Takanosu M, Kagawa Y. Severe osteogenesis imperfecta caused by *CREB3L1* mutation in a cat

Supplemental Table 1. Blood examination of a kitten with severe osteogenesis imperfecta.

Complete blood count	Unit	Patient	RI
Red blood cell count	$\times10^{12}$/L	7.9	6.5–12.2
Hematocrit	L/L	0.33	0.30–0.52
Hemoglobin	g/L	115	98–162
Mean corpuscular volume	fL	42	36–53
Mean corpuscular hemoglobin	pg	14.5	11.8–17.3
Mean corpuscular hemoglobin concentration	g/L	344	281–358
Reticulocyte count	$\times10^9$/L	24	3–50
White blood cell count	$\times10^9$/L	14.5	2.8–17.0
Neutrophil, segmented	$\times10^9$/L	7.4	1.5–10.3
Lymphocyte	$\times10^9$/L	5.2	0.9–6.9
Monocyte	$\times10^9$/L	0.6	0.05–0.7
Eosinophil	$\times10^9$/L	1.2	0.2–1.6
Basophil	$\times10^9$/L	0.1	0.0–0.3
Platelet	$\times10^9$/L	310	151–600
Chemistry			
Glucose	mmol/L	7.3	4.3–8.5
Creatinine	μmol/L	18	53–141
Urea	mmol/L	8.2	5.7–11.8
Phosphorus	mmol/L	2.49	1.45–3.36
Calcium, total	mmol/L	2.63	1.98–2.83
Na	mmol/L	151	150–165
K	mmol/L	5.1	3.7–5.9
Cl	mmol/L	117	115–126
Protein, total	g/L	67	52–82
Albumin	g/L	27	22–39
Globulin	g/L	40	28–48
Alanine aminotransferase	μkat/L	0.77	0.20–1.92
Alkaline phosphatase	μkat/L	1.92	0.23–3.21
γ-glutamyl transferase	μkat/L	0.12	0–0.12

Bilirubin, total	µmol/L	5.13	0.0–15.4
Cholesterol, total	mmol/L	4.56	1.61–4.95
Ammonia	µmol/L	32.1	16.4–55.7
Intact parathyroid hormone	ng/L	7.1	8.0–25.0
Ionized calcium	mmol/L	1.23	1.22–1.50

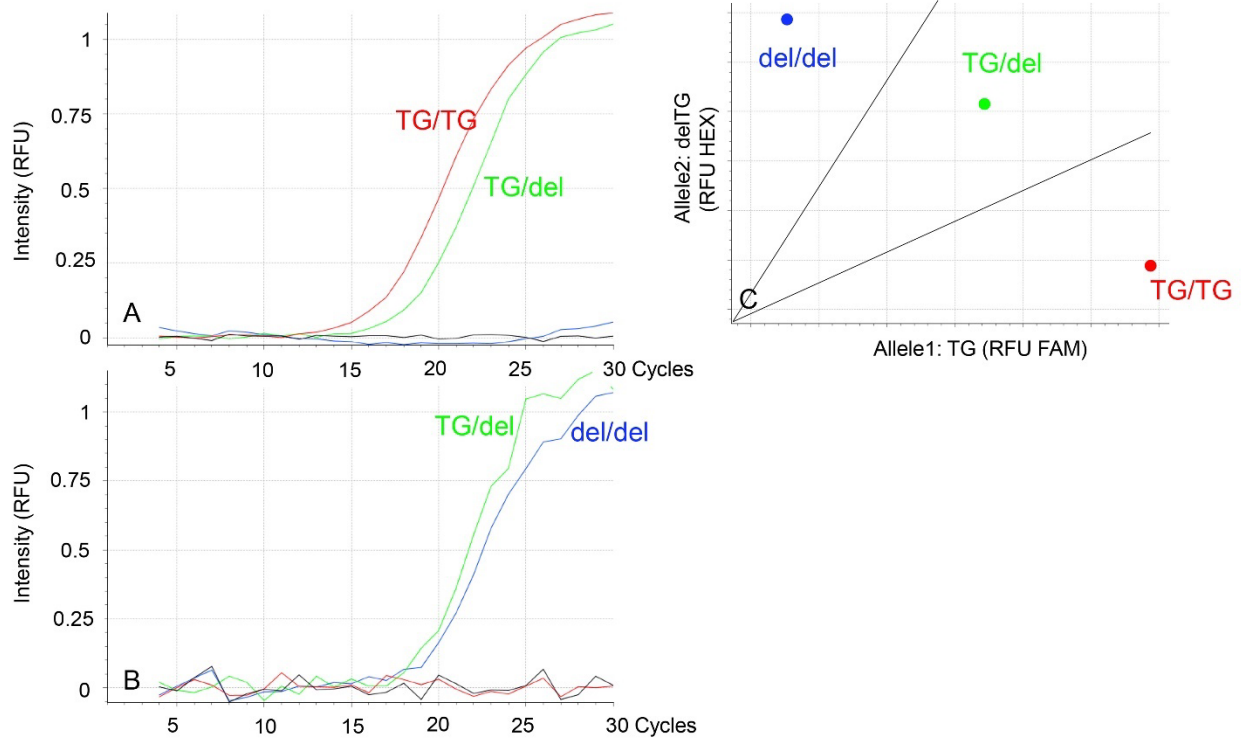**Supplemental Table 2.** Genotyping data of c.370_371delTG of 136 clinically normal cats.

Sample	Breed	Genotype*
1	Crossbred	TGTG
2	Crossbred	TGTG
3	Scottish Fold	TGTG
4	Crossbred	TGTG
5	Crossbred	TGTG
6	Crossbred	TGTG
7	Crossbred	TGTG
8	Crossbred	TGTG
9	Kinkalow	TGTG
10	Crossbred	TGTG
11	Crossbred	TGTG
12	Crossbred	TGTG
13	American Shorthair	TGTG
14	Crossbred	TGTG
15	Crossbred	TGTG
16	Munchkin	TGTG
17	Crossbred	TGTG
18	Crossbred	TGTG
19	Crossbred	TGTG
20	American Shorthair	TGTG
21	American Shorthair	TGTG
22	Crossbred	TGTG
23	Crossbred	TGTG
24	Crossbred	TGTG
25	Crossbred	TGTG
26	Bengal	TGTG
27	Crossbred	TGTG

28	Crossbred	TGTG
29	Scottish Fold	TGTG
30	Crossbred	TGTG
31	Crossbred	TGTG
32	Crossbred	TGTG
33	American Shorthair	TGTG
34	American Shorthair	TGTG
35	American Shorthair	TGTG
36	Crossbred	TGTG
37	American Shorthair	TGTG
38	Crossbred	TGTG
39	American Shorthair	TGTG
40	Crossbred	TGTG
41	Crossbred	TGTG
42	Crossbred	TGTG
43	Crossbred	TGTG
44	Crossbred	TGTG
45	American Shorthair	TGTG
46	Crossbred	TGTG
47	Crossbred	TGTG
48	Crossbred	TGTG
49	Munchkin	TGTG
50	Crossbred	TGTG
51	Crossbred	TGTG
52	Crossbred	TGTG
53	Crossbred	TGTG
54	Scottish Fold	TGTG
55	Crossbred	TGTG
56	Crossbred	TGTG
57	Crossbred	TGTG
58	Crossbred	TGTG
59	Crossbred	TGTG
60	Crossbred	TGTG
61	Crossbred	TGTG
62	Crossbred	TGTG
63	Crossbred	TGTG
64	Crossbred	TGTG
65	Crossbred	TGTG

66	Crossbred	TGTG
67	Crossbred	TGTG
68	Crossbred	TGTG
69	Crossbred	TGTG
70	Crossbred	TGTG
71	Crossbred	TGTG
72	Crossbred	TGTG
73	Crossbred	TGTG
74	Crossbred	TGTG
75	Crossbred	TGTG
76	Crossbred	TGTG
77	Crossbred	TGTG
78	Crossbred	TGTG
79	Crossbred	TGTG
80	Crossbred	TGTG
81	Crossbred	TGTG
82	Crossbred	TGTG
83	Crossbred	TGTG
84	Crossbred	TGTG
85	Crossbred	TGTG
86	Norwegian Forest Cat	TGTG
87	Crossbred	TGTG
88	Crossbred	TGTG
89	Crossbred	TGTG
90	Crossbred	TGTG
91	Crossbred	TGTG
92	Crossbred	TGTG
93	Crossbred	TGTG
94	Crossbred	TGTG
95	Crossbred	TGTG
96	Crossbred	TGTG
97	Crossbred	TGTG
98	Crossbred	TGTG
99	American Shorthair	TGTG
100	Crossbred	TGTG
101	Crossbred	TGTG
102	Crossbred	TGTG
103	Crossbred	TGTG

104	Crossbred	TGTG
105	Crossbred	TGTG
106	Crossbred	TGTG
107	Crossbred	TGTG
108	Crossbred	TGTG
109	Crossbred	TGTG
110	Scottish Fold	TGTG
111	Crossbred	TGTG
112	Crossbred	TGTG
113	Crossbred	TGTG
114	Crossbred	TGTG
115	Crossbred	TGTG
116	Crossbred	TGTG
117	Crossbred	TGTG
118	Crossbred	TGTG
119	Crossbred	TGTG
120	Crossbred	TGTG
121	Crossbred	TGTG
122	Crossbred	TGTG
123	Crossbred	TGTG
124	Crossbred	TGTG
125	Munchkin	TGTG
126	American Shorthair	TGTG
127	American Curl	TGTG
128	Munchkin	TGTG
129	Crossbred	TGTG
130	Main Coon	TGTG
131	Crossbred	TGTG
132	Crossbred	TGTG
133	American Shorthair	TGTG
134	Crossbred	TGTG
135	Crossbred	TGTG
136	Crossbred	TGTG

* TGTG means homozygous for TG. There is no deletion of TG.

Supplemental Figure 1. Genotyping of the *CREB3L1* mutation by PCR allelic competitive extension (PACE). The heterozygous sample contained equal amounts of genomic DNA obtained from the normal cat and from the patient. **A.** Detection of the normal allele; the wild-type (TG/TG) and heterozygous (TG/del) samples are amplified but the mutant sample is not. **B.** Detection of the mutant allele; the homozygous and heterozygous mutants are amplified, but the normal sample is not. **C.** Cluster analysis; 3 genotypes are distinguished successfully.

See also **Supplemental Video.** Video of the patient walking.