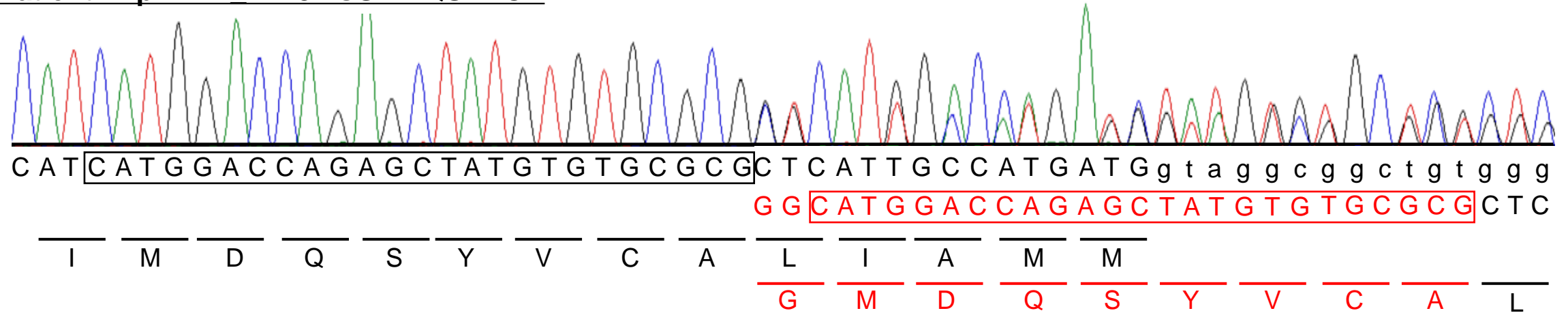


## Supplemental Figure S1. Image of the electropherogram of Sanger sequencing

**Patient 1: p.A427 L428insGMDQSYVCA**



Red characters indicate the 27-bp insertion, which is consisted with the 25-bp duplication in addition with GG. Black and red rectangles indicate the original and duplicated 25-bp nucleotide segments, respectively. By adding “GG”, the inserted 25-bp nucleotide segment caused 9 amino-acid insertion reading to in-frame insertion.