CASE REPORT

# VACTERL association after infertility treatment - case report

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## Abstract

VACTERL association includes at least three of the following congenital anomalies: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal and limb abnormalities. The objective of article is to demonstrate the perinatal outcome of VACTERL case in 38-year-old pregnant woman with a history of infertility. Pregnancy has occurred after intracytoplasmic sperm injection and embryoscopy before embryo transfer. The 1st trimester ultrasound demonstrated right forearm anomaly-aplasia of the right radius and thumb. The normal female karyotype (46XX) was obtained. Increasing amniotic fluid index and invisible stomach from 26 weeks confirmed the suspicion of VACTERL association. After reduction of 900 ml of amniotic fluids at $34^{+4}$ weeks of gestation the symptoms of premature separation of placenta occurred. The newborn weight was 2,360 g according to Apgar scale assessment 5/5 at $1^{st}$ and $5^{th}$ minutes. Multiple reconstructive operations were performed after birth. Among them sigmostomy, gastrostomy, ezofageotraheal fistulae plastic, that were followed by bronchoscopy and fibrogastroscopy. VACTERL association is a sporadic phenomenon, but among ICSI pregnancies fetal structural abnormalities occurrence is slightly higher. Our case demonstrates that the different components included into VACTERL association can be visualized at different gestational age, allowing to suspecting this association later in the pregnancy and often not prior to delivery. The family must has to understand the consequences of complications and their impact on the quality of child life.

### Keywords

Fetal anomaly, Infertility, VACTERL

## 1 Introduction

The vertebral, anal, cardiac, trachea-esophageal, renal, and limb birth defects or VACTERL association is a rare congenital malformations spectrum [1]. In addition to these mentioned components included into VACTERL the fetus might also have other congenital anomalies. VACTERL association incidence is estimated between 1 in 10,000 to 40,000 births [2]. The number is variable, as different studies have used different diagnostic criteria.

The differential diagnosis for VACTERL association includes a wide variety of syndromes, among which are Baller-Gerold, CHARGE and Currarino syndromes, deletion 22q11.2 syndrome, Fanconi anemia, Feingold and Fryns syndromes,

MURCS association, oculo-auriculo-vertebral and Opitz G/BBB syndrome, as well as Pallister-Hall and Townes-Brocks syndromes [2].

Antenatal diagnosis can be complicated, as certain features can be difficult to ascertain prenatally. The different components included into VACTERL association can be visualized at different gestational age, allowing to suspect this association later in the pregnancy and often not prior to delivery.

The aim of this manuscript is to demonstrate the outcome of patients with VACTERL association in infertile patient after embrioscopy. Embryoscopy is a novel method, when images of the embryos can be acquired every 15 min with a time-lapse system and therefore embryo developmental ability. Embryo time-lapse analysis makes it possible to identify with objectivity poor prognosis markers of implantation. Embryo transfer of the embryo of good quality improves the implantation rate and outcome of ICSI procedures.

## 2 Case presentation

We presented a case of 38-year-old patient with a history of 15 years of infertility in whom pregnancy has occurred after intracytoplasmic sperm injection (ICSI) and embryoscopy before embryo transfer (ET). First trimester ultrasound demonstrated right forearms anomaly with abnormal wrist angle (see Figure 1). The patient refused to performe chorionic villus sampling. At $16^{th}$ weeks of pregnancy the right radius and the thumb aplasia was confirmed. Diagnostic amniocentesis was performed and normal female karyotype (46XX) was obtained. It is known, that radius bone aplasia is associated with a number of syndromes and disorders that may manifest later in pregnancy. The family was informed about that and decided to continue the pregnancy. Starting from $26^{th}$ weeks of gestation the stomach was not filling anymore, it was not possible to visualize the anus as well. Progressively increased amniotic fluid index (AFI). Abnormalities led to think about tracheal-esophageal fistula and confirmed the suspicion of VACTERL association. That followed by new finding at $30^{th}$ weeks of gestation: vertebral malformation (see Figure 2). At $34^{+4}$ weeks of gestation AFI reached 45 cm. To reduce the risk of premature delivery amnioreduction was performed. After reduction of 900 ml of amniotic fluids, contractions started. That followed by amniotomy and 4 L of amniotic fluids was released. The symptoms of premature separation of placenta lead to emergency cesarean section. The newborn weight was 2,360 g. Apgar scale assessment 5/5/6 at $1^{st}$, $5^{th}$ and $10^{th}$ minutes. The girl was intubated and transferred to neonatal intensive care unite (NICU). Catheter into the esophagus could enter only 11 cm in depth. The absence of radius, thumb anus was confirmed (see Figures 3, 4). The child was transfered to the Children's Hospital. Sigmostomy, gastrostomy, tracheoesophageal fistula reconstruction were performed on the second day of life followed by bronchoscopy and fibrogastroscopy. Later on the positive dynamics was observed: sigmostomy started working and feeding with breast milk was started through the gastrostomy. The child was extubated on the $6^{th}$ day after operation. From $15^{th}$ day of life the child was oxygen independent. In the next months the girl survived reconstruction of esophagus and jejunostomy with several episodes of sepsis due to repeated invasive procedures and devices. At 5 months of the life her weight was only 4,540, oral intake was started combining with functioning iejunostomy. According to the plan the reconstruction of anal sphincter is planned in the future.

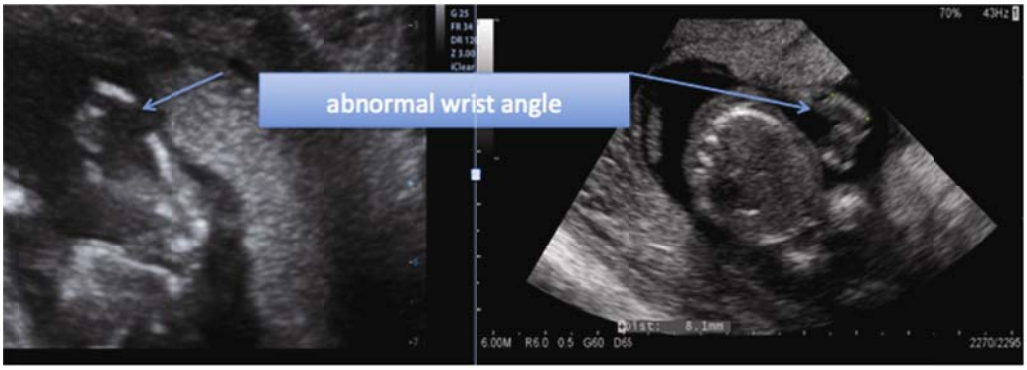


**Figure 1.** Right forearms anomaly with abnormal wrist angle (*arrow*), $12^{+5}$ weeks scan

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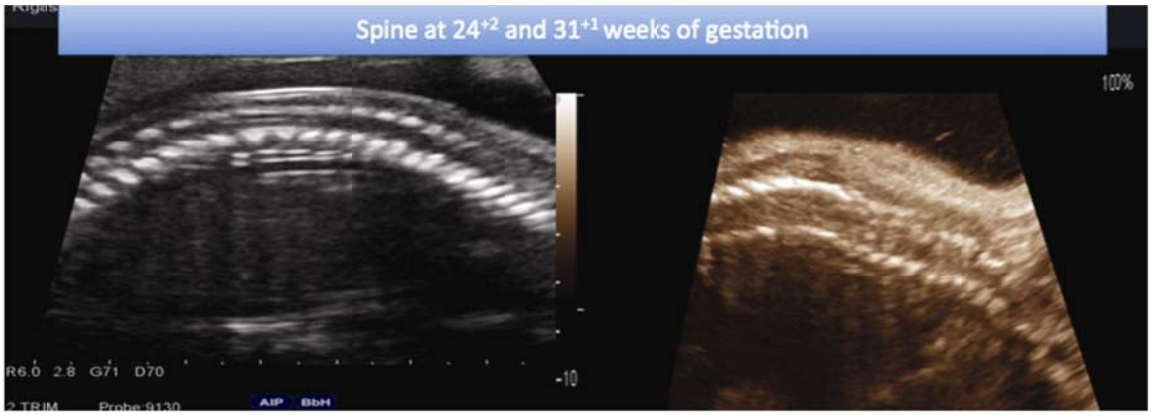


**Figure 2.** Abnormality of spine at $24^{+2}$ and $31^{+1}$ weeks

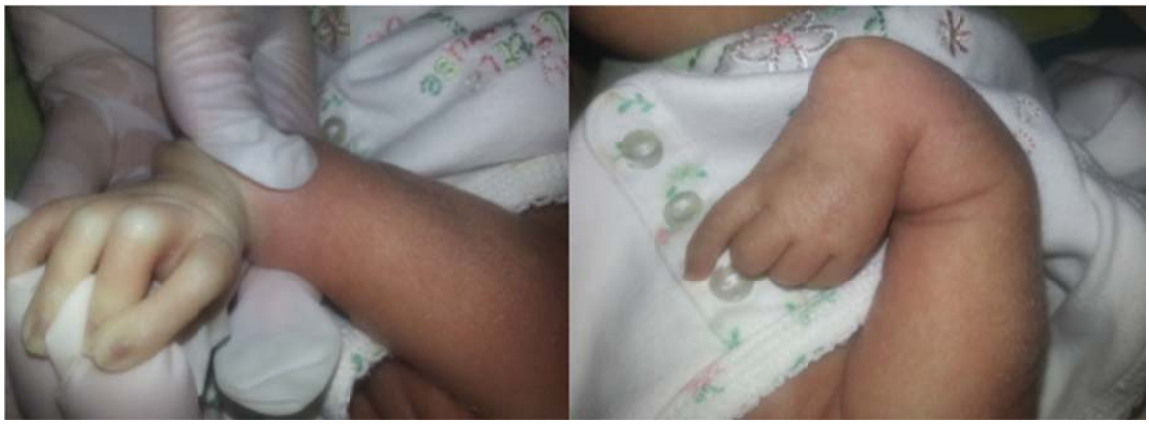

**Figure 3.** The right radius and the thumb aplasia in newborn

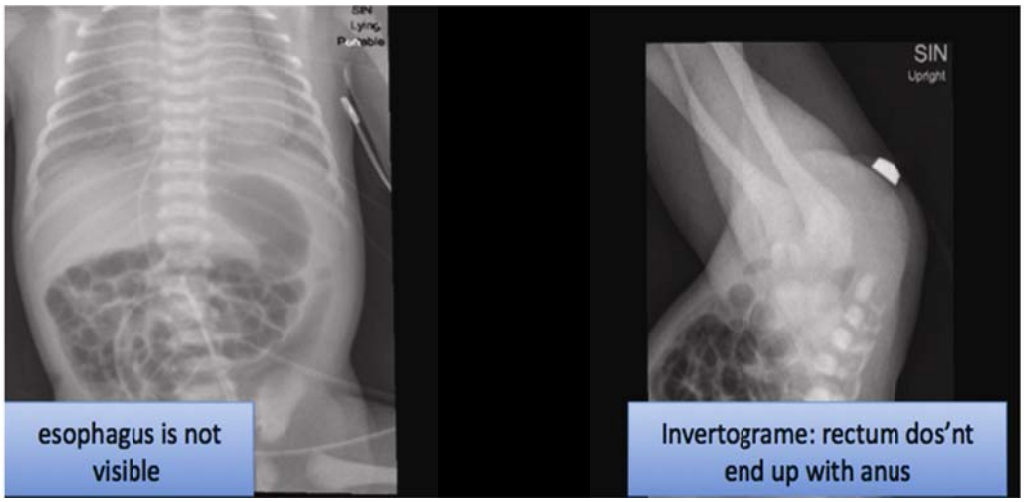


**Figure 4.** X-ray image of abdominal cavity and thorax (gastric tube ends up in stomach) and invertograme

# 3 Discussion

The first description of VACTERL association mentioned in the literature was in 1973 by Quan and Smith, who analyzed seven patients with combined vertebral defects, anal atresia, tracheo-esophageal fistula, radial and renal dysplasia [3]. Later

on extra abnormalities, including cardiac, were reported. In spite of the passed years the etiology of VACTERL association still remains a mystery for scientists [4]. Inherited factors, as well as environmental influences have been reported [5-7].

Antenatal imaging, prenatal echocardiography, as well as MRI should help to make the diagnoses of VACTERL quite early, but unfortunately even with careful and skillful examinations anorectal anomalies and tracheo-esophageal fistula often can not be detected in the second and early third trimester of pregnancy, even with a careful and skillful examinations.

Modern surgical techniques in our days improve prognoses of infants with VACTERL association, but we should remember that the coexistence of VACTERL association adversely affects not only the surgical outcome but also the organ functioning [8]. Further studies are needed to evaluate the quality of life of these patients.

VACTERL association is a sporadic phenomenon, but among ICSI pregnancies fetal structural abnormalities occurrence is slightly higher, mostly related to parental factors. We demonstrated the first case of the VACTERL association among patients after infertility treatment.

Our case demonstrates that various anomalies included into VACTERL association can be visualized ultrasonographicaly at different gestational age consequently allowing to suspect this association later on in pregnancy. And even during anomaly scan at 20-22$^{nd}$ week of gestation, when radius aplasia looks like an isolated pathology, patients should be informed about possible late findings and severity of various pathologies. The family must understand the consequences of complications with the impact on the quality of child's life.

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