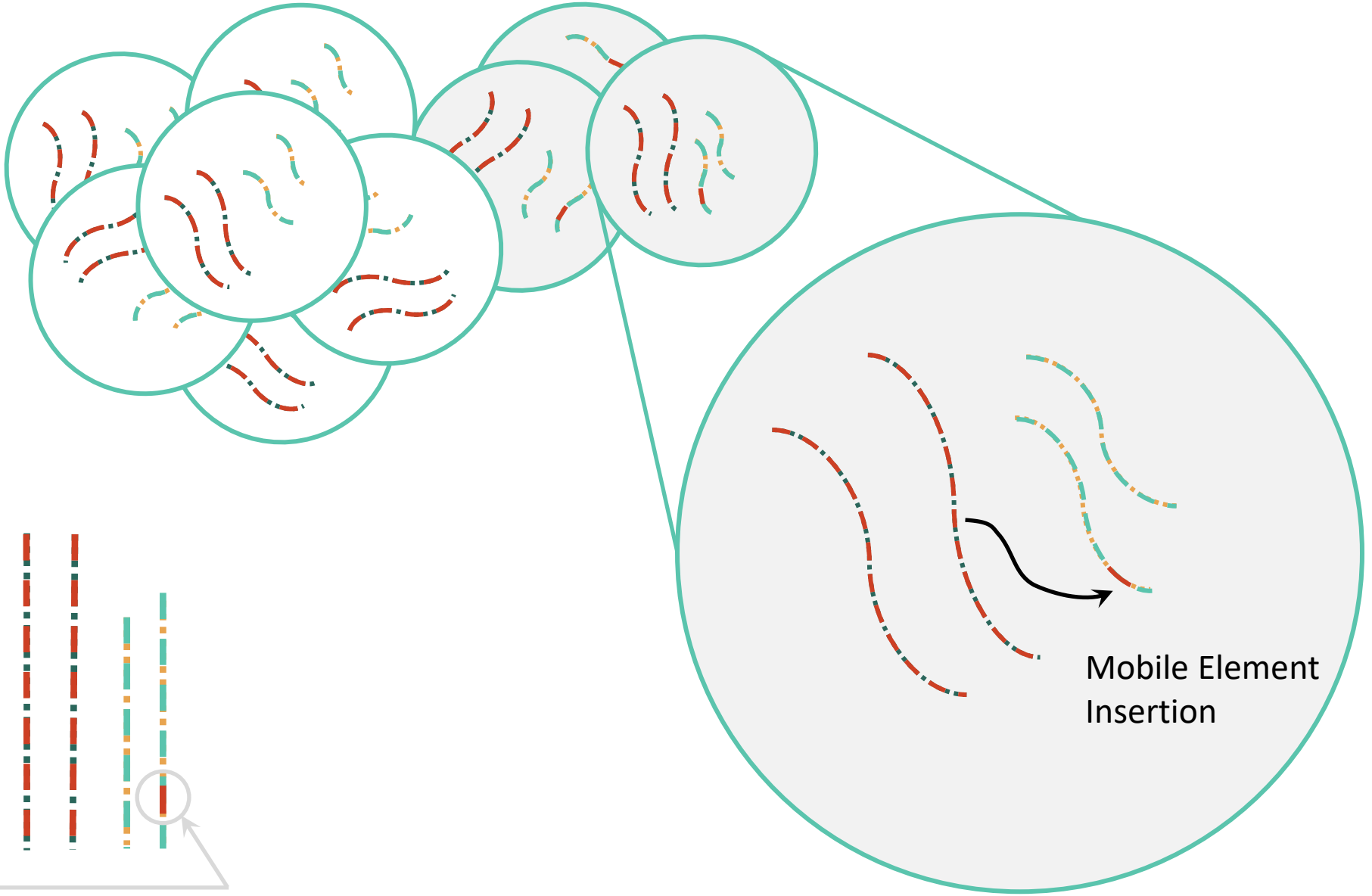
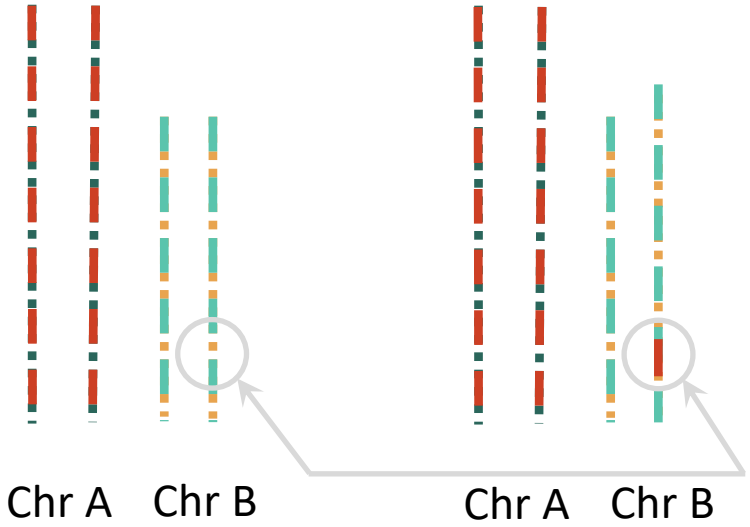
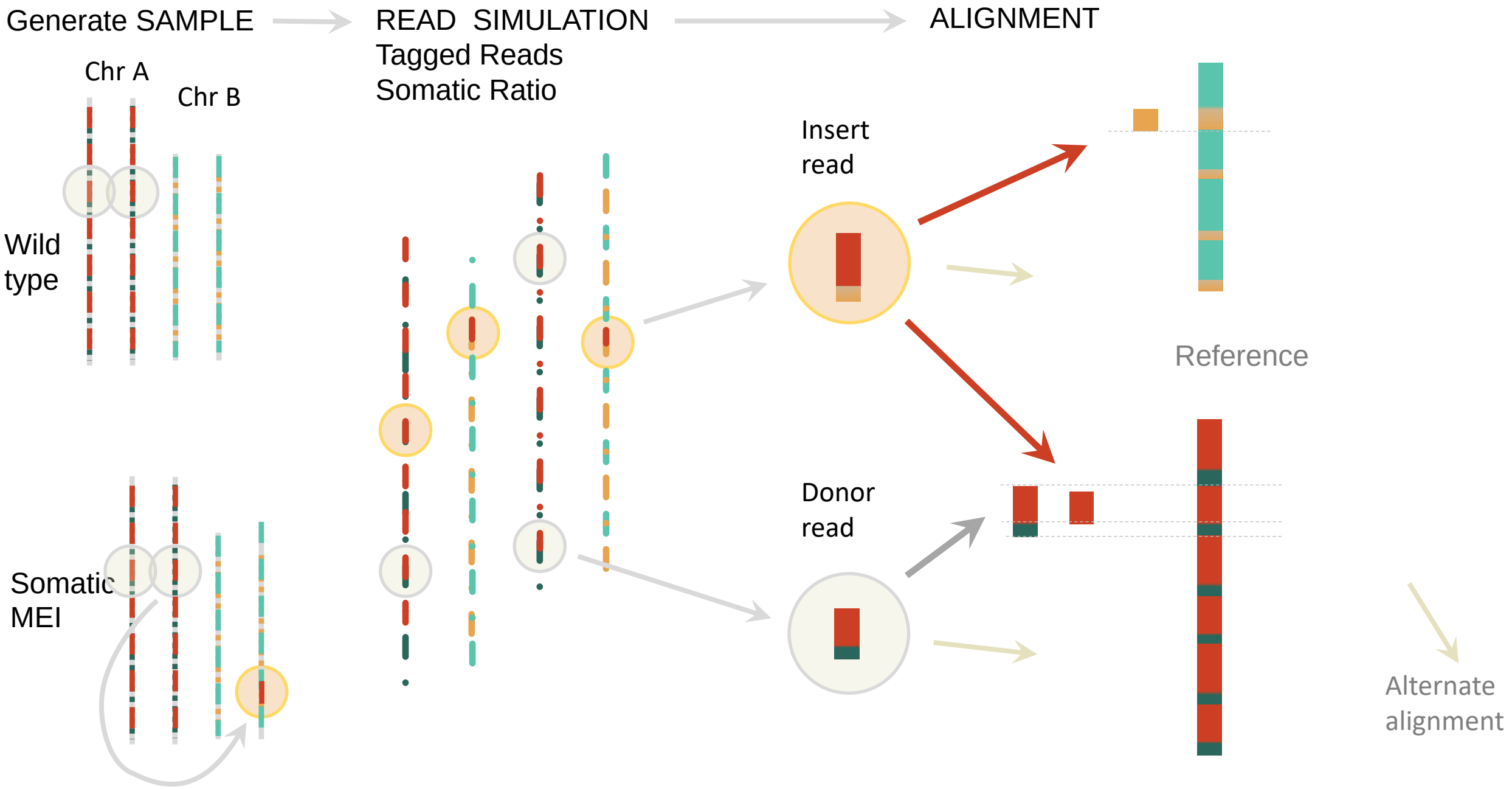
Simulation and Analysis of Somatic Mobile Element Insertion

Sample with
Somatic MEI





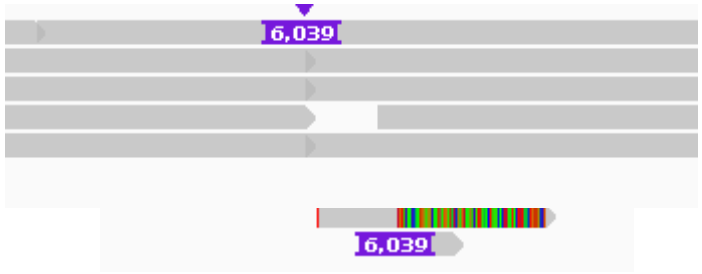
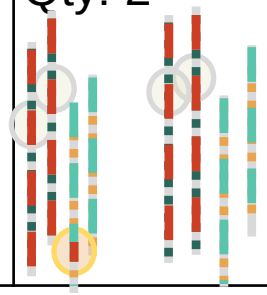

Generate SAMPLE
READ SIMULATION
Tagged Reads
Somatic Ratio
ALIGNMENT
Chr A
Chr B
Wild type
Somatic MEI
Insert read
Donor read
Reference
Alternate alignment

SAMPLE *Human*

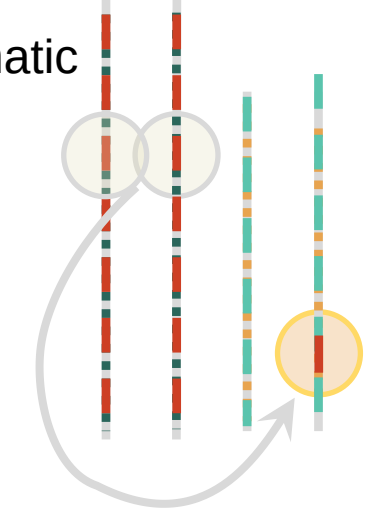
Donor MEI: Chr1 L1HS 118,852,350 6021

Insert Site: ChrX 21,005,740

ALIGNMENT / Mapping + tags!

			LRA	PBMM2
Insertion Site	Somatic Qty: 5	10X		
Donor Site	Somatic Qty: 3 WildType Qty: 2 	6X 4X		Clipping :

Somatic
MEI
10X



		Structural Variant Detector			MEI Classifier	
		Sniffles	SVIM	PBSV	TLDR	xTEA
Aligner	LRA	—	OK	★	OK	—
	PBMM 2	—	dup	OK	—	—

Sniffles does not work well with low coverage.

★ Needs RG tag in BAM file.

Preliminary Data

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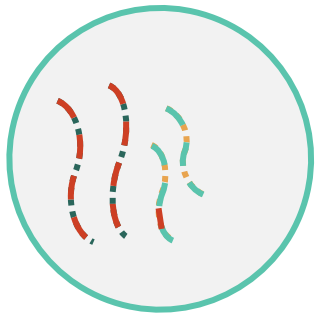
Moving forward investigate:

- Read simulators
- Best combination of tools and classifiers.
- Expanded data sets



Thanks to all from the Adelson Lab.

References:

Source for full length active L1 in human genome:

Characterization of LINE-1 transposons in a human genome at allelic resolution

Lei Yang, Genevieve A. Metzger, Richard N. McLaughlin Jr.

doi: https://doi.org/10.1101/594200

Plus more to come…… tools papers