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Myelomeningocele in a newborn with VACTERL association

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ABSTRACT

VACTERL association refers to the concomitant occurrence of congenital malformations such as vertebral defects, anorectal malformations, cardiac anomalies, tracheoesophageal fistula, renal anomalies, and limb defects. This case describes a newborn girl who presented with anomalies compatible with this association, such as scoliosis, sacral dysgenesis, right lower phocomelia, and multicystic renal dysplasia, also associated with myelomeningocele, which was surprising as it occurred concomitantly with the other congenital malformations.

Although neural tube defects are not part of the classic criteria for VACTERL, their coexistence with the characteristic malformations of this association raises the possibility of broadening its phenotypic spectrum, encourages debate on the inclusion of new criteria to define it, and highlights the importance of considering systematic evaluation of the spinal cord in screening, aspects that have been little explored in Latin America.

Keywords: *congenital anomalies; multicystic dysplastic kidney; spinal dysraphism; meningomyelocele.*

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INTRODUCTION

The acronym VACTERL refers to a combination of congenital malformations first described in 1973, which includes: vertebral defects (V), anal atresia (A), cardiac malformations (C), tracheoesophageal fistula (TE), renal dysplasia (R), and limb malformations (L).[1] The diagnosis is confirmed when three or more of these anomalies are present.[2] It is relevant due to its complexity and implications for management, and also because it constitutes a higher risk of neurodevelopmental disorders.[3] Its etiology is unclear due to the heterogeneity of its phenotypes; however, a genetic role in its causality has been proposed, suggesting a possible direct relationship between the hereditary component and this pathology.[4-6] Epidemiologically, it affects approximately 1 in every 10 000 to 40 000 live births.[2]

Spinal cord abnormalities are not part of the classic definition of VACTERL, however, it has been proposed to modify the acronym to VACTERLS, where the "S" refers to spinal dysraphisms, taking into account that these are reported in 10-40% of patients with this association.[7] Open spinal dysraphisms, such as myelomeningocele, and closed spinal dysraphisms, such as lipomyelomeningocele or myelocystocele, have been described.[7] In this case, we present a newborn with a clinical diagnosis of VACTERL association due to the presence of right lower phocomelia, right multicystic renal dysplasia, and vertebral defects, also associated with a spinal dysraphism type myelomeningocele.

CLINICAL CASE

We present the case of a female patient born in Medellín, Colombia, at 35.2 weeks of gestational age by scheduled cesarean section due to premature rupture of membranes, with a prenatal diagnosis of a myelomeningocele-type neural tube defect, Chiari type II malformation, and right multicystic renal dysplasia. The patient was the product of the fourth pregnancy of a 37-year-old mother with no relevant pathological history or chronic diseases during pregnancy. Her gynecological and obstetric history included one spontaneous abortion and two previous full-term pregnancies with healthy newborns and no perinatal complications.

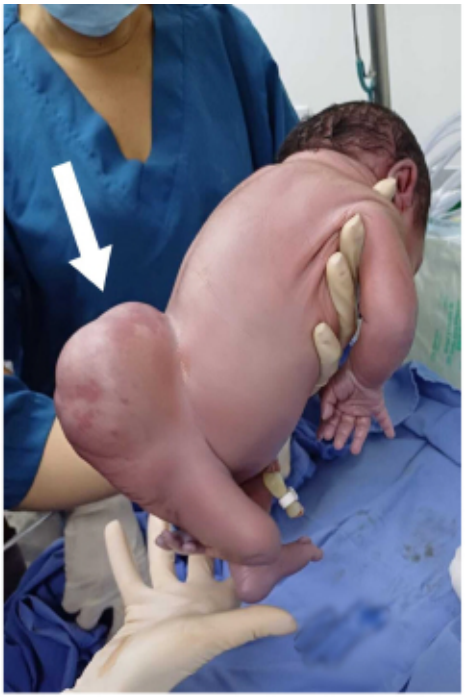
During prenatal care, a genetic screening ultrasound performed at 11 weeks showed an intermediate risk for aneuploidy. At 27 weeks, an obstetric Doppler ultrasound was performed, revealing findings consistent with myelomeningocele and right renal multicystic dysplasia. Subsequently, at 32 weeks, a fetal MRI was ordered, showing an absence of posterior bone elements from the L2 vertebral body level, with a solution of continuity in the skin. It should be noted that, due to socioeconomic limitations and access issues in the parents' place of origin, they did not receive counseling on invasive prenatal studies. At birth, the patient weighed 2585 grams, measured 44 centimeters in length, had a head circumference of 32 centimeters, and had Apgar scores of 8 and 9 at one minute and five minutes, respectively. Physical examination revealed a normal-shaped skull with a normotensive anterior fontanelle, pink conjunctiva, anicteric sclera, intact palate, normally implanted auricles, and patent external auditory canals. The neck was mobile, with no masses or adenomegaly. The chest was symmetrical, with preserved vesicular breath sounds and rhythmic heart sounds without murmurs. The abdomen was soft, with no masses or visceromegaly, and the umbilical cord consisted of one artery and one vein. A smooth, fluctuating mass measuring approximately 13 × 14 cm was identified in the medial and paramedian right lumbosacral region, covered by intact skin and with no evidence of cerebrospinal fluid fistula or bleeding (*Figure 1*). In the right lower extremity, hypoplasia of the femur, tibia, and fibula was evident, associated with ipsilateral oligodactyly, consistent with phocomelia (*Figure 2*); the rest of the extremities were mobile, without edema, and with a capillary refill time of less than 2 seconds. The skin had no lesions, and neurological examination revealed no motor or sensory deficits. Lipomeningocele and lipomyelomeningocele were considered as differential diagnoses for the lumbosacral mass, but these were subsequently ruled out by magnetic resonance imaging. Given the coexistence of vertebral, renal, and limb anomalies, we considered the possibility of a VACTERL association, and we requested a karyotype study with G-banding, which showed no numerical or structural alterations. An echocardiogram was performed, revealing a patent *foramen ovale* with preserved ventricular function and no dilatation of the cavities or signs of pulmonary hypertension. A voiding cystourethrogram was indicated, which reported grade V left vesicoureteral reflux without post-void residual urine. Likewise, total spine MRI

showed left-convex lumbar scoliosis, a defect of the posterior elements from L2 to S1, and spinal cord protrusion at the L2 level, consistent with myelomeningocele (*Figure 3*). Dysplastic changes were also identified in the right sacroiliac joint, dysgenesis of the right gluteal muscles, and a tethered cord.

The Neurosurgery Department performed the resection of extensive bilateral skin tissue, correction of the tethered cord, and limited myelotomy in the non-functional right area, which was successfully achieved. The Nephrology Department performed a bladder catheterization due to sacral dysgenesis to prevent urinary tract infections and preserve renal function.

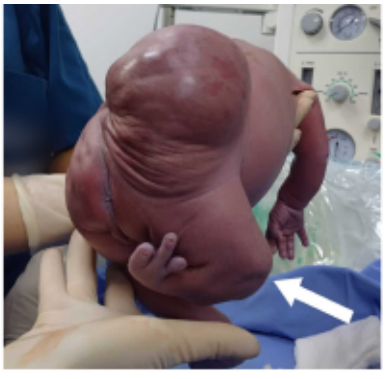
The Medical Genetics Department concluded that, based on the clinical findings, the patient met the diagnostic criteria for VACTERL association, with a significant neural tube defect, a myelomeningocele. In this case, given the limitations of available resources, the associated costs, and the limited clinical benefit of performing advanced genetic studies, we decided not to continue studies at that time.

Figura 1. Lumbosacral tumor



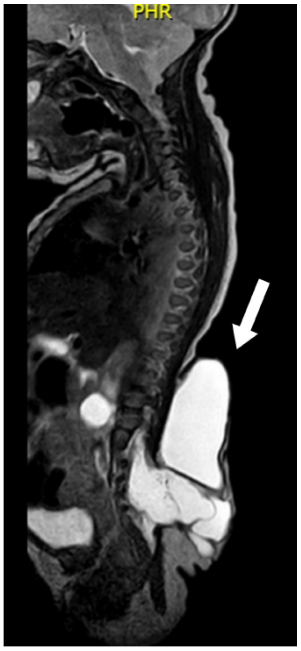
A 13 × 14 cm mass covered with skin, consistent with myelomeningocele, is noted.

Figure 2. Anomalies in the lower right limb



Hypoplasia of the femur, tibia, and fibula, and ipsilateral oligodactyly are observed.

Figure 3. Magnetic resonance imaging of the entire spine



The arrow shows spinal cord protrusion.

One month after birth, the patient was progressing favorably, with no infectious or neurological complications. She was discharged with education on warning signs, scheduled follow-up appointments with pediatric neurosurgery and nephrology, and inclusion in the Kangaroo Mother Care Program in her hometown to ensure comprehensive follow-up.

DISCUSSION

The VACTERL association consists of malformations in multiple systems. It is reported that between 60% and 80% of patients have spinal abnormalities such as scoliosis, hemivertebrae, and butterfly vertebrae.[8] Limb abnormalities are present in 50% of cases, with phocomelia commonly observed on one side.[8,9] Renal abnormalities occur in 50-80% of patients, with cystic and/or dysplastic kidneys, horseshoe kidney, and unilateral or bilateral renal agenesis being common. In some cases, these malformations may be accompanied by genitourinary and ureteral abnormalities.[8] In addition to the VACTERL association anomalies described in this case, a significant neural tube defect is noted: myelomeningocele. This malformation belongs to the group of spinal dysraphisms. Although not included in the current diagnostic criteria for this association, it has been reported to coexist with it, suggesting an etiopathogenic interrelationship. Authors such as Lubinsky (2015) report that this correlation could be explained by alterations in common molecular pathways involved in embryonic development, such as hedgehog signaling, retinoic acid, and genetic or epigenetic factors, whose involvement is not yet fully understood.[10-12]

The finding of vertebral, renal, and limb malformations in this patient, compatible with the VACTERL association, drew attention due to its association with myelomeningocele. The coexistence of both has been little explored in Latin America, and there are possible complications the patient could present. A considerable percentage of patients with VACTERL have been found to have spinal dysraphism. According to Amelot et al. (2020), this supports the need to review the diagnostic criteria for this association to consider the formal inclusion of spinal cord anomalies, achieving a more complete characterization and phenotyping.[7]

From a clinical perspective, incorporating spinal anomalies into the diagnostic criteria for VACTERL would be highly relevant, given that these malformations often go unnoticed in initial screening studies. The expansion of the phenotype represents a crucial opportunity to systematically incorporate spinal evaluation from birth. Although magnetic resonance imaging is the gold standard, spinal ultrasound is a cost-

effective alternative, with sensitivity and specificity greater than 95%, making it an ideal tool to be performed in the first three months of life. This early detection is essential, as it allows for timely multidisciplinary therapeutic planning and prevents complications associated with late diagnosis, impacting healthcare practice and the quality of life of these patients.[13] ■

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