检测报告

**Testing Report**

**方案编号Protocol ID：61186372NSC2002**

**项目编号Project ID：XW0413**

 **送检信息 General Information**

|  |  |
| --- | --- |
| **受试者信息 Patient Information** | |
| **研究中心**Site Name**: {{sample.site\_name}}** | **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}}** |
| **性 别**Gender**:** **{%if sample.gender==”男”%}☑男Male □女Female{%elif sample.gender==”女”%}□男Male ☑女Female {%else%}□男Male □女Female {%endif%}** |  |
| **样本信息 Specimen Information** | |
| **样本编码**Sample ID**: {{sample.specimen\_parent\_id}}** | **访视周期**Visit**：筛选期** Screening |
| **样本类型**Specimen Type**: 血液** Blood | **样本数量**Specimen Amount**:{{sample.blood\_specimen\_amount}}** |
| **采集日期**Specimen Collection Date**: {{sample.blood\_collection\_date}}** | **接收日期**Reception Date**:{{sample.blood\_date\_received}}** |
| **报告日期**Report Date**: {{sample.report\_date}}** |  |

**备注 Notes:**

{%p if qc and qc.dna\_data\_qc%}

{%p if qc.dna\_data\_qc.final == “T” and lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control == “合格” and lib\_quality\_control.lib\_dna\_qc.dna\_final\_library\_quality\_control == “合格” and lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control == “合格”%}

**□** 本检测报告结果为通过。即关键质控点合格（即数据QC合格）。

This is a PASS report. The key quality control parameters are pass (pass data QC).

{%p else%}

{%p if var.var\_somatic.level\_I + var.var\_somatic.level\_II +var.var\_somatic.level\_III%}

{%p if lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control != “合格”%}

**□**本检测报告结果为合格。此样本提取质控低于通过标准但进行了风险检测。本报告可以确认检测出的基因变异，但是不能确认是否有其它变异未能检出。提示原因可能为本患者cfDNA丰度较低，重新送样可能不会增加成功率。

This is a QUALIFIED report. This sample failed the extraction QC standard，but performed testing at risk. We can confirm the presence of genomic alterations detailed in this report, but we cannot confirm the absence of other alterations. It may be due to low abundance of cfDNA in this sample, re-sending samples may not improve the success rate.

{%p elif lib\_quality\_control.lib\_dna\_qc.dna\_final\_library\_quality\_control != “合格” or lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control != “合格”%}

**□**本检测报告结果为合格。此样本核酸含量不足,文库质控低于通过标准但进行了风险检测。本报告可以确认检测出的基因变异，但是不能确认是否有其它变异未能检出。提示原因可能为本患者cfDNA丰度较低，重新送样可能不会增加成功率。

This is a QUALIFIED report. DNA is insufficient in this sample This sample failed the library QC standard，but performed testing at risk . We can confirm the presence of genomic alterations detailed in this report, but we cannot confirm the absence of other alterations. It may be due to low abundance of cfDNA in this sample, re-sending samples may not improve the success rate.

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

**□** 本检测报告结果为合格。此样本数据QC低于合格标准。本报告可以确认检测出的基因突变，但是不能确认是否有其它突变未能检出。提示原因可能为本患者cfDNA丰度较低，重新送样可能不会增加成功率。

This is a QUALIFIED report. This sample failed data QC standard. We can confirm the presence of genomic alterations detailed in this report, but we cannot confirm the absence of other alterations. It may be due to low abundance of cfDNA in this sample, re-sending samples may not improve the success rate.

{%p endif%}

{%p else%}

{%p if lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control != “合格”%}

**□** 本检测报告结果为失败。本例受试者样本核酸含量不足，未通过抽提质控。提示原因可能为该患者cfDNA丰度很低，重新送样可能不会增加成功率。

This is a FAIL report. cfDNA is insufficient in this sample, and failed extraction QC. It may be due to very low abundance of cfDNA in this sample, re-sending samples may not increase the success rate.

{%p elif lib\_quality\_control.lib\_dna\_qc.dna\_final\_library\_quality\_control != “合格” or lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control != “合格”%}

**□** 本检测报告结果为失败。本例受试者样本核酸含量不足，未通过文库质控。提示原因可能为该患者cfDNA丰度很低，重新送样可能不会增加成功率。

This is a FAIL report. cfDNA is insufficient in this sample, and failed library QC. It may be due to very low abundance of cfDNA in this sample, re-sending samples may not increase the success rate.

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

**□** 本检测报告结果为失败。本例受试者样本核酸含量不足，未通过数据质控。提示原因可能为该患者cfDNA丰度很低，重新送样可能不会增加成功率。

This is a FAIL report. cfDNA is insufficient in this sample, and failed data QC. It may be due to very low abundance of cfDNA in this sample, re-sending samples may not increase the success rate.

{%p endif%}

{%p endif%}

{%p endif%}

{%p else%}

{%p if lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control != “合格”%}

**□** 本检测报告结果为失败。本例受试者样本核酸含量不足，未通过抽提质控。提示原因可能为该患者cfDNA丰度很低，重新送样可能不会增加成功率。

This is a FAIL report. cfDNA is insufficient in this sample, and failed extraction QC. It may be due to very low abundance of cfDNA in this sample, re-sending samples may not increase the success rate.

{%p elif lib\_quality\_control.lib\_dna\_qc.dna\_final\_library\_quality\_control != “合格” or lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control != “合格”%}

**□** 本检测报告结果为失败。本例受试者样本核酸含量不足，未通过文库质控。提示原因可能为该患者cfDNA丰度很低，重新送样可能不会增加成功率。

{%p endif%}

{%p endif%}

**** **检测结果 Results**

本研究基于DNA水平, 检测10个基因的特定变异：点突变、小片段插入缺失、基因融合，包含*EGFR，MET，KRAS，ALK，ROS1，RET，HER2，BRAF，NRAS，PIK3CA*。根据HGRAO批准，本项目仅报告*EGFR*基因变异结果。

This study detect specific mutation in 10 gene at DNA level：SNV, Indel and Fusion. 10 genes including *EGFR，MET，KRAS，ALK，ROS1，RET，HER2，BRAF，NRAS，PIK3CA.* But only report *EGFR* mutation according to HGRAO approval.

* 基因突变检测结果：The gene mutation detected:

{%p if qc and qc.dna\_data\_qc%}

{%p if qc.dna\_data\_qc.final == “T” and lib\_quality\_control.lib\_dna\_qc.total\_dna\_quality\_control == “合格” and lib\_quality\_control.lib\_dna\_qc.dna\_final\_library\_quality\_control == “合格” and lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_quality\_control == “合格”%}

{%p if var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| {%tr for a in var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III%} | | | | | |
| *{{a.gene\_symbol}}* | {{a.EGFR\_tag}} | {{a.gene\_region}} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {{a.freq\_str}} |
| {%tr endfor%} | | | | | |

{%p else%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| *EGFR* | 未检出  Not detected | 未检出  Not detected | 未检出  Not detected | 未检出  Not detected | 未检出  Not detected |

{%p endif%}

{%p else%}

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

{%p if var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| {%tr for a in var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III%} | | | | | |
| *{{a.gene\_symbol}}* | {{a.EGFR\_tag}} | {{a.gene\_region}} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {{a.freq\_str}} |
| {%tr endfor%} | | | | | |

{%p else%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| *EGFR* | N/A | N/A | N/A | N/A | N/A |

{%p endif%}

{%p else%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| *EGFR* | N/A | N/A | N/A | N/A | N/A |

{%p endif%}

{%p endif%}

{%p else%}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **基因变异**  **Genetic Alteration** | **外显子/内含子**  **Exon/Intron** | **碱基变化**  **NT Change** | **氨基酸变化**  **AA Change** | **突变丰度**  **Allele Frequency** |
| *EGFR* | N/A | N/A | N/A | N/A | N/A |

{%p endif%}

**检测人**Test by**： 复核人**Review by**： 审批人**Approve 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 doctor.

**检测内容 Description**

* **检测方法 Method**

样本核酸提取后采用“人类10基因突变联合检测试剂盒（可逆末端终止测序法）”（厦门艾德生物医药科技股份有限公司）进行文库构建和目标区域捕获，测序平台为贝瑞和康NextSeq CN500，使用ADXLC10（版本号：v3.0）对检测数据进行分析。检测*EGFR/ALK/ROS1/RET/KRAS/NRAS/PIK3CA/BRAF/HER2/MET*基因变异。

After the sample nucleic acid extraction, the library construction and target region capture are carried out by the AmoyDx® LC10-Essential NGS Panel (Reversible Terminator Sequencing), the sequencing platform is BerryGenomics NextSeq CN500. Using ADXLC10 (version: v3.0) to analyze testing data. Gene list of the assay is EGFR/ALK/ROS1/RET/KRAS/NRAS/PIK3CA/BRAF/HER2/MET.

|  |  |  |
| --- | --- | --- |
| **试剂盒名称**  **Key Reagent** | **货号**  **Cat. No.** | **批号**  **Lot. No.** |
| 人类10基因突变联合检测试剂盒  （可逆末端终止测序法）  AmoyDx® Essential NGS Panel  (Reversible Terminator Sequencing) | 8.0627402X024I | {{lib\_quality\_control.lib\_dna\_qc.lot\_no\_construction}}(Library Construction);  {{ lib\_quality\_control.lib\_dna\_qc.lot\_no\_capture}}(Hybrid Capture) |

**检测局限性 Limitation of the Test**

1.本项检测无法检测超出上述检测范围的突变。

Mutations of other areas outside the scope of the test cannot be detected**.**

2.本检测仅在DNA水平进行检测，检测的突变类型仅为点突变、小片段插入缺失及融合；不包含其他水平(如RNA水平或蛋白水平)的变异或其他类型的突变。

This test can only detect SNV, Indel and Fusion at DNA level, other types of variant and other levels，such as RNA level and protein level, are not included in this test.

3. 如未检出指定的基因变异，可能由于该样本中该突变的丰度低于本项目的检测限（0.3％）。

Allele frequency of a mutation lower than the Limit of Detection(0.3％) may leads to false-negatives result.

**数据质控结果 QC Result**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **质控内容**  **QC parameter** | | **合格标准**  **QC Standard** | **质控结果**  **QC Result** | **是否合格**  **Pass/Risk/Fail** |
| 提取质控  Extraction QC | 样品DNA总量  DNA Amount | ＞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”, “”)}}  {%p else%}  N/A  {%p endif%} | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_qty%}  {%p if lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num > 10 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 文库质控  Library QC | 捕获前文库总量  Library Input Before Capture | ＞0.5µg | {%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\_num / 1000}}  {%p else%}  N/A  {%p endif%} | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_qty%}  {%p if lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_qty\_num > 500 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 捕获后文库浓度  Library Conc After Capture | ＞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”, “”)}}  {%p else%}  N/A  {%p endif%} | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_concentration%}  {%p if lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_concentration\_num > 2.5 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 数据质控  Output QC | Q30 | ＞75％ | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.cleandata\_q30%}  {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.cleandata\_q30%}  {%p if qc.dna\_data\_qc.cleandata\_q30\_num > 0.75 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 覆盖度  Coverage | ＞98％ | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.cover\_ratio%}  {{qc.dna\_data\_qc.cover\_ratio|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.cover\_ratio%}  {%p if qc.dna\_data\_qc.cover\_ratio\_num > 0.98 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 平均原始深度  Mean Depth | ＞10000X | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.depth\_mean%}  {{qc.dna\_data\_qc.depth\_mean|replace(“.00”, “”)}}  {%p else%}  N/A  {%p endif%} | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.depth\_mean%}  {%p if qc.dna\_data\_qc.depth\_mean\_num > 10000 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |
| 平均有效深度  Mean Effective Depth | ＞1500X | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.depth\_ssbc%}  {{qc.dna\_data\_qc.depth\_ssbc|replace(“.00”, “”)}}  {%p else%}  N/A  {%p endif%} | {%p if qc and qc.dna\_data\_qc and qc.dna\_data\_qc.depth\_ssbc%}  {%p if qc.dna\_data\_qc.depth\_ssbc\_num > 1500 %}  合格Pass  {%p else%}  不合格Fail  {%p endif%}  {%p else%}  N/A  {%p endif%} |

**名词解释 Noun Interpretation**

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.

平均原始深度：目标区域每个碱基被覆盖到的次数的平均值

Mean Depth: Average of the number of times each base in the target area is covered

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

Mean Effective Depth: The average of the depth of all the individual base of target region, after single strand base calibration