

Supplementary Table

Supplementary Table S1 Clinical and biochemical characteristics of 95 GS patients (non-significant)

Parameter	All	Younger Group (≤16 years)	Older Group (>16 years)	P value	Male	Female	P value
Number	95	49	46		51	44	
Fatigue (n, %)	60 (63.2%)	30 (61.2%)	30 (65.2%)	0.832	32 (62.7%)	28 (63.6%)	1.000
Cramps (n, %)	11 (11.6%)	6 (12.2%)	5 (10.9%)	1.000	5 (9.8%)	6 (13.6%)	0.750
Muscle weakness (n, %)	9 (9.5%)	2 (4.1%)	7 (15.2%)	0.085	5 (9.8%)	4 (9.1%)	1.000
Abdomina pain (n, %)	6 (6.3%)	5 (10.2%)	1 (2.2%)	0.205	3 (5.9%)	3 (6.8%)	1.000
Dizziness (n, %)	5 (5.3%)	1 (2.0%)	4 (8.7%)	0.195	2 (3.9%)	3 (6.8%)	0.660
Myalgia (n, %)	4 (4.2%)	1 (2.0%)	3 (6.5%)	0.351	2 (3.9%)	2 (4.5%)	1.000
Diarrhaea (n, %)	3 (3.2%)	1 (2.0%)	2 (4.3%)	0.609	2 (3.9%)	1 (2.3%)	1.000
Nocturia (n, %)	2 (2.1%)	1 (2.0%)	1 (2.2%)	1.000	0 (0.0%)	2 (4.5%)	0.212
Salt craving (n, %)	2 (2.1%)	2 (4.1%)	0 (0.0%)	0.495	1 (2.0%)	1 (2.3%)	1.000
Polyuria (n, %)	2 (2.1%)	1 (2.0%)	1 (2.2%)	1.000	2 (3.9%)	0 (0.0%)	0.497
Thirst, polydipsia (n, %)	2 (2.1%)	2 (4.1%)	0 (0.0%)	0.495	1 (2.0%)	1 (2.3%)	1.000
Paralysis (n, %)	1 (1.1%)	0 (0.0%)	1 (2.2%)	0.484	1 (2.0%)	0 (0.0%)	1.000
Tinnitus (n, %)	1 (1.1%)	0 (0.0%)	1 (2.2%)	0.484	1 (2.0%)	0 (0.0%)	1.000
Foam urine (n, %)	1 (1.1%)	0 (0.0%)	1 (2.2%)	0.484	1 (2.0%)	0 (0.0%)	1.000
Constipation (n, %)	1 (1.1%)	0 (0.0%)	1 (2.2%)	0.484	0 (0.0%)	1 (2.3%)	0.463
Hypokalemia (n, %)	91 (95.8%)	46 (93.9%)	45 (97.8%)	0.618	49 (96.1%)	42 (95.5%)	1.000
Hypomagnesemia (n, %)	64 (69.6%)	30 (62.5%)	34 (77.3%)	0.174	35 (71.4%)	29 (67.4%)	0.821
Serum sodium level (mmol/L)	138.06±3.36	137.69±3.21	138.47±3.50	0.266	138.17±3.79	137.93±2.79	0.734
Serum chloride level (mmol/L)	97.00 (94.00,100.00)	96.50±3.99	96.45±4.92	0.965	96.60 (93.98,99.21)	97.00±4.44	0.424

N (%) represents categorical variables, compared using Fisher's exact test; Mean (SD) or median (IQR25, IQR75) represents continuous variables, respectively compared using Student's t-test or Mann-Whitney U test; P<0.05 was considered statistically significant. Normal reference value of serum potassium 3.5-5.5 mmol/L; Normal reference value of serum magnesium 0.7-1.0 mmol/L; Normal reference value of serum sodium 135-145 mmol/L; Normal reference value of serum chloride 96-106 mmol/L.

Supplementary Table S2 The list of variants in patients

Patient number	Variant 1	Variant 2	Variant 3
1	c.1077C>G		
2	c.1456G>A		
3	c.1946C>T		
4	c.179C>T		
5	c.1946C>T		
6	c.2761C>T		
7	c.179C>T		
8	c.1456G>A		
9	c.1456G>A		
10	c.1456G>A		
11	c.1195C>T		
12	c.179C>T		
13	c.506-1G>A		
14	c.2661_2747del		
15	c.506_852del		
16	c.965-1_976delinsACCGAAAATTTT		
17	c.2129C>A	c.2129C>A	
18	c.1456G>A	c.602-16G>A	
19	c.1049C>T	c.2548+253C>T	
20	c.506-1G>A	c.179C>T	
21	c.179C>T	c.742-2A>G	
22	c.1924C>T	c.2660+1G>A	
23	c.506-1G>A	c.179C>T	
24	c.506-1G>A	c.1456G>A	
25	c.2129C>A	c.2582G>A	
26	c.1456G>A	c.1452C>A	
27	c.1930C>T	c.836T>G	
28	c.1670-191C>T	c.2129C>A	
29	c.179C>T	c.179C>T	
30	c.179C>T	c.179C>T	
31	c.1964G>A	c.1964G>A	
32	c.1456G>A	c.1456G>A	
33	c.179C>T	c.179C>T	
34	c.1195C>T	c.1195C>T	
35	c.179C>T	c.179C>T	
36	c.1163A>G	c.1163A>G	
37	c.2936T>C	c.2936T>C	
38	c.179C>T	c.179C>T	
39	c.1077C>G	c.1567G>A	
40	c.488C>T	c.800C>T	
41	c.1456G>A	c.473G>A	
42	c.179C>T	c.1964G>T	
43	c.1456G>A	c.626G>A	
44	c.179C>T	c.1964G>T	
45	c.1964G>A	c.1456G>A	
46	c.1456G>A	c.1315G>A	

47	c.1077C>G	c.1195C>T	
48	c.1108G>C	c.1925G>A	
49	c.593G>T	c.1322G>T	
50	c.179C>T	c.1456G>A	
51	c.2099T>C	c.1077C>G	
52	c.1022C>T	c.1077C>G	
53	c.1108G>C	c.1925G>A	
54	c.179C>T	c.1084G>A	
55	c.1844C>T	c.2029G>A	
56	c.179C>T	c.1919A>G	
57	c.1077C>G	c.1322G>T	
58	c.179C>T	c.1698C>A	
59	c.911C>T	c.1077C>G	
60	c.179C>T	c.1124C>A	
61	c.1456G>A	c.2963T>C	
62	c.1456G>A	c.1195C>T	
63	c.179C>T	c.1294T>G	
64	c.179C>T	c.2717G>A	
65	c.179C>T	c.1456G>A	
66	c.2548+253C>T	c.805_806ins18	
67	c.1718T>G	c.2969insGCT	
68	c.1718T>G	c.2969insGCT	
69	c.2660+2dupT	c.2186G>T	
70	c.965-1_976delinsACCGAAAATTTT	c.625C>T	
71	c.965-1_976delinsACCGAAAATTTT	c.1850A>G	
72	c.965-1_976delinsACCGAAAATTTT	c.2717G>A	
73	c.965-1_976delinsACCGAAAATTTT	c.1268G>T	
74	c.965-1_976delinsACCGAAAATTTT	c.602-16G>A	
75	c.486_490delinsA	c.536T>A	
76	c.486_490delinsA	c.1456G>A	
77	c.179C>T	c.486_490delinsA	
78	c.1456G>A	c.1625_1627del	
79	c.1077C>G	c.2369_2883del	
80	c.1456G>A	c.2875_2876del	
81	c.1567G>A	c.2420_2446del	
82	c.179C>T	c.346_353del	
83	c.1322 G>T	c.2875_2876del	
84	c.381delG	c.863T>G	
85	c.179C>T	c.1384del	
86	c.1294T>G	c.346_353del	
87	c.342_343dupTG	c.911C>T	
88	c.2492del	c.2369_2883del	
89	c.784_785insTCATTGGCGTGGTCTCGG	c.965-1_976delinsACCGAAAATTTT	
90	c.965-1_976delinsACCGAAAATTTT	c.965-1_976delinsACCGAAAATTTT	
91	c.953T>G	c.1196G>A	c.1664C>T
92	c.1456G>A	c.1964G>A	c.2738G>A

Supplementary Table S3 The correlation analysis between phenotype and genotype

	Heterozygous	Compound heterozygous	Homozygous	P
Lowest serum potassium level (mmol/L)	2.87±0.47	2.78±0.41	2.66±0.40	0.438
Lowest serum magnesium level (mmol/L)	0.56±0.09	0.65±0.17	0.51±0.07	0.004
Serum sodium level (mmol/L)	138.07±3.98	138.17±3.17	137.39±3.09	0.771
Serum chloride level (mmol/L)	94.71±4.80	97.05 (94.85, 100.00)	95.83±6.78	0.150

Supplementary Table S4 The typical clinical indicators of 19 patients without biallelic SLC12A3 variants

Patient number	Age at diagnosis (year)	Blood pressure (mmHg)	Serum potassium (mmol/L)	Serum magnesium (mmol/L)	Serum creatinine (μmol/L)	Urinary potassium (mmol/L)	Urinary chloride (mmol/L)	Urinary calcium (mmol/L)	Urinary magnesium (mmol/L)	Urinary creatinine (μmol/L)	pH	HCO ₃ ⁻ (mmol/L)
1	16	106/61	2.97	0.42	55.00	25.22	86.30	0.15	2.63	5011.00	7.47	29.6
2	22	94/60	1.82	0.74	82.00	68.52	99.60	0.04	3.14	6352.00	7.46	29.3
3	52	111/77	2.50	0.76	52.00	49.78	96.00	0.09	3.52	5846.78	7.47	29.9
4	6	118/81	3.83	0.54	67.00	27.10	98.60	0.07	2.16	5552.00	7.46	28.0
5	7	101/59	3.36	0.47	76.00	28.53	93.60	0.08	2.57	5432.00	7.50	30.1
6	15	115/72	2.49	0.56	49.00	79.53	99.50	0.26	5.92	9940.73	7.45	28.3
7	17	113/69	2.30	0.56	68.00	88.43	87.40	0.28	3.92	8690.70	7.50	30.0
8	18	120/81	2.70	0.45	59.00	52.95	90.00	0.18	2.35	5323.00	7.46	27.4
9	19	127/82	3.10	0.60	71.00	43.89	90.30	0.27	1.55	6443.00	7.47	29.4
10	24	133/87	2.60	0.59	71.00	50.78	95.80	0.09	2.82	5346.78	7.47	28.6
11	24	113/79	2.40	0.51	83.00	51.93	99.60	0.07	2.16	6352.00	7.50	29.0
12	29	106/57	2.75	0.68	96.00	49.21	90.40	0.16	1.55	5443.00	7.46	29.0
13	30	106/61	2.75	0.68	103.52	48.32	88.50	0.45	4.92	10940.00	7.45	29.8
14	30	110/70	2.70	0.60	63.86	68.53	99.50	0.57	5.92	14930.74	7.46	28.2
15	52	128/75	3.10	0.47	67.00	43.91	90.40	0.05	1.55	5443.00	7.48	29.2
16	4	95/63	2.32	0.52	39.00	53.78	96.00	0.08	3.92	5245.82	7.45	28.0
17	2	90/60	3.74	0.65	36.60	30.78	95.80	0.09	4.06	5336.78	7.45	28.5
18	5	96/65	3.09	0.65	80.20	41.53	99.60	0.06	3.46	6352.00	7.46	29.7
19	10	113/80	2.70	0.37	58.00	52.91	90.40	0.10	2.55	5443.00	7.47	28.5