

SUPPLEMENTARY FIGURES AND TABLES

Supplementary Figures

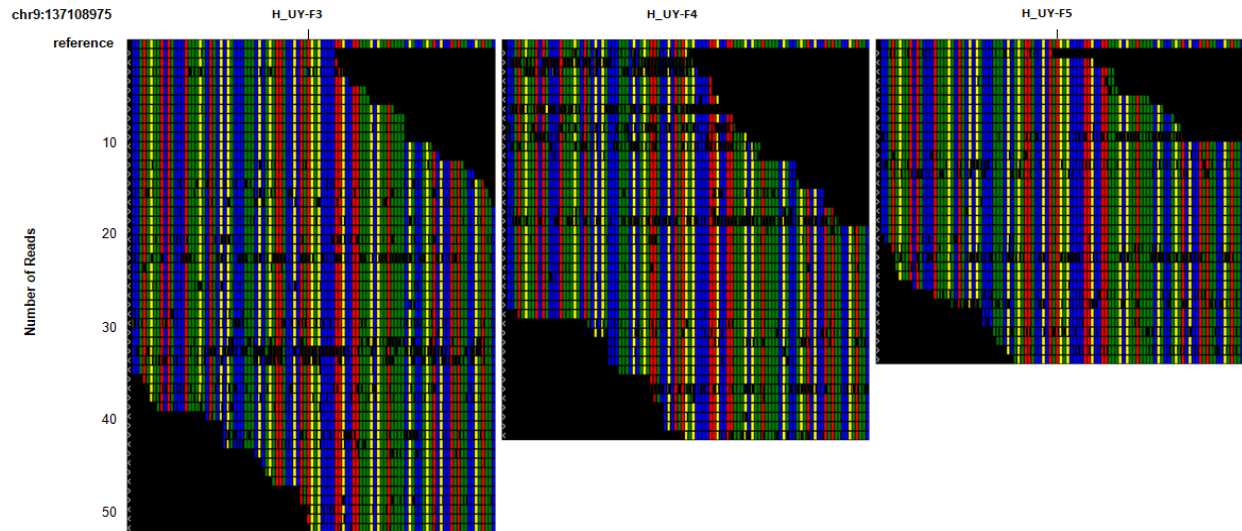


Fig. S1. Integrative Genomics Viewer (IGV) visualization of read alignments for MAN1B1 rs4567 in trio of three siblings, showing a consistent A/A genotype in all (unaffected siblings H_UY-F3 and H_UY-F4 and index subject H_UY-F5). The vertical axis represents the sequencing depth (number of independent reads) and the horizontal axis represents the genomic coordinates with the rs4567 at the center. The colors yellow, blue, red and green correspond to the nucleotides T, C, A and G, respectively.

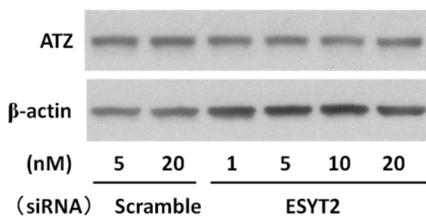


Fig. S2. Effect of knocking down ESYT2 expression using siRNA on ATZ and β-actin levels in HTO/Z cell line.

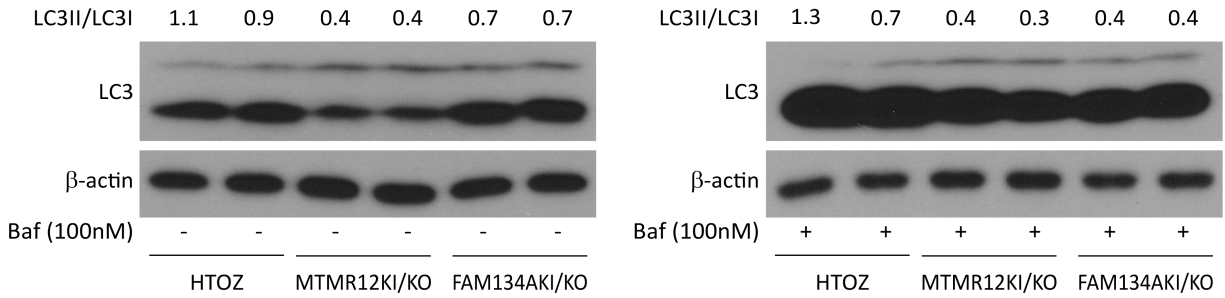


Fig S3. Effect of MTMR12 and FAM134A variants on autophagic flux in HTO/Z cell line. Immunoblot analysis for LC3 and β -actin in HTO/Z after 24 hr incubation in the absence or presence of bafilomycin 100 nM. Densitometric analysis of LC3II/LC3I ratio is shown at the top.

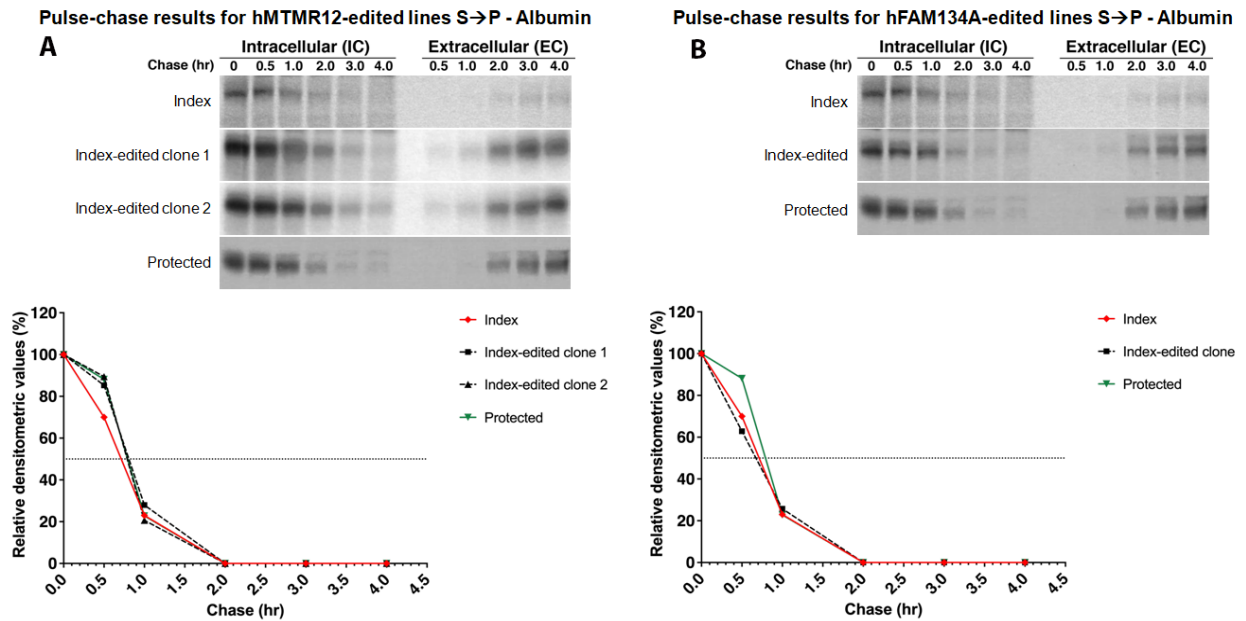


Fig. S4. Effect of correcting MTMR12 (A) and FAM134A (B) variants in index subject on kinetics of albumin fate.