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

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S2 Table. Clinical and biological characteristics of type III BS patients

Patient	*Sex*	*Age at Dx (years)*	*Weight (SDS)*	*Height (SDS)*	*Gest Age (weeks)*	*PH*	*pH blood*	*P. HCO_3^-* (mEq/L)	*P. Na* (mEq/L)	*P. K* (mEq/L)	*P. Cl* (mEq/L)	*P. Creat* (mg/dl)	*P. Mg* (mg/dl)	*P. Ca* (mg/dl)	*P. renin activity* (ng/ml/h)	*P. aldost* (pg/ml)	*FE Na (%)*	*FE K (%)*	*FE Cl (%)*	*TTKG*	*U Ca/Cr (mg/mg)*	*U Ca (mg/kg/d)*	*NC*
p.[Ala204Thr];[Ala204Thr]																							
*SOR0003	F	6.8	-1.29	-0.11	36	+	7.53	35	134	1.4	100	0.67	0.9	NA	20.9	945	1.2	49.9	2.9	NA	0.43	11	-
*SOR0005	F	3	-1.63	-0.97	NA	+	7.5	27.9	130	2	85	0.5	2.2	9.4	60.5	484	2.8	50.5	4	10.8	0.5	9.7	+
*SOR0008	F	2	-4.88	-3.16	40	-	7.47	28	134	2	88	0.68	1.5	9.7	NA	NA	1.5	62.3	2.12	NA	0.31	5.9	+
*SOR0009	F	2	-4.50	-3.48	36	+	7.31	20	143	1.7	100	0.75	NA	10	13.6	NA	1.33	76.8	2.37	NA	0.33	9.2	-
*SOR0023	M	1.8	-3.18	-3.66	NA	-	7.47	24.3	139	3.3	85	0.36	1.9	5.2	53.4	267	1.2	27.5	2.09	NA	0.73	NA	-
*SOR0025	F	2.3	-3.73	-1.96	40	-	7.57	26.7	132	2.3	95	0.46	1.5	10.7	215	5610	1.2	34.2	2	NA	0.23	NA	-
*SOR0026	F	0.7	-3.73	-1.92	40	+	7.42	36.9	135	3.3	90	0.2	2.5	10.6	13.6	1654	0.14	7.2	0.19	10.8	0.2	2.2	-
*SOR0039	F	1.4	-4.05	-2.34	41	-	7.49	28	138	1.6	93	0.3	2.8	10.1	86.6	1281	2.3	54	3.5	NA	0.2	NA	-
*SOR0045	M	0.7	2.02	-2.3	NA	+	7.48	39.9	133	2.3	79	0.45	2.3	NA	NA	670	0.18	13.4	0.31	NA	NA	NA	-
*SOR0047	M	0.7	-3.46	-1.90	NA	-	7.65	31	137	2.5	83	0.28	3.1	10.8	NA	760	0.15	17.7	0.28	12.1	0.15	NA	-
*SOR0048	F	2	-2.33	-1.23	42	+	7.38	27	126	2.7	82	0.16	2.9	10.5	77.4	282	0.6	14.2	0.6	8.2	0.23	NA	-
*SOR0050	M	0.7	-5.26	-5.35	41	-	7.5	31.4	140	2.2	99	0.3	2.4	8.9	NA	NA	0.25	20.8	NA	16.9	0.03	NA	-
*SOR0051	M	25	NA	NA	NA	NA	7.34	23.6	134	2.3	98	2.68	2.6	9.6	NA	NA	NA	NA	NA	NA	NA	NA	+
*SOR0062	M	3	-2.67	-1.89	40	-	NA	30.7	130	1.9	89	0.33	2.4	9.7	80.4	1608	0.9	42	1.4	NA	1.1	17.5	+
*SOR0073	F	0.7	-3.19	-0.98	40	-	7.47	29.9	139	3.3	97	0.4	2.2	10.4	8.2	1370	0.001	25	0.3	14.5	0.38	NA	-
SOR0084	M	0.5	-2	-0.82	38	+	7.46	27.1	137	3.7	83	0.4	2.4	10.8	40	1900	0.31	34.8	0.62	NA	0.12	NA	-
MEAN		**3.1**	**-2.93**	**-2.23**	**39.5**	**7/15**	**7.47**	**29.2**	**135**	**2.4**	**90**	**0.56**	**2.2**	**9.7**	**60.9**	**1403**	**0.94**	**35.35**	**1.62**	**12.2**	**0.35**	**9.2**	**4/16**
SD		**6.1**	**1.79**	**1.34**	**2**	**-**	**0.09**	**5**	**4.4**	**0.7**	**7**	**0.59**	**0.6**	**1.4**	**58.6**	**1434**	**0.83**	**20.17**	**1.28**	**3.1**	**0.28**	**5.1**	**-**
p.[Ala204Thr];[0]																							
*SOR0011	F	11	NA	-2.33	NA	-	7.45	29.4	138	2	93	0.5	1.7	10	31.1	1015	0.81	31.4	1.11	NA	NA	2.2	-
*SOR0024-1	F	17	1.00	1.83	40	-	NA	33	132	2.6	134	0.4	1.3	9.8	195	114	NA	NA	NA	NA	NA	NA	-
*SOR0024-2	F	0.9	-4.97	-1.2	NA	NA	NA	NA	NA	1.8	NA	0.5	NA	NA	NA	NA	NA	12	NA	NA	NA	0.5	-
MEAN		**9.6**	**-1.98**	**-0.6**	**40**	**0/2**	**7.45**	**31.2**	**135**	**2.1**	**113**	**0.47**	**1.5**	**9.9**	**113.1**	**564**	**0.81**	**21.7**	**1.11**	**-**	**-**	**1.4**	**0/3**
SD		**8.1**	**4.23**	**2.2**	**-**	**-**	**-**	**2.5**	**4.2**	**0.4**	**29**	**0.06**	**0.3**	**0.1**	**115.9**	**637**	**-**	**13.7**	**-**	**-**	**-**	**1.2**	**-**
p.[Ala204Thr];[Glu442Gly]																							
*SOR0054-1	M	0.3	-2.36	-0.27	NA	-	7.51	32.6	131	2.3	80	0.51	2.2	10.6	NA	NA	0.29	42.1	0.6	NA	0	NA	-
*SOR0054-2	F	0.5	-0.77	0.58	NA	-	7.4	26.5	145	2.2	81	0.5	NA	NA	60.6	220	0.22	30.9	NA	NA	0.53	NA	-
*SOR0054-3	F	0.3	-2.23	-2.06	NA	-	7.46	28.4	134	2.9	86	0.66	2.2	NA	21	185	0.47	25	1.19	NA	0	NA	-
*SOR0057	F	0.7	-6.19	-3.49	36	+	7.62	35.3	134	2.3	87	0.29	2.8	10.7	39	473	0.14	24.3	0.49	16.6	0.43	NA	+
MEAN		**0.5**	**-2.89**	**-1.31**	**36**	**1/4**	**7.5**	**30.7**	**136**	**2.4**	**83**	**0.49**	**2.4**	**10.7**	**40.2**	**293**	**0.28**	**30.6**	**0.76**	**16.6**	**0.24**	**-**	**1/4**
SD		**0.2**	**2.32**	**1.82**	**-**	**-**	**0.09**	**4**	**6.2**	**0.3**	**3.5**	**0.15**	**0.4**	**0.07**	**19.8**	**157**	**0.14**	**8.2**	**0.38**	**-**	**0.28**	**-**	**-**
p.[Ala204Thr];[Val170Met]																							
*SOR0064	M	0.8	-2.57	-1.69	41	+	7.55	43	134	3.1	81	0.33	2.2	9.4	19.9	1584	0.17	8.2	0.23	8	0.54	1.5	-

Patient	*Sex*	*Age at Dx (years)*	*Weight (SDS)*	*Height (SDS)*	*Gest Age (weeks)*	*PH*	*pH blood*	*P. HCO_3^-* (mEq/L)	*P. Na* (mEq/L)	*P. K* (mEq/L)	*P. Cl* (mEq/L)	*P. Creat* (mg/dl)	*P. Mg* (mg/dl)	*P. Ca* (mg/dl)	*P. renin activity* (ng/ml/h)	*P. aldost* (pg/ml)	*FE Na (%)*	*FE K (%)*	*FE Cl (%)*	*TTKG*	*U Ca/Cr (mg/mg)*	*U Ca (mg/kg/d)*	*NC*
p.[Ile398_Thr401del];[0]																							
*SOR0063	F	0.4	-2.91	-1.05	41	-	7.59	33.5	133	2.7	94	0.33	2.3	11.8	30	384	0.26	35.2	1.3	NA	0.12	NA	-
p.[Ala210Val];[?]																							
*SOR0076	F	1	1.53	-1.61	NA	-	7.46	28	138	2.7	NA	0.6	NA	NA	50	480	NA	NA	NA	NA	NA	NA	-
p.[(Ser343Alafs*6)];[Glu442Gly]																							
SOR0081	M	4	-2	-1	NA	NA	7.45	NA	129	2.3	86	0.27	2.2	10.4	NA	NA	0.24	23	NA	NA	0.07	NA	+
p.[Arg595*];[Arg595*]																							
SOR0080	M	11	-0.98	-5	40	NA	7.5	29.8	135	2.1	85	0.89	2.1	10.7	NA	33	1.39	38.4	1.05	13.9	0.04	0.8	-
p.[(Leu252fs)];[(Leu252fs)]																							
SOR0090	M	0.5	-5	-2	39	NA	7.74	60	123	1.6	89	0.3	1.4	9.2	19.7	30	3.75	159	15	NA	3	NA	-
p.[0];[0]																							
SOR0097	F	0.1	-1	1	41	NA	7.45	30.9	121	3.4	67	0.35	1.4	11.6	NA	NA	NA	157	10.26	13	4.4	7.6	-

Abbreviations: SD, standard deviation; Dx, diagnostic; NA, not available; SDS, standard deviation score in comparison with an age- and sex-matched reference population; PH, polyhydramnios; P, plasmatic; HCO3-, bicarbonate; Creat, creatinine; Aldost, aldosterone; FE, fractional excretion; TTKG, transtubular potassium gradient; U, urinary; Ca/Cr, calcium/creatinine ratio; NC, nephrocalcinosis.

*Patients included in our previous manuscript [9].