

## Poor phenotype-genotype association in a large series of patients with type III Bartter syndrome.

Alejandro García Castaño<sup>1</sup>, Gustavo Pérez de Nanclares<sup>1</sup>, Leire Madariaga<sup>2,3</sup>, Mireia Aguirre<sup>2</sup>, Álvaro Madrid<sup>4</sup>, Sara Chocrón<sup>4</sup>, Inmaculada Nadal<sup>5</sup>, Mercedes Navarro<sup>6</sup>, Elena Lucas<sup>7</sup>, Julia Fijo<sup>8</sup>, Mar Espino<sup>9</sup>, Zilac Espitaletta<sup>10</sup>, Víctor García Nieto<sup>11</sup>, David Barajas de Frutos<sup>12</sup>, Reyner Loza<sup>13</sup>, Guillem Pintos<sup>14</sup>, Luis Castaño<sup>1,3,15</sup>, RenalTube Group<sup>1,4,11,16</sup>, Gema Ariceta<sup>4</sup>.

1 BioCruces Institute, Ciberer, Cruces University Hospital, Bizkaia, Spain; 2 Pediatric Nephrology, Cruces University Hospital, Bizkaia, Spain; 3 Department of Pediatrics, School of Medicine and Odontology, University of Basque Country UPV/EHU, Bizkaia, Spain; 4 Pediatric Nephrology, Vall d'Hebron University Hospital, Universitat Autònoma, Barcelona, Spain; 5 Pediatric Nephrology, Virgen del Camino Hospital, Pamplona, Spain; 6 Pediatric Nephrology, La Paz University Hospital, Madrid, Spain; 7 Pediatrics, Manises Hospital, Valencia, Spain; 8 Pediatric Nephrology, Virgen del Rocío Hospital, Sevilla, Spain; 9 Pediatric Nephrology, Fundación Alcorcón University Hospital, Madrid, Spain; 10 San Ignacio University Hospital, Bogotá, Colombia; 11 Pediatric Nephrology, Nuestra Señora de Candelaria University Hospital, Tenerife, Canarias, Spain; 12 Pediatric Nephrology, Virgen de las Nieves Hospital, Granada, Spain; 13 Nephrology Unit, Cayetano Heredia University, Cayetano Heredia Hospital, Lima, Peru; 14 Germans Trias i Pujol University Hospital, Badalona, Spain; 15 Centro de Investigación Biomédica en Red de Diabetes y Enfermedades Metabólicas Asociadas (CIBERDEM), Instituto de Salud Carlos III, Madrid, Spain; 16 Pediatric Nephrology, Asturias Central University Hospital, Oviedo, Asturias, Spain.

**Corresponding author:** gariceta@vhebron.net

**S1 Table. Comparison of clinical and biochemical characteristics of different cohorts of patients with confirmed type III BS.**

	This study (n=30)		Simon <i>et al.</i> (n=17) [2]		Konrad <i>et al.</i> (n=36) [10]		Peters <i>et al.</i> (n=20) [14]		Bettinelli <i>et al.</i> (n=13) [11]		Lee <i>et al.</i> (n=23) [13]	
	n	%	n	%	n	%	n	%	n	%	n	%
<b>Polyhydramnios</b>	9/24	37	NA	NA	8/25	32	5/20	25	2/13	15	NA	±22
<b>Diagnosis ≤ 1 year</b>	16/30	53	12/17	70	22/34	65	11/20	55	10/13	77	20/23	87
<b>Hypokalemia*</b>	30/30	100	17/17	100	36/36	100	13/20	65	13/13	100	23/23	100
<b>Hyponatremia*</b>	17/29	59	NA	NA	NA	NA	NA	NA	7/13	54	NA	NA
<b>Hypochloremia*</b>	21/28	75	NA	NA	NA	NA	5/20	25	13/13	100	23/23	100
<b>Metabolic alkalosis*</b>	27/29	93	17/17	100	NA	100	4/20	20	13/13	100	23/23	100
<b>Magnesemia*</b>	6/26 (Low)	23	17/17 (Normal)	100	9/23 (Low)	39	NA (Low)	±50	3/13 (Low)	23	5/23 (Low)	22
<b>Hyperreninemia*</b>	20/20	100	6/6	100	NA	100	NA	NA	13/13	100	23/23	100
<b>Hyperaldosteronism*</b>	19/22	86	6/6	100	NA	100	NA	NA	13/13	100	23/23	100
<b>Calciuria*</b>	6/26 (Low) 8/26 (High)	23 31	11/17 ( High )	65	2/25 (Low) / 7/25 (High)	8/ 28	10/20 (High)	50	2/13 (Low) / 1/13 (High)	15/ 8	13/23 (High)	56
<b>Nephrocalcinosis</b>	6/30	20	0/17	0	4/25	16	2/20	10	0/13	0	4/23	17

Abbreviations: n, number of patients; NA, not available. \*According to our laboratory reference values.