



Gene by Gene serial:

Targeted mutation analysis of the CDKN2A gene

Tested individual surname: NA	Sample: Extracted DNA	Date Received:
Tested individual name: NA	Customer serial:	Date Reported:
Tested individual I.D.: NA	Gene by Gene serial:	
Sex: NA	Date of Birth: NA	

Results

1. The following pathogenic variant was detected within the targeted regions
 - a. Exon 02: g.21971057C>A; c.301G>T; p.Gly101Trp; rs104894094; heterozygote state.

Targeted genomic regions

1. CDKN2A: Chr9: g.21971057C>A; c.301G>T; p.Gly101Trp; rs104894094. Results are reported with respect to NM_000077.4 and the NCBI annotation 105: GRCh37/hg19.

Discussion

1. The variant c.301G>T was previously suggested to be causative for Melanoma by Roberts et al. (2016). Whole Genome Sequencing Defines the Genetic Heterogeneity of Familial Pancreatic Cancer. Cancer Discov. 2016 Feb;6(2):166-75.

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Gene by Gene is a College of American Pathologists (CAP) accredited and Clinical Laboratory Improvement Amendments (CLIA) certified clinical laboratory qualified to perform high-complexity testing. This test was developed and its performance characteristics determined by Gene by Gene. It has not been cleared or approved by the FDA. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. Only the genomic regions listed above were tested; there is a possibility that the tested individual is a carrier for additional, undetected mutations. Direct Sanger sequencing of a gene is unable to identify large deletions of a gene or part of it. Although molecular tests are highly accurate, rare diagnostic errors may occur that interfere with analysis. Sources of these errors include sample mix-up, trace contamination, and other technical errors. The presence of additional variants nearby may interfere with mutation detection. Genetic counseling is recommended to properly review and explain these results to the tested individual.