OncoPro检测报告

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| --- | --- | --- | --- |
| **方案编号：** | **HSK46575-101** | **厦维项目编号：** | **XW6002** |

**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：** | **前列腺癌** |
| 性别： | **男** | **出生年份：** | **{{sample.birthday}}** |

|  |  |  |  |
| --- | --- | --- | --- |
| 样本信息 | | | |
| 样本编码： | **{{sample.specimen\_parent\_id}}** | **样本类型：** | **全血** |
| 临床研究分期： | **{{sample.clinical\_trial\_phase}}** | **访视周期：** | **{{sample.visit\_name}}** |
| 采样日期： | **{{sample.blood\_collection\_date}}** | **接收日期：** | **{{sample.blood\_date\_received}}** |
| 报告日期： | **{{sample.report\_date}}** | | |

**检测结果**

* **检测结果总结**

|  |  |
| --- | --- |
| **检测项** | **检测结果** |
| **AR检测结果** | **{%if var.special.tXW6002.AR%}检出*{{var.special.tXW6002.AR}}*{%else%}未检出{%endif%}** |
| **HRR相关基因检测结果** | **{%if var.special.tXW6002.HRR%}检出*{{var.special.tXW6002.HRR}}*{%else%}未检出{%endif%}** |
| **其他基因检测结果** | **{%if var.special.tXW6002.other%}检出*{{var.special.tXW6002.other}}*{%else%}未检出{%endif%}** |

* **详细检测结果**

1. **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** | **变异分类** |
| {%tr if qc.dna\_data\_qc.final!=”F”%} | | | | | | | |
| {%tr if (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_snvindel%} | | | | | | | |
| {%tr for a in (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_snvindel%} | | | | | | | |
| *{{a.gene\_symbol}}* | SNV/Indel | {{a.transcript\_primary}} | {{a.gene\_region}} | {{a.hgvs\_c}} | {{a.hgvs\_p}} | {{[a, sample]|freq\_stran}} | {%p if a.clinic\_num\_s==5%}  I类变异  {%p else%}  II类变异  {%p endif%} |
| {%tr endfor%} | | | | | | | |
| {%tr endif%} | | | | | | | |
| {%tr if (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_snvindel%} | | | | | | | |
| {%tr for b in (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_snvindel%} | | | | | | | |
| *{{b.gene\_symbol}}* | SNV/Indel | {{b.transcript\_primary}} | {{b.gene\_region}} | {{b.hgvs\_c}} | {{b.hgvs\_p}} | {{[b, sample]|freq\_stran}} | III类变异 |
| {%tr endfor%} | | | | | | | |
| {%tr endif%} | | | | | | | |
| {%tr if not (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_snvindel%} | | | | | | | |
| / | / | / | / | / | / | / | / |
| {%tr endif%} | | | | | | | |
| {%tr else%} | | | | | | | |
| N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | | | | |

1. **CNV检测结果**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因** | **突变类型** | **转录本** | **拷贝数/突变丰度** | **变异分类** |
| {%tr if qc.dna\_data\_qc.cnv\_noise\_num<=0.2%} | | | | |
| {%tr if (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_cnv%} | | | | |
| {%tr for a in (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_cnv%} | | | | |
| *{{a.gene\_symbol}}* | {%p if a.cnv\_type==”loss”%}  Loss  {%p else%}  Amp  {%p endif%} | {{a.transcript\_primary}} | {{[a, sample]|freq\_stran}} | {%p if a.clinic\_num\_s==5%}  I类变异  {%p elif a.clinic\_num\_s==4%}  II类变异  {%p endif%} |
| {%tr endfor%} | | | | |
| {%tr endif%} | | | | |
| {%tr if (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_cnv%} | | | | |
| {%tr for b in (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_cnv%} | | | | |
| *{{b.gene\_symbol}}* | {%p if b.cnv\_type==”loss”%}  Loss  {%p else%}  Amp  {%p endif%} | {{b.transcript\_primary}} | {{[b, sample]|freq\_stran}} | III类变异 |
| {%tr endfor%} | | | | |
| {%tr endif%} | | | | |
| {%tr if not (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_cnv%} | | | | |
| / | / | / | / | / |
| {%tr endif%} | | | | |
| {%tr else%} | | | | |
| N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | |

1. **Fusion检测结果**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因** | **融合类型** | **转录本** | **拷贝数/突变丰度** | **变异分类** |
| {%tr if qc.dna\_data\_qc.final!=”F”%} | | | | |
| {%tr if (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_sv%} | | | | |
| {%tr for a in (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II)|filter\_sv%} | | | | |
| *{{a.gene\_symbol}}* | {{a.five\_prime\_gene}}:{{a.five\_prime\_cds}}-{{a.three\_prime\_gene}}:{{a.three\_prime\_cds}} | {{a.five\_prime\_transcript}}/{{a.three\_prime\_transcript}} | {{[a, sample]|freq\_stran}} | {%p if a.clinic\_num\_s==5%}  I类变异  {%p elif a.clinic\_num\_s==4%}  II类变异  {%p endif%} |
| {%tr endfor%} | | | | |
| {%tr endif%} | | | | |
| {%tr if (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_sv%} | | | | |
| {%tr for b in (var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_sv%} | | | | |
| *{{b.gene\_symbol}}* | {{b.five\_prime\_gene}}:{{b.five\_prime\_cds}}-{{b.three\_prime\_gene}}:{{b.three\_prime\_cds}} | {{b.five\_prime\_transcript}}/{{b.three\_prime\_transcript}} | {{[b, sample]|freq\_stran}} | III类变异 |
| {%tr endfor%} | | | | |
| {%tr endif%} | | | | |
| {%tr if not (var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II+var.var\_somatic.level\_onco\_nodrug+var.var\_somatic.level\_III)|filter\_sv%} | | | | |
| / | / | / | / | / |
| {%tr endif%} | | | | |
| {%tr else%} | | | | |
| N/A | N/A | N/A | N/A | N/A |
| {%tr endif%} | | | | |

**注：**

1. 来源于肿瘤组织的变异解读遵循美国病理学会（AMP）、美国临床肿瘤学会（ASCO）和美国病理学家学会（CAP）共同参与制定的《肿瘤变异解读及报告指南（2017年版）》与中国专家共识《二代测序临床报告解读指引》，根据变异在不同癌种中对应的药物敏感性、诊断及预后证据分为四个等级：A级、B级、C级、D级。基因变异按照其临床意义的重要性分为四个等级：I类变异（具有强临床意义，具有A或B级证据）、II类变异（具有潜在临床意义，具有C或D级证据）、III类变异（临床意义不明）和IV类变异（良性和可能良性变异，已知无临床意义）。
2. 变异结果中仅报告I类变异、II类变异和III类变异的检测结果。
3. 变异包含检出的点突变（SNV）、插入/缺失突变（Indel）、拷贝数变异（CNV）和融合（Fusion）。
4. “/”表示未检出，没有相关结果输出；“N/A”表示不适用或无法评估。

* **检出变异的临床意义提示**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **基因** | **检测结果** | | **丰度/拷贝数/基因型** | **临床提示（耐药/敏感，证据等级）** |
| {%tr if var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II%} | | | | |
| {%tr for a in var.var\_for\_regimen.level\_I+var.var\_for\_regimen.level\_II%} | | | | |
| {{a|gene\_symbol}} | {{a|var\_info}} | | {{[a,sample]|freq\_stran}} | {{a|significance\_regimen}} |
| {%tr endfor%} | | | | |
| {%tr else%} | | | | |
| - | - | - | | - |
| {%tr endif%} | | | | |

**注：**

1. 检出变异与临床意义相关性的证据水平分为A、B、C、D四个等级，A级：对应癌种中FDA/NMPA批准或指南推荐的治疗、诊断或预后的相关标志物；B级：专家共识或III/IV期临床试验研究表明对患者肿瘤治疗有敏感或耐药、或具有诊断、预后意义的生物标志物；C级: FDA/NMPA批准或专业指南推荐的在其他癌种对某个治疗方案敏感或耐药的标志物；或者是作为临床试验入组标准的标志物；或者是多个小型研究结果证实具有诊断或预后意义的标志物；D级: 临床前研究表明具有潜在的治疗意义，或基于小型研究或多个案例报告可能作为辅助疾病诊断或预后的标志物（结论未形成共识）。具有明确临床意义的I类变异，对应药物敏感性证据级别为A级和B级；具有潜在临床意义的II类变异，对应药物敏感性证据级别为C级和D级；不会对临床意义尚不明确的III类变异做药物敏感性分析。

* **具有临床意义的基因变异详细解读**

**{%p for a in [var\_brca, var, sample]|somatic\_level12\_inter%}**

|  |  |
| --- | --- |
| **{%p if a.bio\_category==”Snvindel”%}**  ***{{a.gene\_symbol}}* {{a.gene\_region}} {{a.hgvs\_c}}{%if a.hgvs\_p!=”p.?”%} {{a.hgvs\_p}}{%endif%}**  **{%p elif a.bio\_category==”Cnv”%}**  **{%p if a.cnv\_type==”loss”%}**  ***{{a.gene\_symbol}}*缺失**  **{%p else%}**  ***{{a.gene\_symbol}}* 扩增**  **{%p endif%}**  **{%p elif a.bio\_category==”Sv” or a.bio\_category==”PSeqRnaSv”%}**  **{%p if a.five\_prime\_gene == “MET” and a.three\_prime\_gene == “MET”%}**  ***MET* exon14 skipping**  **{%p else%}**  **{%p if “CRC12” in sample.prod\_names or “Classic” in sample.prod\_names%}**  ***{{a.five\_prime\_gene}}* -*{{a.three\_prime\_gene}}* 融合**  **{%p else%}**  ***{{a.five\_prime\_gene}}* : {{a.five\_prime\_cds}}-*{{a.three\_prime\_gene}}* : {{a.three\_prime\_cds}} 融合**  **{%p endif%}**  **{%p endif%}**  **{%p endif%}**  **{%p if (“CRC12” in sample.prod\_names or “Classic” in sample.prod\_names) and a.judge\_mergeMET %}**  **（MET exon14 skipping）**  **{%p endif%}** | |
| **基因介绍** | {%p if “,” in a.gene\_symbol and (a.bio\_category==”Sv” or a.bio\_category == “PSeqRnaSv”)%}  {%p if a.five\_prime\_gene != a.three\_prime\_gene %}  {{a.five\_prime\_gene\_function|e}}  {{a.three\_prime\_gene\_function|e}}  {%p else%}  {{a.five\_prime\_gene\_function|e}}  {%p endif%}  {%p else%}  {{a.gene\_function|e}}  {%p endif%} |
| **变异解读** | {% if (“CRC12” in sample.prod\_names or “Classic” in sample.prod\_names) and a.bio\_category == “Sv”%}{%if a.five\_prime\_gene ==”MET” and a.three\_prime\_gene ==”MET”%}{{a.variant\_desc\_cn|e}}{{a.variant\_interpret\_cn|e}}{%else%}本次检测到融合突变为{{a.five\_prime\_gene}}-{{a.three\_prime\_gene}}融合，融合模式为{{a.var\_desc\_merge}}。{%endif%}{%else%}{{a.variant\_desc\_cn|e}}{{a.variant\_interpret\_cn|e}}{%endif%}{% if (“CRC12” in sample.prod\_names or “Classic” in sample.prod\_names) and a.judge\_mergeMET%}本次实验在RNA水平也检测到MET exon14 skipping。{%endif%} |
| **治疗策略** | {%p if “TC21” in sample.prod\_names%}  {%p if a.evi\_sum.evi\_split and a.evi\_sum.evi\_split.Predictive\_merge %}  {%p if a.evi\_sum.evi\_split.Predictive\_merge%}  {%p for b in a.evi\_sum.evi\_split.Predictive\_merge%}  **{{b.regimen\_name}}：**  {{b.evi\_interpretation|e}}  {%p endfor%}  {%p endif%}  {%p else%}  目前关于该变异的临床治疗实践尚不明确。  {%p endif%}  {%p elif sample.prod\_names in [“BRCA1/BRCA2（全血）”, “BRCA1/BRCA2（组织）”, “BRCA1/BRCA2（组织 全血）”] and “complete” in sample.report\_name %}  FDA或NMPA已批准奥拉帕利等PARP抑制剂用于携带BRCA1和BRCA2基因有害或疑似有害突变（胚系或体细胞）的卵巢癌、乳腺癌、胰腺癌、前列腺癌等肿瘤患者的治疗。  {%p else%}  {%p for b in a.evi\_sum|evi\_sum%}  **{%if b.regimen%}{{b.regimen}}：{%endif%}**  {{b.inter|e}}  {%p endfor%}  {%p endif%} |

{%p endfor%}

{%p if not [var\_brca, var, sample]|somatic\_level12\_inter%}

|  |  |
| --- | --- |
| **-** | |
| **基因介绍** | - |
| **变异解读** | - |
| **治疗策略** | - |

**{%p endif%}**

**注：**

1. 本部分仅对解读为I类-强临床意义、II类-潜在临床意义的变异进行详细解读。

**编制人： 复核人：**

**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**。**

**检测内容**

* **检测方法**

样本核酸提取后采用“人类泛实体瘤液体活检（OncoPro）基因检测试剂盒（高通量测序法）”进行文库构建和目标区域捕获，测序平台为Illumina NovaSeq 6000，采用厦门艾德“ADXOncoProPlus-bDNA”分析模块对检测数据进行分析，并报告全部基因的检测结果。

|  |  |
| --- | --- |
| **试剂盒名称** | **货号** |
| 人类泛实体瘤液体活检（OncoPro）基因检测试剂盒 | 8.06.0170 |

* **检测局限性**

1. 本检测在DNA水平进行检测，检测的突变类型仅为目标区域基因的点突变（SNV）、插入/缺失（InDel）、拷贝数变异（CNV）、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基因的存在，血浆样本中肿瘤游离核酸含量过少，低于检测限亦可造成阴性结果。

**数据质控**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **质控内容** | | **合格标准** | **风险标准** | **质控结果** |
| 提取质控 | cfDNA总量 | ≥30 ng | ≥5 ng | {%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%} |
| cfDNA浓度 | ≥0.6 ng/μL | ≥0.1 ng/μL | {%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%} |
| 文库质控 | 预文库总量 | ≥500 ng | / | {%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|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |
| 捕获文库总量 | ≥75 ng | / | {%p if lib\_quality\_control and lib\_quality\_control.lib\_dna\_qc and lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_qty%}  {{lib\_quality\_control.lib\_dna\_qc.dna\_fnl\_library\_qty|replace(“.00”,””)}}  {%p else%}  N/A  {%p endif%} |
| 数据质控 | Clean Q30 | ≥75% | / | {{qc.dna\_data\_qc.cleandata\_q30|replace(“.00%”,”%”)}} |
| 1000X有效深度覆盖率 | ≥80% | / | {{qc.dna\_data\_qc.cover1000\_ratio|replace(“.00%”,”%”)}} |
| 中位有效测序深度 | ≥2000x | / | {{qc.dna\_data\_qc.depth\_median|replace(“.00”,””)}} |
| CNV背景噪声 | ≤0.2 | / | {{qc.dna\_data\_qc.cnv\_noise}} |

**注：**

1. CNV变异检测的测序数据质控参数为CNV背景噪声，当此质控数值不合格，不输出CNV的检测结果。
2. 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，影响结果的准确性。

* **名词解释**

Clean Q30: 测序的准确率高于99.9%的碱基的比例

1000X有效深度覆盖率：靶向区域内超过uniq深度超过1000X的比例

中位有效测序深度：靶向区域内的中位uniq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）

**检测范围**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **AR和HRR相关基因列表（49个基因）** | | | | | | |
| *AR* | *BRCA1* | *PALB2* | *BRCA2* | *XRCC2* | *RAD51C* | *RAD51D* |
| *RAD51B* | *RAD51* | *ATM* | *NBN* | *RAD50* | *BRIP1* | *MRE11* |
| *BARD1* | *ATR* | *CHEK1* | *XRCC1* | *FANCL* | *TP53* | *POLE* |
| *POLD1* | *SLX4* | *GEN1* | *ABRAXAS1* | *RAD52* | *ERCC1* | *ERCC4* |
| *AURKA* | *RAD54L* | *EGFR* | *FGFR1* | *CHEK2* | *BRAF* | *KRAS* |
| *MAPK1* | *ERCC3* | *FANCD2* | *ERBB2* | *AKT1* | *AKT2* | *FANCM* |
| *TSC2* | *FANCA* | *NPM1* | *TSC1* | *CD274* | *MSH6* | *SETD2* |

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **其他基因列表（103个基因）** | | | | | | |
| *AKT3* | *ALK* | *APC* | *ARAF* | *ARID1A* | *B2M* | *BAP1* |
| *BCL2L11* | *CCND1* | *CCND2* | *CCNE1* | *CDH1* | *CDK12* | *CDK4* |
| *CDK6* | *CDKN2A* | *CDKN2B* | *CTNNB1* | *CYP2D6* | *DDR2* | *DPYD* |
| *EPCAM* | *ERBB3* | *ERBB4* | *ERCC2* | *ESR1* | *EZH2* | *FBXW7* |
| *FGF19* | *FGF3* | *FGFR2* | *FGFR3* | *FGFR4* | *FH* | *GATA3* |
| *GNA11* | *GNAQ* | *GNAS* | *HDAC2* | *HNF1A* | *HRAS* | *IDH1* |
| *IDH2* | *JAK1* | *JAK2* | *JAK3* | *KDR* | *KEAP1* | *KIT* |
| *MAP2K1* | *MAP2K2* | *MAP2K4* | *MAPK3* | *MDM2* | *MDM4* | *MET* |
| *MLH1* | *MPL* | *MSH2* | *MTAP* | *MTOR* | *MUTYH* | *MYC* |
| *MYCN* | *NF1* | *NF2* | *NFE2L2* | *NOTCH1* | *NRAS* | *NRG1* |
| *NTRK1* | *NTRK2* | *NTRK3* | *PDCD1* | *PDGFRA* | *PIK3CA* | *PIK3CB* |
| *PIK3R1* | *PMS2* | *PPARG* | *PTCH1* | *PTEN* | *PTPN11* | *RAF1* |
| *RASA1* | *RB1* | *RET* | *RHEB* | *RHOA* | *RICTOR* | *RIT1* |
| *RNF43* | *ROS1* | *SF3B1* | *SMAD4* | *SMARCA4* | *SMO* | *STK11* |
| *TERT* | *TMPRSS2* | *TSHR* | *UGT1A1* | *VHL* |  |  |