TheXValidator

Input and results

Input:

ONT based SV callset using cuteSV

hg37

TrioCanu assemblies.

Method:

For each SV defined by chrom, pos, variant:
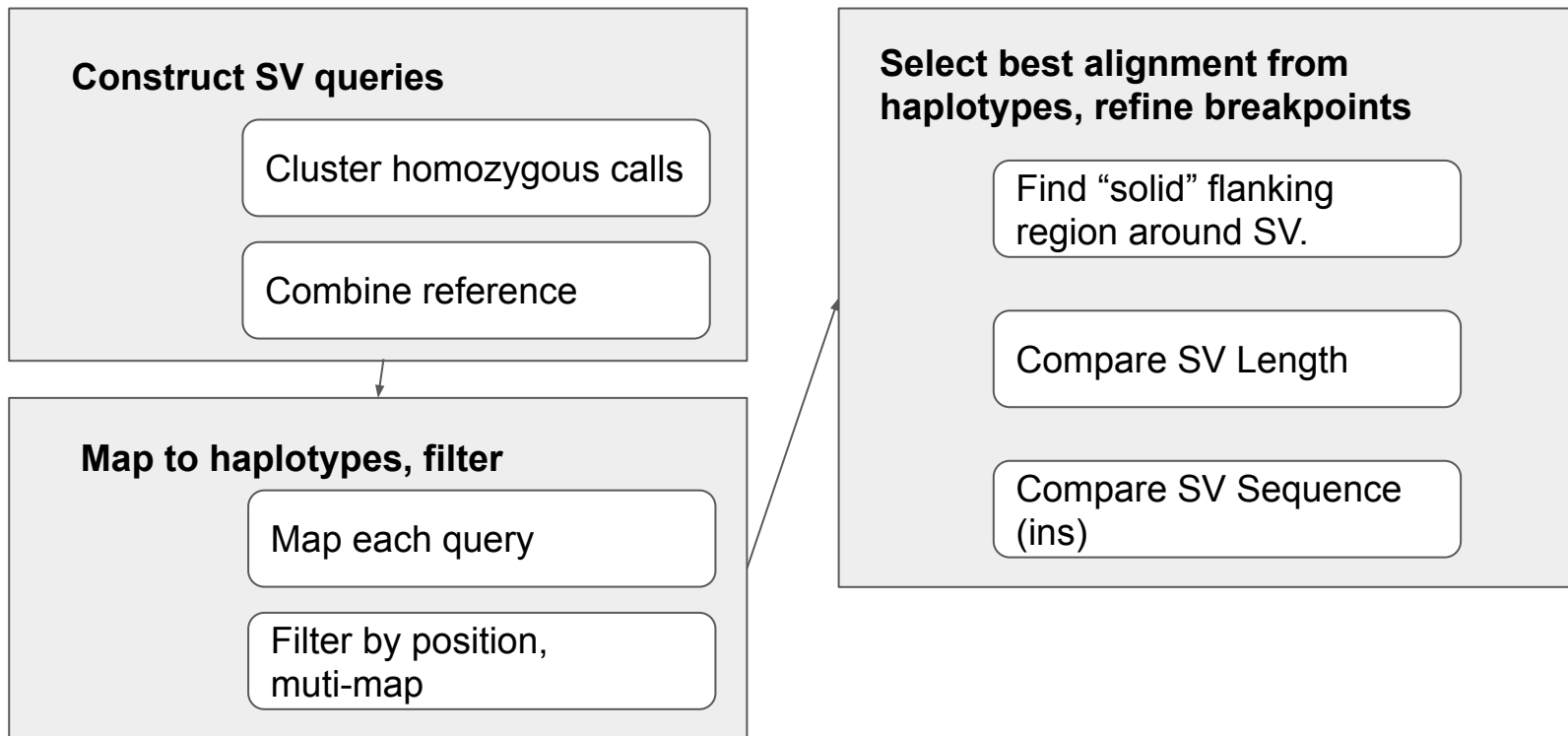
Construct a query as: ref[chrom][pos-prefix:pos] + SV + ref[chrom][pos:pos+suffix]

Query:

prefix	SV	suffix

Map all queries to hap1 and hap2 assemblies.

Workflow

Construct SV queries

Cluster homozygous calls

Combine reference

Map to haplotypes, filter

Map each query

Filter by position, muti-map

Select best alignment from haplotypes, refine breakpoints

Find "solid" flanking region around SV.

Compare SV Length

Compare SV Sequence (ins)

Truvari annotation of variants.