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Vacterl Association with Caudal Regression Syndrome and Complex CHD

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Abstract: ***A rare case of a 1 year 7 months old boy presenting with multiple congenital anomalies including anorectal malformation, double outlet right ventricle with pulmonary atresia, bilateral hydronephrosis, and limb defects. The child fulfilled criteria for VACTERL association and also demonstrated features of caudal regression syndrome. He underwent multiple surgical interventions during infancy including staged anorectal reconstruction, PDA stenting, and colostomy closure. Despite ongoing supportive management, the child continued to experience cyanosis, recurrent urinary tract infections, and developmental delay. This report underscores the challenges of diagnosis and multidisciplinary care in children with overlapping syndromic anomalies.***

Keywords: VACTERL, caudal regression syndrome, anorectal malformation, congenital heart disease, limb anomaly

1. Case Report

A 1 year 7 months old male child, born of first-degree consanguinity, presented on 13 August 2025 with cyanosis on crying, increased urinary frequency with dysuria, and delayed milestones related to limb deformities. Antenatal history was uneventful. No similar family history was reported. At birth, absent anal opening was noted and a sigmoid colostomy was performed; he was later diagnosed with double outlet right ventricle (DORV) with pulmonary atresia and Patent Ductus Arteriosus (PDA). During infancy, he underwent colostomy revision (5 months), PDA stenting (7 months), abdominoperineal posterior sagittal anorectoplasty (9 months), and colostomy closure (11 months). At 10 months, he developed acute urinary retention with recurrent urinary tract infections; ultrasonography revealed bilateral hydronephrosis. At 12 months, intestinal obstruction was managed conservatively. On current admission, the child was drowsy, weighing 8.3 kg, with heart rate 134/min and oxygen saturation 64% on room air. Clinical examination revealed cyanosis, clubbing, surgically reconstructed anus requiring bowel regulation, right phocomelia, and a deformed left foot. Cardiovascular examination showed normal S1 and S2 with a systolic murmur. Hemoglobin was 19 g/dL, hematocrit 59.1%, creatinine 0.29 mg/dL, and chest radiograph showed cardiomegaly with increased bronchovascular markings. Based on the constellation of anomalies, a diagnosis of VACTERL association with caudal regression syndrome and cyanotic congenital heart disease was made. The child was managed supportively and planned for surgical correction of tetrology of fallot.

2. Definition

Caudal regression syndrome is a rare congenital disorder characterized by developmental failure of the caudal portion of the spine and spinal cord. It represents a spectrum of anomalies ranging from partial agenesis of the coccyx to complete absence of the lumbosacral spine.

Etiology and Risk Factors:

- Exact cause unknown; multifactorial.
- Strongly associated with maternal diabetes mellitus (15–25% of cases).
- Genetic predisposition and vascular hypo perfusion during embryogenesis have been implicated.

Pathogenesis:

Abnormal development of the caudal mesoderm during the 3rd–7th week of gestation leads to varying degrees of sacral and lumbar vertebral agenesis. This disruption may also affect the spinal cord, genitourinary, gastrointestinal, and musculoskeletal systems.

Clinical Features:

- **Spinal/Neurological:** Sacral agenesis, absent deep tendon reflexes, neurogenic bladder, lower limb weakness or deformities.
- **Musculoskeletal:** Clubfoot, phocomelia, hip dislocation, contractures.
- **Urogenital:** Vesicoureteral reflux, hydronephrosis, recurrent UTIs, renal agenesis.
- **Gastrointestinal:** Anorectal malformations, imperforate anus.

Classification (Renshaw's)

- **Type I**: Partial/total unilateral sacral agenesis.
- **Type II:** Partial sacral agenesis with bilaterally symmetrical defect.

- **Type III:** Total sacral agenesis with variable lumbar involvement.
- **Type IV:** Total sacral agenesis with fusion of iliac bones.

Diagnosis
- **Antenatal:** Ultrasound/MRI can detect sacral agenesis.
- **Postnatal:** X-ray (absent sacrum/lumbar spine), MRI for spinal cord abnormalities, USG KUB for renal anomalies.

Management
Supportive and multidisciplinary.
- **Orthopedic:** Limb deformity correction, mobility aids.
- **Urological:** Clean intermittent catheterization, prophylactic antibiotics, surgical correction of reflux or obstruction.
- **Surgical:** Management of anorectal malformations, colostomy, PSARP.
- **Neurological**: Rehabilitation for motor deficits.

3. Nursing Management

1) Respiratory and Cardiac Support
- Monitor oxygen saturation and vital signs regularly (especially if associated congenital heart disease).
- Provide oxygen supplementation and position child to ease breathing.
- Assist in pre- and post-operative care for cardiac surgeries.

2) Nutritional and Gastrointestinal Care
- Monitor feeding tolerance, weight gain, and hydration status.
- Support colostomy/PSARP care: stoma site care, prevention of infection, bowel training using suppositories/enemas.
- Educate caregivers on diet modifications to prevent constipation and obstruction.

3) Genitourinary Management
- Strict monitoring of urine output.
- Support for clean intermittent catheterization (CIC) and hygiene to prevent urinary tract infections.
- Encourage adequate fluid intake, monitor for dysuria or retention, and report early signs of infection.

4) Skin and Mobility Care
- Prevent pressure sores in children with immobility/limb deformities.
- Physiotherapy and range-of-motion exercises to promote motor development.
- Support use of orthotic devices or splints as prescribed.

5) Infection Prevention
- Aseptic technique during catheterization, stoma care, and wound dressing.
- Early recognition of fever, irritability, or cloudy urine.

6) Psychosocial and Developmental Support
- Encourage play therapy and stimulation activities suitable for developmental age.
- Provide counseling and emotional support to parents, addressing feelings of guilt (especially in consanguineous marriages).
- Facilitate parental participation in daily care to enhance bonding and confidence.

7) Health Education & Discharge Planning
- Train parents in CIC, colostomy/PSARP aftercare, and recognizing signs of complications.
- Educate on importance of follow-up with cardiology, nephrology, orthopedics, and rehabilitation teams.
- Stress long-term multidisciplinary care to improve quality of life.

8) Prognosis
- Highly variable, depending on the extent of spinal agenesis and involvement of renal and cardiac systems.
- Patients with preserved renal function and manageable orthopedic issues may survive into adulthood.
- Severe cases with multiorgan involvement (as in your case child) carry poor prognosis.

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