

소아재활

게시일시 및 장소 : 10 월 19 일(토) 08:30-12:30 Room G(3F)

질의응답 일시 및 장소 : 10 월 19 일(토) 11:00-11:30 Room G(3F)

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Significance of Skeletal Imaging for Early Diagnosis of VACTERL association

Soyoung Lee^{1**}, Yong Min Choi¹, Seung Wook No¹

Keimyung University, School of Medicine, Daegu, Korea , Department of Rehabilitation Medicine¹

Introduction

VACTERL association specifically refers to the abnormalities in structures derived from the embryonic mesoderm. Some of the features of VACTERL association can be subtle and are not identified until late in childhood or adulthood, making a diagnosis of this condition difficult and realizing the need for rehabilitation too late. This case was presented to identify the significance of musculoskeletal imaging and to show how it was used for clinical diagnosis of VACTERL association.

Case report

The eight months old, full-term baby was referred to the Rehabilitation department for delayed development. She just underwent major surgery with the imperforated anus and had an anovaginal fistula. She was diagnosed with Atrial septal defect in pediatric cardiology and hydronephrosis in urology. The swallowing study was performed due to cricopharyngeal incoordination. The pediatrician conducted genetic studies to determine the cause of various congenital anomalies of the baby from birth but there was no abnormal finding. On physical examination, the baby shows an abnormal postural reaction on axillary hanging, side-lying, prone position, and abnormal Landau reaction. She preferred side lying position one direction only. The chest X-rays have been taken periodically about her heart problem in the pediatric department and showed Rt. thoracic scoliosis after 5 months old. We ordered x-rays of the spine, hand, and foot and finally, she was confirmed to be a VACTERL association. Approximately 90% of cases of VACTERL association appear to be sporadic, we could not find any abnormality on a genetic test.

Conclusion

VACTERL association is clinically defined by the presence of a cluster of congenital malformations. Abnormal spinal curvature due to underlying costovertebral anomalies is common. Clinical signs of scoliosis may be the first one of vertebral anomalies if imaging studies are not performed when the VACTERL association is suspected. The pediatric rehabilitation physician should pay attention to the musculoskeletal structures of an infant with multiple congenital abnormalities and delayed development before they show clinical

signs or symptoms. We could make an early diagnosis by careful review of the musculoskeletal imaging of the VACTERL association with delayed development.



Fig. 1 Chest X-ray on the day of birth.



Fig 2. Chest X-ray shows Rt. thoracic scoliosis on 5-month-old age.



Fig 3. Telegram of the upper trunk shows R. thoracic scoliosis with supernumerary ribs on 10-month-old age.