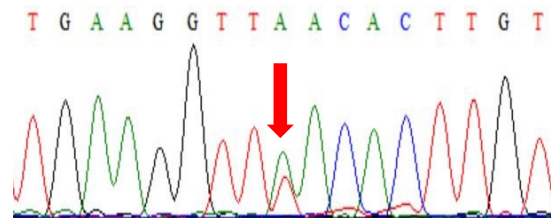
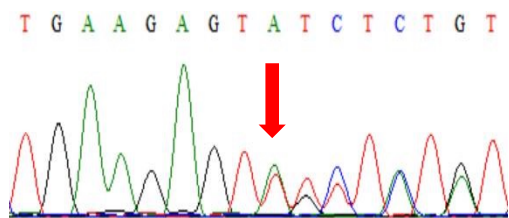


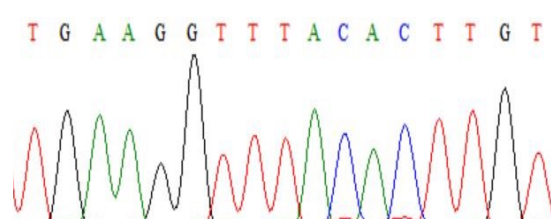
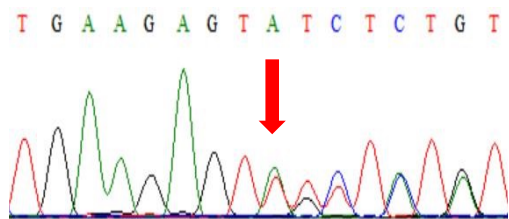
(a)

c.1122_1123insAG (p.Phe375fs)* c.2546T>A (p.Leu849Ter)

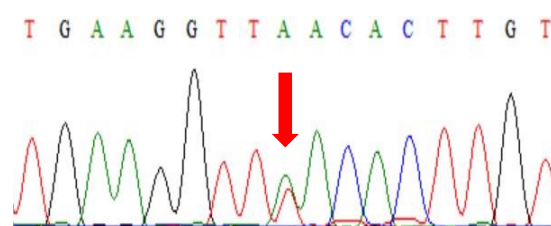
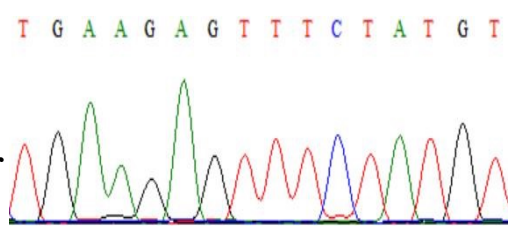
Patient



Father



Mother



*: novel mutation

(b)

