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REVIEW ARTICLE

Signs and symptoms of developmental abnormalities of the genitourinary tract☆

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KEYWORDS
Urinary tract malformations;
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Abstract
Objective: The abnormalities of the genitourinary tract development are the leading cause of chronic kidney disease (CKD) in children. The diagnosis of this disease in Brazil is late and incomplete, which results in increased morbidity and mortality in this age group. Early diagnosis of this condition is the prerogative of generalist pediatricians, and the aim of this study was to review the clinical signs and symptoms associated with developmental abnormalities of the genitourinary tract.
Data sources: Based on the description of a symbolic clinical case, the authors conducted a non-systematic review of medical literature.
Data synthesis: The results suggest that the following data should be used as a warning for early diagnosis of affected children: (a) combined urinary tract abnormalities (chromosomal abnormalities; sequence of malformations [VACTERLand Prune-Belly]; and musculoskeletal, digestive tract, heart, and nervous system malformations); (b) previous history (congenital anomalies of the kidney and urinary tract [CAKUT] in the family, low birth weight, and oligoamnios); (c) clinical signs (polyuria/nocturia, urinary tract infection, systemic arterial hypertension, failure to thrive, weak urinary stream, difficulty to start urination, distended bladder, non-monosymptomatic enuresis, urinary/urge incontinence, and bowel and bladder dysfunction); and (d) pre- and postnatal ultrasonographic alterations (increased anteroposterior diameter of the renal pelvis, mainly in the third trimester of pregnancy; single kidney; hydronephrosis associated with other abnormalities; and hydronephrosis with parenchymal involvement in the post-neonatal assessment).
Conclusion: The suggestions shown here can help the pediatrician to establish clinical hypotheses for the early diagnosis of developmental abnormalities of the genitourinary tract without resorting to expensive and invasive procedures.

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PALAVRAS-CHAVE
Malformação do trato urinário;
Doença renal crônica;
Diagnóstico precoce;
Criança

Sinais e sintomas das anormalidades do desenvolvimento do trato geniturinário

Resumo
Objetivo: As anormalidades do desenvolvimento do trato geniturinário são a principal causa de doença renal crônica (DRC) em crianças. O diagnóstico dessa doença no Brasil é formulado de maneira incompleta e tardia, o que resulta em aumento na morbi-mortalidade nessa faixa etária. O diagnóstico precoce dessa condição é prerrogativa dos pediatras generalistas e o objetivo desse trabalho foi revisar os sinais e sintomas clínicos associados às anormalidades do desenvolvimento do trato geniturinário.
Fontes dos dados: A partir da descrição de um caso clínico simbólico, realizamos uma revisão não sistemática da literatura médica.
Síntese dos dados: Os resultados sugerem que os seguintes dados devem ser utilizados como alerta para o diagnóstico precoce das crianças acometidas: a) anomalias do trato urinário compostas (anomalias cromossômicas, sequências de malformações – VACTERL e Prune-Belly, malformações musculoesqueléticas, do trato digestivo, cardíacas e do sistema nervoso), b) antecedentes (anomalias congênitas do rim e trato urinário (CAKUT) na família,baixo peso ao nascer e oligoâmnio), c) sinais clínicos (polaciúria/noctúria, infecção urinária, hipertensão arterial sistêmica, baixo ganho de peso, jato urinário fraco, dificuldade para iniciar a micção, bexigoma, enurese não monossintomática, urge/incontinência urinária, disfunção do intestino e da bexiga)e d) alterações ultrassonográficas ante e pós-natais (diâmetro ântero-posterior da pélvis renal aumentado principalmente no terceiro trimestre da gestação, rim único, hidronefrose associada a outras anomalias e hidronefrose com comprometimento de parênquima na avaliação pós-neonatal).
Conclusão: As sugestões apresentadas podem ajudar o pediatra a estabelecer hipóteses clínicas para o diagnóstico precoce das anormalidades do desenvolvimento do trato geniturinário sem utilização de metodologias caras e invasivas.

Clinical case

The patient, H.O.R.S., aged 12 years and 3 months, was born at term with no complications during childbirth. He was discharged 72 h after delivery in the mother's company, who was instructed to carry on the follow-up with a specialist, as the prenatal ultrasonography had shown the presence of bilateral hydronephrosis with anteroposterior renal pelvis diameter of approximately 12 mm. However, after birth, the parents were lost to follow-up with the nephrologist.

The consultations with the pediatrician were irregularly attended. At 8 years old, the child was referred for consultation with a hematologist and an endocrinologist due to stunting and anemia unresponsive to treatment with oral iron; these specialists did not find the cause for the alterations.

Only at the age of 10 years and 11 months was the child diagnosed with chronic kidney disease (CKD) secondary to bilateral ureteropelvic junction stenosis. At the time of diagnosis, he had already shown signs of chronic nephropathy with bilateral tapering of the renal parenchyma at the ultrasonographic assessment. The parents stated that at the age of 2 years the child already had daytime sphincter control, but they observed polyuria, nocturia, and excessive water consumption. There were no alterations in urinary stream or episodes of urinary tract infection.

At 11 years and 3 months, he underwent a pyeloplasty with double J catheter implantation on the right in order to prolong the conservative treatment and delay the onset of renal replacement therapy.

At the age of 12 years and 3 months, one year and four months after the diagnosis, he underwent preemptive kidney transplantation from a living donor (his mother).

Discussion

The case described above is factual and common, evidencing the disturbing problem of late diagnosis of CKD in children in Brazil. CKD is defined as the existence of any kidney injury, associated with varying degrees of glomerular filtration rate decrease, as shown in Table 1.[1] Stage 5 CKD, requiring renal replacement therapy (RRT; whether dialysis or transplantation), is a public health problem and there is evidence of the increasing incidence and prevalence of this condition.[2] In 2010, the number of individuals receiving RRT worldwide was estimated at 2.7 million, but it is also estimated that the similar number of patients who had the indication for RRT did not receive it due to lack of access to the treatment, especially for financial reasons.[3] It is further estimated that the proportion of CKD patients in stages 2 to 4 is much higher than that of CKD patients in stage 5.[4]

Fortunately, the proportion of children with CKD is much lower than that of adults, but CKD in children has devastating consequences on growth, weight, height, and intellectual development, resulting in increased morbidity