

Wallace Schwann cells (of variable purity) with heterozygous NF1 germline mutations

c.2542 G>T G848W
c.2350 T>C W784R
whole gene deletion
c.5941 C>T Q1981X
c.3826 C>T R1276X
c.1019-1020delCT
c.7285 C>T R2429X
c.2446 C>T R816X
c.647 C>T L216P
c.1756delACTA
c.4537 C>T R1513X
c.1527+2 delTAAG
c.3826 C>T R1276X
c.6788delTTAC
c.7258+1 G>T splice
c.8216 C>A S2739Y
c.7688delT
c.3357-3360delCTCA
c.6791insA
c.7543delA
c.1381 C>T R461X
c.1237 T>C S413P
c.1627 C>T Q543X
c.2446 C>T R816X
c.1521dupG
c.1748 A>G cryp spl
c.565 A>T K189X
c.4868 A>T D1623V
c.5839 C>T R1947X
c.5242 C>T R1748X
c.5529insT
c.3456_3457insA
c.4269 G>A splice
c.2252-1 G>C spl
R1276Q
c.6709 C>T R2237X
c.31 C>T Q11X
c.7774_7775C

Wallace fibroblast cells (skin or from tumor sample) with heterozygous NF1 germline mutations

c.6788delTTAC
c.2350 T>C W784R
c.6335_6339TGTGTGT
c.4900delA
c.7191delT
c.2542 G>T G848W
whole gene deletion
c.1527+2 delTAAG
c.1722-1723GTGT
c.3456_3457insA
c.2446 C>T R816X
c.7774_7775C
c.7285 C>T R2429X
c.1466 A>G Y489C cryptic splice
c.5010 delG
c.4900delA
c.2970-2972 delAAT (possibly)

c.8335 2 bp insertion

C.4900delA

c.4760 A>C Y1587S

c.1722-1723GTGT splice

c.3113+5 G>C