

PI3K α (E542K/+) Cell line Engineering Summary

| Genotype | rs # | Cosmic ID | Position | | | |
|----------------------------|-------------|-----------|-------------------|--|------------|------------|
| | | | Chromosome Number | Chromosome | Gene | Transcript |
| PI3K α (E542K/+) | rs121913273 | COSM760 | 3q26.3 | g.178936082G>A (GRCh37) g.179218294G>A (GRCh38) | g.74772G>A | c.1624G>A |

| Protein | Base change | Context | AA change |
|-------------|-------------------------------------|---|-------------------------------|
| p.Glu542Lys | GAA \Rightarrow AAA | TCCTCTCTCT (G>A) AAATCACTGA | E [Glu] \Rightarrow K [Lys] |

Reference sequence: Ensembl transcript PIK3CA-001 ENST00000263967

gaaaataaagtcttgcaatgaaaataaattatTTTtacaacagTTaattagcaatgtaaaatttattgaaaatgta
 ttgctTTTTctgtaaatca] [tctgtgaatccagaggggaaaaatatgacaaagaaagctatataagatatta
 tttatTTTtacagAGTAACAGACTAGCTAGAGACAATGAATTAAGGGAAAATGACAAAGAACAGCTCAAAGCAAT
 TTCTACACGAGATCCTCTCTCT (**G>A**) AAATCACTGAGCAGGAGAAAGATTTTCTATGGAGTCACAGgtaagt
 gctaaaatggagatttctctgtttctTTTTctttattacagaaaaaataactgaatttggctgatctcagcatggt
 ttaccataacctattggaataaataaagcagaattttacatgatttttaactataaacattgcctTTTTtaaaaac
 aatggttgtaaaattgatatttTgtggaaaatcatactacattggtagttggcacattaatgctTTTTct

Lowercase blue sequence indicates intron

Uppercase BLACK sequence indicates exon coding sequence

]] indicates the site of engineering

(**G>A**) indicates the E542K mutation

Contact Horizon Diagnostics

Please note that primers designed upstream or across the engineering site will have a reduced efficiency and your assay may fail. Please contact us at technical@horizondx.com if you have any questions.

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