Targeted mutation analysis of the CDKN2A gene

Tested individual surname: NA
Tested individual name: NA
Tested individual I.D.: NA
Sex: NA

Sample: Extracted DNA
Customer serial:
Gene by Gene serial:
Date of Birth: NA

Date Received:
Date Reported:

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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