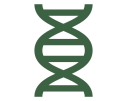
检测报告

|  |  |  |  |
| --- | --- | --- | --- |
| **方案编号：** | **JAB-21822-2001** | **厦维项目编号：** | **XW2404** |

**送检信息**

|  |  |  |  |
| --- | --- | --- | --- |
| 受试者信息 | | | |
| 中心名称 | **{{sample.site\_name}}** | | |
| 受试者筛选号 | **{{sample.subject\_ID}}** | **疾病类型** | **{{sample.primary\_disease}}** |
| 性别 | **{%if sample.gender==”男”%}☑男 □女{%elif sample.gender==”女”%}□男 ☑女 {%else%}□男 □女 {%endif%}** | **出生年份** | **{{sample.birthday}}** |

|  |  |  |  |
| --- | --- | --- | --- |
| 样本信息 | | | |
| 样本编码 | **{{sample.specimen\_parent\_id}}** | **样本类型** | **{{sample.specimen\_type}}** |
| 访视周期 | **{{sample.visit\_name}}** | **采集日期** | **{{sample.tissue\_collection\_date}}** |
| 接收日期 | **{{sample.tissue\_date\_received}}** | **报告日期** | **{{sample.report\_date}}** |

{%p if lib\_quality\_control.lib\_dna\_qc.rna\_qty\_qc != ‘合格’ or lib\_quality\_control.lib\_dna\_qc.rna\_concn\_qc != ‘合格’%}

**备注：RNA提取质控不合格，检测结果仅包含点突变（SNV）、插入缺失（****InDel）、拷贝数扩增（CNA）。**

{%p endif%}

**检测内容**

* **检测方法**

样本核酸提取后采用“人类癌症多基因突变检测试剂盒（高通量测序法）”（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。采用ADXHS-Classic模块进行数据分析，分析版本号ADXHS-Classic v2.0.0。

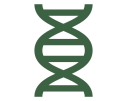
|  |  |
| --- | --- |
| **试剂盒名称** | **货号** |
| 人类癌症多基因突变检测试剂盒（高通量测序法） | 8.06.0046 |

* **检测范围**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **检测点突变与插入缺失变异的基因列表（36个基因）** | | | | | | | |
| *AKT1* | *ALK* | *BRAF* | *CTNNB1* | *DDR2* | *DPYD* | *EGFR* | *ERBB2* |
| *ESR1* | *FGFR1* | *FGFR2* | *FGFR3* | *FGFR4* | *HRAS* | *IDH1* | *IDH2* |
| *KEAP1* | *KIT* | *KRAS* | *MAP2K1* | *MET* | *NFE2L2* | *NRAS* | *NTRK1* |
| *NTRK2* | *NTRK3* | *PDGFRA* | *PIK3CA* | *POLE* | *PTEN* | *RB1* | *RET* |
| *ROS1* | *STK11* | *TP53* | *UGT1A1* |  |  |  |  |
| **检测拷贝数扩增的基因列表（6个基因）** | | | | | | | |
| *CDK4* | *EGFR* | *ERBB2* | *MET* | *MYC* | *NKX2-1* |  |  |
| **检测融合变异的基因列表（10个基因）** | | | | | | | |
| *ALK* | *FGFR1* | *FGFR2* | *FGFR3* | *NRG1* | *NTRK1* | *NTRK2* | *NTRK3* |
| *RET* | *ROS1* |  |  |  |  |  |  |

**检测局限性**

产品的检测性能与样品质量密切相关，样品质控结果为风险时，存在检测灵敏度降低以及检测结果准确性降低的风险。

**检测结果**

* **KRAS G12C入组标准检测结果**

{%p if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_G12C%}

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **变异位点** | **突变丰度** | **检测结果** |
| {%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)|filter\_G12C%} | | | |
| KRAS | G12C | {{a.freq\_str}} | 检出 |
| {%tr endfor%} | | | |

{%p else%}

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **变异位点** | **突变丰度** | **检测结果** |
| 未检出 | 未检出 | 未检出 | 未检出 |

{%p endif%}

* **SNV及InDel检测结果**

{%p if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_snvindel%}

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **基因** | **变异类型** | **转录本** | **外显子/内含子** | **碱基变化** | **氨基酸变化** | **突变丰度** |
| {%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)|filter\_snvindel%} | | | | | | |
| {{a.gene\_symbol}} | {%p if ‘>’ in a.hgvs\_c%}  SNV  {%p else%}  InDel  {%p endif%} | {{a.transcript\_primary}} | {{a.gene\_region}} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {{a.freq\_str}} |
| {%tr endfor%} | | | | | | |

{%p else%}

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **基因** | **变异类型** | **转录本** | **外显子/内含子** | **碱基变化** | **氨基酸变** | **突变丰度** |
| 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 |

{%p endif%}

* **CNA检测结果**

{%p if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_cnv%}

|  |  |  |
| --- | --- | --- |
| **基因** | **变异类型** | **拷贝数** |
| {%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)|filter\_cnv%} | | |
| {{a.gene\_symbol}} | CNA | {{a.cn\_mean}} |
| {%tr endfor%} | | |

{%p else%}

|  |  |  |
| --- | --- | --- |
| **基因** | **变异类型** | **拷贝数** |
| 未检出 | 未检出 | 未检出 |

{%p endif%}

* **Fusion检测结果**

{%p if lib\_quality\_control.lib\_dna\_qc.rna\_qty\_qc == ‘合格’ and lib\_quality\_control.lib\_dna\_qc.rna\_concn\_qc == ‘合格’ and qc.dna\_data\_qc.depth\_rna\_ctrl\_num >= 20%}

{%p if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_sv%}

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **融合类型** | **拷贝数** |
| {%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)|filter\_sv%} | | | |
| *{{a.gene\_symbol}}* | {{a.three\_prime\_transcript}} | {{a.five\_prime\_gene}}:{{a.five\_prime\_cds}}-{{a.three\_prime\_gene}}:{{a.three\_prime\_cds}} | {{a.copies}} copies |
| {%tr endfor%} | | | |

{%p else%}

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **融合类型** | **拷贝数** |
| 未检出 | 未检出 | 未检出 | 未检出 |

{%p endif%}

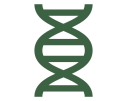
{%p else%}

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **融合类型** | **拷贝数** |
| N/A | N/A | N/A | N/A |

{%p endif%}

**检测人： 复核人： 审批人：**

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**。**

**数据质控**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **质控内容** | | | **质控标准** | | **质控结果** | **合格/风险/失败** |
| **合格** | **风险** |
| 病理质控 | 肿瘤细胞含量 | | ≥20％ | 5%≤肿瘤细胞含量＜20% | {%p if lib\_quality\_control.lib\_dna\_qc.macrodissection\_performed==”是”%}  {{lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed\_num }}  {%p else%}  {{lib\_quality\_control.lib\_dna\_qc.tumor\_content\_num}}  {%p endif%} | {%p if lib\_quality\_control.lib\_dna\_qc.macrodissection\_performed==”是”%}  {%p if lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed\_num >= 0.2%}  合格  {%p elif lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed\_num >= 0.05%}  风险  {%p else%}  失败  {%p endif%}  {%p else%}  {%p if lib\_quality\_control.lib\_dna\_qc.tumor\_content\_num >= 0.2%}  合格  {%p elif lib\_quality\_control.lib\_dna\_qc.tumor\_content\_num >= 0.05%}  风险  {%p else%}  失败  {%p endif%}  {%p endif%} |
| 提取质控 | 样本DNA | 总量 | ≥50ng | / | {{lib\_quality\_control.lib\_dna\_qc.dna\_qty|replace(“.00”, “”)}} | {%p if lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num>=50%}  合格  {%p else%}  失败  {%p endif%} |
| 浓度 | ≥6.25ng/µL | / | {{lib\_quality\_control.lib\_dna\_qc.dna\_concn|replace(“.00”, “”)}} | {%p if lib\_quality\_control.lib\_dna\_qc.dna\_concn\_num>=6.25%}  合格  {%p else%}  失败  {%p endif%} |
| 样本RNA | 总量 | ≥30ng | / | {{lib\_quality\_control.lib\_dna\_qc.rna\_qty|replace(“.00”,””)}} | {%p if lib\_quality\_control.lib\_dna\_qc.rna\_qty\_num>=30%}  合格  {%p else%}  失败  {%p endif%} |
| 浓度 | ≥4ng/µL | / | {{lib\_quality\_control.lib\_dna\_qc.rna\_concn|replace(“.00”,””)}} | {%p if lib\_quality\_control.lib\_dna\_qc.rna\_concn\_num>=4%}  合格  {%p else%}  失败  {%p endif%} |
| 文库质控 | 文库浓度 | | ≥10ng/µL | 5ng/µL≤文库浓度＜10ng/µL | {{lib\_quality\_control.lib\_dna\_qc.library\_concn|replace(“.00”,””)}} | {%p if lib\_quality\_control.lib\_dna\_qc.library\_concn\_num>=10%}  合格  {%p elif lib\_quality\_control.lib\_dna\_qc.library\_concn\_num>=5%}  风险  {%p else%}  失败  {%p endif%} |
| 数据质控 | Q30 | | ＞75％ | / | {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”, “%”)}} | {%p if qc.dna\_data\_qc.cleandata\_q30\_num>=0.75%}  合格  {%p else%}  失败  {%p endif%} |
| DNA文库平均有效深度 | | ≥400X | / | {{qc.dna\_data\_qc.depth\_ssbc|replace(“.00”,””)}} | {%p if qc.dna\_data\_qc.depth\_ssbc\_num>=400%}  合格  {%p else%}  失败  {%p endif%} |
| RNA内参拷贝数 | | ≥20X | / | {{qc.dna\_data\_qc.depth\_rna\_ctrl|replace(“.00”,””)}} | {%p if qc.dna\_data\_qc.depth\_rna\_ctrl\_num>=20%}  合格  {%p else%}  失败  {%p endif%} |

{%p if lib\_quality\_control.lib\_dna\_qc.macrodissection\_performed == ’是’%}

{%p if lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed\_num >= 0.2 and qc.dna\_data\_qc.final == ’T’ and lib\_quality\_control.lib\_dna\_qc.library\_concn\_num>=10 and lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num>=50 and lib\_quality\_control.lib\_dna\_qc.dna\_concn\_num>=6.25 and lib\_quality\_control.lib\_dna\_qc.rna\_qty\_num>=30 and lib\_quality\_control.lib\_dna\_qc.rna\_concn\_num>=4%}

本检测报告结果为：☑通过Passed □合格Qualified □失败Failed

{%p elif lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed\_num < 0.05 or qc.dna\_data\_qc.final != ’T’ or lib\_quality\_control.lib\_dna\_qc.library\_concn\_num < 5 or lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num < 50 or lib\_quality\_control.lib\_dna\_qc.dna\_concn\_num < 6.25 or lib\_quality\_control.lib\_dna\_qc.rna\_qty\_num < 30 or lib\_quality\_control.lib\_dna\_qc.rna\_concn\_num < 4%}

本检测报告结果为：□通过Passed □合格Qualified ☑失败Failed

{%p else%}

本检测报告结果为：□通过Passed ☑合格Qualified □失败Failed

{%p endif%}

{%p else%}

{%p if lib\_quality\_control.lib\_dna\_qc.tumor\_content\_num >= 0.2 and qc.dna\_data\_qc.final == ’T’ and lib\_quality\_control.lib\_dna\_qc.library\_concn\_num>=10 and lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num>=50 and lib\_quality\_control.lib\_dna\_qc.dna\_concn\_num>=6.25 and lib\_quality\_control.lib\_dna\_qc.rna\_qty\_num>=30 and lib\_quality\_control.lib\_dna\_qc.rna\_concn\_num>=4%}

本检测报告结果为：☑通过Passed □合格Qualified □失败Failed

{%p elif lib\_quality\_control.lib\_dna\_qc.tumor\_content\_num < 0.05 or qc.dna\_data\_qc.final != ’T’ or lib\_quality\_control.lib\_dna\_qc.library\_concn\_num < 5 or lib\_quality\_control.lib\_dna\_qc.dna\_qty\_num < 50 or lib\_quality\_control.lib\_dna\_qc.dna\_concn\_num < 6.25 or lib\_quality\_control.lib\_dna\_qc.rna\_qty\_num < 30 or lib\_quality\_control.lib\_dna\_qc.rna\_concn\_num < 4%}

本检测报告结果为：□通过Passed □合格Qualified ☑失败Failed

{%p else%}

本检测报告结果为：□通过Passed ☑合格Qualified □失败Failed

{%p endif%}

{%p endif%}

注：

通过：病理质控、提取质控、文库质控及数据质控均合格。

合格：病理质控合格/风险、提取质控合格、文库质控风险/合格及数据质控均合格。

失败：病理质控或提取质控或文库质控或数据质控质控失败。

* **名词解释**

Q30: 测序的准确率高于99.9%的碱基的比例

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