

ctDNA计算流程

循环肿瘤DNA (ctDNA) 是血液中肿瘤衍生的片段化DNA，是一种具有特征性的肿瘤标志物。通过ctDNA的相关检测，可以检测到肿瘤在患者血液中的踪迹信息，同时可以提供患者体内肿瘤的全景数据。ctDNA作为一项非侵入性的检测方法在肿瘤的筛查、预后、治疗等各个方面都具有重要的意义。

存在一些客户缩小gene list筛选突变的情况，这种情况下需要重新计算ctDNA fraction。

1. 重新注释，通过一个**示例json文件**和一个制作好的**file.list**可以完成自动注释任务投递（作者XCW）

```
python /mnt/vol2_ws/test/xcw/scripts/run_json_batch.py
/mnt/vol2/rendong/test/xcw_test/results/mutect2_anno/file.list
/mnt/vol2/rendong/test/xcw_test/results/mutect2_anno/json/test_anno.json
```

json文件内容如下：

```
{
  "InputDataSet": {
    "workflowInput": {
      "pair_or_single.anno_type": "cf",
      "pair_or_single.outdir":
"vol1@xtao:test/wbzhao/test/anno_0528/1926221",
      "pair_or_single.panel": "66",

      "pair_or_single.vcf": "vol1_root@xtao:rendong/project/66gene/1926221/3.somatic_VC/mutect.vcf",

      "pair_or_single.genelist": "vol1@xtao:rendong/project_info/bed_file/50-panel/50gene.list"
    }
  },
  "Name": "MUTECT2_ANNO_test_anno",
  "Pipeline": "MUTECT2_ANNO",
  "Priority": 8
}
```

cf指样本类型为**cfDNA**（组织算不了ctDNA fraction），指定**输出路径**、**测序以及初次分析真实的panel**——pair_or_single.panel，样本的**mutect.vcf**的路径，以及客户指定的**gene list**的位置。

file.list应为如下格式：

```
tumor anno_type outdir panel vcf genelist
1926221 cf test/wbzhao/test/anno_0528/1926221 66 rendong/project/66gene/1926221/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1823339 cf test/wbzhao/test/anno_0528/1823339 66 rendong/project/66gene/1823339/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2001147 cf test/wbzhao/test/anno_0528/2001147 66plus rendong/project/66plusgene/2001147/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2027395 cf test/wbzhao/test/anno_0528/2027395 66 rendong/project/66gene/2027395/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2001755 cf test/wbzhao/test/anno_0528/2001755 642 rendong/project/620geneV3/2001755/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1921629 cf test/wbzhao/test/anno_0528/1921629 66 rendong/project/66gene/1921629/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1924929 cf test/wbzhao/test/anno_0528/1924929 66 rendong/project/66gene/1924929/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1924255 cf test/wbzhao/test/anno_0528/1924255 66 rendong/project/66gene/1924255/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2002715 cf test/wbzhao/test/anno_0528/2002715 66plus rendong/project/66plusgene/2002715/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1929237 cf test/wbzhao/test/anno_0528/1929237 66 rendong/project/66gene/1929237/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1822535 cf test/wbzhao/test/anno_0528/1822535 66 rendong/project/66gene/1822535/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1822539 cf test/wbzhao/test/anno_0528/1822539 66 rendong/project/66gene/1822539/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1924777 cf test/wbzhao/test/anno_0528/1924777 66 rendong/project/66gene/1924777/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2023351 cf test/wbzhao/test/anno_0528/2023351 642 rendong/project/620geneV3/2023351/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1924779 cf test/wbzhao/test/anno_0528/1924779 66 rendong/project/66gene/1924779/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2027107 cf test/wbzhao/test/anno_0528/2027107 642 rendong/project/620geneV3/2027107/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1924437 cf test/wbzhao/test/anno_0528/1924437 642 rendong/project/620geneV3/1924437/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1929577 cf test/wbzhao/test/anno_0528/1929577 642 rendong/project/620geneV3/1929577/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1900981 cf test/wbzhao/test/anno_0528/1900981 66 rendong/project/66gene/1900981/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1929581 cf test/wbzhao/test/anno_0528/1929581 66 rendong/project/66gene/1929581/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
1921855 cf test/wbzhao/test/anno_0528/1921855 66 rendong/project/66gene/1921855/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
2022227 cf test/wbzhao/test/anno_0528/2022227 66 rendong/project/66gene/2022227/3.somatic_VC/mutect.vcf rendong/project_info/bed_file/50-panel/50gene.list
```

2. 通过注释流程产生的**ctdna_pre.xls**计算 ctDNA fraction

```
python
/mnt/vol1/rendong/project_info/pipeline/scripts/docker_contain/wdl_python/BioBin
/ct_dna.py -i
/mnt/vol2_ws/test/wbzhao/test/anno_0528/1926221/3.somatic_VC/ctdna_pre.xls -c
/mnt/vol1_root/rendong/project/66gene/1926221/5.somatic_CNV/1926221.called.seg -
p 50 -st cf -o /mnt/vol2_ws/test/wbzhao/test/anno_0528/1926221/ctdna.xls
```

- -i 输入文件
- -c segment文件
- -p 筛选后的gene list
- -st sample type
- -o 输出文件