OncoPro NGS检测报告

**OncoPro NGS Test Report**

**方案编号（ Protocol ID ）：FCN-159-003**

**厦维项目代码（ Project ID ）：XW4302**

* **送检信息 General Information**

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| --- | --- | --- | --- |
| **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}}** | | **疾病类型**Disease Type: | **{{sample.primary\_disease}}** |
| **出生年份** Year of Birth:**{{sample.birthday}}** | | **性别**Gender: | {%p if sample.gender==”男”%}  **☑男**Male **□女**Female  {%p else%}  **□男**Male **☑女**Female  {%p endif%} |
| **样本信息 Specimen Information** | | | |
| **样本类型及数量**  Specimen Type Amount: | **{%p if “石蜡” in sample.sample\_type%}**  **☑石蜡组织切片**FFPE：**{{sample.tissue\_specimen\_amount}}**  **{%p else%}**  **□石蜡组织切片**FFPE：  **{%p endif%}** | **{%p if sample.control\_sample\_id %}**  **☑ 全血（2mL/管）**Blood：**{{sample.blood\_specimen\_amount }}**  **{%p else%}**  **□ 全血（2mL/管）**Blood：  **{%p endif%}** | **□ 其它**Other： |
| **样本编号**Sample ID： | **{{sample.specimen\_parent\_id}}** | **接收日期**Reception Date: | **{{sample.receive\_data }}** |
| **组织采集日期**Tissue Collection Date: | **{{sample.tissue\_collection\_date}}** | **切片日期**Slice date: | **{{sample.section\_date}}** |
| **全血采集日期**Blood Collection Date: | **{{sample.blood\_collection\_date}}** | **报告日期**Report Date: | **{{sample.report\_date}}** |
| **检测实验室**Testing laboratory： | **上海厦维医学检验实验室有限公司**Shanghai Xiawei Medical Laboratory Co., Ltd. | | |

* **检测内容 Test Description**
* **检测项目简介Test Introduction**

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| --- | --- |
| **检测方法**  **Detection Method** | 基于目标区域捕获富集的高通量测序 |
| **检测平台**  **Detection Platform** | 贝瑞和康 NextSeq CN500 |
| **检测内容**  **Detection Content** | 采用人类100基因突变联合检测试剂盒（高通量测序法）检测，人类100基因突变软件分析 |
| **报告范围**  **Report Scope** | 检测BRAF V600E突变、KIAA1549-BRAF融合、及NF1突变在内的体细胞和胚系100 基因突变 |
| **试剂盒名称**  **Key Reagent** | 人类100基因突变联合检测试剂盒AmoyDx® OncoPro Solid Tumor Panel |
| **试剂盒货号**  **Cat. No.** | 8.06.0154 |

* **检测结果Test Results**
  + **体系突变结果 Somatic mutation results**

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| --- | --- | --- | --- | --- | --- | --- |
| **基因**  **Gene** | **转录本号Transcript ID** | **外显子**  **Exon** | **突变类型**  **Mutation Type** | **核苷酸变化**  **NT Changes** | **氨基酸变化**  **AA Change** | **突变频率（%）**  **Mutation Frequency** |
| {%tr if qc.dna\_data\_qc and qc.dna\_data\_qc.final != “F”%} | | | | | | |
| {%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}}* | {{a.transcript\_primary}} | {{a.gene\_region}} | {%p if a.type\_cn != “--”%}  {{a.type\_cn}}  {%p else%}  {{a.type|replace(“Intronic”, “内含子区突变”)|replace(“UTR”, “UTR区突变”)|replace(“FlankingRegion5”, “基因上游突变”)|replace(“FlankingRegion3”, “基因下游突变”)}}  {%p endif%} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {{a.freq\_str}} |
| {%tr endfor%} | | | | | | |
| {%tr else%} | | | | | | |
| 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 |
| {%tr endif%} | | | | | | |
| {%tr else%} | | | | | | |
| N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | | | |

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| --- | --- | --- | --- | --- | --- | --- | --- |
| **融合基因**  **Fusion genes** | **转录本号**  **Transcript ID** | **伴侣基因Companion genes** | **伴侣基因转录本Companion genes transcript ID** | **融合类型Fusion Type** | **外显子 Exon** | **断点位置** **Breakpoint Location** | **拷贝数**  **Copies** |
| {%tr if qc.rna\_data\_qc and qc.rna\_data\_qc.final != “F”%} | | | | | | | |
| {%tr if (var.var\_somatic.level\_I + var.var\_somatic.level\_II + var.var\_somatic.level\_onco\_nodrug )|filter\_sv %} | | | | | | | |
| {%tr for a in (var.var\_somatic.level\_I + var.var\_somatic.level\_II + var.var\_somatic.level\_onco\_nodrug)|filter\_sv %} | | | | | | | |
| {%p if “,” in a.gene\_symbol%}  *{{a.five\_prime\_gene}}*  {%p else%}  *{{a.gene\_symbol}}*  {%p endif%} | {%p if “,” in a.gene\_symbol%}  {{a.five\_prime\_transcript}}  {%p else%}  {%p if a.gene\_symbol == a.five\_prime\_gene%}  {{a.five\_prime\_transcript }}  {%p else%}  {{a.three\_prime\_transcript}}  {%p endif%}  {%p endif%} | {%p if “,” in a.gene\_symbol%}  *{{a.three\_prime\_gene}}*  {%p else%}  {%p if a.gene\_symbol == a.five\_prime\_gene%}  *{{a.three\_prime\_gene}}*  {%p else%}  *{{a.five\_prime\_gene}}*  {%p endif%}  {%p endif%} | {%p if “,” in a.gene\_symbol%}  {{a.three\_prime\_transcript}}  {%p else%}  {%p if a.gene\_symbol == a.five\_prime\_gene%}  {{a.three\_prime\_transcript}}  {%p else%}  {{a.five\_prime\_transcript }}  {%p endif%}  {%p endif%} | {{a.five\_prime\_gene}}-{{a.three\_prime\_gene}} | {{a.five\_prime\_gene}}: {{a.five\_prime\_cds}}-{{a.three\_prime\_gene}}: {{a.three\_prime\_cds}} | {{a.five\_prime\_chr}}:{{a.five\_prime\_chr\_pos}}-{{a.three\_prime\_chr}}:{{a.three\_prime\_chr\_pos}} | {{a.rnasv\_reads|replace(“.0”, “”)}} |
| {%tr endfor%} | | | | | | | |
| {%tr else%} | | | | | | | |
| 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 |
| {%tr endif%} | | | | | | | |
| {%tr else%} | | | | | | | |
| N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | | | | |

* + **胚系突变结果 Germline mutation results**

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| --- | --- | --- | --- | --- | --- | --- |
| **基因**  **Gene** | **转录本号Transcript ID** | **外显子**  **Exon** | **突变类型**  **Mutation Type** | **核苷酸变化**  **NT Changes** | **氨基酸变化**  **AA Change** | **纯合/杂合**  **Homozygous**  **/Heterozygous** |
| {%tr if qc.dna\_data\_qc and qc.dna\_data\_qc.final != “F”%} | | | | | | |
| {%tr if var.var\_germline.level\_5 +var.var\_germline.level\_4 %} | | | | | | |
| {%tr for a in var.var\_germline.level\_5 +var.var\_germline.level\_4 %} | | | | | | |
| *{{a.gene\_symbol}}* | {{a.transcript\_primary}} | {{a.gene\_region}} | {%p if a.type\_cn != “--”%}  {{a.type\_cn}}  {%p else%}  {{a.type|replace(“Intronic”, “内含子区突变”)|replace(“UTR”, “UTR区突变”)|replace(“FlankingRegion5”, “基因上游突变”)|replace(“FlankingRegion3”, “基因下游突变”)}}  {%p endif%} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {%p if a.freq|float >= 0.8%}  纯合  {%p else%}  杂合  {%p endif%} |
| {%tr endfor%} | | | | | | |
| {%tr else%} | | | | | | |
| 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 | 未检出 |
| {%tr endif%} | | | | | | |
| {%tr else%} | | | | | | |
| N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | | | |

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| **检测人**  Tested by | **复核人**  Reviewed by | **审批人**  Approved 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.

* **检测局限性 Limitation of the Test**

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| 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.本检测在DNA、RNA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 and RNA level, other types of variant and other levels，such as protein level, are not included in this test. |
| 3. 若基因融合发生在高度重复区域中，则存在漏检的可能。  If gene fusion occurs in highly replicate regions, there is a risk of missed detection. |

* **数据质控结果 QC result**

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| --- | --- | --- | --- | --- |
| **质控内容**  **QC parameter** | | | **合格**  **Pass** | **质控结果**  **QC results** |
| 病理质控\*  Pathology QC | 肿瘤细胞含量Tumor cell content | | ≥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%} |
| <20%，富集后≥20% | {%if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed%}{{lib\_quality\_control.lib\_dna\_qc.tumor\_content\_macrodissection\_performed}}{%else%}N/A{%endif%} |
| 提取质控  Extraction QC | DNA | 浓度Concentration | ≥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Amount | ≥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Concentration | ≥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Amount | ≥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%} |
| 胚系DNA | 总量Amount | ≥60ng | {%if lib\_quality\_control and lib\_quality\_control.control\_lib\_dna\_qc and lib\_quality\_control.control\_lib\_dna\_qc.dna\_qty%}{{lib\_quality\_control.control\_lib\_dna\_qc.dna\_qty|replace(“.00”,””)}}{%else%}N/A{%endif%} |
| 文库质控  Library QC | DNA文库总量  DNA Library Amount | | ≥ 500 ng | {%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”,””)}}{%else%}N/A{%endif%} |
| RNA文库总量  RNA Library Amount | | ≥ 1000 ng | {%if lib\_quality\_control and lib\_quality\_control.rna\_lib\_qc and lib\_quality\_control.rna\_lib\_qc.rna\_pre\_library\_qty%}{{lib\_quality\_control.rna\_lib\_qc.rna\_pre\_library\_qty|replace(“.00”,””)}}{%else%}N/A{%endif%} |
| DNA数据质控  DNA Data QC | Q30 | | >75% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.cleandata\_q30}}  {%p else%}  N/A  {%p endif%} |
| 覆盖度Coverage | | ≥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UNIQDepth | | ≥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的热点占比  CoverageRatioUNIQ300 | | ≥80% | {%p if qc.dna\_data\_qc %}  {{qc.dna\_data\_qc.coverage\_ratio\_uniq\_hot|replace(“.00%”, “%”) }}  {%p else%}  N/A  {%p endif%} |
| RNA数据质控  RNA Data QC | 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  RNARef.-Coverage | | ≥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  RNARef.-UNIQDepth | | ≥250x | {%p if qc.rna\_data\_qc %}  {{qc.rna\_data\_qc.rnaref\_depth\_mean\_uniq|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |

备注：如富集，肿瘤细胞含量报告富集前和富集后的含量。

Notes: We report tumor cell content before and after macro-dissection here if to be enriched.

* **名词解释 Noun Interpretation**

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| **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. |
| **有效测序深度UNIQ Depth：** 目标区域测序去重平均深度。  UNIQ Depth: The average clean depth of sequencing coverages. |
| **有效测序深度≥300x的热点占比：** 去重平均深度≥300x的热点区域占目标热点区域的比例。  Coverage RatioUNIQ300: The proportion of hotspot regions with coverage depth ≥ 300×. |
| **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。  RNARef. -Coverage: The proportion of the RNA reference genes sequencing data mapped region to the designed RNA reference genes target region. |
| **内参基因有效测序深度：**RNA内参基因测序去重平均深度。  RNARef.-UNIQ Depth: The average clean depth of the RNA reference genes. |