X-SVLen

Input:

ONT based SV callset using cuteSV

Truvari run using default and permissive params

hg37

TrioCanu assemblies

Method:

For each SV defined by chrom, pos, variant:

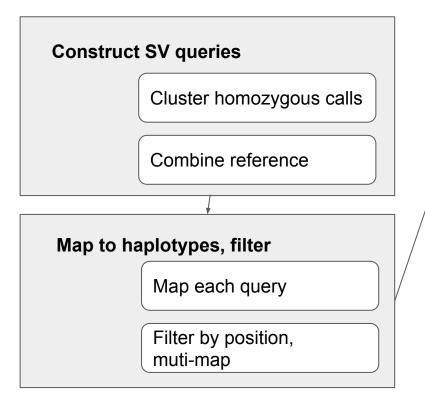
Construct a query as:

ref[chrom][pos-prefix:pos] + SV + ref[chrom][pos:pos+suffix]

prefix SV suffix

Map all queries to hap1 and hap2 assemblies.

Workflow



Select best alignment from haplotypes, refine breakpoints

Find "solid" flanking region around SV.

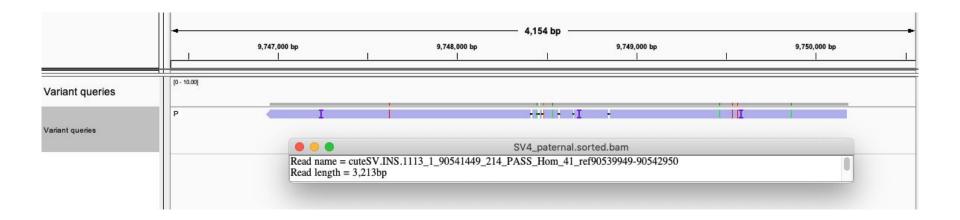
Compare SV Length

Compare SV Sequence (ins)

Assembly calls that overlap contigs.

SVType	INS		DEL	
SVLen	<50	>=50	<50	>=50
number of these calls that overlap				
assembly contigs	16054	17481	30494	6475

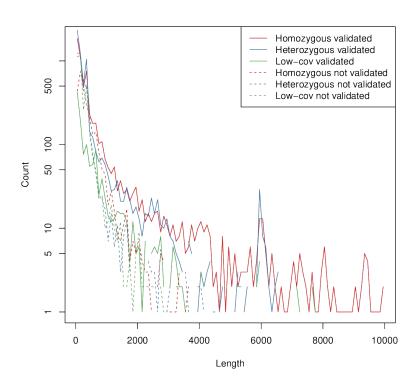
Mapped query sequence



Example difficult to validate call.



Annotation of true positives



Comparison against Truvari

SVType	INS		DEL	
SVLen	<50	>=50	<50	>=50
	3351 FP	2835 FP	880 FP	1571 FP
number of truvari	7200 TP	4095 TP	1925 TP	2099 TP
TP/FP/no-call	15974	80	30196	298
	not_processed	not_processed	not_processed	not_processed
number of	5220 FP	8979 FP	23202 FP	3444 FP
assembly TP/FP/no-call	10834 TP	8502 TP	7292 TP	3031 TP

Thank you!!

Rocio Esteban

Debapriya Sinha

Chunxiao Liao

And all the organizers of the 2020 Hackathon YOU ARE AWESOME