Supplementary file 2 Demographic features, genotype, and outcome of 41 Vietnamese patients with beta-ketothiolase deficiency.

Patient*	Age at diagnosis / Onset	Sex	Family history	*ACAT1* mutations#		Outcome						
				Maternal	Paternal	Survival	Follow-up duration	Present age	Recurrent episodes	DQ	Height / Weight	Brain imaging abnormalities
1 (GK70)	34m/34m	M	—	c.163_167delinsAA (Phe55_Leu56delinsLys)	c.622C>T (p.Arg208*)	D (5.5y)	2.5y		—	60	NA	+
2 (GK76)	12m/12m	M	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	10y	11y	1 (15m)	90	+0.5/-1.5 SD	—
3 (GK72)	12m/12m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	10y	11y	1 (15m)	90	+2/+1.5 SD	—
4 (GK73)	11m/11m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	10y	11y	—	95	-1/-1.5 SD	—
5 (GK74)	18m/18m	M	—	c.622C>T (p.Arg208*)	c.1006-1G>C (p.Val336fs)	D (2.5y)	1y		1 (25m)	NA	NA	NA
6 (GK75)	11m/11m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	8y	9y	1 (15m)	80	median (both)	—
7 (GK75b)	15m/6m	M	+	NA	NA	D (2y)	9m		1 (2y)	NA	NA	NA
8 (GK79)	12m/12m	F	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	9y	10y	2 (12,36m)	100	+1/+1 SD	—
9 (GK79b)	3d/6m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	4y	4y	—	90	-2/-1 SD	—
10 (GK80)	7m/7m	F	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	7.5y	8y	—	90	-2/-2 SD	—
11	17m/17m	F	—	NA	NA	NA	NA	NA	NA	NA	NA	NA
12	11m/11m	M	—	NA	NA	NA	NA	NA	NA	NA	NA	NA
13 (GK86)	10m/10m	F	—	c.1006-1G>C (p.Val336fs)	c.1006-1G>C (p.Val336fs)	S	6y	7y	5 (11,15,32, 48,53m)	90	median/+1.5 SD	—
14 (GK87)	11m/11m	F	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	6y	7y	3 (18,28,36m)	80	-0.5 SD/median	—
15 (GK89)	12m/12m	F	—	c.1006-1G>C (p.Val336fs)	c.1006-1G>C (p.Val336fs)	S	6y	7y	1 (12m)	90	-1.5/-0.5 SD	—
16 (GK90)	7m/7m	M	—	c.622C>T (p.Arg208*)	c.1229C>T (p.Ala410Val)	S	5.5y	6y	2 (9,11m)	90	-1.8/-1 SD	—
17 (GK91)	9m/9m	M	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	5y	5.5y	1 (15m)	90	-2/-1 SD	—
18 (GK93)	17m/17m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	5y	6.5y	1 (22m)	95	-1.5/-1.5 SD	—
19 (GK93b)	22m/22m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	1.2y	3y	—	90	-2/+1 SD	—
20 (GK100)	9m/9m	F	—	g.20623_29833delins GTAA (exons 6-11 del)	c.1006-1G>C (p.Val336fs)	S	4.5y	5.1y	1 (10m)	95	-2/-1.5 SD	—
21 (GK101)	14m/14m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	4y	5y	—	55	-2/-1.5 SD	+
22 (GK101b)	5y/10m	M	+	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	4y	9y	—	60	NA/median	+

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				Maternal	Paternal	Survival	Follow-up duration	Present age	Recurrent episodes	DQ	Height / Weight	Brain imaging abnormalities
23 (GK102)	20m/20m	M	—	c.851G>A (p.Ser284Asn)	c.622C>T (p.Arg208*)	S	4y	5.5y	1 (2y)	80	-0.5/-0.5 SD	—
24 (GK103)	11m/11m	M	+	c.1006-1G>C (p.Val336fs)	c.622C>T (p.Arg208*)	S	3.5y	4.5	1 (11m)	90	-2/-2 SD	—
25 (GK103b)	4.5y/—	M	+	NA	NA	S	3.5y	8y	—	95	median/-0.5 SD	NA
26 (GK104)	16m/16m	F	+	c.622C>T (p.Arg208*)	c.1032dupA (p.Glu345fs)	S	3.5y	5y	—	100	-5/+0.5 SD	—
27 (GK104b)	25d/—	M	+	c.622C>T (p.Arg208*)	c.1032dupA (p.Glu345fs)	S	3y	3y	—	95	-2/-1.5 SD	NA
28	24/24m	F	—	NA	NA	S	3y	5y	—	90	-2/-1 SD	—
29 (GK105)	11m/11m	M	—	c.622C>T (p.Arg208*)	c.1006-1G>C (p.Val336fs)	S	2.5y	3.5y	—	95	-2/-2 SD	—
30	18m/18m	F	—	NA	NA	S	2.5y	4y	—	85	NA	NA
31	9m/7m	F	—	NA	NA	S	2y	3y	—	90	NA	NA
32 (GK117)	8m/8m	F	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	D (8m)			—	NA	NA	NA
33	8m/8m	M	—	NA	NA	D (8m)			—	NA	NA	+
34	7m/7m	F	—	NA	NA	S	2.5y	3y	—	92	-2/-1 SD	—
35 (GK115)	39m/24m	F	—	c.622C>T (p.Arg208*)	c.1006-1G>C (p.Val336fs)	S	2y	5y	2 (24,28m)	100	-1.8/-1.5 SD	—
36 (GK116)	15m/15m	M	—	c.622C>T (p.Arg208*)	c.1006-1G>C (p.Val336fs)	S	1.5y	3y	—	95	median/-0.5 SD	—
37 (GK118)	36m/36m	F	+	c.1A>G	c.1A>G	S	2y	5y	—	95	-1.8/-1 SD	—
38 (GK118s)	18m/12m	F	+	c.1A>G	c.1A>G	S	1y	2y	—	100	-1.7/-0.2 SD	NA
39 (GK120)	14m/14m	F	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	2m	1.5y	—	100	median/+0.5 SD	NA
40 (GK119)	23m/23m	M	—	c.1006-1G>C (p.Val336fs)	c.1006-1G>C (p.Val336fs)	S	7m	2.5y	—	95	-1/-1.8 SD	NA
41 (GK121)	6m/6m	F	—	c.622C>T (p.Arg208*)	c.622C>T (p.Arg208*)	S	6m	1y	—	95	median/-1.8 SD	NA

*All patients were born to nonconsanguineous parents of Kinh ancestry. 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.

Mutations are described according to the latest HGVS recommendations (version 15.11) using *ACAT1* NCBI Reference Sequences: **NM_000019.3** and **NG_009888.1**.

Abbreviations: D, died; DQ, developmental quotient; d, day; F, Female; M, Male; m, month; NA, not applicable; S, survived; SD, standard deviation; y, year.