# A successful pregnancy outcome in a case of maternal VACTERL*

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**ABSTRACT**

Vertebral anomalies-anal atresia-cardiac abnormalities-tracheoesophageal fistula-renal agenesis-limb (VACTERL) defects association is a rare congenital disease. While most scientific literature focus on the clinical presentation and management of pediatric patients with this condition, this paper focuses on the challenges faced by a 22-year-old primigravid, who was able to carry a pregnancy to term, despite the many anomalies associated with being afflicted with VACTERL.

*Keywords: VACTERL, VACTERL association, VATER*

## INTRODUCTION

VATER association is a rare congenital malformation initially described by Quan and Smith (1973) referring to a non-random co-occurrence of vertebral defects, anal atresia, traceheo-esophageal fistula with esophageal atresia, renal defects and radial limb dysplasia[1]. Since then, the description of the association has been extended to include cardiac malformations, limb anomalies (VACTERL)[2]. The etiologic cause of the disease is not yet elucidated. Some researchers have hypothesized that it is from an abnormality in the mesodermal cell line development during embryogenesis that accounts for the occurrence of the association[3]. To this date, there is limited data on the medical sequelae among patients that survive into adulthood. This paper focuses on the challenges faced by a pregnant individual with VACTERL association.

## CASE

This is the case of a 22-year-old single, primigravid, Roman Catholic from Las Piñas City.

She is an undiagnosed case of multiple congenital anomaly. She was born to a then 40-year-old multigravida at a local hospital. Her mother narrated that after she was born, the doctors in the said hospital did not stress the need for further assessment or consults for her congenital anomalies. As she was growing up, her mother noted that she did not have any problem and that she did not acquire any sickness necessitating a consult with a physician. She did not receive any formal schooling and received home schooling from her elder siblings due to the difficulty in ambulating to and from their home. They have an unremarkable family medical history with no note of hypertension, diabetes mellitus, bronchial asthma in both the maternal and paternal lineage. No one else in their family has any deformity.

She had her menarche at the age of 13 with a regular 30 day interval lasting 5 days soaking her shorts with no note of dysmenorrhea and intermenstrual bleeding. Due to the limb anomaly of her lower extremities, the patient reported that she is unable to wear any undergarment and that she resorts to wearing shorts in lieu of it.

She had her first coitus at the age of 20 and has had 1 non-promiscuous sexual partner. There is no note of any sexually transmitted disease and no use of any form of contraceptive.

Eight months prior to admission, patient noted cessation of menses but no consults and diagnostic examinations were done.

Seven months prior to admission, a pregnancy test was done which yielded a positive test result, prompting consult at a local lying in clinic in Las Pinas. She was then advised to seek further consult at a tertiary hospital due to her birth defects.

Two and one-half months prior to admission, she was first seen at this hospital's Out Patient Department (OPD), where she was diagnosed to have VACTERL-like association on the basis of vertebral, anal and limb components. There was note of a prominent right scapula (Figure 1), absence of the 2nd digit, middle and distal phalanx of the 3rd digit on the right hand, absence of the 2nd and 3rd digits with fusion of the 4th and 5th digits of the left

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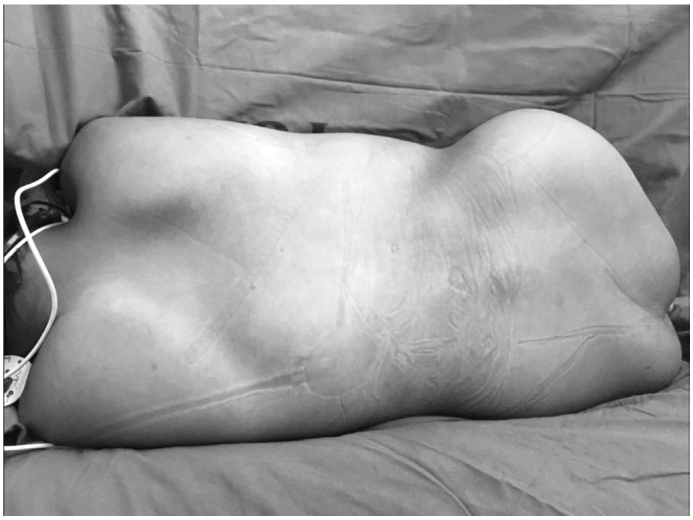

**Figure 1.** Posterior view with note of prominent right scapula

hand (Figure 2A). Her heels are fused together and a soft tissue attaches them to her left gluteal area (Figure 2B). At the time, the initial plan was to refer her to the high risk clinic for evaluation and co-management and for a second trimester ultrasound to be done.

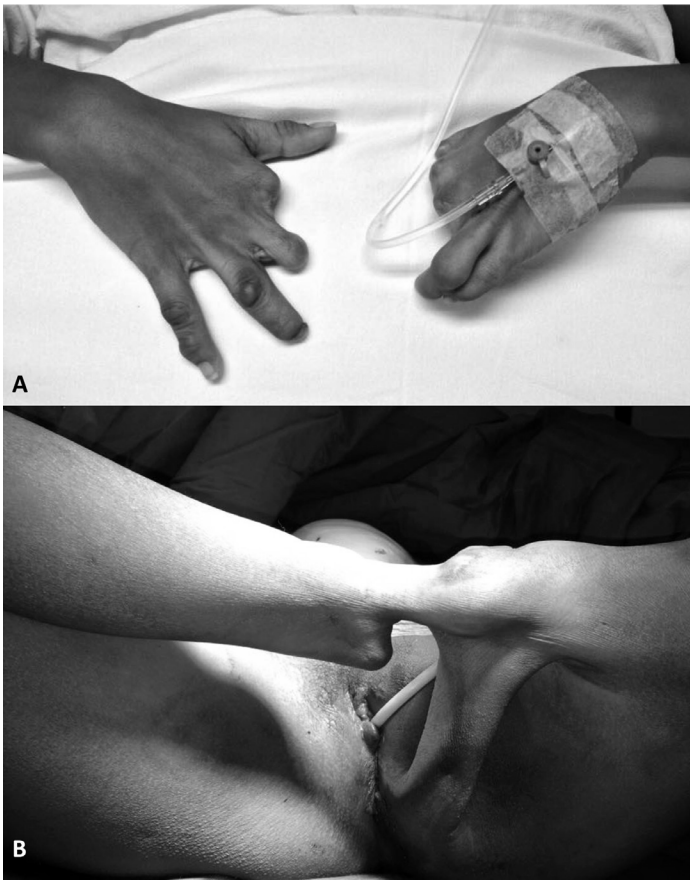


**Figure 2.** (A) Right hand: Absent 2nd digit, middle and distal phalanx of the 3rd digit; Left hand: Absent 2nd and 3rd digits with fusion of the 4th and 5th digits; (B) Heels ajoined with an attachment anchoring it to the left glutteal area; lateral and posterior to the anal canal

She had one prenatal follow-up at the High Risk Clinic during the course of her pregnancy where she had a biophysical profile, biometry and congenital anomaly scan done which revealed that she had a single live intrauterine pregnancy, transverse, with good cardiac and somatic activities, 28 4/7 weeks by composite sonar aging; Placenta fundal, grade II; biophysical score10/10 with adequate amniotic fluid volume at 12.9 cm, appropriate for gestational age (AGA) at 1312 + 192grams, confirmed negative congenital anomaly scan with a consideration of uterine didelphys.

She was lost to follow-up due to the difficulty in travelling from her home to the hospital due to both financial constraints and the availability of a vehicle as a means of transportation. She narrated that in order to arrive to the hospital she would have to walk out of her house through an unevenly cemented street which was often littered with garbage in order to get to the main street. She would propel herself by walking using her arms as crutches and her knees as her feet. After arriving at the main street she would then hail a pedicab to get to the main road in order to hire a cab to get to the hospital.

Patient consulted at the admitting section of the hospital due to labor pains. On examination, she had stable vitals with a blood pressure of 110/70 mmHg, heart rate of 88 beats per minute, respiratory rate of 18 breaths per minute and was afebrile at 36.5 degrees Celsius. She weighed 26.5kg and with a height of 87.5cm. She had an anicteric sclerae, pink conjunctivae, no anterior neck mass, no cervical lymphadenopathy and no jugular vein distention. She had equal chest expansion and clear breath sounds, an adynamic precordium with distinct heart sounds, normal rate and regular rhythm, no murmurs were appreciated. She had pink nail beds, full and equal pulses and there was no note of edema. No sensorimotor deficits were noted and she had intact cranial nerves.

On abdominal examination she had a fundic height 32 cm, estimated fetal weight 2-2.8kg, breech in presentation with fetal heart rate of 130's at the right umbilical area (Figure 3). On speculum examination, she had normal external genitalia nulliparous vagina, the cervix cannot be visualized due to anatomic variation in positioning. On pelvic examination, she had normal external genitalia, sparse pubic hair, nulliparous vagina, cervix is 0.5x0.5cm knobby and deviated to the left, corpus was enlarged to age of gestation, lax sphincter tone, intact rectal vault, no intraluminal masses, bilateral parametria smooth and pliable (Figure 4).

She was admitted with a primary working impression of pregnancy uterine 36 weeks and 6 days age of gestation by amenorrhea cephalic in preterm labor, to consider maternal VACTERL (vertebral, anal and limb) association gravida 1 para 0. Upon admission, her preterm labor was

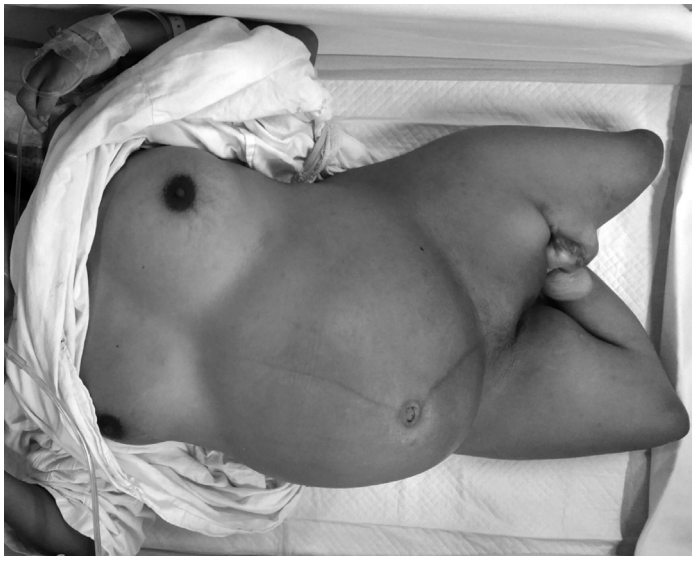

**Figure 3.** Full body: fundic height 32 cm, estimated fetal weight 2.-2.8kg, breech in presentation with fetal heart rate of 130's at the right umbilical area.

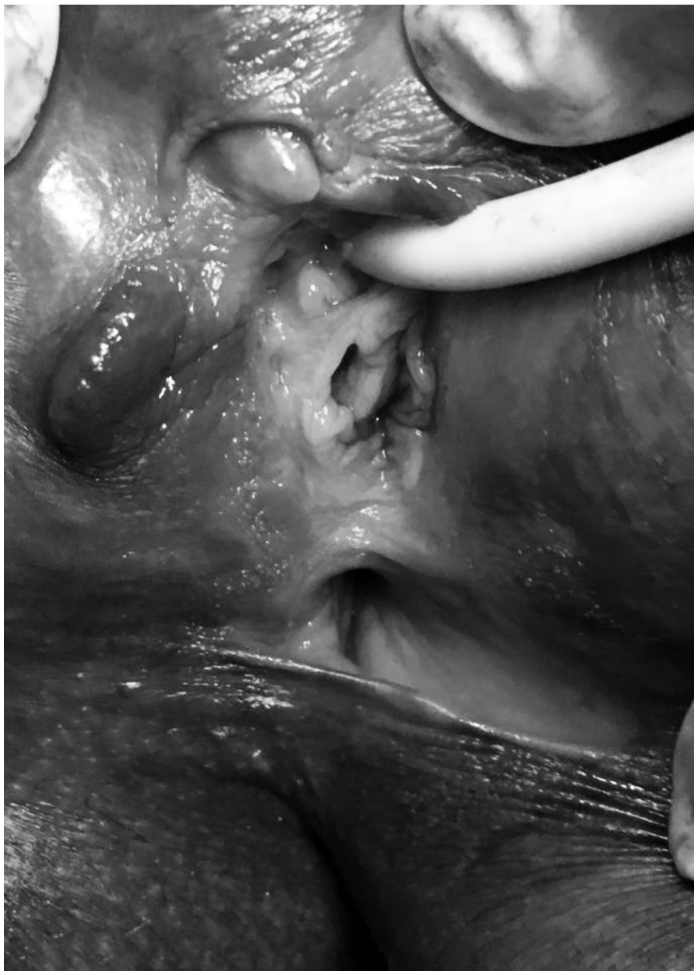

**Figure 4.** Pelvic examination. Normal external genitalia nulliparous vagina, cervix is 0.5x0.5cm knobby and deviated to the left corpus enlarged to age of gestation, lax sphincter tone, intact rectal vault, no intraluminal masses, bilateral parametria smooth and pliable.

controlled with the Nifedipine 30mg loading dose and was maintained on 20mg every 4 hours. Diagnostic work-ups were requested which included a chest antero-posterior radiograph that revealed thoracic dextroscoliosis, prominent right scapula. The lumbosacaral view noted a free window between L4 to L5 for anesthetic insertion of a spinal needle (Figure 5). A transabdominal ultrasound was done which revealed that her right kidney was displaced to the level of her anterior superior iliac spine. Two-dimensional echocardiography done on the patient revealed that she had an atrial septal defect, with predominantly left to right shunting, concentric left ventricular hypertrophy with adequate wall motion and contractility and preserved overall systolic function (66%), normal left atrium, right atrium and right ventricular dimensions with normal contractility and systolic function, normal aortic root and pulmonary artery dimensions, mitral sclerosis with mild mitral regurgitation, structurally normal aortic valve, structurally normal tricuspid and pulmonic valves, mild tricuspid regurgitation, normal arterial pressure with pulmonic regurgitation.

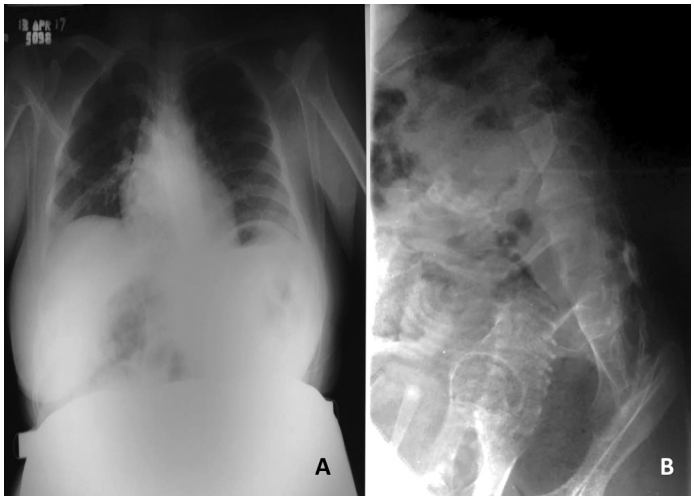


**Figure 5.** (A) Chest anteroposterior radiography (B) Lumbosacral radiography, There is note of a free space between L4-L5.

After the necessary work-ups were completed, the multidisciplinary team which included the sections of obstetrics and gynecology, anesthesiology, cardiovascular medicine reached a consensus that the best approach for the delivery was to have a scheduled cesarean section. During the multidisciplinary meeting it was pointed out that due to the presence of a spinal deformity the anesthesiology service recommended that during the elective cesarean section, general anesthesia was to be done. The cardiovascular service did not have any objections to the operative plan. The patient and her family were made aware of the consensus and the risks and benefits were explained to them.

The patient was then scheduled for elective cesarean section on her 5th hospital day. While awaiting the scheduled date, the patient was referred to other services which included orthopedics and rehabilitation medicine. The section of orthopedics noted that they would see the patient in an OPD setting for evaluation for possible corrective surgery and prosthetics fitting. The department of rehabilitation medicine saw the patient and evaluated her for possible occupation al therapy post delivery.

Intraoperatively, the patient was placed in supine position and placed under general anesthesia. A vertical skin incision was done and a low vertical cesarean section was done. A live baby girl was delivered weighing 2685 grams, 38 weeks by pediatric aging, appropriate for gestational age, via complete breech extraction with an APGAR score of 7 becoming 9. After placental delivery, the uterus was then noted to be bicornuate in nature (Figure 6). Carbetocin 100mcg was given intramuscularly in anticipation of a delay in uterine tone. The estimated blood loss was 500cc which did not necessitate any blood transfusion.

The patient tolerated the procedure well. After a few hours in the recovery room she was transferred back to the ward once she was fully awake. The nursing attendants of the ward taught her how to properly carry and position the infant during breastfeeding. There was no note of difficulty in positioning and handling the infant while admitted.

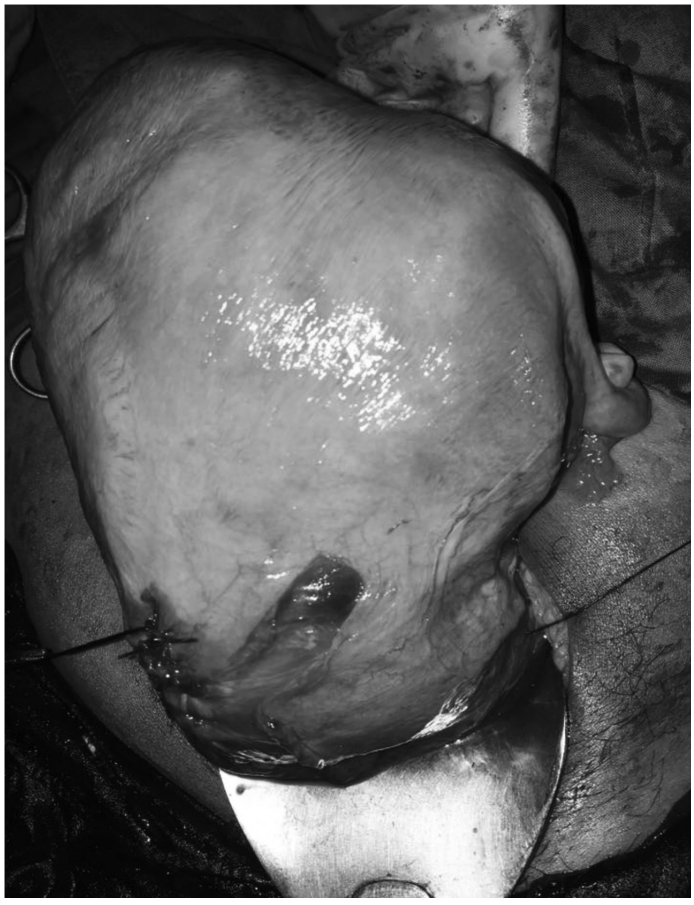

**Figure 6.** Bicornuate Uterus.

Four months post-operatively, our patient is able to care for her child requiring help only in carrying the infant to the lavatory for bathing. Otherwise, she is able to care for and nurse her child on her own.

She was scheduled to have follow-up with the orthopedics section on OPD basis but has failed to follow-up with them due to financial difficulties in acquiring transportation back and forth from the hospital.

## DISCUSSION

VACTERL association is a rare disease and much of the literature written and available has been focused on pediatric patients diagnosed with the disease. Little is written on the long term outcomes and other medical or surgical sequelae faced by patients with the disease.

Currently there is no available objective laboratory test, for its diagnosis. VACTERL association is diagnosed based on the clinical manifestations4 that presents in combination in a patient. In the study done by Raam, Pineda-Alvareza, Hadleyc and Solomon, they noted that significant malformations cannot be ascertained until after the patient's care has moved beyond the pediatric realm[5]. With the advancements in the field of medicine and technology, more and more medical practitioners and researchers are now able to discuss and share their findings and report on them on a digital platform but despite this, only a few literature have been written following up on the status of patients diagnosed with VACTERL association after they have reached adulthood. In the same study Raam et al noted that after following up on 11 patients diagnosed to have VACTERL, they noted that there were no evidence of neurocognitive and developmental delays. This implies that the prognosis of patients with VACTERL is promising if the necessary work-up and intervention is provided for them.

For parturient patients with VACTERL association an elective cesarean section under general anesthesia is the ideal approach given that 60-90% of patients with the disease present with vertebral anomalies which may vary in severity[6,7]. It is notable that VACTERL association is rarely combined with genital anomalies[8]. Our patient had a hypoplastic cervix which aside from the fused limbs prevented her from having a possible vaginal delivery.

A major concern that will be faced by parturient individuals with disabilities is in how they would care for their newborn infant. In the study done by Lin, Lin, Lin et al, they reported that most children with VACTERL association have a degree of functional independence similar to that of unaffected children[9,10]. The level of functional independence however would greatly vary based on the degree of the associated congenital

anomalies present in an individual. Depending on the needs of an individual patient, it would be ideal to have a team of specialists which should include members from rehabilitation medicine, orthopedics and psychiatry in order to approach and counsel the patient and her family members with regards on how to go about the daily functions needed prior, during and after delivery.

## SUMMARY

VACTERL association is a rare disease and the occurrence of having a parturient patient with VACTERL association is even rarer given the complex combination of congenital anomalies which may accompany the disease. As such, when a parturient patient with VACTERL association presents herself, a multidisciplinary committee should be tapped in order to fully maximize the help that the medical field can offer such individuals. The family members of such a patient should also be prepared for the additional role they would play once the patient delivers and the child is born. They should also be assured given the assurance during the pregnancy that the possibility that the unborn child would have the same congenital anomaly is even rarer than its occurrence. ■

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