NGS检测报告

**NGS Test Report**

 **送检信息 General Information**

|  |  |  |  |
| --- | --- | --- | --- |
| **受试者信息 Patient Information** | | | |
| **研究中心** Site Name：**{{sample.site\_name}}** | | | |
| **疾病类型**Disease Type：**{{sample.primary\_disease}}** | | **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 □女{%else%}□男 ☑女{%endif%}** | |
| **样本信息 Specimen Information** | | | |
| **样本编****号:**  Sample ID | **{{sample.specimen\_parent\_id}}** | **组织类型****:**  Specimen Type: | **{%p if “手术” in sample.specimen\_type%}**  **☑手术**Surgical **活检**Biopsy  **{%p else%}**  **手术**Surgical **☑活检**Biopsy  **{%p endif%}** |
| **采集日期:**  Collection Date | **{{sample.tissue\_collection\_date}}** | **切片日期:**  Slide Section Date | **{{sample.section\_date}}** |
| **接收日期:**  Reception Date | **{{sample.receive\_data }}** | **报告日期:**  Report Date | **{{sample.report\_date}}** |

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**检测结果Results**

* **检测结果总结**Summary of test results

|  |  |
| --- | --- |
| **检测项**  **Test item** | **检测结果**  **Detection results** |
| **基因变异检测结果**  **Gene mutation detection results** | {%if qc.dna\_data\_qc.final!=”F”%}{{[var,sample]|var\_s\_summary}}{%else%}N/A{%endif%} |
| **纯合缺失检测结果**  **Results of homozygous deletion detection** | {{[qc,hd]|filter\_hd\_xw1402}}。 |
| **微卫星状态（MSS/MSI）**  **Microsatellite status** | {%p if qc.dna\_data\_qc.depth\_ssbc\_num>=400%}  {%if msi.var\_id==”MSS”%}微卫星稳定（MSS）{%else%}微卫星不稳定（MSI-H）{%endif%}  {%p else%}  N/A  {%p endif%} |

**注：**

1. 变异结果中仅统计与临床意义相关的致癌性/疑似致癌性、致病性/疑似致病性和意义不明变异。

Only likely oncogenic, pathogenic or likely pathogenic, and uncertain significance variants are included in the variant interpretation results.

1. DNA用于检测点突变、插入/缺失、微卫星不稳定性、拷贝数扩增和纯合缺失变异；RNA用于检测融合和MET exon14 skipping 等；

DNA is used to detect Single Nucleotide Variations (SNV), Insertion and Deletion (InDel), Microsatellite Instability (MSI), Copy Number Amplification (CNA) and Homozygous Deletion (HD) variations; RNA is used to detect fusions and MET exon 14 skipping.

1. 检测微卫星不稳定状态时只区分MSI-H 和非 MSI-H，不能区分 MSI-L 和MSS 样本。

When detecting microsatellite Instability (MSI), the results can only distinguish between MSI-high (MSI-H) and non-MSI-high (non-MSI-H) samples; it does not differentiate between MSI-low (MSI-L) and microsatellite stable (MSS) samples.

1. 检出点突变和插入缺失时提示变异频率，指基因位点突变型占野生型和突变型之和的比例；检出拷贝数变异时提示拷贝数，正常细胞中基因拷贝数为2；检出融合或MET exon14 skipping变异时提示拷贝数，这里指测序数据中支持基因融合或MET exon14 skipping变异的reads数；

When identifying Single Nucleotide Variations (SNV) and Insertion and Deletion (InDel), the report includes the mutation frequency, which is the ratio of mutated alleles to the total of both wild-type and mutated alleles at specific gene loci. For Copy Number Amplification (CNA), the report indicates the copy number, with normal cells typically having two copies of each gene. When detecting gene fusions or MET exon 14 skipping mutations, the report provides the number of reads supporting these alterations in the sequencing data.

* **详细检测结果** Detailed test results

1. **基因变异检测结果**Gene variation results

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因名称**  **Gene** | **变异类型**  **Variation type** | **检测结果**  **Detection result** | **频率/拷贝数Frequency/Copies** | **临床解读**  **Clinical Interpretation** |
| {%tr if qc.dna\_data\_qc.final!=”F”%} | | | | |
| {%tr if var.var\_somatic.level\_I+var.var\_somatic.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III%} | | | | |
| {%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}}* | {%p if a.bio\_category==”Snvindel”%}  {%p if a.judge\_mergeMET==”yes”%}  SNV/Indel  MET exon14 skipping  {%p else%}  SNV/Indel  {%p endif%}  {%p elif a.bio\_category==”Cnv”%}  CNA  {%p elif a.bio\_category==”Sv”%}  {%p if a.five\_prime\_gene==”MET” and a.three\_prime\_gene==”MET”%}  MET exon14 skipping  {%p else%}  Fusion  {%p endif%}  {%p endif%} | {%p if a.bio\_category==”Snvindel”%}  {%p if a.judge\_mergeMET==”yes”%}  {{a.transcript\_primary}} {{a.gene\_region}} {{a.hgvs\_c}}{%if a.hgvs\_p!=”p.?”%} {{a.hgvs\_p}}{%endif%}  {{a.var\_hgvs\_2}}  {%p else%}  {{a.transcript\_primary}} {{a.gene\_region}} {{a.hgvs\_c}}{%if a.hgvs\_p!=”p.?”%} {{a.hgvs\_p}}{%endif%}  {%p endif%}  {%p elif a.bio\_category==”Cnv”%}  扩增  {%p elif a.bio\_category==”Sv”%}  {{a.five\_prime\_gene}}:{{a.five\_prime\_cds}}-{{a.three\_prime\_gene}}:{{a.three\_prime\_cds}}  {{a.five\_prime\_transcript}}/{{a.three\_prime\_transcript}}  {%p endif%} | {%p if a.bio\_category==”Snvindel” and a.judge\_mergeMET==”yes”%}  {{a.freq\_str}}  {{a.freq\_2}}  {%p else%}  {{[a, sample]|freq\_stran}}  {%p endif%} | {%p if a.clinic\_num\_s==5%}  致癌性变异  oncogenic variation {%p elif a.clinic\_num\_s==4%}  疑似致癌性变异  Likely oncogenic variation  {%p else%}  意义不明变异  uncertain significance  {%p endif%} |
| {%tr endfor%} | | | | |
| {%tr else%} | | | | |
| / | / | / | / | / |
| {%tr endif%} | | | | |
| {%tr else%} | | | | |
| N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | |

1. **纯合缺失检测结果**Homozygous Deletion test results

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因**  **Gene** | **外显子**  **Exon** | | | **结果判定**  **Result** **Judgment** |
| {%tr if hd%} | | | | |
| {%tr if qc.dna\_data\_qc.snp\_cover\_ratio\_num >= 0.9%} | | | | |
| {%tr for a in hd%} | | | | |
| {%tr if a.var\_auto\_result==”T”%} | | | | |
| *{{a.gene\_symbol}}* | {{a.region}} | | | 阳性Positive |
| {%tr else%} | | | | |
| *{{a.gene\_symbol}}* | / | | | 阴性Negative |
| {%tr endif%} | | | | |
| {%tr endfor%} | | | | |
| {%tr else%} | | | | |
| *MTAP* | N/A | | | N/A |
| *CDKN2A* | N/A | | | N/A |
| {%tr endif%} | | | | |
| {%tr else%} | | | | |
| {%tr if qc.dna\_data\_qc.snp\_cover\_ratio\_num >= 0.9%} | | | | |
| *MTAP* | | / | 阴性Negative | |
| *CDKN2A* | | / | 阴性Negative | |
| {%tr else%} | | | | |
| *MTAP* | | N/A | N/A | |
| *CDKN2A* | | N/A | N/A | |
| {%tr endif%} | | | | |
| {%tr endif%} | | | | |

**注：**

1. “/”表示阴性，没有相关结果输出；“N/A”表示不适用或无法评估。

"/" indicates negative, no relevant results are output; "N/A" indicates not applicable or unable to assess.

编制人 Reported by**：** 复核人Reviewed by**：**

**注：本报告仅针对本次送检标本。**

Note: This report is only for the samples received this time.

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**质控标准Quality Control Standard**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **质控参数**  **Quality control（QC） content** | | **合格标准**  **Pass quality control standard** | **风险标准**  **Risk quality control standard** | **质控数值**  **Quality control value** |
| 病理评估  Pathology QC | 肿瘤细胞含量占比  Tumor cell content | ≥30% | 5~30% | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and 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  Nucleic acid QC | DNA浓度  Con.（ng/μL） | ≥4.3 | 1.43 ≤DNA浓度Con.<4.3，三管建库reconstruct three-tube libraries with the same index and merge them | {%p 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”,””)}}  {%p else%}  N/A  {%p endif%} |
| DNA 总量  Amount (ng) | ≥30 | N/A | {%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%} |
| RNA浓度  Con.（ng/μL） | ≥4 | N/A | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.rna\_concn%}  {{lib\_quality\_control.lib\_dna\_qc.rna\_concn|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |
| RNA 总量  Amount (ng) | ≥30 | N/A | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.rna\_qty%}  {{lib\_quality\_control.lib\_dna\_qc.rna\_qty|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |
| 文库  library QC | 文库浓度  library Con.  （ng/μL） | ≥5 | N/A | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.library\_concn%}  {{lib\_quality\_control.lib\_dna\_qc.library\_concn|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |
| 测序数据  Sequencing  Data QC | Clean Q30 | ≥75% | N/A | {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”,”%”)}} |
| 平均有效测序深度（Depth\_CDS） | ≥400× | N/A | {{qc.dna\_data\_qc.depth\_ssbc|replace(“.00”, “”)}}× |
| RNA内参拷贝数（RNA-Control） | ≥10× | N/A | {{qc.dna\_data\_qc.depth\_rna\_ctrl|replace(“.00”, “”)}}× |
| SNP区域50 ×覆盖率（Coverage(50x)\_SNP） | ≥90% | N/A | {{qc.dna\_data\_qc.snp\_cover\_ratio|replace(“.00%”,”%”)}} |

**注：**

1. 点突变、插入/缺失突变、微卫星不稳定性、拷贝数扩增、融合和MET exon14 skipping等变异类型检测要求样本肿瘤细胞含量不低于20%。纯合缺失检测要求样本肿瘤细胞含量不低于30%。

The detection of mutation types such as Single Nucleotide Variations (SNV), Insertion and Deletion (InDel), Microsatellite Instability (MSI), Copy Number Amplification (CNA), Fusion and MET exon14 skipping requires that the tumor cell content in the sample is at least 20%. For detecting Homozygous Deletion (HD), the tumor cell content must be no less than 30%.

1. 点突变、插入/缺失突变、微卫星不稳定性、拷贝数扩增变异的测序数据质控参数为平均有效测序深度，融合和和MET exon14 skipping变异的测序数据质控参数为RNA内参拷贝数，纯合缺失的测序数据质控参数为SNP区域50 ×覆盖率，若变异类型对应的测序数据质控参数的质控数值不合格，则不输出相应变异类型的检测结果。

The quality control parameters for sequencing data related to Single Nucleotide Variations (SNV), Insertion and Deletion (InDel), Microsatellite Instability (MSI), and Copy Number Amplification (CNA) are based on the Depth\_CDS. For the Fusions and MET exon 14 skipping variants, the quality control parameter is the RNA-Control. In the case of Homozygous Deletion (HD), the quality control parameter is the Coverage (50x)\_SNP. If the quality control values for the sequencing data corresponding to these mutation types do not meet the required standards, the detection results for those specific mutation types will not be reported.

1. 样本质控数值低于合格标准时，检测结果可能存在突变位点假阴性结果，以及影响拷贝数扩增、微卫星不稳定性和纯合缺失结果的准确性。

When the quality control values of the sample are below the acceptable standards, the test results may have false negative results for mutation sites, as well as affect the accuracy of Copy Number Amplification (CNA), Microsatellite Instability (MSI), and Homozygous Deletion(HD) results.

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**检测内容 Test Description**

* **检测方法 Test Method**

样本核酸提取后采用“人类实体瘤多基因突变检测试剂盒”进行文库构建，测序平台为ADx-SEQ200 Plus，采用ADx-SEQ200 Plus配套的分析软件对检测数据进行分析。

After nucleic acid extraction, the library is constructed using the "AmoyDx® HANDLE OncoPro NGS Panel" and the sequencing platform is the ADx-SEQ200 Plus. The analysis is performed using the ADx-SEQ200 Plus analysis software.

* **检测局限性 Limitation of the Test**

1. 本检测仅在DNA/RNA水平进行检测，检测的变异类型仅为点突变(SNV)、插入/缺失(InDel)、微卫星不稳定性(MSI)、拷贝数扩增(CNA）、融合、MET exon14 skipping及纯合缺失（HD）；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 Single Nucleotide Variations,Insertion and Deletion, Microsatellite Instability,Copy Number Amplification,Fusion,MET exon14 skipping and Homozygous Deletion at DNA/RNA level, other types of variant and other levels，such as protein level, are not included in this test.

1. 未检出的特定基因变异，可能由于该样本关键质控点低于合格标准。

Failing to meet the key quality control parameter may lead to false-negative result.

1. 未检出的特定基因变异，可能由于该样本中变异的丰度低于本项目的检测限。

Allele frequency of a mutation lower than the Limit of Detection may lead to false-negatives result.