组织FGFR基因突变检测报告

**FGFR Gene Mutation Detection Report for Tissue**

**方案编号 Protocol：****42756493BLC3004(TAR-210)**

**实验室项目编号Laboratory Project ID：XW0417**

 **送检信息 General Information**

|  |  |
| --- | --- |
| **受试者信息 Subject Information** | |
| **中心编号** Site ID： **{{sample.site\_ID}}** | **受试者编号** Subject ID： **{{sample.subject\_ID}}** |
| **出生年份** Year of Birth**：{{sample.birthday}}** | **病理诊断**Pathological Diagnosis：**{{sample.pathol\_diagn}}** |
| **肿瘤类型**Tumor Type**：{{sample.specimen\_type}}** | **取材部位**Anatomic Site：**{{sample.anatomic\_site}}** |
| **样本信息 Specimen Information** | |
| **样本编号**Sample ID**：{{sample.specimen\_parent\_id}}** | **样本类型**Sample Type**：组织切片** Tissue slides |
| **采集日期**Sample Collection Date**：{{sample.tissue\_collection\_date|replace(“-”, ”.”)}}** | **切片日期**Slide Section Date**：{{sample.section\_date|replace(“-”, ”.”)}}** |
| **接收日期**Reception Date**：{{sample.tissue\_date\_received|replace(“-”, ”.”)}}** | **报告日期**Report Date**：{{sample.report\_date|replace(“-”, ”.”)}}** |

**检测结果Testing Results**

在本研究中利用的检测方法是基于DNA和RNA水平, 检测*FGFR*基因突变及融合。突变的检测范围为 *FGFR1/FGFR2/FGFR3/FGFR4* 基因全外显子区域及外显子端部≤5bp 的内含子区域；融合突变检测范围为包含*FGFR1/FGFR2/FGFR3/FGFR4* 激酶区的融合类型。本报告基于42756493BLC3004(MoonRISe-1)研究方案的入组标准仅报告*FGFR2/FGFR3*基因突变及融合结果。

The assay utilized in this study detects *FGFR* gene mutations and fusions at both DNA and RNA levels. The mutation (SNV/Indel) detection scope includes the entire exon sequences and intronic regions within 5 base pairs of the exon termini for SNV/Indel types. The fusion mutation detection focuses on fusions that involve the kinase domains of *FGFR1/FGFR2/FGFR3/FGFR4*. The report focuses solely on the *FGFR2/FGFR3* gene variant and fusion results, in accordance with inclusion criteria of the study 42756493BLC3004(MoonRISe-1).

{%p if qc.dna\_data\_qc.final == “F”%}

* 入组标准基因融合结果Gene fusion results as per the inclusion criteria:

|  |  |  |  |
| --- | --- | --- | --- |
| **基因名称**  **Gene** | **融合名称**  **Fusion ID** | **融合类型**  **Fusion Type** | **检测结果**  **Test Result** |
| *FGFR2* | FGFR2-BICC1 | FGFR2:exon17-BICC1:exon3 | N/A |
| FGFR2-CASP7 | FGFR2:exon17-CASP7:exon2 | N/A |
| *FGFR3* | FGFR3-TACC3v1 | FGFR3:exon17-TACC3:exon11 | N/A |
| FGFR3-TACC3 v3 | FGFR3:exon17-TACC3:exon10 | N/A |
| FGFR3-BAIAP2L1 | FGFR3:exon17-BAIAP2L1:exon2 | N/A |

* 入组标准基因突变结果Gene variant results as per the inclusion criteria：

|  |  |  |  |
| --- | --- | --- | --- |
| **基因名称**  **Gene** | **氨基酸突变**  **CDS Mutation** | **核酸变异**  **Amino Acid Variant** | **检测结果**  **Test Result** |
| *FGFR3* | p.R248C | c.742C>T | N/A |
| p.G370C | c.1108G>T | N/A |
| p.S249C | c.746C>G | N/A |
| p.Y373C | c.1118A>G | N/A |

{%p else%}

* 入组标准基因融合结果Gene fusion results as per the inclusion criteria:

|  |  |  |  |
| --- | --- | --- | --- |
| **基因名称**  **Gene** | **融合名称**  **Fusion ID** | **融合类型**  **Fusion Type** | **检测结果**  **Test Result** |
| *FGFR2* | FGFR2-BICC1 | FGFR2:exon17-BICC1:exon3 | {%p if var.special.xw0417.BICC1%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| FGFR2-CASP7 | FGFR2:exon17-CASP7:exon2 | {%p if var.special.xw0417.CASP7%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| *FGFR3* | FGFR3-TACC3v1 | FGFR3:exon17-TACC3:exon11 | {%p if var.special.xw0417.TACC3v1%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| FGFR3-TACC3 v3 | FGFR3:exon17-TACC3:exon10 | {%p if var.special.xw0417.TACC3v3%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| FGFR3-BAIAP2L1 | FGFR3:exon17-BAIAP2L1:exon2 | {%p if var.special.xw0417.BAI%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |

* 入组标准基因突变结果Gene variant results as per the inclusion criteria：

|  |  |  |  |
| --- | --- | --- | --- |
| **基因名称**  **Gene** | **氨基酸突变**  **CDS Mutation** | **核酸变异**  **Amino Acid Variant** | **检测结果**  **Test Result** |
| *FGFR3* | p.R248C | c.742C>T | {%p if var.special.xw0417.R248%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| p.G370C | c.1108G>T | {%p if var.special.xw0417.G370%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| p.S249C | c.746C>G | {%p if var.special.xw0417.S249%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |
| p.Y373C | c.1118A>G | {%p if var.special.xw0417.Y373%}  阳性Positive  {%p else%}  阴性 Negative  {%p endif%} |

{%p endif%}

{%p if qc.dna\_data\_qc.final == “T”%}

{%p if lib\_quality\_control.lib\_dna\_qc.pathology\_qc == “风险” or lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control == “风险” or lib\_quality\_control.rna\_lib\_qc.rna\_qty\_qc == “风险” or lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control == “风险”%}

说明：此样本进行了风险检测，不排除有漏检的可能性。

Note: This sample is tested at risk, and the potential for missed mutation detections is not excluded.

{%p endif%}

{%p endif%}

{%p if qc.dna\_data\_qc.final == “F”%}

说明：此样本数据质控不合格，检测失败。“N/A“表示没有相关突变结果输出。

Note: This sample didn’t pass the data quality control and the test has failed. “N/A” indicate the mutation result is not available.

{%p endif%}

**编制人**Report by**： 复核人**Review by**：**

注：本报告仅针对本次送检标本，该检测为肿瘤患者个体化治疗提供参考，治疗方案由医生决策。

Note: This report is only for the samples received this time. This test provides a reference for precision treatment of cancer patients, and the treatment plan is decided by the physician.

**检测内容Description**

* **检测方法 Assay**

提取FFPE组织样本DNA和RNA，使用厦门艾德生物医药科技股份有限公司的人类*FGFR1/FGFR2/FGFR3/FGFR4*基因突变检测试剂盒(可逆末端终止测序法)，进行文库构建和目标区域捕获和富集，捕获后的文库进行高通量测序。测序完成后，采用厦门艾德“人类 *FGFR1/FGFR2/FGFR3/FGFR4* 基因突变分析软件”对检测数据进行分析，得到相关多基因的检测结果。

DNA and RNA are extracted from formalin-fixed, paraffin-embedded (FFPE) tissue samples. Subsequently, library construction and target region capture and enrichment are performed using the AmoyDx® *FGFR1-4* NGS panel (Reversible Terminator Sequencing) developed by Amoy Diagnostics Co., Ltd. Next Generation Sequencing is then conducted on the captured library. Upon completion of sequencing, the generated data is analyzed using Mutation Analysis Software of Human *FGFR*s Gene (ADXFGFR-tMut) developed by Amoy Diagnostics Co., Ltd to obtain comprehensive mutation (SNV/Indel) and fusion (Fusion) results for the relevant multigene panel.

* **检测范围 Scope of Detection**

*FGFR1、FGFR2、FGFR3、FGFR4*基因融合突变、点突变、插入缺失突变。

*FGFR1, FGFR2, FGFR3, FGFR4* gene fusion, single nucleotide variant, insertion and deletion.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因**  **Gene** | **转录本**  **Transcript** | **外显子**  **Exon** | **检测突变类型**  **Types** | **阳性判断值**  **Positive Cut Off** |
| *FGFR1* | NM\_023110 | 2~18 | 点突变/插入/缺失/融合  SNV/Insertion/Deletion/  Fusion | 1）点突变、插入、缺失：  ①突变比例（Freq）不低于1.8％  ②突变绝对拷贝数(Var\_DS)不低于2  1)SNV/Indel  ①Frequency of variant (Freq)≥1.8％  ②Original reads（Var\_DS）≥2  2）融合突变  融合支持reads数（Uniqreads）≥6  2）Fusion  Unique reads of fusion（Uniqreads）≥6 |
| *FGFR2* | NM\_000141 | 2~18 | 点突变/插入/缺失/融合  SNV/Insertion/Deletion/  Fusion |
| *FGFR3* | NM\_000142 | 2~18 | 点突变/插入/缺失/融合  SNV/Insertion/Deletion/  Fusion |
| *FGFR4* | NM\_213647 | 2~18 | 点突变/插入/缺失/融合  SNV/Insertion/Deletion/  Fusion |

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**质控结果 QC Result**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **质控项QC Items** | | **质控标准限QC Standard Limits** | | | **质控结果**  **QC Result** |
| **合格Pass** | **风险Risk** | **不合格 Fail** |
| 病理质控  Pathology QC | 肿瘤细胞含量  Tumor Content | ≥20% | 1%~20% | ＜1% | {%p if lib\_quality\_control.lib\_dna\_qc.macrodissection\_performed==”是”%}  {{lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed}}  {%p else%}  {{lib\_quality\_control.lib\_dna\_qc.tumor\_content}}  {%p endif%} |
| 核酸质控  Nucleic Acid QC | DNA总量  DNA Amount | ≥30ng | ≥10ng | < 10ng | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_qty%}  {{lib\_quality\_control.lib\_dna\_qc.dna\_qty|replace(“.00”, “”)}}ng  {%p else%}  N/A  {%p endif%} |
| RNA总量  RNA Amount | ≥20ng  (DV200≥30%) | ≥10ng  (DV200≥30%) | < 10ng  (DV200≥30%) | {%p if lib\_quality\_control.rna\_lib\_qc.dv200\_num >=30%}  {{lib\_quality\_control.rna\_lib\_qc.rna\_qty|replace(“.00”, “”)}}ng  {%p else%}  N/A  {%p endif%} |
| ≥200ng  (DV200＜30%) | ≥50ng  (DV200＜30%) | < 50ng  (DV200＜30%) | {%p if lib\_quality\_control.rna\_lib\_qc.dv200\_num <30%}  {{lib\_quality\_control.rna\_lib\_qc.rna\_qty|replace(“.00”, “”)}}ng  {%p else%}  N/A  {%p endif%} |
| 文库质控  Library QC | 捕获前文库总量  Pre-library Amount | ≥450ng | ≥150ng | <150ng | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_qty%}  {{lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_qty|replace(“.00”, “”)}}ng  {%p else%}  N/A  {%p endif%} |
| 捕获后文库浓度  Post-library Con. | ＞2.5ng/μL | N/A | ≤2.5ng/μL | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_concentration%}  {{lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_concentration|replace(“.00”, “”)}} ng/μL  {%p else%}  N/A  {%p endif%} |
| 数据质控  Data QC | Q30 | ≥75% | N/A | <75% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”,”%”)}}  {%p else%}  N/A  {%p endif%} |
| 覆盖度 Coverage | ≥98% | N/A | <98% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cover\_ratio|replace(“.00%”,”%”)}}  {%p else%}  N/A  {%p endif%} |
| 平均有效深度  SSBCDepth | ≥500X | N/A | <500X | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.depth\_ssbc|replace(“.00”,””) }}X  {%p else%}  N/A  {%p endif%} |
| CDS 区覆盖均一性  SSBC100 | ≥99% | N/A | <99% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.ssbc\_100x\_ratio|replace(“.00%”,”%”) }}  {%p else%}  N/A  {%p endif%} |
| RNA来源Reads覆盖深度 RNADP | ≥150X | N/A | <150X | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.depth\_rna\_ctrl|replace(“.00”,””) }}X  {%p else%}  N/A  {%p endif%} |
| DNA来源Reads的覆盖深度 DNADP | ≥200X | N/A | <200X | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.depth\_dna\_ctrl|replace(“.00”,””) }}X  {%p else%}  N/A  {%p endif%} |

备注：N/A表示不适用

Note: N/A indicate not applicable**名词解释 Noun Interpretation**

**DV200**：片段长度大于等于200bp以上RNA的比例

DV200: Fragment length greater than or equal to 200bp of RNA by Agilent 2100

**Q30：**测序的准确率高于99.9%的碱基的比例

Q30: This means that the base call accuracy (i.e., the probability of a correct base call) is 99.9%.

**覆盖度：**检测到的区域占目标区域的比例

Coverage: The proportion of the sequencing data mapped region to the designed target region.

**CDS区覆盖均一性：**目标区域中CDS区覆盖深度大于100×的区域的比例

CDS region uniformity (SSBC100): The proportion of CDS region with over 100x depth coverage in the target region.

**平均有效深度：**对所有reads进行校正后，目标区域每个碱基被覆盖到的次数的平均值

SSBC depth: The average of the depth of all the individual base of target region, after single strand base calibration.

**RNA来源reads覆盖深度：**RNA内参质控点被覆盖到的次数的平均值

RNADP: Average reads counts of unique RNA reads from each internal RNA control loci.

**DNA来源reads的覆盖深度：**DNA内参质控点被覆盖到的次数的平均值

DNADP: Average reads counts of unique DNA reads from each internal DNA control loci.