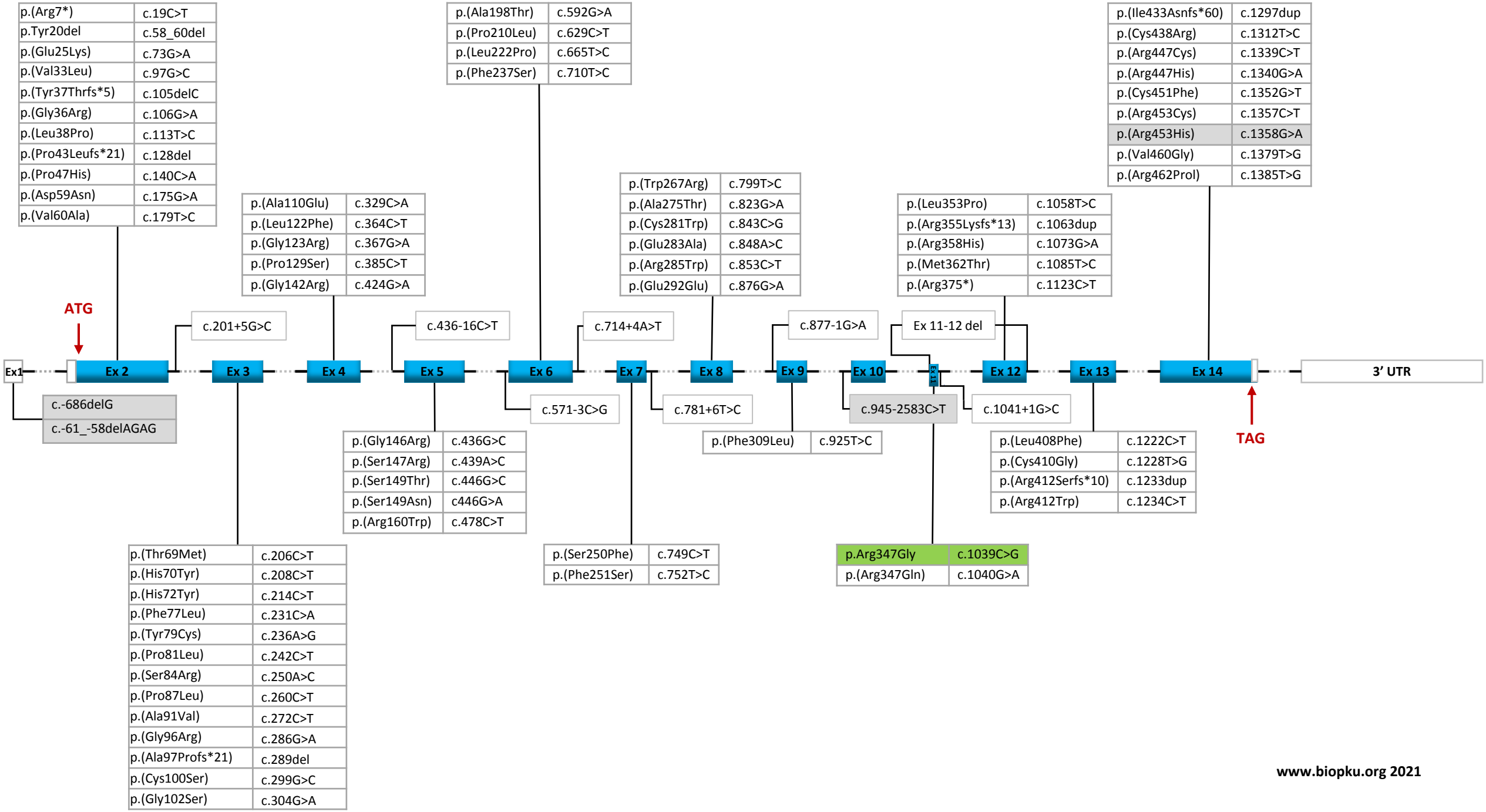
p.(Arg7*) c.19C>T
p.Tyr20del c.58_60del
p.(Glu25Lys) c.73G>A
p.(Val33Leu) c.97G>C
p.(Tyr37Thrfs*5) c.105delC
p.(Gly36Arg) c.106G>A
p.(Leu38Pro) c.113T>C
p.(Pro43Leufs*21) c.128del
p.(Pro47His) c.140C>A
p.(Asp59Asn) c.175G>A
p.(Val60Ala) c.179T>C
p.(Ala198Thr) c.592G>A
p.(Pro210Leu) c.629C>T
p.(Leu222Pro) c.665T>C
p.(Phe237Ser) c.710T>C
p.(Ile433Asnfs*60) c.1297dup
p.(Cys438Arg) c.1312T>C
p.(Arg447Cys) c.1339C>T
p.(Arg447His) c.1340G>A
p.(Cys451Phe) c.1352G>T
p.(Arg453Cys) c.1357C>T
p.(Arg453His) c.1358G>A
p.(Val460Gly) c.1379T>G
p.(Arg462Prol) c.1385T>G
p.(Ala110Glu) c.329C>A
p.(Leu122Phe) c.364C>T
p.(Gly123Arg) c.367G>A
p.(Pro129Ser) c.385C>T
p.(Gly142Arg) c.424G>A
p.(Trp267Arg) c.799T>C
p.(Ala275Thr) c.823G>A
p.(Cys281Trp) c.843C>G
p.(Glu283Ala) c.848A>C
p.(Arg285Trp) c.853C>T
p.(Glu292Glu) c.876G>A
p.(Leu353Pro) c.1058T>C
p.(Arg355Lysfs*13) c.1063dup
p.(Arg358His) c.1073G>A
p.(Met362Thr) c.1085T>C
p.(Arg375*) c.1123C>T
ATG
c.201+5G>C
c.436-16C>T
c.714+4A>T
c.877-1G>A
Ex 11-12 del
Ex1
Ex 2
Ex 3
Ex 4
Ex 5
Ex 6
Ex 7
Ex 8
Ex 9
Ex 10
Ex 11
Ex 12
Ex 13
Ex 14
3' UTR
c.-686delG
c.-61_-58delAGAG
c.571-3C>G
c.781+6T>C
c.945-2583C>T
c.1041+1G>C
TAG
p.(Gly146Arg) c.436G>C
p.(Ser147Arg) c.439A>C
p.(Ser149Thr) c.446G>C
p.(Ser149Asn) c446G>A
p.(Arg160Trp) c.478C>T
p.(Phe309Leu) c.925T>C
p.(Leu408Phe) c.1222C>T
p.(Cys410Gly) c.1228T>G
p.(Arg412Serfs*10) c.1233dup
p.(Arg412Trp) c.1234C>T
p.(Thr69Met) c.206C>T
p.(His70Tyr) c.208C>T
p.(His72Tyr) c.214C>T
p.(Phe77Leu) c.231C>A
p.(Tyr79Cys) c.236A>G
p.(Pro81Leu) c.242C>T
p.(Ser84Arg) c.250A>C
p.(Pro87Leu) c.260C>T
p.(Ala91Val) c.272C>T
p.(Gly96Arg) c.286G>A
p.(Ala97Profs*21) c.289del
p.(Cys100Ser) c.299G>C
p.(Gly102Ser) c.304G>A
p.(Ser250Phe) c.749C>T
p.(Phe251Ser) c.752T>C
p.Arg347Gly c.1039C>G
p.(Arg347Gln) c.1040G>A
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