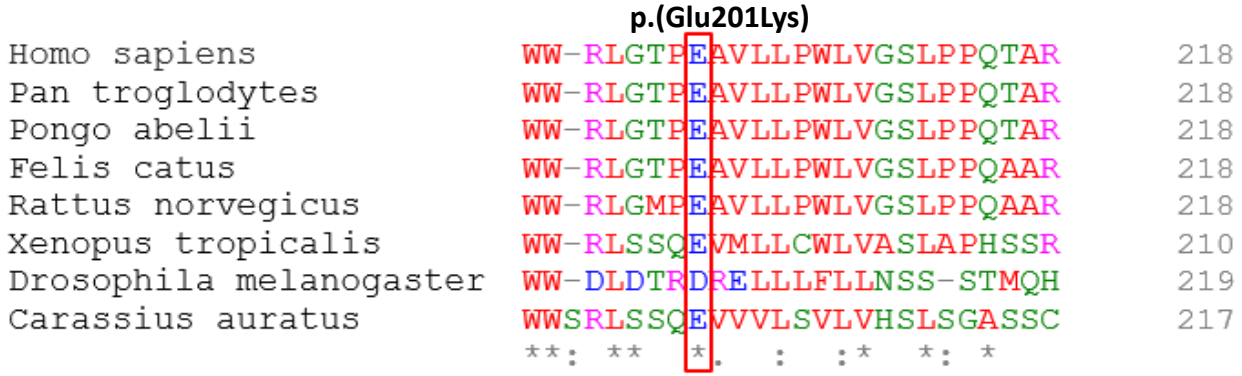
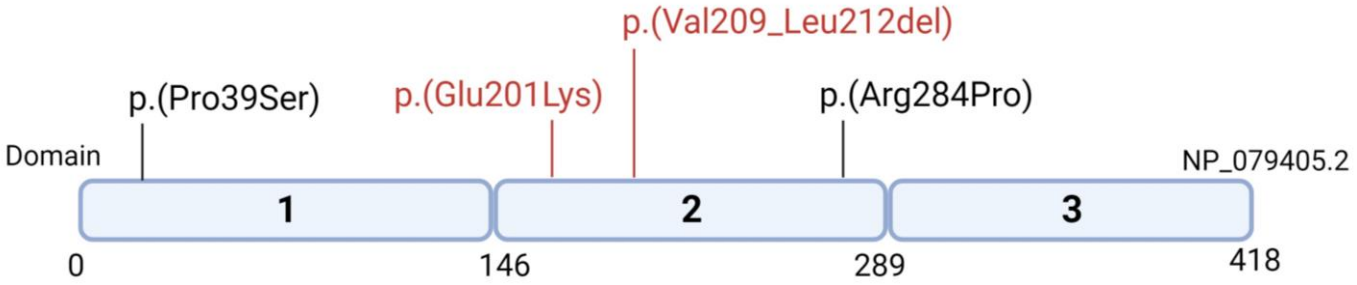


Supplementary figure 1: Multiple sequence alignment of p.(Glu201Lys) in *FUZ* indicates that glutamine residue at 201 position is conserved among different vertebrates. An asterisk (*) represents fully conserved residue, colon (:) represents residues with strongly similar properties, and period (.) represents residues with weakly similar properties.

A)

B)



Supplementary figure 2: A) Cartoon illustration of structure of the gene (*FUZ*) and (B) domains of FUZ protein with disease-causing variants identified in individuals with FUZ related disorders. Novel biallelic variants identified in this study are coded in red.