductal stenosis in the literature (1,2,3,4,5). But according to our knowledge there is no report of the coexistence of horseshoe kidney and spinal lipoma in VATER association, and this is the first report of this rare

occurrence.

Horseshoe kidney, which has an estimated incidence of 1 to 4 in 1000 persons, is the result of fusion of metanephric buds between the fourth and eighth weeks of embryogenesis, blocking their cephalic migration and normal rotation (9, 10). Aberrant migration of posterior nephrogenic cells to form a connection between the two developing kidneys might be the cause of fusion (9). Most often, this fusion consists of just the lower poles connected by normal renal parenchyma, but on occasion this fusion can be more extensive or the connection can be a fibrotic band of tissue. The fused kidneys fail to ascend and rotate into the usual position. Horseshoe kidneys are associated with ureteropelvic junction obstruction, ureterovesical reflux, urinary calculi, and urinary tract infections (11). Typically the renal pelves and ureters are anteriorly displaced, with the ureters lying anterior to the isthmus with a tendency to kink, often resulting hydronephrosis.

There is a risk of cancer development in HK.

Hypernephroma, renal pelvic tumors, Wilms' tumor, rhabdomyosarcoma and teratoma are among the reported neoplasias arising from HK (12). The increased incidence of tumors, especially of Wilms' tumor, may be attributable to the predisposition due to embryologic lesion which causes HK. Histological analysis of resected tissue in our case yielded no tumor.

In the literature distal spinal cord pathologies in the VATER association were reported which indicate that concordance exists between spinal dysraphism and VATER association (13, 14). Pathologically spinal structures in the reported series indicate primary mesodermal disorder of closure, which than involves the neuroectoderm to varying degrees. Although the exact incidence of spinal cord lipoma or spinal dysraphism in VATER association is not known, developments in neuroimaging techniques will help early recognition of distal spinal cord pathologies.

Due to the indubitable benefits of early corrective urologic surgery in children with VATER association we suggest a careful and dedicated genitourinary radiological examination and appropriate urologic surgery and emphasize the importance of neuroimaging of the spinal canal with VATER association even in the absence of clinically apparent neurological damage.

References

1. Barnes JC, Smith WL: The VATER Association. *Radiology* 1978; 126:445-449

2. Werner W, Beintker M, Schubert J, Kaiser WA: The VATER syndrome from the urologic viewpoint. *Urologe A* 1998; 37:203-205
3. Botto LD, Khoury MJ, Mastroiacovo P, et al.: The spectrum of congenital anomalies of the VATER association: an international study. *Am J Med Genet* 1997; 11; 71:8-15
4. Say B, Gerald PS: A new polydactyly/imperforate-anus/vertebral-anomalies syndrome? *Lancet* 1968;2:688
5. Quan L, Smith DW. The VATER association: vertebral defect, anal atresia, T-E fistula with esophageal atresia, radial and renal dysplasia: a spectrum of associated defects. *J Pediatr* 1973;82:104
6. Erkan D, Lam LA, Yazici Y, Magid SK. VATER association: is it recognized by rheumatologists ? *Clin Rheumatol* 2001;20:128
7. Kallen K, Mastroiacovo P, Castilla EE, Roberts E, Kallen B. VATER non-random association of congenital malformations: Study based on data from four malformation registers. *Am J Med Genet* 2001;101:26
8. Beasley SW, Pardo JD, Qi BQ, Tovar JA, Xia HM. The contribution of the adriamycin-induced rat model of the VATER association to our understanding of congenital abnormalities and their embryogenesis. *Pediatr Surg Int* 2000;16:465
9. Lee CT, Hilton S, Russo P. Renal mass within a horseshoe kidney: preoperative evaluation with three-dimensional helical computed tomography. *Urology* 2001;57:168
10. Kehagias DT, Gouliamos AD, Vlahos LJ. Horseshoe kidney associated with anomalous inferior vena cava. *Eur Radiol* 1999;9:935
11. Bradshaw A, Donnelly LF, Foreman JW. Thrombocytopenia and absent radii (TAR) syndrome associated with horseshoe kidney. *Pediatr Nephrol* 2000;14:29
12. Talpallikar MC, Sawant V, Hirugade S, Borwankar SS, Sanghani H. Wilms' tumor arising in a horseshoe kidney. *Pediatr Surg Int* 2001;17:465
13. James HE, Chesnut R, Krous H, Billman G, Jones KL, Jones M. Distal spinal cord pathology in the VATER association. *J Pediatr Surg* 1994;29:1501
14. Chestnut R, James HE, Jones KL. The Vater association and spinal dysraphia. *Pediatr Neurosurg* 1992;18(3):144-8

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