

GENOME BUILDER USING SNPS AND INDELS

Requirements:

First, you will need to parse the VCF file and extract the relevant information. The VCF file contains information about genetic variations, including single nucleotide polymorphisms (SNPs), insertions, deletions, and structural variants. Each line in the file represents a variant, and the fields in the line contain information about the variant, such as its position in the genome, the reference and alternate alleles, and any additional annotations.

Next, you will need to read the reference genome into memory. The reference genome is typically stored in a file in FASTA format, which consists of a series of DNA sequences with associated labels.

Once you have extracted the variants from the VCF file and the reference genome, you can iterate over the variants and apply the necessary edits to the reference genome. For example, if the variant is a SNP, you can simply replace the base at the specified position with the alternate allele. If the variant is an insertion or deletion, you will need to insert or delete the appropriate number of bases at the specified position.

Finally, you can write the edited genome to a new file or return it as a string, depending on your requirements.

Sample files:

If this is a Reference Genome:

>chr1

```
CCCTGCCAGGGCTGCTGGTGATTCTCCACATCCTTAGGCTCCGCGGTGCTTACCTTCAGG
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCCATTT
TTTAAGAGAGGACGAAGGTGAGAGGGAGACTACAATGAAAAGGTTGGGAGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTATG
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
```

>chr2

```
ACCTGCCAGGGCTGCTGGTGATTCTCCACATCCTTAGGCTCCGCGGTGCTTACCTTCAGG
```

```
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCCATT
TTTAAGAGAGGACGAAGGTGAGAGGGAGACTACAATGAAAAGGTTGGGAGGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTATG
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
>chr3
TCCTGCCAGGGCTGCTGGTGATTCTCCACATCCTTAGGCTCCGCGGTGCTTACCTTCAG
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCCATT
TTTAAGAGAGGACGAAGGTGAGAGGGAGACTACAATGAAAAGGTTGGGAGGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTATG
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
```

THIS IS A VCF FILE CONTAINING ALL THE VARIANTS

```
##fileformat=VCFv4.2
##fileDate=20090805
##source=myImputationProgramV3.1
##reference=file:///seq/references/1000GenomesPilot-NCBI36.fasta
##contig=<ID=20,length=62435964,assembly=B36,md5=f126cdf8a6e0c7f379d618ff66beb2da,species="
Homo sapiens",taxonomy=x>
##phasing=partial
##INFO=<ID=NS,Number=1,Type=Integer,Description="Number of Samples With Data">
##INFO=<ID=DP,Number=1,Type=Integer,Description="Total Depth">
##INFO=<ID=AF,Number=A,Type=Float,Description="Allele Frequency">
##INFO=<ID=AA,Number=1,Type=String,Description="Ancestral Allele">
##INFO=<ID=DB,Number=0,Type=Flag,Description="dbSNP membership, build 129">
##INFO=<ID=H2,Number=0,Type=Flag,Description="HapMap2 membership">
##FILTER=<ID=q10,Description="Quality below 10">
##FILTER=<ID=s50,Description="Less than 50% of samples have data">
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
```

```
##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
##FORMAT=<ID=DP,Number=1,Type=Integer,Description="Read Depth">
##FORMAT=<ID=HQ,Number=2,Type=Integer,Description="Haplotype Quality">
#CHROM POS ID REF ALT QUAL FILTER INFO FORMAT NA00001
chr1 3 . C A 100 PASS . GT 0/1
chr1 30 . A GT 100 PASS . GT 0/1
chr1 59 . GG T 100 PASS . GT 0/1
chr1 118 . TTTTTT T 100 PASS . GT 1/0
chr1 240 . G TAC 100 PASS . GT 0/1
chr2 120 . T AC 100 PASS . GT 0/1
chr3 5 . G C 100 PASS . GT 0/1
chr3 60 . G AAAAAA 100 PASS . GT 0/1
```

IF there is a 0/0 found in the sample (NA00001) coloum then that means the alternate is homozygous and it can be skipped else the variant is applied (0/1 , 1/0 , 1/1)

Sometimes there are Ns on the reference. If we encounter an N on the reference. Just continue and don't apply the variant.

VCF

```
##fileformat=VCFv4.2
##contig=<ID=2,length=51304566>
##INFO=<ID=AC,Number=A,Type=Integer,Description="Allele count in genotypes">
##INFO=<ID=AN,Number=1,Type=Integer,Description="Total number of alleles in called genotypes">
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=DP,Number=1,Type=Integer,Description="Read Depth">
##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
#CHROM POS ID REF ALT QUAL FILTER INFO FORMAT SAMPLE1 SAMPLE2 SAMPLE3 SAMPLE4 SAMPLE5 SAMPLE6 SAMPLE7
2 81170 . C T . . AC=9;AN=7424 GT:DP:GQ 0/0:4:12 0/0:3:9 0/1:1:3 0/1:9:24 1/0:4:12 0/0:5:15 0/0:4:12
2 81171 . G A . . AC=6;AN=7446 GT:DP:GQ 0/1:4:12 0/0:3:9 0/0:1:3 0/0:9:24 0/1:4:12 0/1:5:15 0/0:4:12
2 81182 . A G . . AC=5;AN=7506 GT:DP:GQ 0/0:5:15 0/0:4:12 0/0:5:15 0/0:9:24 0/0:4:12 0/0:4:12 0/0:4:12
2 81204 . T G . . AC=2;AN=7542 GT:DP:GQ 1/0:5:15 0/0:9:27 0/0:10:30 0/0:15:39 0/0:9:27 1/0:13:39 0/1:14:42
```

Variant files are tricky so be careful.

This is the successful outcome of after applying the program you created.

>chr1

```
CCATGCCAGGGCTGCTGGTGATTCTCCACGTTTCCTTAGGCTCCGCGGTGCTTACCTTCAT
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCCA
AAGAGAGGACGAAGGTGAGAGGGGAGACTACAATGAAAAGGTTGGGAGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTAT
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
```

>chr2

```
ACCTGCCAGGGCTGCTGGTGATTCTCCACATCCTTAGGCTCCGCGGTGCTTACCTTCAGG
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCATT
TTAAGAGAGGACGAAGGTGAGAGGGGAGACTACAATGAAAAGGTTGGGAGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTATG
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
```

>chr3

```
TCCTCCAGGGCTGCTGGTGATTCTCCACATCCTTAGGCTCCGCGGTGCTTACCTTCAG
ACTCTCCAGTTGTAACCCCTTTGTTGGGATGCCTGGGAGCCAGACAAGGTCACCCATT
TTAAGAGAGGACGAAGGTGAGAGGGGAGACTACAATGAAAAGGTTGGGAGGGGCCCCAGG
CATGGCCCCTGTGTGTGGAAAACACAGGTGACCACCGGCACCCAGACTGTCTACACTATG
CCTCCAGAAGGCACTTTGCCTAGCAACAGGCCTGACCATGCAGCGCTGGTCCAATCTCTC
```

The actual Sequence is very Huge. In billions of bases and Gbs in Size.