Result SummaryPositive

Result- The following heterozygous alteration was identified: Amino Acid change: p.R497PfsX6 (Arg497ProfsX6) DNA change: c.1489dupC (g.37070354) Classification: PATHOGENIC.

Interpretation - The c.1489dupC (p.R497PfsX6) alteration is a known pathogenic mutation. This result is consistent with a diagnosis of Lynch syndrome for this individual.

Method â□□Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and intron/exon boundaries of the MLH1 gene. Array comparative genomic hybridization (aCGH) was used to test for the presence of large deletions and duplications in this gene.