Contents

SNV+CNV

```
dragen -f -r ${1} \
   -1 ${2} -2 ${3} --output-directory ${4} --output-file-prefix ${5} \
   --RGID Illumina_RGID --RGSM ${5} \
   --enable-map-align true \
   --enable-map-align-output true \
   --output-format BAM \
   --enable-duplicate-marking true \
   --enable-sort true \
   --enable-variant-caller true \
   --vc-enable-vcf-output true \
   --enable-vcf-compression true \
   --enable-cnv true \
   --cnv-enable-self-normalization true \
```

SV

```
--enable-sv true
```

repeat

```
--repeat-genotype-enable true
--repeat-genotype-specs /opt/edico/repeat-specs/hg19_expanded/
```

gvcf

```
--vc-emit-ref-confidence GVCF
```

DRAGEN-ML:typically removes 30-50% of SNP FPs, with smaller gains on INDELS. FN counts are reduced by 10% or more.

```
--vc-ml-dir=/opt/edico/resources/ml_model/hg19 --vc-ml-enable-recalibration=true
```

Star Allele Caller 星号等位基因：用于药物基因组

```
--enable-star-allele true
```

Targeted callers: 输出.targeted.vcf.gz and .targeted.json

```
--enable-targeted=true
```

High Sensitivity Mode

```
--vc-enable-high-sensitivity-mode=true
```

Pharmacogenomics (PGx) grouping

```
--enable-pgx=true
```

CNV 分析参数

```
--enable-cnv true
--cnv-filter-copy-ratio 0.2     #   The default value is 0.2, leading to calls less than CR=0.8 or
 ↪  greater than CR=1.2.
--cnv-filter-length 10000       #   Specifies the minimum event length in bases at which a reported
 ↪  event is marked as PASS in the output VCF file. The default is 10000
--cnv-filter-qual 10            #   PASS in the output VCF file
```

CNV 解析度

WGS_Coverag_per_Sample	Recommended_Resolution(bp)
5X	10000
10X	5000
>=30X	1000

–*cnv-interval-width* 用来控制解析度，WES 默认是 500，WGS 默认是 1000 该参数在分析是需要设置，如果设置变小会增加分析时间