

VACTERL Association: Second Level Care Management

Hilberth Cristhian López Mestas¹, Tania de la Rosa Tavera², María Carrasquel Valecillos³, Cheryl Diaz Barrientos⁴

¹Residente de tercer año de cirugía general del Hospital Universitario de Puebla, Puebla, Mexico (Corresponding author)

²Medico interno de pregrado del Hospital Universitario de Puebla, Mexico

³Medico adscrito del servicio de cirugía pediátrica del Hospital Universitario de Puebla, Mexico

⁴Profesora titular de cirugía general del Hospital Universitario de Puebla, Mexico

Abstract: *The VACTER or VACTERL association (vertebral, anal, cardiovascular, tracheoesophageal, esophageal, renal, and Limbs anomalies) is a multifactorial pathology characterized by congenital anomalies whose name is due to the acronym of the defects that occur; the morbidity and mortality and the prognosis of these patients depend on immediate surgical corrections in the newborn and its subsequent surgical clinical management. The prevalence of this condition is 1/10, 000 live births. In Mexico, only 141 cases have been reported, of which 14 have been in the state of Puebla, so the objective of this article is to report the first clinical case that is presented. at the University Hospital of Puebla and its surgical management.*

Keywords: VACTERL Association; VACTER; Congenital Anomalies

1. Introduction

The VACTER or VACTERL association is a multifactorial pathology [1, 2] characterized by a series of congenital anomalies whose name is due to the acronym of the defects present [3] (Table 1), it has a prevalence of 1/10, 000 live births with a higher incidence in males 2: 6 [4] and its diagnosis is made when there are 3 or more of these anomalies [5].

Table 1: Defects occurring in the VACTERL association

V	Vertebral Malformations
A	Atresia Anal
C	Cardiovascular Anomalies
T	Tracheoesophageal fistula
E	Esophageal Atresia
R	Kidney Malformations
L	Limb Dysplasia

The etiology of this association is considered multifactorial, however, it has been suggested that these simultaneous alterations are due to generalized damage of the mesenchymal tissue during the fourth week of intrauterine life. In addition, genetically, the heterozygous deletion of chromosome 13, trisomy in chromosome 18, changes in specific genes such as FOXF1, HOXD13, and mitochondrial alterations may influence these malformations [6, 7].

The morbimortality and prognosis of these patients will depend on the need for surgical management of life - threatening anomalies such as esophageal and/or anal atresia, which, depending on the patient's evolution, can be corrected surgically as required [8].

VACTERL association is a rare entity; only 141 cases have been reported in Mexico [9], of which 14 were documented in the state of Puebla [7]. The aim of this article is to describe

the first case of VACTERL association and the surgical management required at the University Hospital of Puebla.

2. Case Presentation

This is a female newborn son of a mother with poorly controlled gestational diabetes, with poor adherence to prenatal care, maternal history of substance abuse, obtained by cesarean section at 35 weeks of gestation with a weight of 2280 grams, height of 44 cm, APGAR 8/9, with prenatal ultrasonographic diagnosis of umbilical cord with 2 vessels, ectrodactyly of the left foot, mesomelia of the left pelvic limb, probable duodenal atresia, intestinal loop dilatation and thoracic hemivertebra by magnetic resonance imaging of the uterus. At birth, physical examination revealed imperforate anus (Figure 1), single umbilical artery, esophageal atresia, ectrodactyly of the left foot, mesomelia of the right pelvic limb (Figure 2) and bilateral triphalangeal thumb, for which VACTERL association was diagnosed.



Figure 1: Perianal region showing agenesis of genitalia and imperforate anus.



Figure 2: Image showing ectrodactyly of left foot and mesomelia of right pelvic limb.

After his initial care, he was taken to the neonatal intensive care unit to continue the diagnostic approach and to regulate the most appropriate therapeutic conduct. An abdominal ultrasound was performed, reporting a smaller left kidney with respect to the collateral one and an absence of vascularity in color Doppler mode, diagnosing renal hypoplasia; the echocardiogram identified interatrial communication vs. foramen ovale, large patent ductus arteriosus and moderate pulmonary arterial hypertension.

Surgical management was decided, where under general anesthesia exploratory laparotomy was performed; Patient in dorsal decubitus with previous asepsis and antisepsis of the neck, thorax, abdomen, genitals and thighs, the approach was performed by initial transverse incision to the left side at umbilical level of approximately 3 cm dissecting by planes until reaching the abdominal cavity, large and small intestine was exteriorized to rule out fistula sites, ruling out fistula, proceeding to perform colostomy identifying descending colon, securing proximal and distal end, dissecting mesocolon (Figure 3). Using a scalpel, a colectomy is performed, exteriorizing and fixing the colostomy on the left flank (Figure 4). Stoma maturation was performed with modified Brooke technique, stoma functionality was confirmed, without complications.

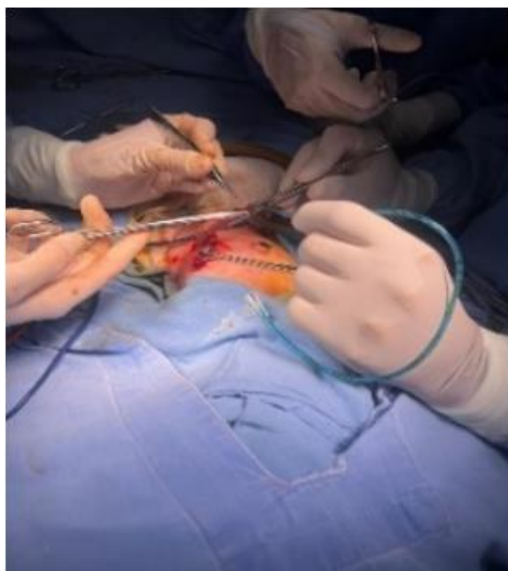


Figure 3: Externalization of transverse colon to perform colostomy

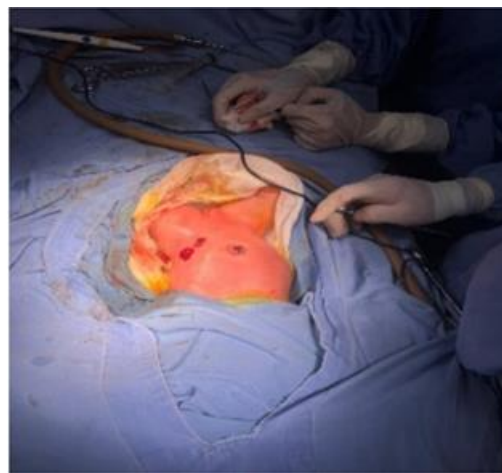


Figure 4: Colostomy externalized and fixed in left flank

As a second surgical time, a transverse incision of approximately 5 centimeters is made at the level of the right hypochondrium, dissecting by planes until reaching the cavity, identifying the biliary tract with a widely distended stomach, preventing adequate visualization of the duodenum, Kocher maneuver is performed to release duodenum, a flange is observed at the level of the first duodenal portion, which causes stenosis site, blunt dissection is performed with mosquito forceps and release of duodenum (Figure 5). 5 cm at the level of the gastric pylorus, covering all its layers, an orotracheal feeding tube number 8 is introduced caudally with impossibility of passage to the duodenum, a transverse duodenotomy of approximately 1 cm in length distal to the site of stenosis is performed; performing gastroduodenum anastomosis by diamond technique with vycryl 5 - 0, corroborating functionality through the passage of the feeding tube and passage of the instilling of physiological solution.



Figure 5: Release of atresic zone of duodenum

When continuing surgical exploration retroperitoneum is reviewed finding right renal hydronephrosis and left renal agenesis, at the level of pelvic hollow female annexes were evidenced (right and left ovary and uterus) confirming sex of the patient, abdominal surgical time is terminated, with closure of the abdominal cavity in one plane with vycryl 4 - 0 with continuous surgete and skin with 5 - 0 nylon ashiff points thus ending the surgical time without complications, The third surgical procedure at thoracic level was started, however the patient presented with respiratory support through invasive

mechanical ventilation and sudden desaturation up to 10% without recovery, resuscitation maneuvers were started without response, giving time of death 17: 07 hours.

3. Discussion

The VACTERL association consists of different malformations that can be presented in different combinations, however, in spite of the fact that 3 or more of these are needed to make the diagnosis, they can appear in different proportions, anal atresia occurs in up to 90% of the cases that present this association, cardiac malformations between 40 - 80%, esophagotracheal fistula from 50 to 80%, renal anomalies up to 80%, defects in the limbs up to 50%, vertebral defects from 60 to 95% and only 1% of the cases present the complete spectrum [5]; in the case of this patient, she presented with all the abnormalities.

Among the etiology of the VACTERL association are not only genetic alterations, but also underlying maternal diseases, in this case, the mother had a history of uncontrolled gestational diabetes, being considered by some authors as a risk factor for this entity, as well as Fanconi's anemia, maternal use of sex hormones in the first trimester of pregnancy, benzodiazepines and lead [10].

The diagnosis of VACTERL association is made from prenatal control mainly with the use of imaging tests that show characteristics of malformations, in this clinical case MRI was requested at 32.2 SDG where, in addition to only one umbilical artery, stomach dilatation and double bubble image were found, which with the severe polyhydramnios presented by the mother was associated with esophageal atresia; only 5% of the cases with VACTERL association present esophageal atresia and duodenal atresia together, and the incidence is even lower when they are presented together with imperforate anus [11]. With respect to the extremities, shortening of the left pelvic limb was identified with a decrease in femoral length, and neither tibia nor fibula could be identified, so mesomelia of the left pelvic limb was diagnosed, as well as disruption of the spine in the thoracic segment, so a diagnosis of hemivertebra was suggested.

The treatment of patients with VACTERL association focuses on surgical corrections of congenital anomalies, mainly respiratory as in this case, cause of death of this patient, followed by long - term medical management of other alterations or sequelae, finally the prognosis of the subjects will depend on the severity of the picture, however, adult survivors with this pathology may present a cognitive development without alterations [12, 13].

4. Conclusion

The VACTERL association is a rare condition whose prognosis depends on the severity and medical management, because these abnormalities should be treated immediately in the newborn, it is important that there is adequate prenatal care and timely referral to specialized third level centers, to reduce morbidity and mortality, through an appropriate clinical and surgical approach.

References

- [1] Siebel S, Solomon BD (2013) Mitochondrial factors and VACTERL association - related congenital malformations. *Mol Syndromol* 4 (1 - 2): 63 - 73.
- [2] Chen Y, Liu Z, Chen J, Zuo Y, Liu S, et al. (2016) The genetic landscape and clinical implications of vertebral anomalies in VACTERL association. *J Med Genet* 53 (7): 431 - 7.
- [3] Solomon BD, Baker LA, Bear KA, Cunningham BK, Giampietro PF, et al. (2014) An approach to the identification of anomalies and etiologies in neonates with identified or suspected VACTERL (vertebral defects, anal atresia, tracheo - esophageal fistula with esophageal atresia, cardiac anomalies, renal anomalies, and limb anomalies) association. *Journal of Pediatrics* 164 (3).
- [4] Solomon BD (2011) VACTERL/VATER association. *Orphanet Journal of Rare Diseases*.6: 56.
- [5] Santos J, Nogueira R, Pinto R, Cerveira I, Pereira S (2013) First Trimester Diagnosis of VACTERL Association. *Clin Pract* 3 (1): e5.
- [6] Huamán - Rodríguez M, Cortez - Espejo C, Chavarry - Rodríguez L, Castañeda - Guzmán I, Cortez - Ponce de León C, et al. (2022) Case report: Newborn with VACTERL association and tracheomalacia. *Revista Médica de Trujillo* 17 (1): 38 - 42.
- [7] García - Guzmán AE, Aparicio - Rodríguez JM (2014) Asociación congénita VACTERL: reporte de catorce casos en hospital pediátrico de tercer nivel. *Rev Mex Cir Ped*.
- [8] Ávila - Iglesias MC, Rojas - Maruri CM (2017) VACTERL association. Presentation of an anatomoclinical case and general considerations. *Acta Pediátrica de México* 38 (5): 330 - 6.
- [9] Maricela Guadalupe Blas Concha. Descripción de las anomalías morfológicas congénitas encontradas en pacientes diagnosticados con asociación VACTERL.2020.
- [10] Rodríguez Z E (2013) Asociación VACTER: reporte de un caso. *Revista Mexicana de Pediatría*.80: 109 - 13.
- [11] Torres Valencia NA, Pardo M, Llanos J, Sierra L, Alarcón Y (2020) Asociación VACTERL de presentación inusual: reporte de caso. *Revista Med* 28 (1): 59 - 67.
- [12] Wheeler PG, Weaver DD (2005) Adults with VATER association: Long - term prognosis. *Am J Med Genet* 138 A (3): 212 - 7.
- [13] Lin HY, Lin SP, Lin HY, Hsu CH, Chang JH, et al. (2012) Functional independence of Taiwanese children with VACTERL association. *Am J Med Genet A* 158 A (12): 3101 - 5.