OncoPro检测报告

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| **方案编号：** | **SHR-1316-307** | **厦维项目编号：** | **XW0248** |

**送检信息**

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| --- | --- | --- | --- |
| 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}}** |

**检测内容**

* **检测方法**

样本核酸提取后采用“人类100基因突变联合检测试剂盒”进行文库构建和目标区域捕获，测序平台为贝瑞和康NextSeq CN500，采用厦门艾德“人类100基因突变分析软件”对检测数据进行分析。

|  |  |
| --- | --- |
| **试剂盒名称** | **货号** |
| 人类100基因突变联合检测试剂盒 | 8.06.0154 |

* **检测范围**

|  |  |  |  |  |  |  |  |  |  |
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| **点突变与插入缺失变异的基因列表（99个基因）** | | | | | | | | | |
| *AKT1* | *AKT2* | *AKT3* | *ALK* | *APC* | *AR* | *ARAF* | *ARID1A* | *ATM* | *B2M* |
| *BARD1* | *BCL2L11* | *BRAF* | *BRCA1* | *BRCA2* | *BRIP1* | *CCND1* | *CCNE1* | *CDH1* | *CDK12* |
| *CDK4* | *CDK6* | *CDKN2A* | *CHEK1* | *CHEK2* | *CTNNB1* | *DDR2* | *EGFR* | *ERBB2* | *ERBB3* |
| *ESR1* | *EZH2* | *FANCA* | *FANCL* | *FBXW7* | *FGFR1* | *FGFR2* | *FGFR3* | *FGFR4* | *GNA11* |
| *GNAQ* | *GNAS* | *HDAC2* | *HRAS* | *IDH1* | *IDH2* | *JAK1* | *JAK2* | *JAK3* | *KDR* |
| *KEAP1* | *KIT* | *KRAS* | *MAP2K1* | *MAP2K2* | *MAPK1* | *MAPK3* | *MDM2* | *MET* | *MLH1* |
| *MSH2* | *MSH6* | *MTAP* | *MTOR* | *MYC* | *NF1* | *NF2* | *NOTCH1* | *NRAS* | *NTRK1* |
| *NTRK2* | *NTRK3* | *PALB2* | *PDGFRA* | *PIK3CA* | *PIK3R1* | *PMS2* | *POLD1* | *POLE* | *PTEN* |
| *PTPN11* | *RAD51B* | *RAD51C* | *RAD51D* | *RAD54L* | *RAF1* | *RB1* | *RET* | *RICTOR* | *ROS1* |
| *SMAD4* | *SMO* | *STAT3* | *STK11* | *TERT* | *TP53* | *TSC1* | *TSC2* | *VHL* |  |
| **拷贝数变异的基因列表（2个基因）** | | | | | | | | | |
| *KEAP1* | *STK11* |  |  |  |  |  |  |  |  |
| **检测融合变异的基因列表（21个基因）** | | | | | | | | | |
| *ALK* | *AR* | *BCL2L11* | *BRAF* | *EGFR* | *ERBB2* | *ESR1* | *FGFR1* | *FGFR2* | *FGFR3* |
| *FGFR4* | *KIT* | *MET* | *NRG1* | *NTRK1* | *NTRK2* | *NTRK3* | *PDGFRA* | *RAF1* | *RET* |
| *ROS1* |  |  |  |  |  |  |  |  |  |

* **检测局限性**

1. 本检测在DNA、RNA水平进行检测，DNA样本检测的突变类型为点突变（SNV）、插入/缺失（InDel）、拷贝数变异（CNV），RNA样本检测融合（Fusion）；不包含其他水平（如蛋白水平）的变异或其他类型的突变。
2. 阴性结果不能完全排除突变或融合基因的存在，样本中肿瘤细胞过少，过度降解或文库中突变或融合的DNA或RNA浓度低于检测限亦可造成阴性结果。
3. 若基因融合发生在高度重复区域中，则存在漏检的可能。

**检测结果**

* **SNV及InDel检测结果**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **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 if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug)|filter\_snvindel%} | | | | | | |
| {%tr for a in (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug)|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%} | | | | | | |
| {%tr else%} | | | | | | |
| 未检出相关变异 | | | | | | |
| {%tr endif%} | | | | | | |

* **CNV检测结果**

**{%p if qc.dna\_data\_qc.cnv\_noise and qc.dna\_data\_qc.cnv\_noise\_num<=0.25%}**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **基因** | **转录本** | | **拷贝数** | | | **突变类型** |
| {%tr if (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug)|filter\_cnv%} | | | | | | |
| {%tr for a in (var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug)|filter\_cnv%} | | | | | | |
| *{{a.gene\_symbol}}* | {%p if a.gene\_symbol==’STK11’%}  NM\_000455.5  {%p else%}  NM\_203500.2  {%p endif%} | | {{a.cn\_mean}} | | | {{a.cnv\_type}} |
| {%tr endfor%} | | | | | | |
| {%tr else%} | | | | | | |
| 未检出 | | 未检出 | | 未检出 | 未检出 | |
| {%tr endif%} | | | | | | |

**{%p elif not qc.dna\_data\_qc.cnv\_noise%}**

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **拷贝数** | **突变类型** |
| 未检出 | 未检出 | 未检出 | 未检出 |

**{%p else%}**

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **拷贝数** | **突变类型** |
| N/A | N/A | N/A | N/A |

**{%p endif%}**

* **Fusion检测结果**

|  |  |  |  |
| --- | --- | --- | --- |
| **基因** | **转录本** | **融合类型** | **拷贝数/突变丰度** |
| {%tr if (var.var\_somatic\_rna\_sv.level\_I+var.var\_somatic\_rna\_sv.level\_II+var.var\_somatic\_rna\_sv.level\_onco\_nodrug)|filter\_sv%} | | | |
| {%tr for a in (var.var\_somatic\_rna\_sv.level\_I+var.var\_somatic\_rna\_sv.level\_II+var.var\_somatic\_rna\_sv.level\_onco\_nodrug)|filter\_sv%} | | | |
| *{{a.gene\_symbol}}* | {{a.five\_prime\_transcript}}  {{a.three\_prime\_transcript}} | {{a.five\_prime\_gene}}:{{a.five\_prime\_region}}-{{a.three\_prime\_gene}}:{{a.three\_prime\_region}} | {{a.rnasv\_reads}} |
| {%tr endfor%} | | | |
| {%tr else%} | | | |
| 未检出 | 未检出 | 未检出 | 未检出 |
| {%tr endif%} | | | |

**注：“N/A”表示不适用或无法评估。**

**编制人： 复核人：**

注：

1. 本报告仅报告致癌性和疑似致癌性位点。

2. 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** |
| 病理质控 | 肿瘤细胞含量 | | ≥20％ | {%if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.tumor\_content%}{{lib\_quality\_control.lib\_dna\_qc.tumor\_content}}{%else%}N/A{%endif%} |
| 提取质控 | DNA | 浓度 | ≥1.5ng/µL | {%if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_concn%}{{lib\_quality\_control.lib\_dna\_qc.dna\_concn|replace(“.00”,””)}}{%else%}N/A{%endif%} |
| 总量 | ≥50ng | {%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”,””)}}{%else%}N/A{%endif%} |
| RNA | 浓度 | ≥5 ng/µL | {%if lib\_quality\_control and lib\_quality\_control.rna\_lib\_qc and lib\_quality\_control.rna\_lib\_qc.rna\_concn%}{{lib\_quality\_control.rna\_lib\_qc.rna\_concn|replace(“.00”,””)}}{%else%}N/A{%endif%} |
| 总量 | ≥20ng | {%if lib\_quality\_control and lib\_quality\_control.rna\_lib\_qc and lib\_quality\_control.rna\_lib\_qc.rna\_qty%}{{lib\_quality\_control.rna\_lib\_qc.rna\_qty|replace(“.00”,””)}}{%else%}N/A{%endif%} |
| 文库质控 | 预文库浓度 | DNA | ≥16.7 ng/µL | {%if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_concentration%}{{lib\_quality\_control.lib\_dna\_qc.dna\_pre\_library\_concentration|replace(“.00”,””)}} {%else%}N/A{%endif%} |
| RNA | ≥33.3 ng/µL | {%if lib\_quality\_control and lib\_quality\_control.rna\_lib\_qc and lib\_quality\_control.rna\_lib\_qc.rna\_pre\_library\_concentration%}{{lib\_quality\_control.rna\_lib\_qc.rna\_pre\_library\_concentration|replace(“.00”,””)}} {%else%}N/A{%endif%} |
| 捕获文库浓度 | DNA | ≥2.5ng/µL | {%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”,””)}} {%else%}N/A{%endif%} |
| RNA | ≥1.2ng/µL | {%if lib\_quality\_control and lib\_quality\_control.rna\_lib\_qc and lib\_quality\_control.rna\_lib\_qc.rna\_fnl\_library\_concentration%}{{lib\_quality\_control.rna\_lib\_qc.rna\_fnl\_library\_concentration|replace(“.00”,””)}} {%else%}N/A{%endif%} |
| 数据质控 | DNA | Q30 | ＞75％ | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} |
| 覆盖度 | ≥95% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cover\_ratio|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} |
| 有效测序深度 | ≥400x | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.depth\_mean\_uniq|replace(“.00”,””) }}  {%p else%}  N/A  {%p endif%} |
| 有效测序深度≥300x的热点比例 | ≥80% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.coverage\_ratio\_uniq\_hot|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} |
| CNVNoise | ≤0.25 | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cnv\_noise}}  {%p else%}  N/A  {%p endif%} |
| RNA | Q30 | ＞75％ | {%p if qc.rna\_data\_qc %}  {{qc.rna\_data\_qc.cleandata\_q30|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} |
| 内参基因覆盖度 | ≥98% | {%p if qc.rna\_data\_qc%}  {{qc.rna\_data\_qc.rnaref\_cover\_ratio|replace(“.00%”, “%”)}}  {%p else%}  N/A  {%p endif%} |
| 内参基因有效测序深度 | ≥250x | {%p if qc.rna\_data\_qc %}  {{qc.rna\_data\_qc.rnaref\_depth\_mean\_uniq|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |

* **名词解释**

Q30: 测序的准确率高于99.9%的碱基的比例

覆盖度：检测到的区域占目标区域的比例

有效测序深度：目标区域测序去重平均深度

有效测序深度≥300x的热点比例：去重平均深度≥300x的热点区域占目标热点区域的比例

内参基因覆盖度：检测到的RNA内参区域占目标RNA内参区域的比例

内参基因有效测序深度：RNA内参基因测序去重平均深度

CNV背景噪声：同一个基因内探针的归一化深度的波动程度（该项质控仅针对CNV变异检测，当仅有CNVNoise质控不合格时，不影响其他变异类型的检出）