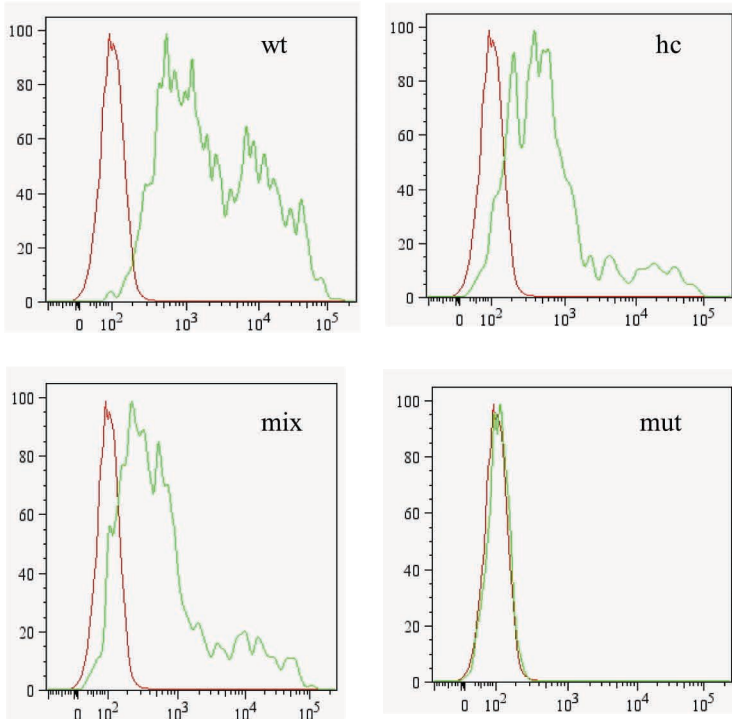
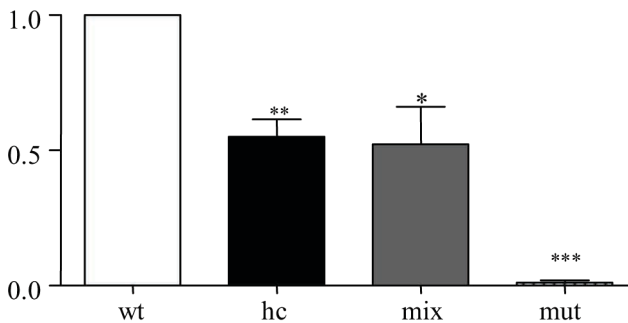


Anti- $\alpha 1$  Alexa 647

A

Surface  $\alpha 1$  subunit protein expression of  
 $\alpha 1/\alpha 1(S326fs328X)\beta 2\gamma 2S$ 

B

Relative surface expression (fluorescence index) of  
the  $\alpha 1$  subunit protein in HEK 293T cells  
 $\alpha 1/\alpha 1(S326fs328X)$  subunit minigenes

**Supplementary Figure 2. The more quantitative technique of flow cytometry was used to determine the relative surface expression of  $\alpha 1$  subunit protein.** HEK 293T cells were cotransfected with  $\beta 2$  and  $\gamma 2S$  subunit cDNAs and the wild-type  $\alpha 1$  subunit minigene (ratio of 1:1:1, (wt) or 1:1:0.5:0.5 (empty vector) (hc), mixed expression of  $\alpha 1/\alpha 1(S326fs328X)$  subunit minigenes (ratio of 1:1:0.5:0.5 (mix)) and mutant  $\alpha 1(S326fs328X)$  subunit minigene (ratio of 1:1:1 (mut)) using Fugene. **(A)** The flow cytometry histograms depict the expression pattern of surface  $\alpha 1$  subunit in different experiment conditions as detected with fluorescently conjugated anti human  $\alpha 1$  antibody ( $\alpha 1$ -Alexa 647). **(B)** The expression of the total wild-type protein was arbitrarily taken as 1, and the  $\alpha 1$  subunit surface expression in other conditions was normalized to the wild-type each time (\* $p = 0.05$ , \*\*  $p = 0.005$ , \*\*\* $p < 0.001$  vs wild type  $n = 4$ ). There was no difference of  $\alpha 1$  subunit surface expression between the haploinsufficiency control (hc) and mixed (mix) conditions ( $p = 0.8636$ ).