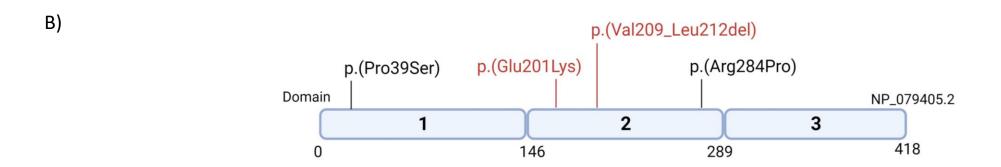
```
p.(Glu201Lys)
Homo sapiens
                        WW-RLGTPEAVLLPWLVGSLPPQTAR
                                                       218
Pan troglodytes
                        WW-RLGTPEAVLLPWLVGSLPPQTAR
                                                       218
Pongo abelii
                        WW-RLGTPEAVLLPWLVGSLPPQTAR
                                                       218
                        WW-RLGTPEAVLLPWLVGSLPPQAAR
Felis catus
                                                       218
Rattus norvegicus
                        WW-RLGMPEAVLLPWLVGSLPPQAAR
                                                       218
Xenopus tropicalis
                        WW-RLSSQEVMLLCWLVASLAPHSSR
                                                       210
Drosophila melanogaster WW-DLDTRDRELLLFLLNSS-STMQH
                                                       219
Carassius auratus
                        WWSRLSSQEVVVLSVLVHSLSGASSC
                                                       217
```

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.







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.