

50 Tumor Gene Analysis Report

50个肿瘤相关基因的突变筛查结果

Date: 08 / 20 /2016

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|-------------|---|
| Case ID: | 08112016-AB |
| DNA Source: | DNA of Human Liver Cancer |
| Method: | Ion AmpliSeq Cancer Hotspot Panel V2 by PGM |
| History: | Liver Concer (9T) and Control (9N) |

Sample -9 Control of Liver Cancer

| Chrom | Position | Ref | Variant | Allele Call | Frequency | Type | Allele | Gene ID |
|-------|-----------|-----|---------|--------------|-----------|------|--------|---------|
| chr4 | 55946354 | G | T | Heterozygous | 51.6 | SNP | Novel | KDR |
| chr4 | 55980239 | C | T | Heterozygous | 42.9 | SNP | Novel | KDR |
| chr7 | 55249063 | G | A | Heterozygous | 53.5 | SNP | Novel | EGFR |
| chr13 | 28610183 | A | G | Heterozygous | 49.5 | SNP | Novel | FLT3 |
| chr19 | 1220321 | T | C | Heterozygous | 44.6 | SNP | Novel | STK11 |
| chr4 | 1807894 | G | A | Homozygous | 100 | SNP | Novel | FGFR3 |
| chr4 | 55141055 | A | G | Homozygous | 100 | SNP | Novel | PDGFRA |
| chr5 | 112175770 | G | A | Homozygous | 100 | SNP | Novel | APC |
| chr5 | 149433596 | TG | GA | Homozygous | 100 | MNP | Novel | CSF1R |
| chr17 | 7579472 | G | C | Homozygous | 89.9 | SNP | Novel | TP53 |

Sample -9 Liver Cancer

| Chrom | Position | Ref | Variant | Allele Call | Frequency | Type | Allele | Gene ID |
|--------------|----------------|----------|----------|---------------------|-------------|------------|----------------|-------------|
| chr4 | 55946354 | G | T | Heterozygous | 18.5 | SNP | Novel | KDR |
| chr7 | 55249063 | G | A | Heterozygous | 66 | SNP | Novel | EGFR |
| chr13 | 28610183 | A | G | Heterozygous | 85.4 | SNP | Novel | FLT3 |
| chr17 | 7577547 | C | A | Heterozygous | 67.9 | SNP | Hotspot | TP53 |
| chr19 | 1220321 | T | C | Heterozygous | 24.4 | SNP | Novel | STK11 |
| chr4 | 1807894 | G | A | Homozygous | 100 | SNP | Novel | FGFR3 |
| chr4 | 55141055 | A | G | Homozygous | 100 | SNP | Novel | PDGFRA |
| chr5 | 112175770 | G | A | Homozygous | 100 | SNP | Novel | APC |
| chr5 | 149433596 | TG | GA | Homozygous | 100 | MNP | Novel | CSF1R |
| chr17 | 7579472 | G | C | Homozygous | 100 | SNP | Novel | TP53 |

- Summary:
1. Among 2871 cancer-related variants screened, 10 mutations were identified in the tumor sample.
 2. There are 5 heterozygous mutated alleles and 5 homozygous mutations.
 3. The Hotspot mutation found in tumor sample, suggests high disease risk, which means substantially higher than the general population. *

Notes:

* From the genetic perspective, high disease risk indicates the presence of mutated alleles related to susceptible cancer genes. These mutated alleles may also be present in the genome of your parents, children, brothers and sisters. It is recommended to check their genetic background as well, which is beneficial for early diagnostic and disease prevention.

[**Please check our "50 Cancer Genes" webpage for links to detailed description of these 50 cancer genes at NCBI.](#)

中文提示：

* 每个肿瘤相关基因的介绍, 参考 “<http://www.batj.net/images/gene-50.pdf>”

** **Hotspot** 是经过研究，该基因位点的突变是与肿瘤密切相关。

*** 从遗传角度分析, 高风险疾病表示患病风险高于普通人群, 说明您体内存在与这些疾病发生相关的高风险的易感基因. 这些易感基因也可能存在于您的父母, 子女, 兄弟姐妹的遗传物质中, 建议进行相关基因检测, 以便及早进行针对性预防.