Supplemental Table 1. Minor allele frequency of human single nucleotide variants accompanied with the alteration in the β5t amino acid sequence.

rsID	Residue change	Minor allele frequency
rs34457782	G49S	0.0084
rs149885629	A203T	0.0038
rs572934212	S80H (frame shift)	0.0030
rs200339082	R169C	0.0016
rs201138394	R155H	0.0014
rs181637075	R238H	0.0012
rs202220440	R115Q	0.0008
rs572562943	G152S	0.0008
rs200639481	R169H	0.0006
rs149026126	A212T	0.0004

Listed are the top ten variants of the highest frequency in accordance with the dbSNP database hosted by NCBI. The rs34457782 variant that causes the G49S variation has the highest frequency, as shown in red letters.

Supplemental Table 2. Minor allele frequency of β5t-G49S variation in various human populations.

Population description	Sample no.	Minor allele frequency
African Carribbeans in Barbados	96	0.0000
Americans of African Ancestry in SW USA	61	0.0000
Bengali from Bangladesh	86	0.0116
Chinese Dai in Xishuangbanna, China	93	0.0054
Utah Residents with Northern and Western Ancestry	99	0.0152
Han Chinese in Bejing, China	103	0.0485
Southern Han Chinese	105	0.019
Colombians from Medellin, Colombia	94	0.0000
Esan in Nigeria	99	0.0000
Finnish in Finland	99	0.0303
British in England and Scotland	91	0.0000
Gujarati Indian from Houston, Texas	103	0.0000
Gambian in Western Divisions in the Gambia	113	0.0000
Iberian Population in Spain	107	0.0000
Indian Telugu from the UK	102	0.0000
Japanese in Tokyo, Japan	104	0.0240
Kinh in Ho Chi Minh City, Vietnam	99	0.0303
Luhya in Webuye, Kenya	99	0.0051
Mende in Sierra Leone	85	0.0000
Mexican Ancestry from Los Angeles USA	64	0.0000
Peruvians from Lima, Peru	85	0.0000
Punjabi from Lahore, Pakistan	96	0.0000
Puerto Ricans from Puerto Rico	104	0.0144
Sri Lankan Tamil from the UK	102	0.0000
Toscani in Italia	107	0.0047
Yoruba in Ibadan, Nigeria	108	0.0000

Sample number and minor allele frequency are listed in accordance with the NCBI database.

Supplemental Table 3. Genotypes of rs34457782 variant determined in genomic DNAs from 949 healthy volunteers collected at the University of Tokushima.

Genotype	Number	Frequency (%)
G/G	890	93.78
G/A	59	6.21
A/A	0	0
Major G allele	1,839	96.89
Minor A allele	59	3.11

The observed genotype distribution showed no significant deviation from the Hardy-Weinberg equilibrium ($\chi 2$=0.98, df=1, p=0.32).

Figure 1A

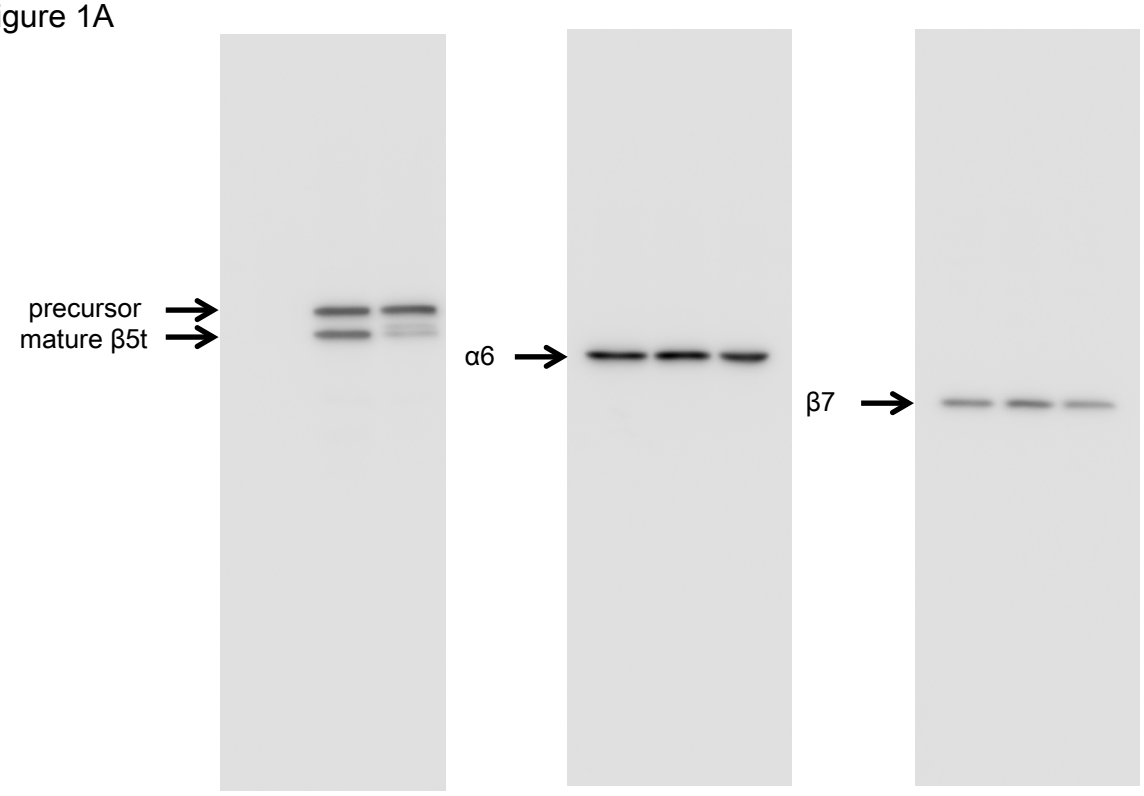

Figure 1B

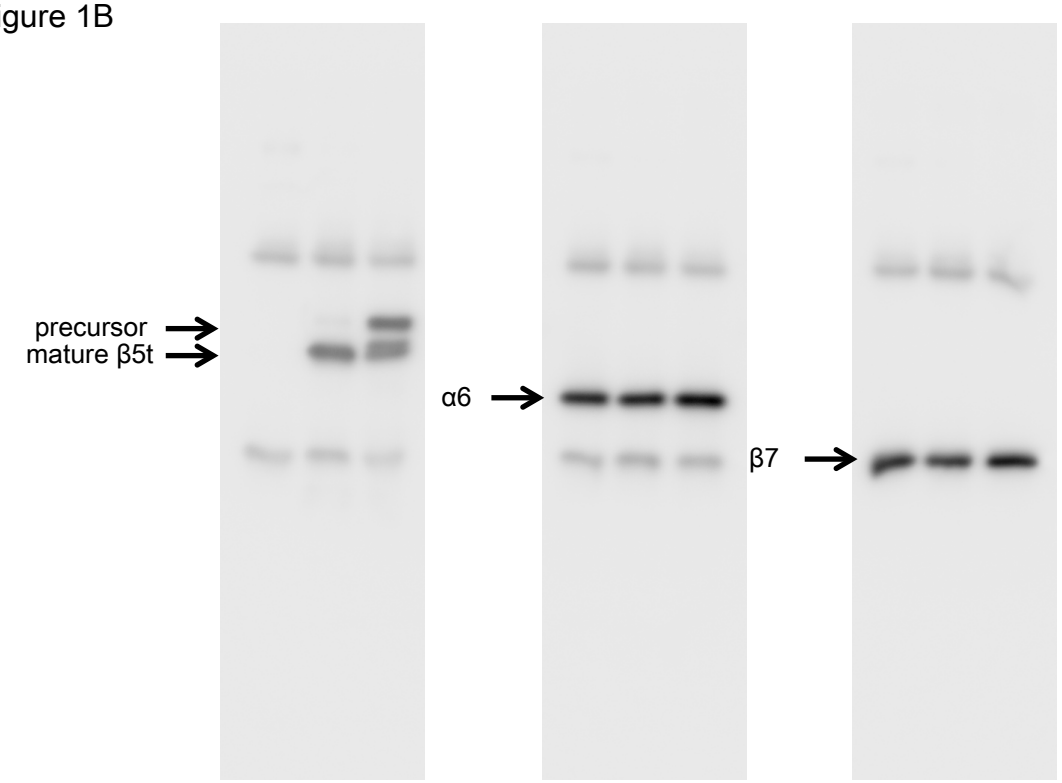

Figure 1C

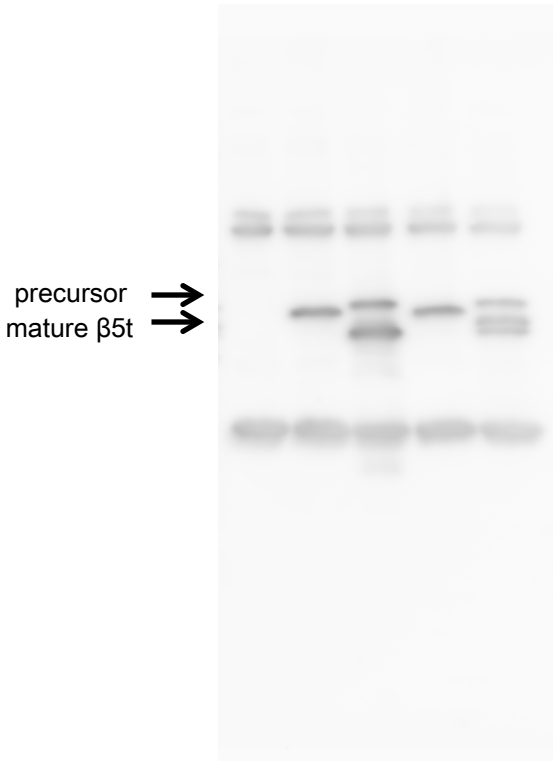

Figure 2B

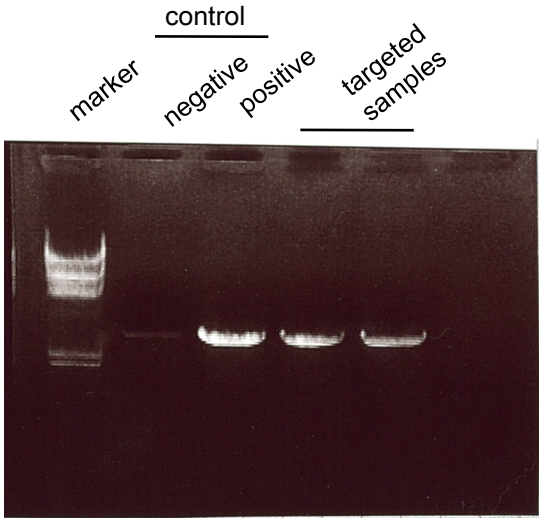

Figure 2C

These two lanes are shown in Fig. 2C.