A RARE CASE OF POLYCYSTIC KIDNEY DISEASE AND MULTICYSTIC DYSPLASTIC KIDNEY IN A PEDIATRIC PATIENT

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Abstracts

Introduction Cystic kidney disease (CyKD) is one of the most important causes of chronic kidney disease (CKD) in children. Multiple kidney cysts can occur unilaterally (e.g. multicystic dysplastic kidney – MCDK) or bilaterally (e.g. autosomal dominant or autosomal recessive polycystic kidney disease) due to genetic or non-genetic (developmental or acquired) disorders. Here we present a rare case of combined polycystic kidney disease (PCKD) and MCDK in a pediatric patient.

Case Report The patient was admitted to our hospital for the first time at the age of two. She was normally developed (weight: 10.8 kg – 19th percentile, height: 90 cm – 80th percentile) with blood pressure (BP) within normal limits.

Estimated glomerular filtration rate (eGFR) at that time was 70.2 mL/min with elevated albumin/creatinine ratio 5.2 mg/mmol and she was diagnosed with G2A2 stage of CKD. During her prenatal period oligohydramnios and polycystic kidneys were detected. At birth a palpable mass of the left abdomen was found. First sonography after birth showed multiple kidney cysts of various sizes on the left kidney and lesser number of small cysts. At the age of four arterial hypertension (130/70 mmHg) was diagnosed and ACE inhibitor was introduced. Due to parental disapproval, genetic testing wasn’t performed.

Conclusion We conclude that, although rare, different types of CyKD can be associated and we should consider it when setting the diagnosis. Due to vast differential diagnosis and overlapping clinical presentations of CyKD genetic testing should be performed whenever possible.

358 RECURRENT MULTI DRUG RESISTANT URINARY TRACT INFECTIONS IN A THREE-YEAR-OLD HOSPITALIZED CHILD WITH HYPOTONIC CEREBRAL PALSY


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Background Catheter associated urinary tract infection (CAUTI) is a common device-acquired infection and represent a potentially harmful reservoir of resistant uropathogens. Guidelines recommend limitation of catheter use, aseptic catheter insertion, sterile equipment, strict hand hygiene, use of smallest catheter possible and maintenance of a closed drainage system.

Klebsiella Pneumoniae is a non-mobile aerobic rod causing a large spectrum of hospital-acquired infections, especially pneumonia or urinary tract infections (UTI), developing intrinsic resistance genes. Treating multi drug resistant(MDR) gram negative pathogens becomes a challenge for the caregiver.

Vesicoureteral reflux(VUR) consists of backflow of urine from the bladder into the ureters. It can be primary or secondary due to abnormal lower urinary tract function and elevated intravesical pressure. Post void residual(PVR) is a hallmark of detrusor underactivity(DUA) in children.

Case Presentation Summary We present the case a three years old boy, hospitalized for viral encephalitis, undergoing artificial respiratory support and urine catheterization for 6 weeks. Neurological status was hypotonic cerebral palsy and secondary urinary incontinence in a previously toilet trained child.

First febrile UTI developed two days after removing urine catheter. High resistant Klebsiella pn. (ESBL+, AAC(3)-II) was treated with a ten-day course of Cephraxine and Amikacin. Second febrile UTI was accompanied by febrile seizures, and urine culture was positive with MDR Klebsiella (+ESBL or +HL AmpC, Carbapenem impermeability). Treatment with high dose Meropenem (40mg/kg/dose) for twelve days was successful. Third febrile UTI occurred five days after finishing treatment with same MDR Klebsiella strain. Once again, fourteen-day high dose Meropenem course was successful. Fourth Follow-up ultrasound showed progressive involution of the MCDK left and multiple very small cysts that generate abnormal parenchymal echogenicity (eg, salt-and-pepper sign) on the right kidney. Functional MRI urography showed non-functional MCDK left and functional right kidney with lesser number of small cysts. At the age of four arterial hypertension (130/70 mmHg) was diagnosed and ACE inhibitor was introduced. There were no signs of liver disease. Abdominal ultrasound revealed no cysts of liver, spleen or pancreas. Gynecological ultrasound and ophthalmological examination were also normal. Due to parental disapproval, genetic testing wasn’t performed.

Conclusion We conclude that, although rare, different types of CyKD can be associated and we should consider it when setting the diagnosis. Due to vast differential diagnosis and overlapping clinical presentations of CyKD genetic testing should be performed whenever possible.
febrile UTI with Escherichia Coli occurred two weeks later and was treated successfully with ten-day course of Ciprofloxacin.

Ultrasound examination revealed normal kidneys and bladder, while voiding cystography was evocative for bilateral VUR and post void residual.

Neurologic status improved over a three-month period from almost complete hypotonic palsy to mild hypotonic paraplegia.

**Discussions** Long hospitalization and prolonged urine catheterization increase the risk of CAUTI. High dose Meropenem remains efficient in treating MDR Klebsiella strains. Recurrent UTI and was treated successfully with ten-day course of Meropenem.

Genetic screening has been recommended as the gold standard for the proper clinical diagnosis and understanding the importance of genetic test in diagnosis of inherited kidney disease. Alport Syndrome by genetic screening.

An eight-year-old boy presented with a history of macroscopic hematuria attacks preceded by upper respiratory tract infections. Renal biopsy was compatible with IgAN showing positive staining of IgA in the mesangium and the basement membrane in immunofluorescence.

Sensory neural hearing loss was developed at ten years of age. Also the proband had a family history of hematuria with proteinuria. Despite the positive mesangial IgA staining in immunofluorescence, atypical renal phenotypes for IgAN including persistent hematuria and hearing loss, and positive family history and electron microscopic findings of renal biopsy aroused suspicion of Alport Syndrome. Genetic analysis (whole exon sequencing) demonstrated a homozygous mutation in COL4A4 (chr 2q22.27922261 c.2438delG (p.Gly813AspfsTer56) in the proband. Heterozygous mutation was identified by Sanger sequencing of gene COL4A4 in the carrier of all member of his family.

Due to this very rare coincidence, we emphasize that atypical clinical findings should warn the clinicians for other possible diagnosis. The diagnosis of this case also highlights the importance of genetic test in diagnosis of inherited kidney disease. Genetic screening has been recommended as the gold standard for the proper clinical diagnosis and understanding the mode of inheritance when Alport syndrome has suspected in a patient.

**A NOVEL COL4A4 MUTATION IN THE PROBAND INITIALLY DIAGNOSED AS IGAN WITH AUTOSOMAL RECESSIVE ALPORT SYNDROME**

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**Immunoglobulin** A nephropathy (IgAN) is recognized as the most common form of glomerulonephritis all over the world. The diagnosis of IgAN is generally made according to clinical findings and histologic lesions on renal biopsy. Alport Syndrome is an inherited rare glomerular disorder characterized by hematuria, proteinuria, sensory neural hearing loss and progressive kidney disease. We here present a patient initially diagnosed as IgAN with biopsy findings and then diagnosed as Alport Syndrome by genetic screening.

An eight-year-old boy presented with a history of macroscopic hematuria attacks preceded by upper respiratory tract infections. Renal biopsy was compatible with IgAN showing positive staining of IgA in the mesangium and the basement membrane in immunofluorescence. Furthermore, the thinning and thickened of the glomerular basement membrane and splitting of lamina densa was found in electron microscopic examination. During his follow-up, persistent microscopic hematuria and proteinuria were noticed.

Sensorineural hearing loss was developed at ten years of age. Also the proband had a family history of hematuria with proteinuria. Despite the positive mesangial IgA staining in immunofluorescence, atypical renal phenotypes for IgAN including persistent hematuria and hearing loss, and positive family history and electron microscopic findings of renal biopsy aroused suspicion of Alport Syndrome. Genetic analysis (whole exon sequencing) demonstrated a homozygous mutation in COL4A4 (chr 2q22.27922261 c.2438delG (p.Gly813AspfsTer56) in the proband. Heterozygous mutation was identified by Sanger sequencing of gene COL4A4 in the carrier of all member of his family.

Due to this very rare coincidence, we emphasize that atypical clinical findings should warn the clinicians for other possible diagnosis. The diagnosis of this case also highlights the importance of genetic test in diagnosis of inherited kidney disease. Genetic screening has been recommended as the gold standard for the proper clinical diagnosis and understanding the mode of inheritance when Alport syndrome has suspected in a patient.