

Genetic approaches to human renal agenesis/hypoplasia and dysplasia

Simone Sanna-Cherchi · Gianluca Caridi ·
Patricia L. Weng · Francesco Scolari ·
Francesco Perfumo · Ali G. Gharavi ·
Gian Marco Ghiggeri

Published online: 3 July 2007
© IPNA 2007

Erratum to: *Pediatr Nephrol*
DOI 10.1007/s00467-007-0479-1

Table 2 shows some of the principal human malformation syndromes with kidney hypoplasia/dysplasia. The branchio-oto-renal syndrome is caused by mutations of EYA1, SIX1 or SIX5 genes and not SIX2. We apologize for the mistake in the article.

The online version of the original article can be found at <http://dx.doi.org/10.1007/s00467-007-0479-1>.

S. Sanna-Cherchi · P. L. Weng · A. G. Gharavi
Department of Medicine, Division of Nephrology,
Columbia University College of Physicians and Surgeons,
New York, NY, USA

S. Sanna-Cherchi
Department of Clinical Medicine,
Nephrology and Health Science, University of Parma,
Parma, Italy

G. Caridi · G. M. Ghiggeri (✉)
Laboratory of Pathophysiology of Uremia, Istituto G. Gaslini,
Largo G. Gaslini 5,
16148 Genoa, Italy
e-mail: labnefro@ospedale-gaslini.ge.it

P. L. Weng
Department of Pediatrics, Division of Nephrology,
Mount Sinai School of Medicine,
New York, NY, USA

F. Scolari
Division and Chair of Nephrology, Spedali Civili,
University of Brescia,
Brescia, Italy

F. Perfumo
Division of Nephrology, Istituto G. Gaslini,
Genoa, Italy

Table 2 List of human malformation syndromes with kidney hypoplasia/dysplasia (*MCDK* multicystic dysplastic kidney, *VUR* vesicoureteral reflux)

Gene	Human syndrome	Kidney phenotype	OMIM
JAG1, NOTCH2	Alagille syndrome	MCDK, kidney dysplasia, kidney mesangioliopidosis	#118450, #610205
BBS1-BBS11	Bardet-Biedl syndrome	Renal dysplasia and calyceal malformations	#209900
EYA1, SIX1, SIX5	Branchio-oto-renal syndrome	Renal agenesis/dysplasia	#113650
SOX9	Campomelic dysplasia	Diverse renal malformations	#114290
CHD7	CHARGE syndrome	Diverse urinary tract malformations	#214800
Del. 22q11	Di George syndrome	Renal agenesis, dysplasia, VUR	#188400
GATA3	Hypothyroidism, sensorial deafness, renal anomalies (HDR)	Renal agenesis, dysplasia, VUR	#146255
DNA repair	Fanconi anemia	Renal agenesis	#227650
FRAS1, FREM2	Fraser syndrome	Renal agenesis, dysplasia	#219000
KALL1, FGFR1	Kallman's syndrome	Renal agenesis, dysplasia	#308700, #147950
PAX2	Renal coloboma syndrome	Renal hypoplasia, MCDK, VUR	#120330
TCF2	Renal cysts and diabetes syndrome	Renal dysplasia, cysts	#137920
GPC3	Simpson-Golabi-Behmel syndrome	Renal dysplasia, cysts	#300209
DHCR7	Smith-Lemli-Opitz Syndrome	Renal dysplasia, cysts	#270400
SALL1	Townes-Brocks Syndrome	Renal dysplasia, lower urinary tract malformations	#107480
LMX1B	Nail-patella syndrome	Glomerulus malformation, renal agenesis	#161200
NIPBL	Cornelia de Lange syndrome	Renal dysplasia	#122470
CREBBP	Rubinstein-Taybi syndrome	Renal agenesis	#180849
WNT4	Rokitansky syndrome	Renal agenesis	#277000
PEX-family	Zellweger syndrome	Renal dysplasia, cysts	#214100
GLI3	Pallister-Hall syndrome	Renal agenesis, dysplasia	#146510
p57(KIP2)	Beckwith-Wiedemann syndrome	Renal dysplasia	#130650
SALL4	Okhiro syndrome	Renal ectopia with or without fusion, lower urinary tract malformations	#607323
TBX3	Ulnar-mammary syndrome	Renal agenesis	#181450