

Title: Supplementary data 1:

Description: List of significantly differentially expressed genes in P635L HET, P635L HOM, R636Q HET and R636Q HOM relative to WT

Title: Supplementary data 2:

Description: List of significantly differentially spliced events in P635L HET, P635L HOM, R636Q HET and R636Q HOM relative to WT

Title: Supplementary data 3:

Description: List of significantly differentially expressed genes in different cell types based on snRNA-seq including candidate genes for analysis in Fig. 4e-g

Title: Supplementary data 4:

Description: Mapping statistics of NGS experiments including WGS coverage

Title: Supplementary data 5:

Description: Tissue-specific variants found by WGS Supplementary data 6: Sequences of oligonucleotides used in this study