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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

构建图形基因 Graph references 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

3.hash build shell

hg19

DNA+CNV+RNA

```
dragen --output-directory /staging/hash_table/human/hg19_CNV_RNA/ --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_CNV_RNA/ --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
```

4.genome fasta

hg19:https://ilmn-dragen-giab-samples.s3.amazonaws.com/FASTA/hg19.fa

hg38:https://ilmn-dragen-giab-samples.s3.amazonaws.com/FASTA/hg38.fa

hs37d5:https://ilmn-dragen-giab-samples.s3.amazonaws.com/FASTA/hg19.fa