

NOTCH3 exon 33 variants distribution and associated phenotype

Exon 33



ANK Domain

PEST Domain

c.5962dupC
p.Leu1976Profs11

LoF PHENOTYPE

c.6247A>T
p.K2083Ter

c.6409_6410del
p.Lys2137Glyfs104

c.6461_6486del
p.Gly2154Alafs79

c.6498_6577del
p.Ala2167Profs48

c.6603del
p.Val2202Serfs44

c.6626dup
p.Pro2210Alafs32

c.6659_6660del
p.Glu2220Valfs21

c.6692dup
p.Pro2231Glnfs10

c.6692_6693del
p.Pro2231Glnfs10

c.6663C>G
p.Tyr2221Ter

c.6732C>A
p.Tyr2244Ter

LMS PHENOTYPE