Skeletal anomalies in VACTERL association

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INTRODUCTION

The VACTERL association is a group of developmental anomalies which non-randomly occur together. The name is derived from the first letters of the anomalies or affected organs: **V** – vertebra, **A** – anal atresia, **C** – cardiac anomalies, **TE** – tracheo-esophageal fistula, esophageal atresia, **R** – renal anomalies, **L** – limb anomalies.

When any anomaly of the VACTERL association is encountered in a new-born, other anomalies must be systematically searched for. Three VACTERL component features are required for a diagnosis of the VACTERL association, with no one anomaly taking key importance over the others. Due to the complexity of the clinical presentation, extensive testing needs to be performed to rule out the many overlapping conditions such as Fanconi anaemia, CHARGE syndrome, Baller Gerold syndrome, etc[1]. Various imaging techniques play an important role in confirming the anomalies, as well as planning of possible surgical interventions and long-term observation. The workup consists of radiographic imaging (chest radiograph or radiograph of the whole body of a new-born, i.e. “babygram”) and ultrasound of the abdomen and heart. In cases of more complex anomalies, the workup can be further expanded with radiographic contrast imaging, computed tomography, or magnetic resonance imaging[2].

LIMB ANOMALIES

Initially radial anomalies represented a defining feature of the VACTERL association. Since then, a variety of other limb anomalies have been reported in 40 – 55 % of affected individuals[3]. The severity of malformations varies widely with the most typical being:

- **Radial anomalies:** either complete aplasia or partial hypoplasia with or without a deficiency of the thumb bones.
- **Hypoplastic thumb.**
- **Polydactyly or oligodactyly.**

The initial work-up consists of a physical examination to assess for upper and lower limb anomalies[6]. Upon suspicion or detection of limb anomalies initial plain radiographs can be followed by CT or MRI as needed.
Treatment plan should include early physiotherapy and eventual surgical intervention[7].

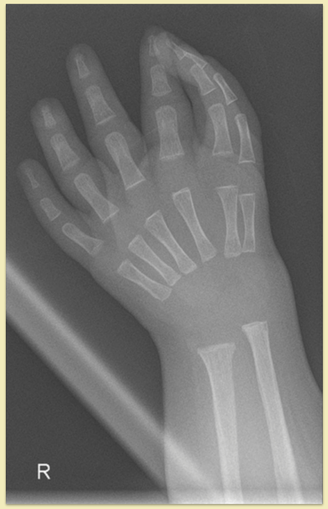

Figure 1. Hand X-Ray of a girl with polydactyly. Exam was preformed in order to exclude radial dysplasia.

VERTEBRAL ANOMALIES

Vertebral anomalies are among the more common anomalies of patients with VACTERL and have been described in 60 – 90 % of affected individuals[3]. One or more of the vertebra may be affected to a varying degree. Vertebral anomalies may present together with rib anomalies, the latter can, however, also present solitarily[2]. The most typical anomalies are:

- **Hemivertebra.** A failed formation of one half of the vertebral body, which can lead to type I congenital scoliosis, kyphosis or lordosis.
- **Vertebral fusion.** May lead to type II congenital scoliosis.
- **Absent or supernumerary vertebra.**
- **Caudal regression or sacral agenesis.** A structural defect of the caudal region of the spine consisting of an absent sacrum and defects of the lumbar spine.
- **Spina bifida.** A defect of the neural tube most often located in the lumbo-sacral region of the spine.

Vertebral anomalies may be associated with spinal cord anomalies. To initially assess a spinal cord anomaly a peripheral neurologic and muscle exam (deep tendon and primitive reflexes, muscle tone, response to tactile stimulation) should be performed. Despite radiation exposure and sedation risks involved, MRI or 3-dimensional CT examinations may be needed in complex malformations. An ultrasound presents a readily available, efficient, and affordable alternative, which avoids sedation, and is considered a valid alternative to MRI up to approximately 3 months of age[4].
Depending on the severity of the malformation orthopedic tracking and surgical interventions may be required[5].

SOURCES:

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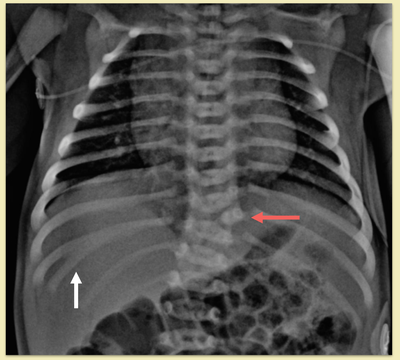

Figure 2. Hemivertebra Th9 (red arrow). On the right side only 11 ribs are present and the 9th rib is bifid (white arrow).

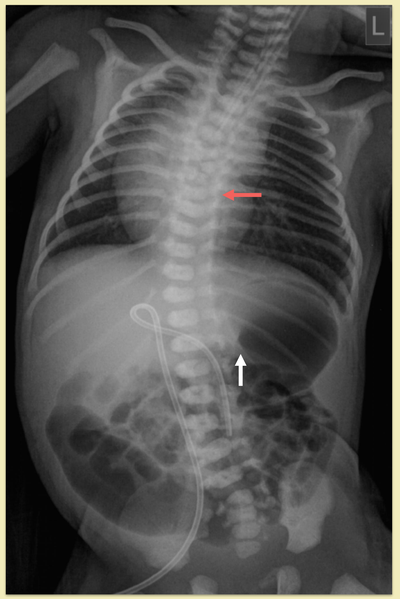

Figure 3. Anomalies of vertebrae Th5-Th8 (red arrow), a hemivertebra at L5/S1 and absent ossification centers of the coccyx can be observed. There is an additional 13th pair or ribs (white arrow). Additionally, in the upper mediastinum the tip of the nasogastric tube curves cranially which indicates another anomaly of the VACTERL association – esophageal atresia. The child also had anal atresia, which can be suspected by the absence of the ossification centers of the coccyx and lack of gas in the rectum.