

**Table S1** PCR primers for amplification of all exons and exon-intron boundaries of the *SLC4A1* gene.

| Location | Primer name | Primer sequence                 | Annealing temperature (°C) | PCR product (bp) |
|----------|-------------|---------------------------------|----------------------------|------------------|
| Promoter | In31L       | 5'-CAGTTTGGGACAAGGGCGTG-3'      | 69                         | 770              |
|          | In32R       | 5'-CGGTGTCGTGAGCTGAAAACC-3'     |                            |                  |
| Exon 1*  | 1L1         | 5'-GTGGGGCGGGCAGATTAG-3'        | 65                         | 199              |
|          | 1R1         | 5'-AGGCACCGAAGACCATCCC-3'       |                            |                  |
| Exon 2*  | 2L          | 5'-CCTCTGGAGGAGGACCAG-3'        | 60                         | 232              |
|          | 2R          | 5'-ACCCCCAACGAAGATAGG-3'        |                            |                  |
| Exon 3*  | 3L          | 5'-CTTCGTTGGGGGTGGAAGAG-3'      | 60                         | 235              |
|          | 3R          | 5'-GTGGAGAAGGGGAGAGGACAA-3'     |                            |                  |
| Exon 4   | 4L          | 5'-GTCTCTGAGGCTCACAGTGGATG-3'   | 63                         | 226              |
|          | 4R          | 5'-ATCCCCTTGCTCCTCTCTTCC-3'     |                            |                  |
| Exon 5   | 5L          | 5'-TGAGCACCCACTATGCCCT-3'       | 63                         | 299              |
|          | 5R          | 5'-CAGCACCCACACAACATCCTC-3'     |                            |                  |
| Exon 6   | 6L          | 5'-AGATGAGGATTGTTGTGGGGT-3'     | 63                         | 261              |
|          | 6R          | 5'-CAAGTGGGCTGGGGAAGTG-3'       |                            |                  |
| Exon 7   | 7L          | 5'-CACCCTGATAGCTCAGCCTG-3'      | 60                         | 243              |
|          | 7R          | 5'-TGAGAAAGCTCTCTCCTTGCCC-3'    |                            |                  |
| Exon 8   | 8L          | 5'-GAGAATGGGAAGGGGAGGATG-3'     | 60                         | 244              |
|          | 8R          | 5'-GGTCCAGGCTGAGGGAAGAC-3'      |                            |                  |
| Exon 9   | 9L          | 5'-TCTTCAGCACCCACCTG-3'         | 60                         | 299              |
|          | 9R          | 5'-TCAGCCACCATGCAGGTCC-3'       |                            |                  |
| Exon 10* | 10L         | 5'-TCCTGGCAATGGGAGCTG-3'        | 65                         | 483              |
|          | 10R         | 5'-GATTGTCTGATGGGAATGGGG-3'     |                            |                  |
| Exon 11  | 11L         | 5'-CCTCACCTCCTCCAGCTACTCC-3'    | 67                         | 318              |
|          | 11R         | 5'-CAGAAGTTGGGGCTGAGACAGAG-3'   |                            |                  |
| Exon 12  | 12L         | 5'-GCTCTATGGGCTCCTGGAAAT-3'     | 62                         | 293              |
|          | 12R         | 5'-AAAGGGTCTTGGGGCAAGG-3'       |                            |                  |
| Exon 13  | 13L         | 5'-CTGTCATGTCCCCCGCAC-3'        | 58                         | 339              |
|          | 13R         | 5'-TGTCTCAGTCTTATACACAACCTCC-3' |                            |                  |
| Exon 14  | 14L         | 5'-TGGTGGTATTTTCCAGCCCAAG-3'    | 60                         | 320              |
|          | 14R         | 5'-GCACTGAGGAATTTGGAGCGG-3'     |                            |                  |
| Exon 15  | 15L         | 5'-AAGGCAGGAGGTGGGGAGTACTG-3'   | 70                         | 201              |
|          | 15R         | 5'-GGAAATGAGGACCTGGGGGGTATC-3'  |                            |                  |
| Exon 16  | 16L         | 5'-TCCTGCTCCCACCTTCCCC-3'       | 68                         | 276              |
|          | 16R         | 5'-TCTGCCTCCCACCTTCCCAG-3'      |                            |                  |
| Exon 17  | 17L         | 5'-TGGAGGAGGCAGGGGAGAAC-3'      | 67                         | 347              |
|          | 17R         | 5'-GGGGCAGGAGGATGGTGAAG-3'      |                            |                  |
| Exon 18  | 18L         | 5'-ATATGGTGCCTGTGTTTATTCCC-3'   | 65                         | 336              |
|          | 18R         | 5'-TGCTATCACACCCAGCAC-3'        |                            |                  |
| Exon 19  | 19L         | 5'-GGTACAGGACCCTTTTCTGG-3'      | 60                         | 334              |
|          | 19R         | 5'-GCCTGCCCTAGTTCTGAGAC-3'      |                            |                  |
| Exon 20* | 20L         | 5'-CAAGACAGCCAGACCCTCTC-3'      | 61                         | 335              |
|          | 20R         | 5'-CTTCCTTCCCCACCCACAG-3'       |                            |                  |
| PolyA*   | PolAL       | 5'-GATGTCCCTGTCTCCCCCTC-3'      | 60                         | 201              |
|          | PolAR       | 5'-CCTAGTTCAGCCAAGACCC-3'       |                            |                  |

PCR polymerase chain reaction, bp base pairs

\* PCR primers were newly designed in this study

**Table S2** Clinical and laboratory data of patient (II:1).

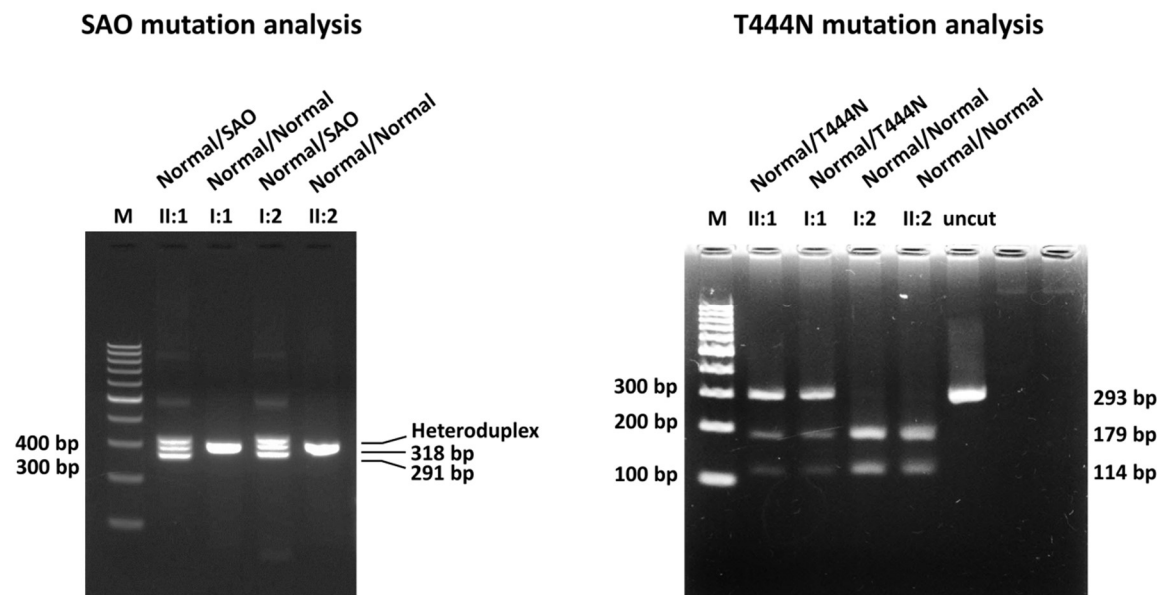
| Patient data                                                       | II:1  | Normal range |
|--------------------------------------------------------------------|-------|--------------|
| <b>Clinical chemistry</b>                                          |       |              |
| Urine pH <sup>a</sup>                                              | 7     | 4.6-8        |
| Serum Bicarbonate <sup>a</sup> (mEq/L)                             | 20    | 22-29        |
| Potassium (K <sup>+</sup> ) (mmol/L)                               | 3.5   | 3.4-4.5      |
| BUN (mg/dl)                                                        | 9.5   | 6-20         |
| Creatinine (mg/dl)                                                 | 0.93  | 0.51-0.95    |
| Creatinine Clearance <sup>a</sup> (cc/minute/1.73 m <sup>2</sup> ) | 70.6  | < 60         |
| <b>Hematology</b>                                                  |       |              |
| Hemoglobin <sup>b</sup> (g/dL)                                     | 11.7  | 12.5-16.8    |
| Hematocrit <sup>b</sup> (%)                                        | 31.7  | 37.1-42.7    |
| Reticulocyte count <sup>b</sup> (%)                                | 3.2   | 0.7-2.3      |
| Absolute count                                                     | 0.101 | 0.023-0.099  |
| RBC (x 10 <sup>6</sup> /μl)                                        | 3.17  | 3.90-6.01    |
| MCV <sup>b</sup> (fL)                                              | 100   | 82-97        |
| MCH (pg)                                                           | 37    | 27-31        |
| MCHC <sup>b</sup> (g/dL)                                           | 37    | 32-36        |
| RDW <sup>b</sup> (%)                                               | 24.5  | 12.5-14.9    |
| White blood cell count (x 10 <sup>3</sup> /μl)                     | 11.5  | 5.3-10       |
| Neutrophils (%)                                                    | 67.8  | 59-69        |
| Lymphocyte (%)                                                     | 23.4  | 34-42        |
| Monocyte (%)                                                       | 4.3   | 4-8          |
| Eosinophils (%)                                                    | 3.7   | 0-5          |
| Basophils (%)                                                      | 0.8   | 0-1          |
| Platelet count (x 10 <sup>3</sup> /μl)                             | 257   | 157-420      |

*BUN* Blood urea nitrogen, *RBC* Red blood cell, *MCV* Mean corpuscular volume, *MCH* Mean corpuscular hemoglobin, *MCHC* Mean corpuscular hemoglobin concentration, *RDW* Red cell distribution width

<sup>a</sup> Diagnostic criteria for patient with dRTA

<sup>b</sup> Diagnostic criteria for patient with anemia

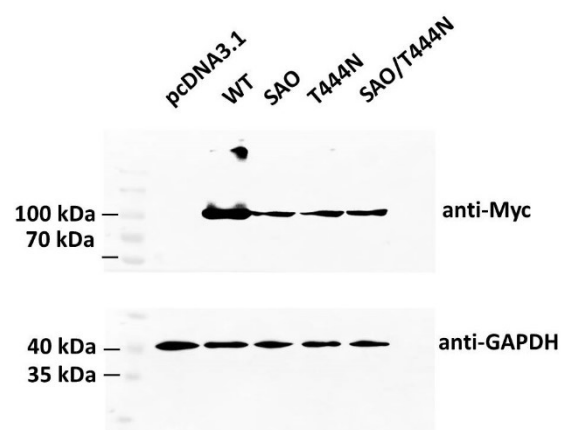
**Fig. S1** The original and unprocessed versions of Fig. 1c.



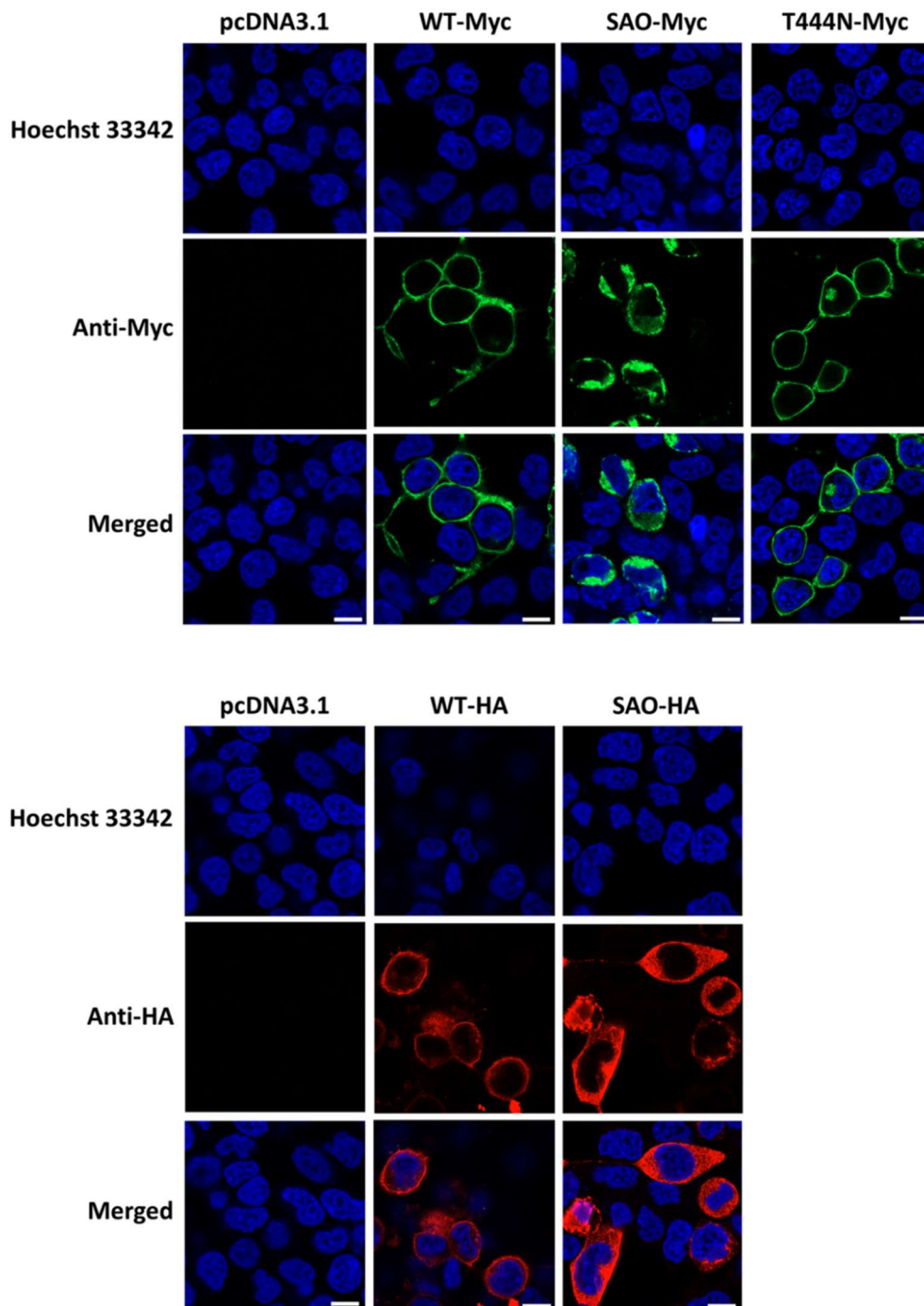
**Table S3** Results of bioinformatic analyses of the T444N mutation using VarCards web-based program.

| <b>Algorithm</b> | <b>Score</b> | <b>Prediction</b> |
|------------------|--------------|-------------------|
| SIFT             | 0.004        | Damaging          |
| Polyphen-2_HDIV  | 1            | Probably damaging |
| Polyphen-2_HVAR  | 0.999        | Probably damaging |
| LRT              | 0            | Deleterious       |
| MutationTaster   | 1.0          | Disease causing   |
| MutationAssessor | 3.675        | High              |
| FATHMM           | -1.4         | Tolerable         |
| PROVEAN          | -4.81        | Damaging          |
| VEST3            | 0.321        | Tolerable         |
| MetaSVM          | 0.769        | Damaging          |
| MetaLR           | 0.776        | Damaging          |
| M-CAP            | 0.124        | Damaging          |
| CADD             | 23.9         | Damaging          |
| DANN             | 0.994        | Damaging          |
| FATHMM MKL       | 0.993        | Damaging          |
| Eigen            | 0.891        | Damaging          |
| GenoCanyon       | 1.0          | Damaging          |
| fitCons          | 0.487        | Tolerable         |
| ReVe             | 0.691        | Tolerable         |
| ClinPred         | 0.88097769   | Pathogenic        |
| REVEL            | 0.759        | Damaging          |

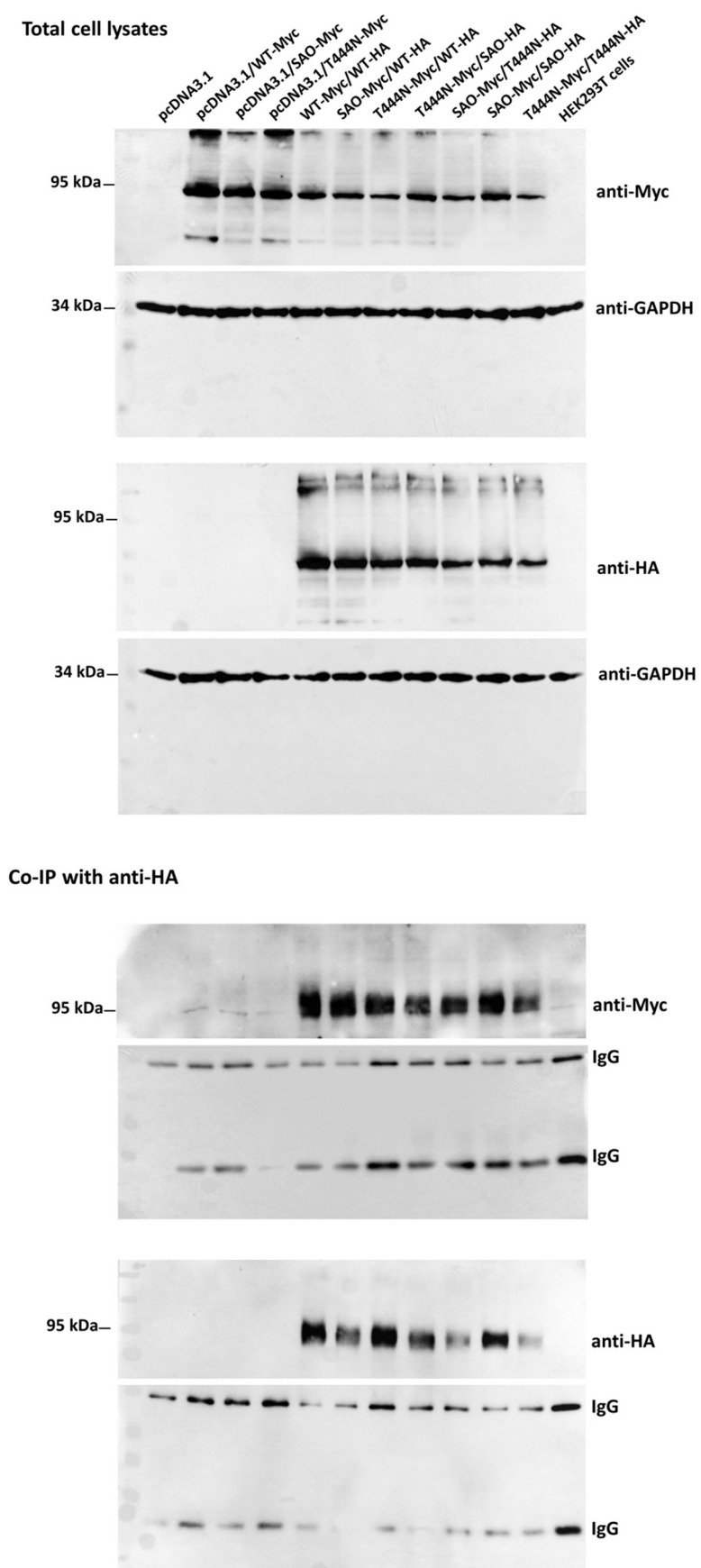
**Fig. S2** The original and unprocessed versions of Fig. 3a.



**Fig. S3** Cellular localization of wild-type and mutant kAE1 proteins in HEK293T cells detected by immunofluorescence assay. The transfected cells were stained with mouse anti-Myc or rabbit anti-HA antibodies, followed by Hoechst 33342 (blue) and AlexaFluor 488-conjugated anti-mouse (green) or AlexaFluor 594-conjugated anti-rabbit (red) antibodies. The cells were visualized by confocal microscopy. Scale bars: 10  $\mu$ m.



**Fig. S4** The original blot and unprocessed versions of Fig. 5a.



**Fig. S5** The original blot and unprocessed versions of Fig. 5b.

