

# Contents

人类参考基因组	1
1. 参考基因组类型选择	1
2. Illumina DRAGEN Reference Genome	1
3. hash build shell	1
hg19	1
hg38	2

## 人类参考基因组

### 1. 参考基因组类型选择

Human		hg19	hs37d5	hg38	chm13	Recommended Reference Type
Germline	SNV	Yes	Yes	Yes	Yes	Graph
	CNV	Yes	Yes	Yes	Yes*	Graph
	SV	Yes	Yes	Yes	Yes*	Graph
	Expansion Hunter	Yes	Yes	Yes	No	Graph
	Targeted Callers	Yes	Yes	Yes	No	Graph
	RNA	Yes	Yes	Yes	Yes*	Non-Graph
	De Novo	Yes	Yes	Yes	Yes*	Graph
	Joint Genotyping	Yes	Yes	Yes	Yes*	Graph
	Biomarkers (HLA)	Yes	Yes	Yes	Yes*	Graph
	Gvcf Genotyper	Yes	Yes	Yes	Yes*	Graph
Somatic	SNV	Yes	Yes	Yes	Yes*	Non-Graph
	UMI SNV	Yes	Yes	Yes	Yes*	Non-Graph
	CNV	Yes	Yes	Yes	Yes*	Non-Graph
	SV	Yes	Yes	Yes	Yes*	Non-Graph
Methylation	Methylation	Yes	Yes	Yes	No	Non-Graph
Annotation	Nirvana	Yes	Yes	Yes	No	n/a

Figure 1: 人类参考基因组

构建图形的参数: `-ht-apply-graph=true`

### 2. Illumina DRAGEN Reference Genome

[https://sapac.support.illumina.com/sequencing/sequencing\\_software/dragen-bio-it-platform/product\\_files.html](https://sapac.support.illumina.com/sequencing/sequencing_software/dragen-bio-it-platform/product_files.html)

### 3. hash build shell

#### hg19

DNA+CNV+RNA

```
dragen --output-directory /staging/hash_table/human/hg19_CNVRNA/ --build-hash-table true
↪ --ht-reference hg19.fa --ht-alt-liftover /opt/edico/liftover/hg19_alt_liftover.sam --ht-decoys
↪ /opt/edico/liftover/hs_decoys.fa --enable-cnv true --ht-num-thread 40 --ht-build-rna-hashtable true
```

methylation

```
dragen --output-directory /staging/hash_table/human/hg19_methylation/ --build-hash-table true
↪ --ht-reference hg19.fa --ht-alt-liftover /opt/edico/liftover/hg19_alt_liftover.sam --ht-decoys
↪ /opt/edico/liftover/hs_decoys.fa --ht-num-thread 40 --ht-methylated true
↪ --ht-methylated-combined=true
```

## hg38

### DNA+CNV+RNA

```
dragen --output-directory /staging/hash_table/human/hg38_CNVRNA/ --build-hash-table true
↪ --ht-build-rna-hashtable true --enable-cnv true --ht-reference hg38.fa --ht-num-threads 40
↪ --ht-alt-liftover /opt/edico/liftover/bwa-kit_hs38DH_liftover.sam --ht-pop-alt-contigs
↪ /opt/edico/liftover/pop_altContig.fa.gz --ht-pop-alt-liftover
↪ /opt/edico/liftover/pop_liftover.sam.gz --ht-pop-snps /opt/edico/liftover/pop_snps.vcf.gz
```

### methylation

```
dragen --output-directory /staging/hash_table/human/hg38_methylation/ --build-hash-table true
↪ --ht-reference hg38.fa --ht-alt-liftover /opt/edico/liftover/bwa-kit_hs38DH_liftover.sam
↪ --ht-decoys /opt/edico/liftover/hs_decoys.fa --ht-num-thread 40 --ht-methylated true
```