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Urogenital and Other Associated Anomalies in Patients With Anorectal Malformations

Mohammad Kazem Sabzehei [1], Seyed Habibollah Mousavi-Bahar [2], Hassan Bazmamoun [1*]

[1] Besat Hospital, Hamedan University of Medical Science, Hamedan, Iran
[2] Department of Urology, Shahid Beheshti Hospital, Hamedan University of Medical Science, Hamedan, Iran

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ABSTRACT

Anorectal malformations (ARM) comprise a wide spectrum of diseases and associated anomalies. ARM are the major factors that contribute to high morbidity and mortality in neonates. The aim of this retrospective study was to review the incidence of urogenital and other associated anomalies in neonates in whom the diagnosis of ARM was confirmed between 2005 and 2010. We found cases of 58 patients with ARM; of these patients, 65.5% were boys and 17.2% were born prematurely. The mean birth weight was 2.8 kg (SD 0.5). The following anomalies associated with ARM were observed in 63.8% of the patients: urogenital (43.1%), cardiovascular (41.4%), sacrospinal (13.8%), limb (10.3%), and tracheoesophageal fistula (TEF; 6.9%) anomaly. Urogenital anomalies were more common than other anomalies among patients with high ARM (25 patients). ARM-associated urologic anomalies were found in 16 of 25 (64%) patients, and hydronephrosis was the most common (62%) urologic anomaly. Genital anomalies were detected in 9 of the 25 (36%) patients, and undescended testes (37.5%) and hypospadias (37.5%) were the most common genital anomalies. Detailed evaluation of associated anomalies, "VACTERL workup," in the neonatal period is essential for newborns with ARM.

Implication for health policy/practice/research/medical education:
Anorectal malformations (ARM) comprise a wide spectrum of diseases and associated anomalies are the major factors that contribut high morbidity and mortality. The aim of this study is to review the incidence of urogenital and other associated anomalies in neonates with ARM.

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Anorectal malformations (ARM) (1) are a complex group of malformations diagnosed because of the absence of an anus or presence of an ectopic anus at birth (2). The incidence of ARM is approximately 1:5000 live births and the cause of these malformations is unknown (2). Approximately 50–60% of neonates with ARM have associated anomalies. The VATER/VACTERL association is a spectrum of ARM-associated defects: vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies. The incidence of associated anomalies is higher in patients with high ARM than in those with low ARM (1-5). The true incidence of urogenital anomalies in patients with high ARM is 50–60% and that in low ARM is 15–20% (6).

Most genital anomalies are visible on clinical examination, but urologic anomalies require further investigations for their detection. Multiple studies have shown that diagnosis of concomitant anomalies, especially urogenital anomalies, in patients with ARM can improve prognosis of the patients. Hence, the objective of this retrospective study was to evaluate the frequency of urogenital and other ARM-associated anomalies in a single population of patients with ARM admitted to our medi-

* *Corresponding author:* Hassan Bazmamoun, Department of pediatrics, Hospital, Hamedan University of Medical Science, Hamedan, Iran. Tel: +98-9121331917, Fax: +98-212667766, *E-mail:* dbazmamoun@yahoo.com

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cal center. Over a period of 5 years, from 2005 to 2010, we reviewed the records of 58 neonates with ARM admitted to Besat Hospital, Hamedan University of Medical Sciences. Variables such as sex, gestational age, birth weight, level of ARM, VACTERL association, and results of physical examination and imaging studies were examined. All patients underwent ultrasonography for urogenital anomalies and radiography of the spine.

Intravenous urography and renal scanning were performed in select cases. Voiding cystourethrogram (VCUG) was performed in the case of patients with abnormal renal ultrasonographic findings. Genital anomalies were diagnosed on the basis of the results of physical examination; imaging studies were performed for the diagnosis of these anomalies only when needed. The presence of other associated anomalies was evaluated as follows. Echocardiography was performed to determine the presence of congenital heart diseases, and lateral lumbar radiography, to determine the presence of vertebral anomalies; further, an infantogram (a nasogastric (NG) tube with a radiopaque line was used in case an attempt to pass the NG tube failed) was obtained to determined the presence of tracheoesophageal fistula (TEF), esophageal atresia, and limb abnormality. VACTERL association was defined if the patient had 2 or more anomalies other than ARM.

In all, 58 neonates were enrolled in the study; 38 (65.5%) were boys and 20 (34.5%) girls. Their average birth weight was 2.839 ± 0.526 kg (range, 1.500–4.250 kg), and 17.2% of them were born prematurely. In all, 63.8% (37) of the neonates had at least 1 congenital anomaly that was a part of the spectrum of the VACTERL association. The most common associated anomalies were urogenital anomalies, observed in 43.1% (25) of the neonates followed by cardiovascular (41.4% [24 neonates]), sacrospinal (13.8% [8 neonates]), limb (10.3% [6 neonates]), and TEF (6.9% [4 neonates]) anomalies.

ARM-associated urologic anomalies were found in 16 of the 25 (64%) patients with urogenital anomalies (12 boys and 4 girls). Hydronephrosis was the most common (62.5%) urologic anomaly, followed by vesicoureteral reflux (VUR) (37.5%), unilateral renal agenesis (31.2%), ureteropelvic junction obstruction (6.2%), horseshoe kidney (6.2%), bladder diverticulum (6.2%), neurogenic bladder (6.2%), renal cyst (6.2%), and posterior urethral valve (6.2%). Genital anomalies were detected in 9 of 25 (36%) patients (7 boys and 2 girls). The most common genital anomalies were undescended testes (37.5%) and hypospadias (37.5%), followed by ambiguous genitalia (22.2%), bifid scrotum (11.1%), urogenital sinus (11.1%), and vaginal agenesis (11.1%). Sacral agenesis, 4 of 8 (50%) patients, was the most common sacrospinal anomaly

Approximately 50% of neonates with ARM have 1 or more abnormalities that affect other systems(7-9). Urogenital anomalies are the most common associated anomalies in patients with ARM, and male patients are more prone to these anomalies than female patients are. In this study, urogenital anomalies were found to be the most common associated anomalies, and this finding is similar to the findings of other studies (1,4,5); however, we did not find any significant difference in the prevalence of urogenital anomalies between the boys and girls in our study. Various studies have shown that the incidence of VUR ranges from 19% to 47% in patients with ARM (10-12), whereas our study showed that this incidence was 37.5%. The incidence of VUR might have been higher in our study if VCUG had been performed for all patients, including those with normal sonographic findings. In the present study, VCUG was performed only when renal sonographic findings were abnormal. Hydronephrosis and unilateral renal agenesis are the most common anomalies of the upper urinary tract, which was similar to other studies (1, 4, 5).

The overall incidence of genital anomalies in our study was 15.5%, which was similar to the overall incidences reported by Metts and Mclorie (16.4% and 16.5%, respectively) (8, 12). The most common genital anomalies were undescended testes and hypospadias, as reported by Vaishali and Srivastava (2, 13). In conclusion, all patients with ARM, irrespective of high or low ARM, should be examined for associated anomalies (infantogram, echocardiography, ultrasonography, and VCUG) to improve treatment outcome.

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