ASIDE Case Reports

Case Report

VACTERL Associated with Lung Hypoplasia and Urinary Anomalies in a 20-Month-Old Boy: A Case Report

Abbas Al Bazzal[1], Mohammad Ali Mtairek[2], Rafaa Dayyoub[2], Rawan Nahle[3], Zahraa Nehme[2], Lina Khouri[4], Mahdi Akouch[1], Hiba Hamdar[5,*]

1-Faculty of Medical Science, Lebanese University, Beirut, Hadath, Lebanon
2-Faculty of Medicine, Damascus University, Damascus, Syrian Arab Republic
3-Toronto Metropolitan University, Toronto, Canada
4-Department of Gastroenterology, Damascus University, Children University Hospital, Damascus, Syria
5-Medical Learning Skills Academy, Beirut, Lebanon

ARTICLE INFO

Article history:
Received 5 Jul. 2025
Received in revised form 9 Sep. 2025
Accepted 9 Sep. 2025
Published 7 Oct. 2025

Keywords:
VACTERL
Lung hypoplasia
Urinary malformation
Rare case

ABSTRACT

The VACTERL association is a rare group of birth defects, including vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb defects, with an incidence of 1 in 10,000 to 40,000 live births. Infants with lung hypoplasia and urinary malformations face significant challenges. VACTERL requires three or more defects for diagnosis, with unclear causes possibly linked to genetic and environmental factors, including consanguinity. While pulmonary and genitourinary anomalies are not typical, they can coexist. This case is unique due to epispadias and bladder inversion, differing from typical urinary anomalies. We present a 20-month-old infant diagnosed with VACTERL along with left lung hypoplasia, bladder inversion, and epispadias. This rare case is the first documented in Syria, emphasizing the importance of recognizing VACTERL variations. Identifying such cases aids in understanding the conditions complexity and guiding better management strategies.

1. Introduction

The acronym VACTERL association refers to a rare class of congenital abnormalities that may manifest concurrently. The elements (V) vertebral anomalies, (A) anal atresia, (C) cardiac defects, (TE) tracheoesophageal fistula, (R) renal anomalies, and (L) limb defects are associated with this association [1]. According to estimates, the incidence is between one in 10,000 and one in 40,000 live births [2].

It is thought to be an uncommon finding that VACTERL is linked to urinary tract malformation and pulmonary hypoplasia [3, 4], as demonstrated by a study where all five neonates with pulmonary agenesis and VACTERL association died during the neonatal period. The morbidity is increased by pulmonary hypoplasia [4]. Despite the lack of a complete understanding of the etiological factors, familial clustering suggests the presence of inherited factors [5]. The presence of the congenital malformations mentioned above is used to make the clinical diagnosis of VACTERL association in addition to imaging, such as MRI for vertebral abnormalities, echocardiography for cardiac defects, and radiological studies for tracheoesophageal and renal anomalies. Prenatal imaging techniques can detect some anomalies; however, technical and resource limitations may hinder early diagnosis in low-resource settings. Surgery remains the primary treatment method [6]. In contrast to numerous other disorders whose hereditary origins have been identified in recent years, the causes of the VACTERL association are still mostly unknown. This can be attributed to various factors, such as the disease's typical sporadic nature, overlapping conditions, and likely clinical and causal heterogeneity. This study aims to present the first documented case of VACTERL association in Syria, which is of particular significance due to the co-occurrence of pulmonary hypoplasia and genitourinary anomalies, features rarely reported in the literature, in addition to highlighting the challenges of managing complex congenital anomalies in resource-limited settings.

2. Case presentation

We discussed the case of a 20-month-old infant boy who was hospitalized due to frequent coughing, lethargy, and nursing difficulties, as reported by his mother when he was 1 month old. Upon clinical examination, the infant showed urethral epispadias, a delay in psychomotor development, and failure to thrive. His surgical history pointed to five surgical interventions: repair of the bladder inversion at one day of life, repair of the tracheal esophageal fistula (TE) at four months of life, repair of the inguinal hernia at nine months of life, repair of the anal atresia in the first few days of life, and repair of the diaphragmatic hernia at fourteen months of life. The mother, who was 23 years old, had a history of one miscarriage and was consanguineous with her husband in the second degree. Still, other than that, her family history was uneventful. A blood test performed upon admission revealed elevations in leukocytes (WBC 32.8×10^9 ↑), CRP (43 mg/dl), and iron-deficiency anemia (Fe: 15 μg/dl ↓; TIBC: 345 μg/dl ↑). Further testing revealed the

* Corresponding author: Hiba Hamdar, Medical Learning Skills Academy, Beirut, Lebanon. Email: Hamdarhiba95@gmail.com

Published in collaboration with the American Society for Inclusion, Diversity, and Equity in Healthcare (ASIDE). ISSN (Print) 3066-7224, ISSN (Online) 3066-7232

Citation: Al Bazzal A, Mtairek MA, Dayyoub R, et al. VACTERL Associated with Lung Hypoplasia and Urinary Anomalies in a 20-Month-Old Boy: A Case Report. ASIDE Case Reports. 2025;2(2):14-19, doi:10.71079/ASIDE.CR.100725161

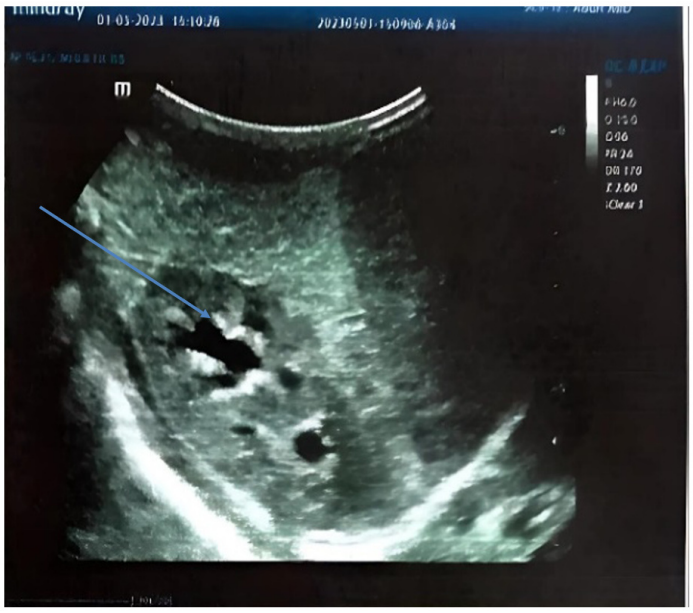

Figure 1: Presents second-degree hydronephrosis in the right kidney with dilated calyces.

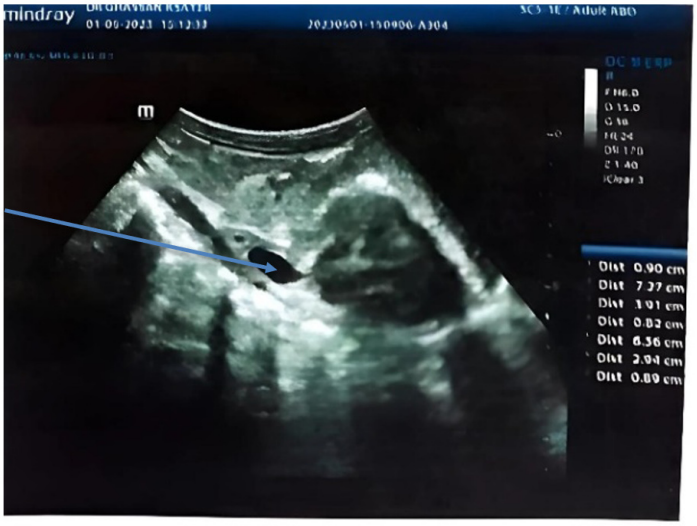

Figure 2: Shows second-degree hydronephrosis in the right kidney with a dilated pelvis.

presence of yeast-like fungi and pyuria, respectively, in the stool culture and urine analysis. After a diagnostic ultrasound of both kidneys, the right kidney's second-degree hydronephrosis was discovered, as seen in **(Figure 1)** and **(Figure 2)**. Using Tc-99m DTPA (diethylene triamine pentaacetate), a dynamic renal scintigraphy was performed to evaluate kidney function. After 15 minutes, the right kidney's cavities and ureter showed signs of severe ascites, which were treated with a Lasix injection. Additionally, there was a protracted and postponed spontaneous emptying time. The kidney responded to the diuretic injection satisfactorily; however, at the end of the imaging, emptying was incomplete, suggesting the presence of vesicoureteral reflux. The glomerular filtration rate was 45 ml/min, and the relative function of the right kidney was 44%. Yet, radiography revealed dysplasia of the acetabular cavity in both legs, as illustrated in **(Figure 3)**. Additionally, an anteroposterior chest radiograph was taken, which suggested pneumothorax because it showed a reduction in the capacity of the left lung. As seen in **(Figure 4)**, the right lung also had larger dendritic fissures or inflammatory infiltrates. Following the injection of 4 mCi of Tc-99m macroaggregated albumin (MAA), lung scintigraphy was performed. The scintigraphy findings were a weakly vascularized left lung (fixation rate of 3%), together with the collapse of the lower segment of the upper lobe of the right lung (fixation rate of 97%). The results indicate that the left lung is hypoplastic. While an echocardiography showed no abnormalities, an upper gastroscopy revealed symptoms of esophagitis and an esophageal stricture in the upper and middle third of the esophagus.

Surgical corrections for congenital anomalies (anal atresia, TEF, bladder inversion, inguinal hernia, diaphragmatic hernia) were performed, and the patient was then treated medically for the sequelae. The management of this complex case addresses both the congenital anomalies and the acute medical issues. Given the patients history of multiple surgical interventions for congenital

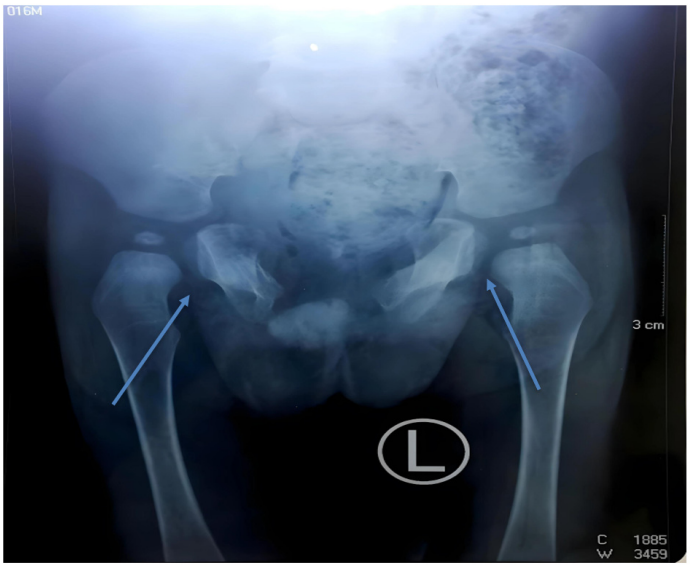

Figure 3: Shows dysplasia of the acetabular cavity in both legs. Both hips are dislocated.

Table 1: Laboratory values for the 20-month-old patient

Test	Value	Reference range	Units	Interpretation
WBC	32.8	5–19	$\times 10^9$/L	Elevated (indicates infection/inflammation)
CRP	43	<10	mg/dL	Markedly Elevated (indicates significant inflammation/infection)
Iron	15	50–120	μg/dL	Low (consistent with iron-deficiency anemia)
TIBC	345	250–310	μg/dL	Elevated (consistent with iron-deficiency anemia)

WBC, white blood cells; CRP, C-Reactive Protein; Fe, Iron; TIBC, Total iron binding capacity; ↑increase; ↓decrease.

malformations (bladder inversion, tracheoesophageal fistula, inguinal hernia, anal atresia, diaphragmatic hernia), surgical and post-surgical care is ongoing and critical. The presence of VACTERL association in conjunction with left lung hypoplasia, bladder inversion, epispadias, and right kidney hydronephrosis requires a multidisciplinary setup involving pediatric surgeons, urologists, nephrologists, pulmonologists, and developmental pediatricians. Unfortunately, the treatment was not completely documented, and the patient was not carefully followed up following the surgery due to financial considerations.

3. Discussion

VACTERL association (VA) is an acronym for a group of non-random co-occurrence congenital malformations defined as three or more of the following defections: vertebral defects (V), anal atresia (A), cardiac malformations (C), tracheoesophageal fistula/esophageal atresia (TEF/EA), renal anomalies (R), limb abnormalities (L) [1]. Prior studies had shown that only < 1% of patients had all 6 anomalies together [8]. There is a slight male predominance, where Fifty-eight percent of cases were males [1]. Vertebral abnormalities are present in 60-80% of cases, atresia in 90%, heart defects in 40-80%, tracheoesophageal fistula in 50 to 80%, and limb abnormalities in half of the cases [9]. Although the exact cause of VACTERL is unknown, a mix of genetic, environmental, or multifactorial abnormalities is considered the cause. The cause of these defects remains idiopathic, but it has been hypothesized that a mesodermal differentiation problem may be involved in the early stages of the first trimester [8]. Some studies have shown some correlation with consanguinity, where these features were found significantly more commonly in first-degree relatives than in the general population [10]. However, no single gene or mutation has been definitively linked to the syndrome, and research continues to explore potential genetic and epigenetic causes. Furthermore, environmental factors, such as maternal diabetes, certain medications, or prenatal infections, have been implicated in increasing the risk of VACTERL [11].

Although VACTERL consists of six malformations, where three must be present for diagnosis, these malformations can co-occur with other pulmonary and genitourinary anomalies [6]. In a study conducted by Knowles et al., all five babies with pulmonary agenesis as a part of the VACTERL association were either stillborn or died in the neonatal period [12]. On the other hand, physicians should be highly suspicious of GU anomalies, even in patients with a VACTERL association who do not have anorectal or renal anomalies. Research on 100 patients with VACTERL association found that GU problems affect 21 percent of the population and are distributed as follows: in males, 20% had hypospadias, 20% had GU fistulae, and 10% of each had ambiguous genitalia (with hypospadias); hypospadias plus more severe defects (not specified); cryptorchidism only; cryptorchidism and a GU fistula; hypospadias and a GU fistula; and hypospadias and hydrocele and of the females, 56% had cloacal anomalies, 33% had GU fistulae, and 11% had a didelphys uterus [6].

In his case report, Yang et al. 2019 reported an unusual VACTERL case compounded by several unusual airway problems, such as the bridging bronchus, airway malacia, and complete tracheal rings [13]. In a 2023 publication, Alwan et al. describe a case of pulmonary agenesis identified in conjunction with several other anomalies. While Alwans case is not directly related to VACTERL, the authors point out that pulmonary agenesis is known to present

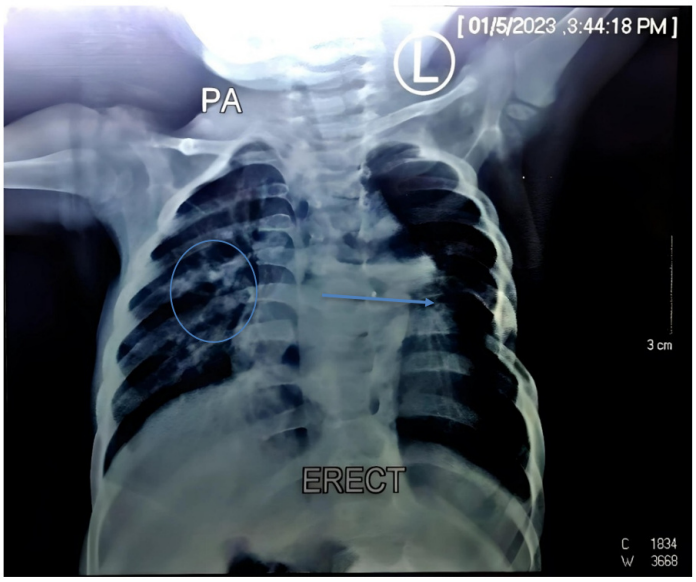

Figure 4: Shows pneumothorax with a reduction in the capacity of the left lung, as well as larger dendritic fissures and inflammatory infiltrates in the right lung. The arrow on the left presents the pneumothorax, and the circle on the right presents the infiltrates.

together with anomalies of multiple other organ systems, including the cardiovascular, skeletal, gastrointestinal, and genitourinary systems [14]. In 2020, Lookzadeh et al. reported a case of VACTERL association in a newborn with all six VACTERL criteria; notably, the limb and kidney defects were asymmetrical on either side of the body [15].

In the described case, three of the six distinctive findings of the VACTERL associationanal atresia, hydronephrosis of the right kidney, and tracheoesophageal fistula (TE) were present. In addition, co-occurrences of pulmonary malformations and hypoplasia of the left lung were present. Moreover, this case shows a unique presentation of genitourinary anomalies, where epispadias instead of hypospadias and bladder inversion were presented.

The diagnosis of VACTERL is primarily based on clinical examination, which may occur after birth, with three malformations sufficient to make the diagnosis [16]. However, diagnostic imaging techniques can be used to detect fetal pathology [7]. Technical and financial constraints prevented us from performing an MRI to assess the vertebral column or a VCU to confirm vesicoureteral reflux. The management of patients with VACTERL/VATER association typically focuses on surgical correction of specific congenital anomalies, such as anal atresia, certain types of cardiac malformations, and/or tracheoesophageal fistula in the immediate postnatal period, followed by long-term medical management of the congenital malformations' sequelae [16], which is exactly what our patient received. Research on the long-term outcomes of patients with VACTERL association is limited, but some studies provide insight into the prognosis and follow-up care for affected individuals. In general, the prognosis depends on the severity of the anomalies present and the success of surgical interventions [10].

4. Conclusion

Given that this is the first case reported in Syria, it is critical to emphasize the importance of conducting thorough examinations and investigations for all VACTERL-related elements when a child exhibits these characteristics. Furthermore, because VACTERL is an association of multiple malformations, healthcare providers must be vigilant for malformations that do not necessarily fit into the typical VACTERL elements. After all, early detection and management are critical for providing appropriate care to patients and avoiding future complications. While the findings provide valuable insights, they are preliminary and based on a single case. Further studies are needed to validate these observations. The literature supports the multifactorial etiology of VACTERL and highlights environmental and genetic factors such as consanguinity, which were also observed in this case. However, more studies will be essential to better understand the interplay of these factors and improve diagnostic and therapeutic approaches.

Conflicts of Interest

The authors declare that they have no competing interests that could have influenced the objectivity or outcome of this investigation.

Funding Source

No funding was received for the conduct of this study or the preparation of this manuscript.

Acknowledgments

None

Informed consent

Patient consent was taken from parents/legal guardians for publication.

Large Language Model

None

Authors Contribution

AB conceptualization and writing the original manuscript. MM conceptualization, data collection and writing original manuscript.

Table 2: Summary of reported cases of VACTERL association

Case	Gender	Age	Family History	Admission History	Diagnosis	VACTERL state	Treatment
Provided Case Report	Male	20 months	Consanguineous parents (second degree), the mother had one miscarriage	Frequent coughing, lethargy, and difficulties nursing at 1 month old. History of 5 surgical interventions: bladder inversion repair (1 day), tracheal esophageal fistula repair (4 months), inguinal hernia repair (9 months), anal atresia repair (first few days), and diaphragmatic hernia repair (14 months).	VACTERL association with left lung hypoplasia, bladder inversion, epispadias, right kidney hydronephrosis	Anal atresia, right kidney hydronephrosis, tracheoesophageal fistula (3 core criteria). Co-occurrence of left lung hypoplasia, bladder inversion, and epispadias.	Surgical corrections for congenital anomalies (anal atresia, TEF, bladder inversion, inguinal hernia, diaphragmatic hernia). Medical management for sequelae.
Yang et al. (2019) [7]	Male	10 months	Non-consanguineous parents, uneventful pregnancy, no family history of congenital malformations	Repeated cough and fever for 2 days	VACTERL association with multiple airway abnormalities (bridging bronchus, airway malacia, complete tracheal rings)	Imperforated anus, rib anomalies, thoracic vertebral anomalies, cardiovascular anomalies (PDA, ASD)	Supplemental oxygen, antibiotics. The patient resolved gradually and was discharged after 10 days. Follow-up showed mild psychomotor retardation, no long-term sequelae, or recurrent pneumonia.
Alwan et al. (2023) [1]	Female	19 years	Not specified	Upper respiratory tract infection and dry cough of a few days' duration	Incidental right-sided pulmonary agenesis with multiple associated anomalies (Klippel-Feil syndrome, bilateral cervical ribs, absent left kidney)	Not explicitly diagnosed with VACTERL, but pulmonary agenesis is noted to be associated with the VACTERL sequence. Associated anomalies include skeletal and genitourinary.	Conservative management with occasional follow-up.
Lookzadeh et al. (2020) [6]	Not specified	Newborn	Not specified	Not specified	VACTERL association with limb and kidney anomaly, severe hydronephrosis, imperforated anus, rectovesical fistula	All 6 VACTERL criteria (spinal anomalies, anorectal anomalies, cardiac disorders, esophageal atresia with tracheoesophageal fistula, renal anomaly, limb anomalies)	Not specified

VACTERL, Vertebral, Anal, Cardiac, Tracheoesophageal, Renal, and Limb anomalies; TEF, Tracheoesophageal Fistula; PDA, Patent Ductus Arteriosus; ASD, Atrial Septal Defect.

RD writing original manuscript. RN writing original manuscript. ZN writing original manuscript. LK writing original manuscript, reviewing and editing. MA writing original manuscript. HH conceptualization, writing original manuscript, reviewing and editing.

Data Availability

All the data is available with the corresponding Author upon request.

References

1. Alwan A, Mohammoud S, Assefa AZ. Incidental pulmonary agenesis with multiple associated anomalies: A case report. Radiol Case Rep. 2023;18(10):3724-8. [PMID: 37636541, PMCID: PMC10447928, https://doi.org/10.1016/j.radcr.2023.08.002].
2. Brown AK, Roddam AW, Spitz L, Ward SJ. Oesophageal atresia, related malformations, and medical problems: A family

study. American Journal of Medical Genetics. 1999;85(1):31-7. [https://doi.org/10.1002/(sici)1096-8628(19990702)85:1<31::Aid-ajmg7>3.0.Co;2-d].
3. Campbell CL, Collins n R T, Zarate YA. Severe neonatal presentation of Kleefstra syndrome in a patient with hypoplastic left heart syndrome and 9q34.3 microdeletion. Birth Defects Res A Clin Mol Teratol. 2014;100(12):985-90. [PMID: 25380126, https://doi.org/10.1002/bdra.23324].
4. Kanu A, Tegay D, Scriven R. Bronchial anomalies in VACTERL association. Pediatr Pulmonol. 2008;43(9):930-2. [PMID: 18671276, https://doi.org/10.1002/ppul.20827].
5. Knowles S, Thomas RM, Lindenbaum RH, Keeling JW, Winter RM. Pulmonary agenesis as part of the VACTERL sequence. Arch Dis Child. 1988;63(7 Spec No):723-6. [PMID: 3415317, PMCID: PMC1590134, https://doi.org/10.1136/adc.63.7_spec_no.723].
6. Lookzadeh MH, Tahghigh F, Amooee A. VACTERL Association in a Newborn A Rare Case Report. World Journal of Peri Neonatology. 2020. [https://doi.org/10.18502/wjpn.v2i1.2811].
7. Yang L, Li S, Zhong L, Qiu L, Xie L, Chen L. VACTERL association complicated with multiple airway abnormalities: A case report. Medicine (Baltimore). 2019;98(42):e17413. [PMID: 31626096, PMCID: PMC6824793, https://doi.org/10.1097/MD.0000000000017413].
8. Salinas-Torres VM, Perez-Garcia N, Perez-Garcia G. Clinical, cytogenetic, environmental and inheritance findings in Mexican neonates with VACTERL association. Indian J Pediatr. 2015;82(1):84-8. [PMID: 25008186, https://doi.org/10.1007/s12098-014-1493-5].
9. Santos J, Nogueira R, Pinto R, Cerveira I, Pereira S. First Trimester Diagnosis of VACTERL Association. Clin Pract. 2013;3(1):e5. [PMID: 24765498, PMCID: PMC3981235, https://doi.org/10.4081/cp.2013.e5].
10. Sharma P, Pawar N, Gupta AK. Congenital intrinsic duodenal obstruction: Clinical study and predictors of neonatal surgical mortality. Journal of Neonatal Surgery. 2022;11. [https://doi.org/10.47338/jns.v11.1034].
11. Shaw S, Raj R. Vacterls association with pulmonary arterial hypertension and least vertebral anomaly.
12. Solomon BD. VACTERL/VATER Association. Orphanet J Rare Dis. 2011;6:56. [PMID: 21846383, PMCID: PMC3169446, https://doi.org/10.1186/1750-1172-6-56].
13. Solomon BD. The etiology of VACTERL association: Current knowledge and hypotheses. Am J Med Genet C Semin Med Genet. 2018;178(4):440-6. [PMID: 30580478, https://doi.org/10.1002/ajmg.c.31664].
14. Solomon BD, Pineda-Alvarez DE, Raam MS, Bous SM, Keaton AA, Velez JI, et al. Analysis of component findings in 79 patients diagnosed with VACTERL association. Am J Med Genet A. 2010;152A(9):2236-44. [PMID: 20683998, PMCID: PMC2930065, https://doi.org/10.1002/ajmg.a.33572].
15. Solomon BD, Pineda-Alvarez DE, Raam MS, Cummings DA. Evidence for inheritance in patients with VACTERL association. Hum Genet. 2010;127(6):731-3. [PMID: 20369369, PMCID: PMC2871973, https://doi.org/10.1007/s00439-010-0814-7].
16. Tonni G, Kocak C, Grisolia G, Rizzo G, Araujo Junior E, Werner H, et al. Clinical Presentations and Diagnostic Imaging of VACTERL Association. Fetal Pediatr Pathol. 2023;42(4):651-74. [PMID: 37195727, https://doi.org/10.1080/15513815.2023.2206905].