

Patient*	Clinical characteristics											Laboratory characteristics										
	Duration of illness before admission	Fever	Vomiting	Diarrhea	Cough / Sneezing	Seizures	Altered mental status	Dehydration	Tachypnea	Hypotension	Mechanical ventilation	pH (mmol/L)	HCO ₃ (mmol/L)	BE (mmol/L)	Anion gap (mmol/L)	Blood glucose (mmol/L)	Serum ammonia (μmol/L)	WBC (×10 ⁹ /μL)	Electrolyte disorders	Characteristic profiles of urinary organic acids/blood acylcarnitines		
26 (GK104)	2 d	+	+	+	—	—	+	+	+	—	—	7.15	2	−25	28	5.1	156	22.5	—	+/+		
27 (GK104b)	No episodes																					+/+
28	1 d	+	+	+	—	+	+	+	+	—	+	7.05	2	−28	28	4.9	NA	NA	—	+/+		
29 (GK105)	4 d	+	+	+	—	—	+	+	+	—	+	7.13	4	−25	26	16.9	108	23.6	—	+/+		
30	1 d	+	+	—	+	—	+	+	+	—	+	6.84	3.6	−30	26	5.2	54	NA	—	+/+		
31	3 d	+	—	—	+	—	+	+	+	—	+	7.1	4.5	−21	26	4.5	NA	18.3	hypokalemia	+/+		
32 (GK117)	2 d	+	—	—	+	+	+	+	+	+	+	6.93	3.1	−29	27	5.5	152	46	—	+/+		
33	4 d	+	+	—	+	—	+	+	+	—	+	7.12	10.7	−17	20	7.84	56	48.5	hypokalemia	+/+		
34	3 d	+	+	+	—	—	+	+	+	—	—	7.02	2	−29	28	5.1	145	NA	NA	+/+		
35 (GK115)	2 d	+	+	—	—	—	+	+	+	—	—	7.2	5	−18	25	4.7	NA	NA	NA	+/+		
36 (GK116)	3 d	+	+	+	—	—	+	+	+	+	+	7.01	3	−30	27	5.8	126	19.8	—	+/+		
37 (GK118)	2 d	+	+	+	—	—	+	+	+	—	—	7.12	5	−22	25	6.8	NA	NA	—	+/+		
38 (GK118s)	2 d	+	+	+	—	—	+	+	+	—	—	7.1	3	−25	27	6	65	15.1	—	—/— [#]		
39 (GK120)	3 d	+	—	—	+	—	+	+	+	—	+	7.09	3	−30	27	4	120	31.1	—	+/+		
40 (GK119)	1 d	+	+	—	—	—	+	+	+	—	—	7.26	6	−20	24	9.4	62	26	—	+/NA		
41 (GK121)	3 d	—	+	+	—	—	+	+	+	—	—	7.2	5	−23	25	2.9	55	14.2	—	+/NA		

* GK number refers to an internal identifier for patients with beta-ketothiolase deficiency whose *ACAT1* mutations were identified at Gifu University (Gifu, Japan). Sib pairs are denoted by b (brother) and s (sister) following patient codes.

[#] Both analyzed under stable conditions.

Abbreviations: d, day; NA, not applicable;