An Pediatr (Barc). 2015;xxx(xx):xxx–xxx

analesdepediatría

www.analesdepediatria.org

ORIGINAL ARTICLE

Unilateral renal agenesis. New arguments about the genetic relationship between kidney malformations and urolithiasis☆

V. Garcia Nieto[a,*], B. Huertes Díaz[b], J. Escribano Subias[c], M.T. Alarcón Alacio[d], J.D. Gonzalez Rodríguez[e], J.E. Cabrera Sevilla[e], C. Peralta Aros[f], M.I. Luis Yanes[a]

[a] *Pediatric Nephrology Section of the Hospital Universitario Nuestra Señora de Candelaria, Santa Cruz de Tenerife, Spain*
[b] *Hospital Universitario de Getafe, Madrid, Spain*
[c] *Hospital Universitari Sant Joan, Reus, Spain*
[d] *Hospital de Fuenlabrada, Madrid, Spain*
[e] *Hospital General Universitario Santa Lucía, Cartagena, Spain*
[f] *Hospital Uyapar, Puerto Ordaz, Venezuela*

Received 4 June 2015; accepted 7 September 2015

KEYWORDS
Hypercalciuria;
Hypocitraturia;
Urolithiasis;
Inheritance;
Unilateral renal agenesis

Abstract
Background: In few previous studies, it has been reported that hypercalciuria is associated with some types of congenital anomalies of the kidney and urinary tract (CAKUT), namely ureteropelvic junction obstruction, vesicoureteral reflux or simple renal cysts. In addition, one higher prevalence of hypercalciuria and/or urolithiasis has been described in their family members compared to the general population. This study was carried out to find out whether children with unilateral renal agenesis (URA) have these features previously described in other CAKUT types.
Methods: In a descriptive and multicenter study we studied the prevalence of hypercalciuria, hypocitraturia and urolithiasis in 67 children (43 males and 24 females) with URA and their families.
Results: The two metabolic anomalies that promote stone formation were observed in 26 children (38.8%), distributed as follows: hypercalciuria in 16, hypocitraturia in 9, and both hypercalciuria and hypocitraturia in 1. Eight children (11.9%) suffered renal colic during follow-up. Familial history of urolithiasis was found in 42/67 children (62.7%): in 12 of the first-degree relatives, in 15 of the second degree relatives and in 15 patients both in the first-degree as in their second degree relatives. In contrast, in historic control group, only in 28.1% of families at least one member had urolithiasis.

☆ Please cite this article as: Garcia Nieto V, Huertes Díaz B, Escribano Subias J, Alarcó Alacio MT, Gonzalez Rodríguez JD, Cabrera Sevilla JE, et al. Agenesia renal unilateral. Nuevos argumentos acerca de la relación genética entre la urolitiasis y las malformaciones renales. An Pediatr (Barc). 2015. http://dx.doi.org/10.1016/j.anpedi.2015.09.023

* Corresponding author.
E-mail address: vgarcianieto@gmail.com (V. Garcia Nieto).

Conclusion: Our results show that the prevalence of hypercalciuria and/or hypocitraturia is greater in pediatric patients with URA than in the general population. Likewise, the prevalence of urolithiasis in the families of these children is also higher than that in the general population.

PALABRAS CLAVE
Hipercalciuria;
Hipocitraturia;
Urolitiasis;
Herencia;
Agenesia renal unilateral

Agenesia renal unilateral. Nuevos argumentos acerca de la relación genética entre la urolitiasis y las malformaciones renales

Resumen
Antecedentes: En escasos trabajos previos, se ha comunicado que puede observarse la presencia de hipercalciuria en pacientes portadores de algunos tipos de CAKUT como estenosis pieloureteral, reflujo vesicoureteral o quistes renales simples. Además, se ha descrito una prevalencia mayor de hipercalciuria y/o urolitiasis en los miembros de las familias de esos niños con algunos tipos de CAKUT, en comparación con la población en general. El presente estudio se llevó a cabo para averiguar si los niños con agenesia renal unilateral (ARU) tienen las características descritas anteriormente en otros tipos de CAKUT.
Métodos: En un estudio descriptivo y multicéntrico se determinó la prevalencia de hipercalciuria, hipocitraturia y urolitiasis en 67 niños (43 hombres y 24 mujeres) con ARU y sus familias.
Resultados: En 26 niños (38,8%) se observaron las dos anomalías metabólicas que favorecen la formación de cálculos renales distribuidos de la siguiente manera: hipercalciuria en 16, hipocitraturia en 9 y tanto hipercalciuria como hipocitraturia en 1. Ocho niños (11,9%) padecieron un cólico renal durante el tiempo total de seguimiento. Una historia familiar de litiasis urinaria se encontró en 42/67 de los niños (62,7%): en familiares de primer grado en 12 de ellos, en familiares de segundo grado en 15 y en ambos grados de familiares en los otros 15. En contraste, en el grupo de control histórico, solamente en 28,1% de las familias, al menos, un miembro había tenido urolitiasis.
Conclusión: Nuestros resultados muestran que la prevalencia de la hipercalciuria y/o hipocitraturia en pacientes pediátricos con ARU es mayor que en la población general. Asimismo, la prevalencia de urolitiasis en las familias de estos niños es también mayor que en la población general.

Introduction

Association between urolithiasis and congenital anomalies of the kidney and urinary tract (CAKUT) has been described in children and adults since the 1920s.[1] The cause of susceptibility to lithiasis was believed to be either: urinary stasis,[2] reduced urinary flow and urinary tract infection in which *Proteus* was the most common microorganism.[3] Urinary stasis is generally assumed to play a major part in the pathogenesis of nephrolithiasis associated with distorted renal anatomy due to a delayed washout of crystals and risk of urinary infections. The reported frequency of genitourinary anomalies in children with urolithiasis is between 19.1%[4] and 29.8%.[5] However, the exact pathogenic relationship between urolithiasis and CAKUT remains unclear.

We reported a higher incidence of hypercalciuria in children with vesicoureteral reflux (VUR),[6] ureteropelvic junction obstruction[7] and simple renal cysts[8] in the general population, and a higher positive family history of urolithiasis.[6–8] This implies that children of parents with urolithiasis, and/or with a family history of urolithiasis, would have increased susceptibility to have a kidney development abnormality. These children also inherit hypercalciuria that could also favor formation of kidney stones.

The present study was conducted to find out if children with unilateral renal agenesis (URA) have a higher prevalence of hypercalciuria, hypocitraturia and/or a familiar history of urolithiasis.

Subjects and methods

Patients

This descriptive and multicenter study included 67 children (43 males, 24 females) with URA who were reviewed at Spanish Pediatric Nephrology Units of ''Hospital Nuestra Señora de Candelaria'' (Tenerife) ($n = 35$), ''Hospital Universitari Sant Joan de Reus'' (Tarragona) ($n = 16$), ''Hospital de Fuenlabrada'' (Madrid) ($n = 10$) and ''Hospital General Universitario Santa Lucía'' (Cartagena) ($n = 8$). The age at the time of the study was 10.0 ± 5.84 years (range: 0.16–21.5). The patients were controlled in each hospital from the