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VACTERL/VATER association—Can a patient with VACTERL association live independently?

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The VACTERL/VATER association is a statistically nonrandom co-occurrence of a group of congenital malformations, including vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistulas, renal anomalies, and limb abnormalities. Because no pathogenetic relation has been found between these malformations, the term "association" is used instead of "syndrome". The incidence of VACTERL association is about 1/10,000–1/40,000 births. No geographical or ethnic differences have been found. The association is defined as at least three component features without clinical or laboratory-based evidence of an alternative diagnosis. Management of VACTERL association is focused on treating issues related to each component feature. The immediate management includes repair of tracheoesophageal fistulas, anal atresia, and severe cardiac defects. Dysphagia, esophageal strictures, gastroesophageal reflux, tracheomalacia, recurrent pneumonia, constipation, fecal incontinence, severe low back pain due to vertebral anomalies, and recurrent nephrolithiasis due to renal anomalies are important issues in long-term management. Fortunately, according to Lin et al's study in 2010, the daily functional skills of Taiwanese children with VACTERL association without brain damage are similar to those of unaffected children. It is vital for the family and society to care for the patient with VACTERL association.

A newborn baby boy with a maternal history of polyhydramnios noted during *antepartum* examination was found to have drooling, respiratory distress, and multiple congenital malformations soon after birth. An oral–gastric (OG) tube could not be inserted. Physical examination showed no dysmorphic facial features, a regular heartbeat without murmur, normal upper limbs, normal spinal curvature, penile hypospadias, and imperforate anus (Fig. 1A). Chest radiography revealed normal vertebrae with extra ribs, coiling of the OG tube and a distended gastric bubble, with a Gross classification type C tracheoesophageal fistula (Fig. 2). Whole body sonography demonstrated corpus callosum dysgenesis, a mild patent foramen ovale, and no renal anomalies. End-to-side anastomosis of the esophagus, repair of the trachea, and an anorectoplasty (Fig. 1B) were done at 4 hours *postpartum*. The patient was stable postoperatively. Feeding with breast milk was started through the OG tube on postoperative Day 7. The anal sutures were removed 13 days postoperatively, and anal dilatation was done daily. The patient was discharged 14 days postoperatively after the parents learned care techniques. At the time of writing, the patient was aged 5 months. After regular rehabilitation, his growth and development were all within normal limits, and his rehabilitation course will be finished shortly.

For primary care medical personnel, there are several major concerns in a child born with a congenital anomaly. These include its severity and whether it is life-threatening, isolated or combined with other anomalies, part of a pattern recognized as a syndrome, or associated with other anomalies. Clinicians need to determine whether any molecular genetic testing is currently available. Care by an interprofessional medical team and the parents' psychosocial concerns and emotional response must also be considered. Explanation of outcomes and genetic counseling to evaluate recurrence risks are mandatory.

For patients, early infant stimulation programs, regular follow-up of neurocognitive milestones, a complete vaccination program, membership in a disease association, and lifelong medical care plans are needed.

The first concern of most parents with a child with a congenital anomaly is how well that child will function in life; for those with

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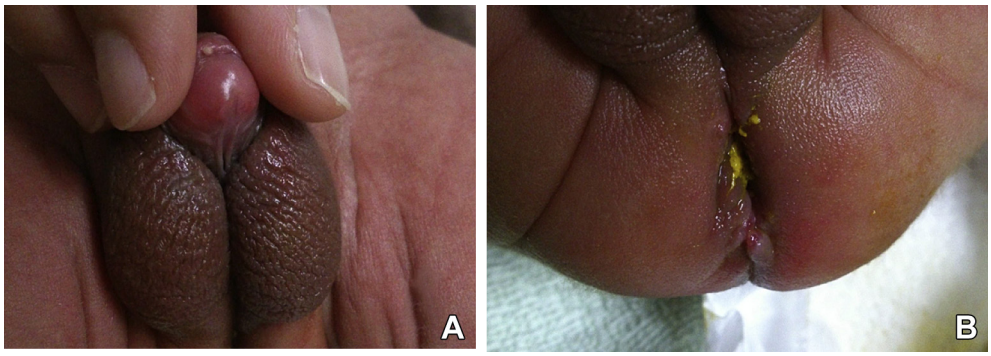

Fig. 1. (A) Penile hypospadias in a newborn boy. (B) The patient could defecate without problems on Day 7 after an operation for imperforate anus.

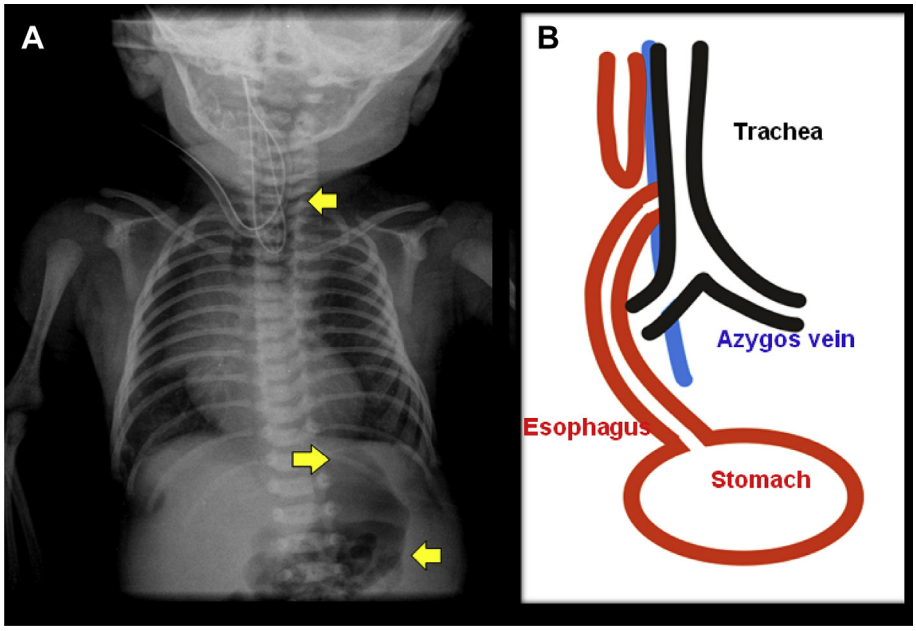

Fig. 2. (A) Preoperative chest radiograph reveals normal vertebrae with extra ribs, coiling of the oral–gastric tube and a distended gastric bubble, and (B) gross classification type C tracheoesophageal fistula (diagram on right).

VACTERL, the prognosis can be relatively positive. Patients with VACTERL association can live independently.

Further reading

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