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A case of VACTERL association with autosomal dominant polycystic kidney disease in children

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Case Study: VACTERL association defects are characterized by vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula with esophageal atresia, renal dysplasia, and limb/radial defects. VACTERL's etiology is unknown and it is both clinically and etiologically heterogeneous. Autosomal dominant polycystic kidney disease (ADPKD) is the most common hereditary kidney disease. However, simultaneous presentation of both the VATERL syndrome and ADPKD is a very rare clinical entity.
A 10-year-old girl admitted to hospital, although she received two days' intravenous antibiotics treatment of acute pyelonephritis, she has been symptoms with costovertebral angle tenderness in left side, continuously. Abdomen CT showed acute pyelonephritis on left kidney and several simple cysts on both kidneys. On her past medical records, she was diagnosed as both hand polydactyly, tracheoesophageal fistula and atrial septal defect in the neonatal period. At 6months of age, she was diagnosed with vesicoureteral reflux grade III in left side. As a results she belongs to VACTERL syndrome and had to no familial history. When several simple cysts in both kidneys were incidentally found in her kidney sonogram at the age of 30months, unfortunately, she did not receive consultations regarding inherited cystic kidney disease. When we reassessed family history, we found out her grandfather was newly diagnosed with chronic kidney disease with cystic lesion. Therefore, we suspected ADPKD and performed the genetic study. The patient was identified to have the pathogenic mutations with a heterozygous c.6273C>T in exon 15 of PKD1[p. Gln2158Ter]. As a result, she was confirmed as ADPKD and nowadays her father was diagnosed with ADPKD, too.
We report a rare case of both VACTERL association and ADPKD presentation. This case shows the importance of careful history taking and clinical and radiologic follow-ups, and genetic confirmation of inherited cystic kidney disease evaluation when cystic kidney lesion is found, in pediatric period.