Case Report

VACTERL-H Syndrome: A Rare Case Presentation with Multisystem Involvement

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Abstract

Background: VACTERL-H syndrome is a rare congenital disorder that involves vertebral, anal, cardiac, tracheoesophageal, renal, and limb anomalies along with hydrocephalus. In this case study, a 3-year-old boy presents with several congenital abnormalities that are consistent with the association between VACTERL and H. The patient also reported having hydrocephalus due to aqueductal stenosis, a right-hand radial ray anomaly, scoliosis, and a history of imperforate anus repair. A horseshoe-shaped kidney and a large dilatation of the ventricles were discovered by radiological examination. Early surgical procedures, such as anorectoplasty and a ventriculo-peritoneal shunt, were successful. This case illustrates the importance of multidisciplinary care and proper clinical assessment for patients with multisystem congenital abnormalities.

Keywords: VACTERL-H syndrome, congenital anomalies, hydrocephalus, radial ray defect, horseshoe kidney, anorectal malformation.

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Introduction

VACTERL association is a well-recognized congenital malformation complex characterized by involvement of multiple body systems, including vertebral defects, anal atresia, cardiac anomalies, tracheoesophageal fistula, renal defects, and limb abnormalities.[1-3] Additionally, it has been documented that hydrocephalus coexists with this condition and is collectively referred to as VACTERL-H syndrome, a more severe and genetically distinct variant.[4] Although the precise cause of the VACTERL association is unknown, most cases are sporadic. A small number of familial cases have been reported, suggesting that environmental or genetic factors may be involved. This syndrome is extremely rare; prior research estimated its incidence at 1-9 per 100,000 live births. This report presents the case of VACTERL-H syndrome in a young male child with involvement of multiple systems.

Case Presentation

A 3-year-old male child was brought to the hospital after having seizures for three days in a row. Laboratory tests showed very low sodium levels. The physical exam revealed scoliosis, macrocephaly, and a right radial ray anomaly. The patient's medical history included surgery to fix an anus that couldn't be opened at six days old. The developmental milestones were appropriate for the child's age, and there was no history of delayed development. The mother's history was normal. The mother did not have a fever, rash, drug use, radiation exposure, or a long-term illness during her pregnancy. There had been no previous stillbirths or abortions, and no scans had been done during pregnancy. The child had never had trouble eating or choking before.

Investigations

- MRI Brain: shows dilated ventricles, a sign of hydrocephalus.
- Ultrasound Abdomen: Revealed a horseshoe kidney.
- Chest X-ray: Demonstrated scoliosis.
- X-ray Right Wrist: Showed radial ray anomaly.

All of the above findings indicated the presence of VACTERL-H syndrome with multiple involvement of different body parts along with the hydrocephalus participation.

Management and Outcome

The patient had low-pressure ventriculoperitoneal (VP) shunt surgery to help with hydrocephalus. The imperforate anus had been fixed by surgery soon after birth. Assistance was given to manage hyponatremia and control seizures. After the surgery, the patient's condition improved, with seizures stopping and sodium levels returning to normal. Long-term multidisciplinary follow-up was recommended to assess renal and musculoskeletal function.

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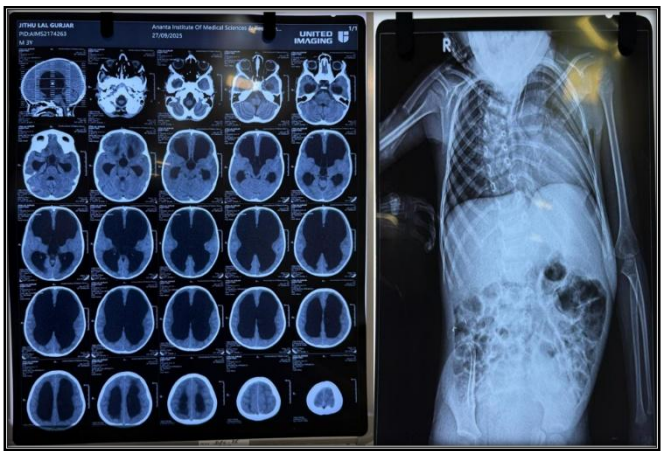

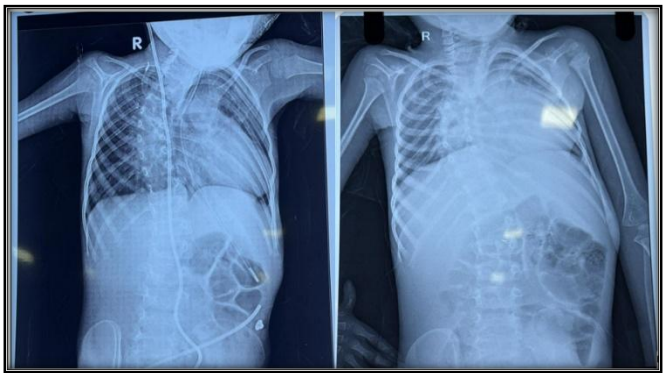

Discussion

VACTERL-H syndrome is a serious form of the VACTERL association that includes hydrocephalus and other congenital disabilities.[7] While the precise mechanism remains unclear, numerous studies indicate that early mesodermal defects play a role in embryogenesis. Diagnosis is chiefly clinical, corroborated by imaging studies.[8] Management prioritizes the rectification of life-threatening malformations, including anorectal and cardiac defects, subsequently addressing renal or limb abnormalities.[9] Improving survival and quality of life requires early diagnosis and coordinated multidisciplinary care, frequently involving pediatric surgeons, neurologists, nephrologists, and orthopedists. In these cases, immediate pediatric and neurosurgical interventions yielded positive outcomes, underscoring the importance of early diagnosis and care.

Conclusion

A thorough physical examination and focused imaging tests are the pillars of diagnosing VACTERL-H syndrome. The severity and combination of anomalies determine the management strategy. To lower morbidity and improve outcomes for those affected, early diagnosis, timely surgical correction, and coordinated multidisciplinary care are crucial.

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Conflicts of interest

There are no conflicts of interest.

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