



# Concordant VACTERL Anomalies in Identical Twins

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## Abstract

Multiple malformation syndromes occur in less than 1/3000 live births. The birth anomalies associated with VACTERL or VATER are well documented in the literature. VACTERL in twin pregnancies has also been documented. An extensive search of the literature reveals this as the first reported case of three shared anomalies in identical twins.

This is a case report of naturally conceived identical twin boys born at 33 and 1/7 weeks to a 26 year old primigravida mother. Boy A born at 1790 grams and Boy B at 2015 grams. Both were noted to have imperforate anus and hypospadias after birth. Both were found to have colocutaneous fistulas. Both underwent operative repair of their anorectal anomalies.

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## Case Presentation

This is a case report of naturally conceived identical twin boys born at 33 and 1/7 weeks to a 26-year-old primigravida mother. Boy A was born at 1790 grams and Boy B at 2015 grams. Both were noted to have imperforate anus and hypospadias after birth. Both were found to have colocutaneous fistulas, that of Boy B being slightly larger than that of Boy A. Ultrasound of both heart and kidneys did not reveal any abnormalities in either child. Skeletal survey likewise was negative. Both underwent transverse loop colostomy on Day 2 of life for diversion with planned anoplasty at a later time. Difficulty in intubation prompted concern for tracheal stenosis and therefore rigid bronchoscopy was also performed with subsequent computed tomography and 3-dimensional reconstruction. Boy A was found to have narrowing in the mid tracheal region with the tightest area measuring approximately 2.0 - 2.6 mm in the AP plain, 2.3 mm in the coronal plain extending over 5 to 10 mm. Boy B was found to have narrowing of the mid trachea with the narrowest portion approaching 1.2 mm as well as focal stenosis of the left mainstem bronchus to 1.7 mm. Both boys were monitored in the neonatal intensive care unit and did well without supplemental oxygen. Urology planned delayed reconstruction of the genitalia. Boy A was discharged to home on Day 15 of life and Boy B on Day 27 due to delayed feeding tolerance. At 5 months of age both returned for rectal exams under anesthesia, and both were confirmed to have colocutaneous fistulas anterior to their rectums. Anoplasty and subsequent colostomy takedown was successful in both patients. Neither has experienced any respiratory compromise since initial discharge.

## Discussion

VACTERL associated anomalies include those of the vertebral, anorectal, cardiac, tracheoesophageal, and renal organ systems as well as the limbs. These defects are likely to occur together in almost any combination of two or more and usually represent a sporadic occurrence in an otherwise normal family [1]. Sporadic anomalies in twins have been reported as there is a known increased risk of congenital malformation with twin pregnancies. Often the anomalies are discordant. Sunagawa et al. [2] recently reported a case of twins conceived after intracytoplasmic sperm injection and embryo transfer each born with VACTERL anomalies, but the children had different anomalies. Cox et al. [3] reported a case of twins with VACTERL anomalies in the same organ systems, but ultimately discordant as each possessed different malformations in these organ systems along with several unique anomalies isolated to each twin. Ardiet et al, Kanasugi et al, and Sandal et al. [4-6] have all reported cases of VACTERL in twins where only one child was born with anomalies. On the other hand, isolated concordant anomalies have been reported. Kubiak et al. [7] reported a case of isolated imperforate anus in monozygotic twins. Their case represented the fifth case of twins concordant for isolated anorectal anomalies.

Multiple identical VACTERL anomalies in twins, however, have never been reported. A case

of mirror image posterior urethral valves and mirror image facial hemihypoplasia was reported in monozygotic twins by Morini et al. [8]. A report of twins each with esophageal atresia and tracheoesophageal fistula, as well as PDA was made in the European literature by Faquhar et al. [9]. Although both had tracheoesophageal anomalies, the PDA does not represent an anomaly per se. Our case, therefore, represents the first case of multiple concordant VACTERL anomalies in twins, involving tracheoesophageal, anorectal, and urinary tract anomalies.

## Conclusion

While VACTERL association anomalies have been observed in twin gestations, there have been no reports to date demonstrating identical anomalies. Here we present such a case with twins having identical anomalies in the tracheoesophageal and anal domains of the VACTERL association. This report lends support to the role of genetics in this rare congenital malformation grouping.

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