Improving Genome Assemblies With Syntenic Alignments

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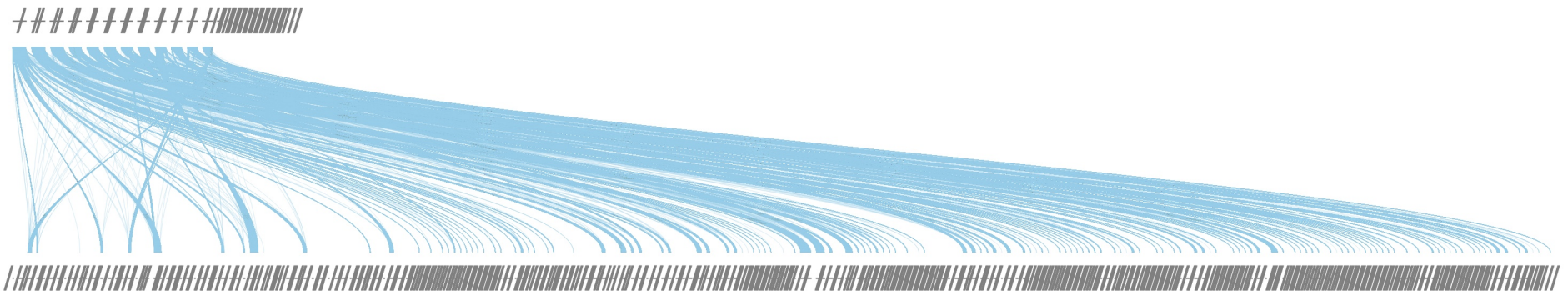
Context

- Synteny alignments provide signal for reference-guided contig scaffolding



Context

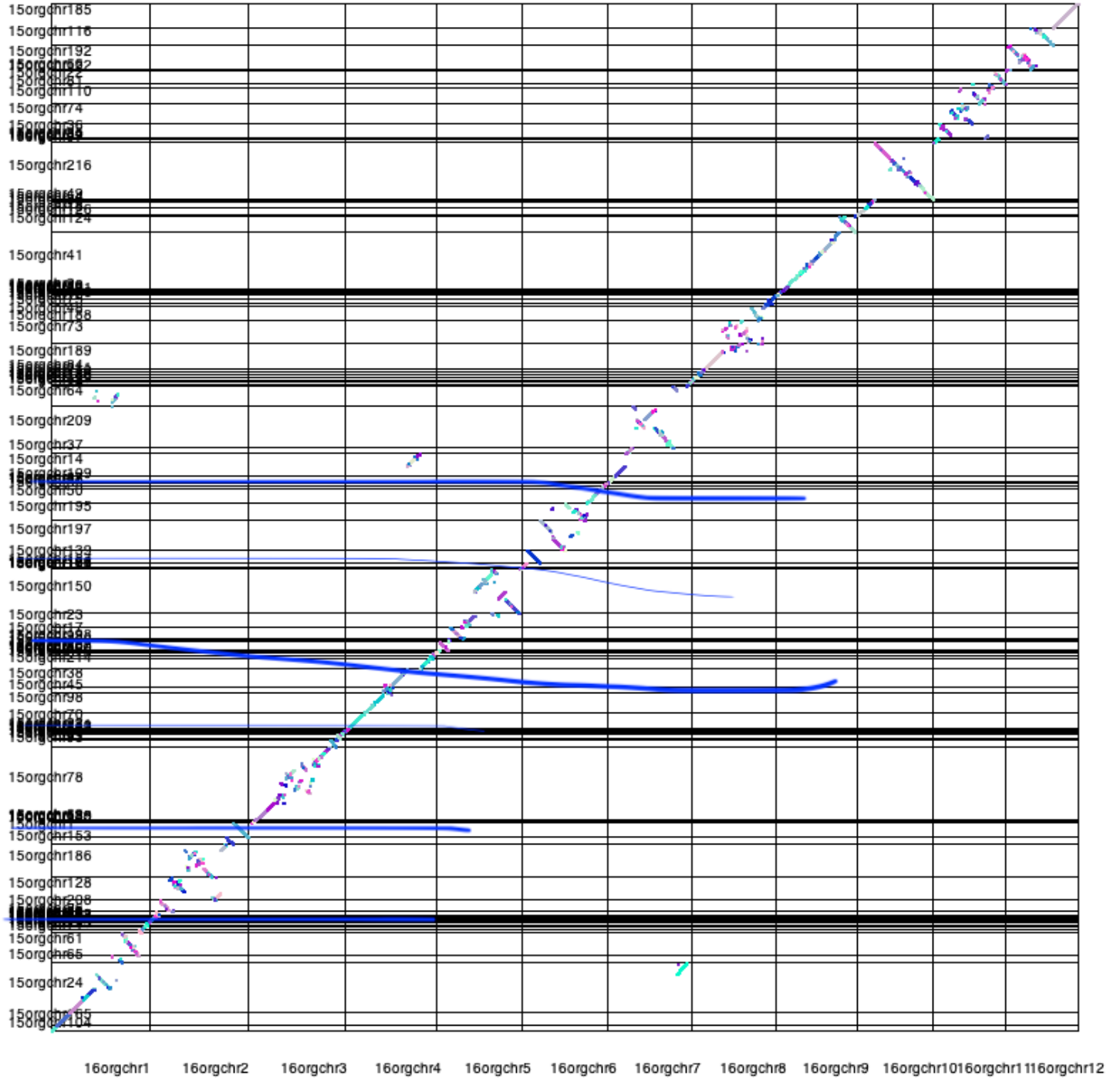
- Synteny alignments provide signal for reference-guided contig scaffolding
- note: not classical reference guided assembly but contig reordering
- tools exist: e.g. Ragout2 , MeDuSa, (Multi-)CSAR
- all use some kind of alignments + optimization to reduce number of rearrangements

Lets give it a try

- need some model for evolutionary events
- would be nice to be able to include multiple references
- example: Solanum stellatiglandulosum - kindly assembled by Thomas

Lets give it a try

- need some model evolutionary events:
  - simple model - regard reference as fully assembled and contigs as reliable
  - enumerate where and which way anchors on target contigs align on reference chromosomes
  - orient contig according to the majority of alignments (and enumerate possible inversion events) - corresponds to minimizing inversion events
  - define the order of target contigs on reference chromsomes based on majority of alignments - corresponds to minimizing translocations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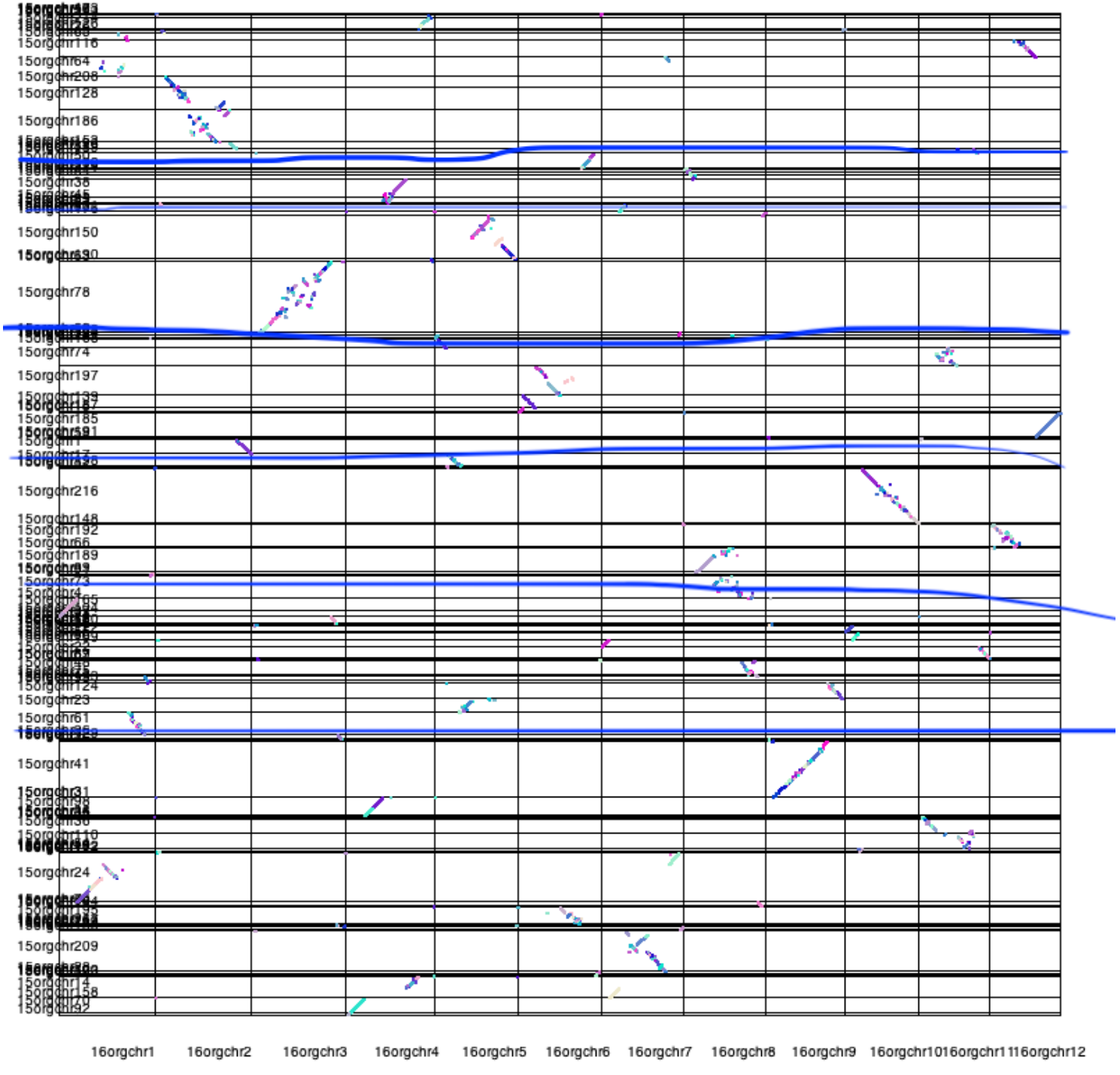
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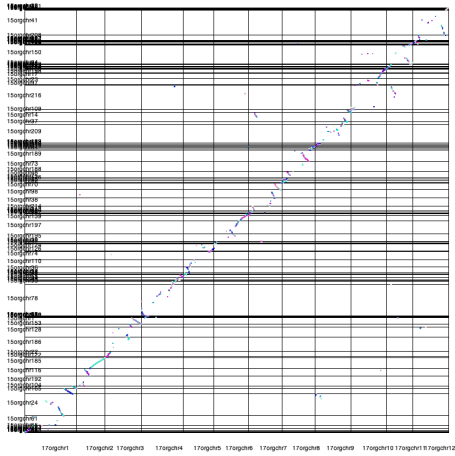
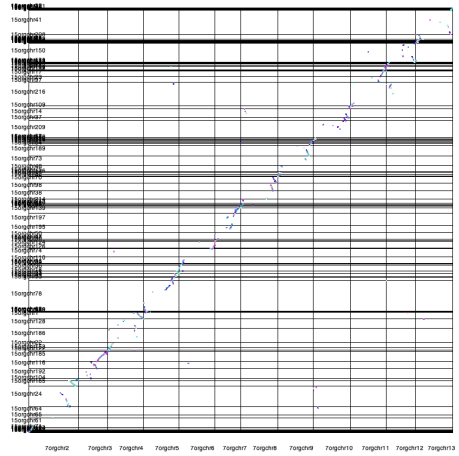
Lets give it a try

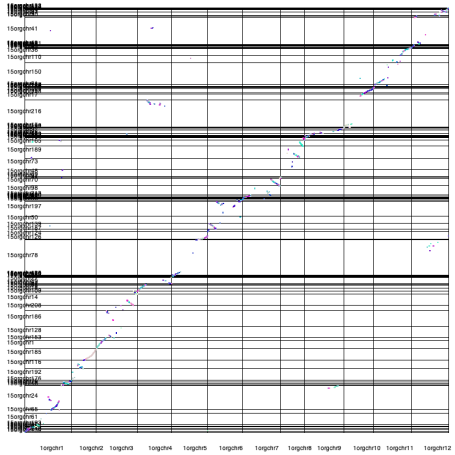
- would be nice to be able to include multiple references:
  - followed Multi-CSAR algorithm



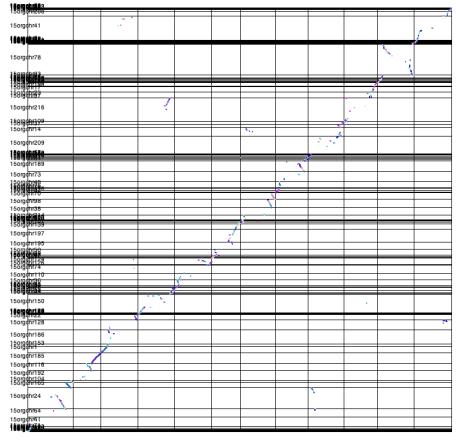
(a) Solanum tuberosum



(b) Solanum lycopersicum



(c) Capsicum annuum



(d) Solanum americanum

(a)

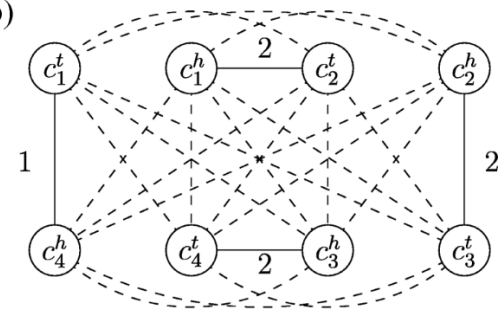
$T = \{c_1, c_2, c_3, c_4\}$

$S_1 = (+c_1, +c_2, +c_3)$

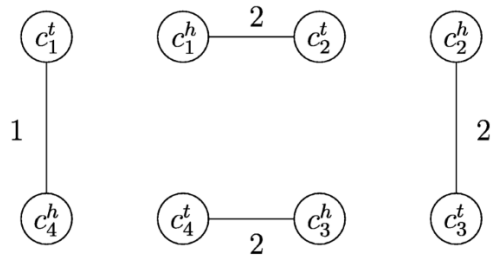
$S_2 = (+c_2, +c_3, +c_4)$

$S_3 = (-c_2, -c_1, -c_4, -c_3)$

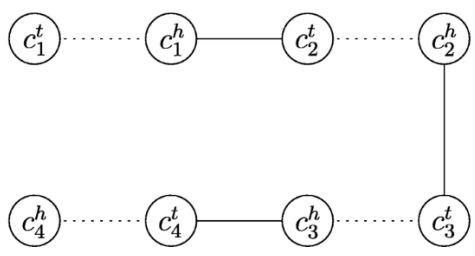
(b)



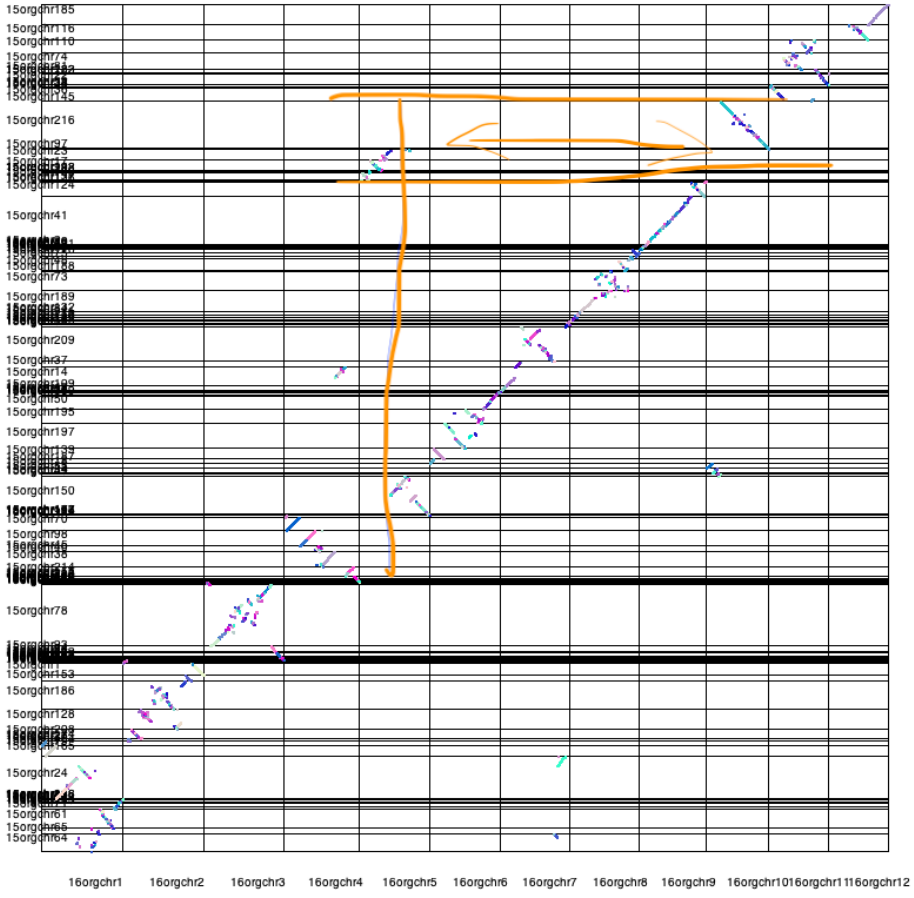
(c)



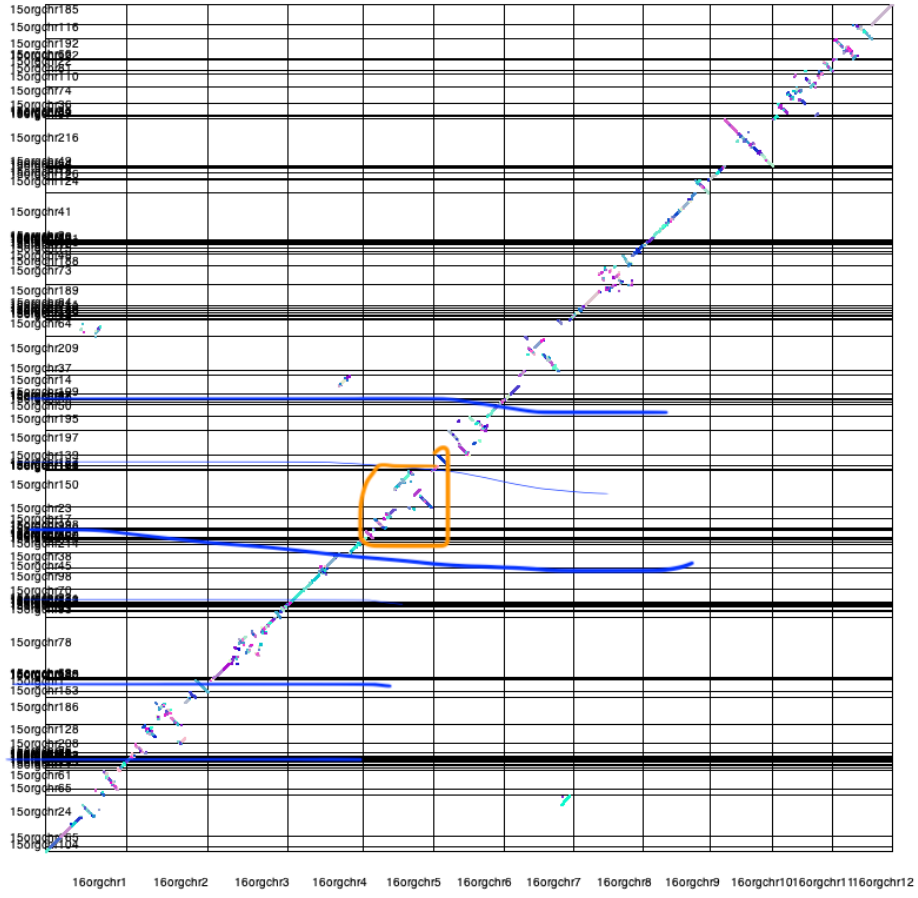
(d)



(e) Scaffold of $T = (+c_1, +c_2, +c_3, +c_4)$



(a) Consensus vs. S.torvum



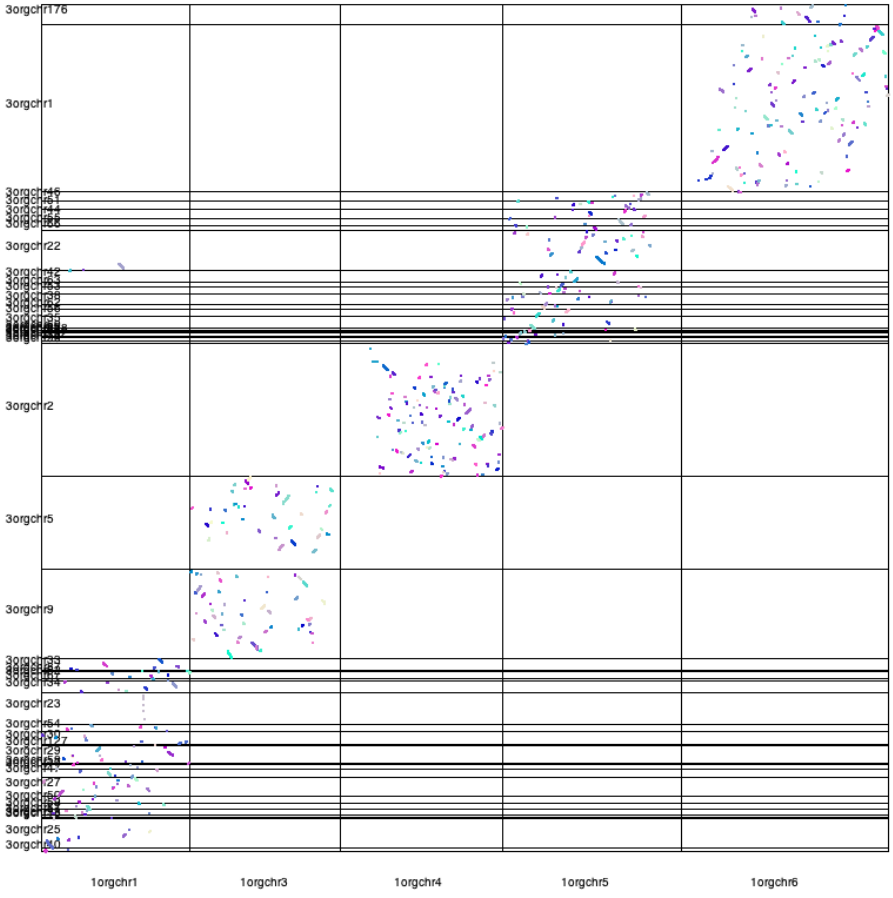
(b) Solanum Torvum

Table: Improved N50 from original 28 MB

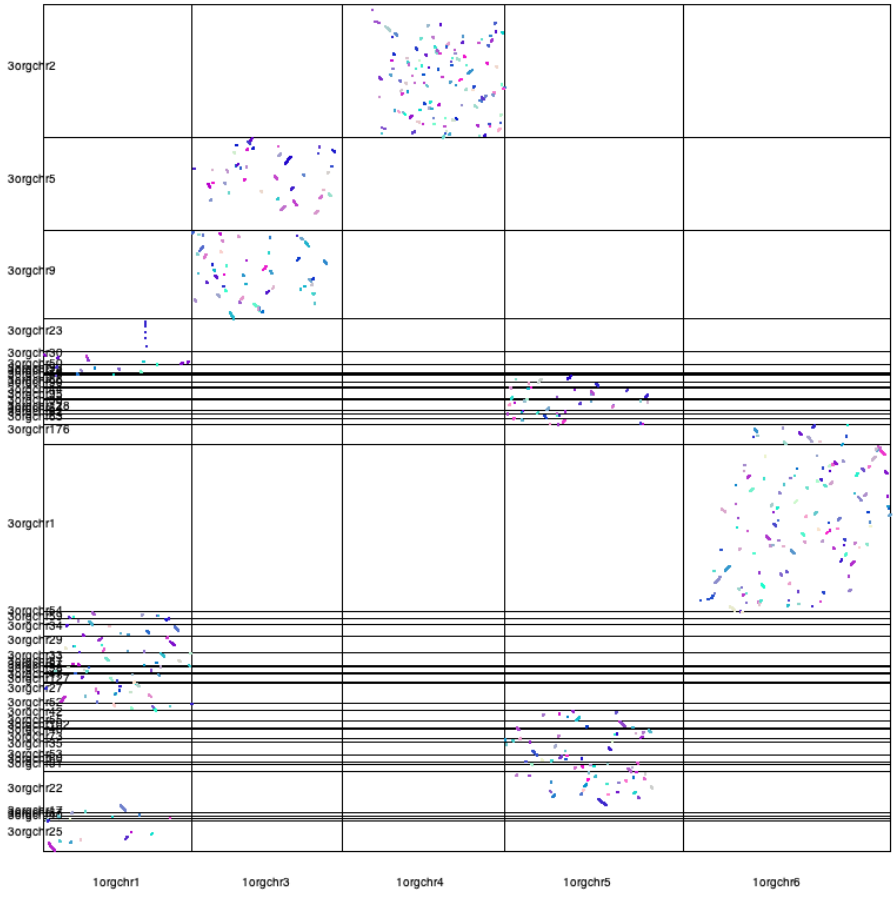
Reference Genome	scaffolded N50 all	scaffolded N50 limited
C.annuum	119 MB	92 MB
S.americanum	107 MB	95 MB
S.candolleanum	83 MB	57 MB
S. dulcamara	104 MB	87 MB
S. galapagense	107 MB	100 MB
S. laciniatum	51 MB	35 MB
S. lycopersicum	107 MB	107 MB
S. lyratum	108 MB	104 MB
S. melongena	107 MB	100 MB
S. pimpinellifolium	107 MB	100 MB
S. pinnatisectum	107 MB	95 MB
S. pseudocapsicum	79 MB	63 MB
S. retroflexum	77 MB	31 MB
S. spirale	73 MB	28 MB
S. torvum	108 MB	108 MB
S. tuberosum	107MB	100 MB
S. viarum	107 MB	99 MB
Consensus neutral weights	100 MB	102 MB
Consensus informed weights	101 MB	100 MB

Take home

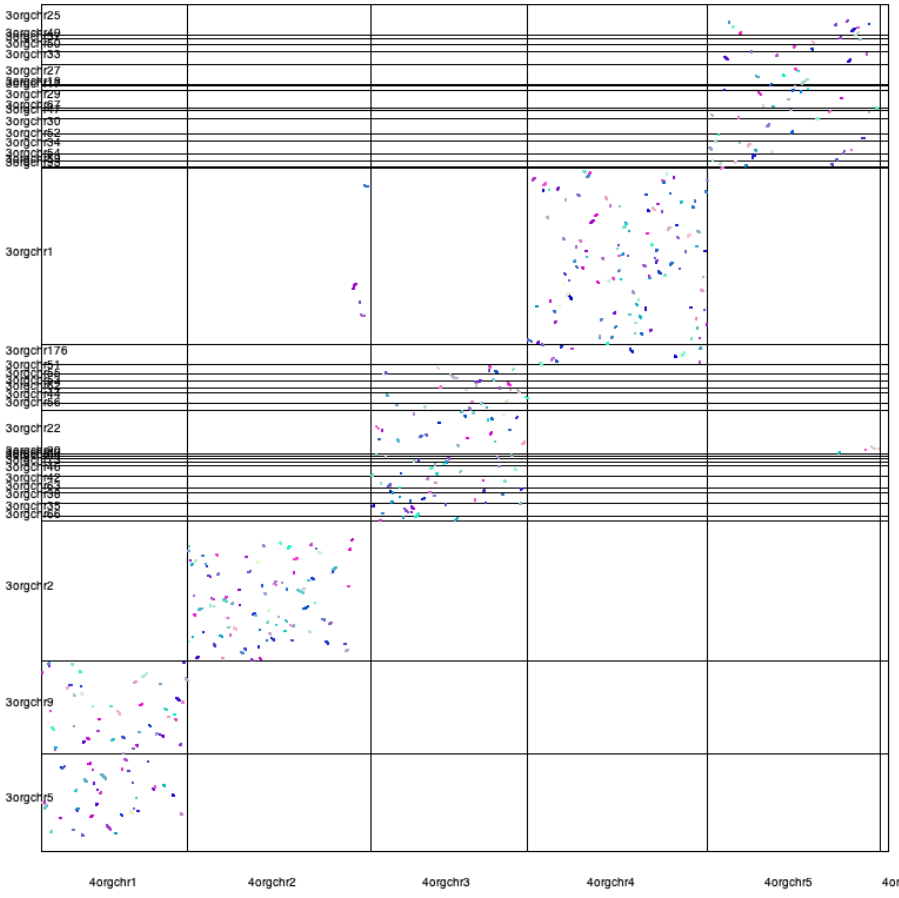
- reference-guided contig scaffolding possible without MSA or gene homology data
- way more benchmarking needed to see within what boundaries it can work
- potentially faster and more scalable
- really interesting bit are the potential breakpoints



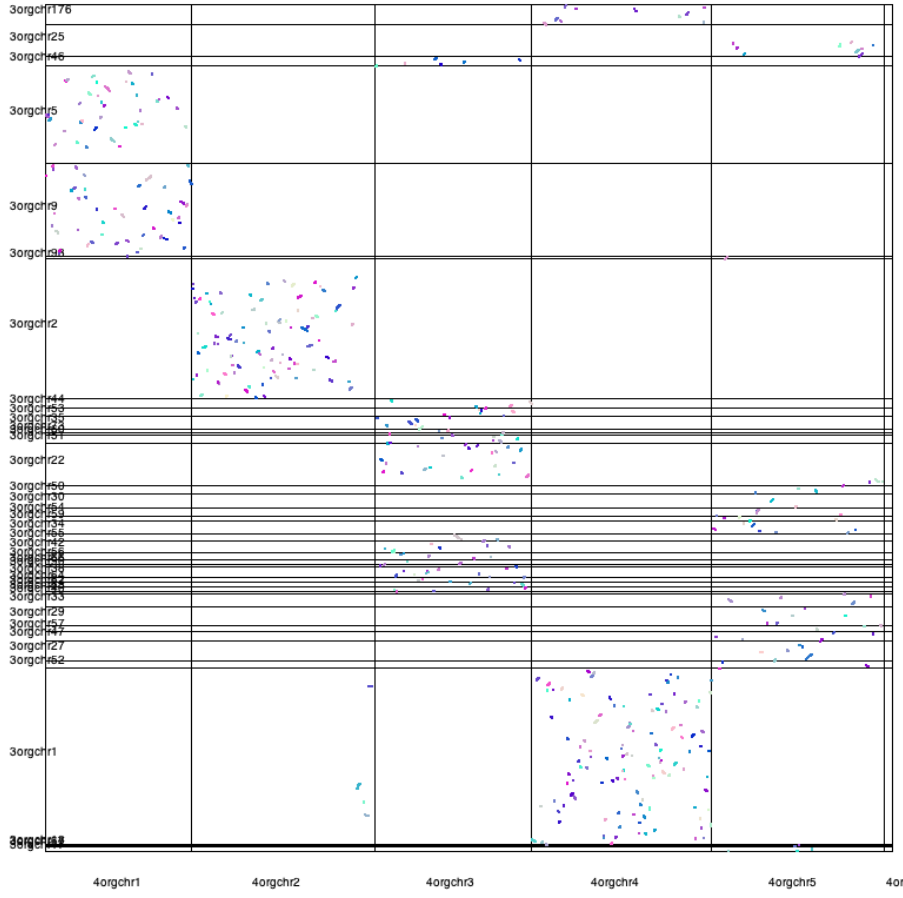
(a) D.mel



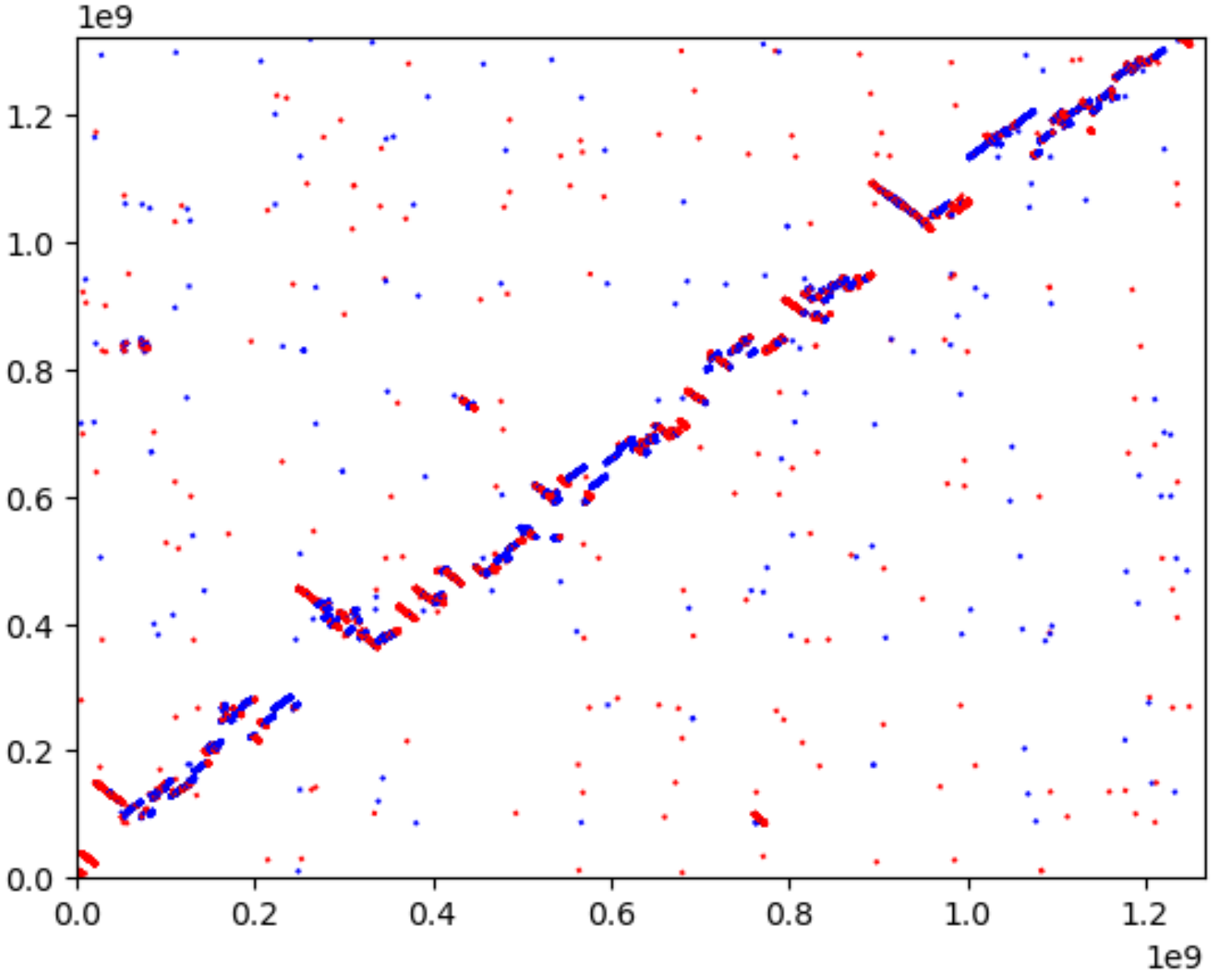
(b) D.mel CSAR



(a) D.busckii



(b) D.busckii CSAR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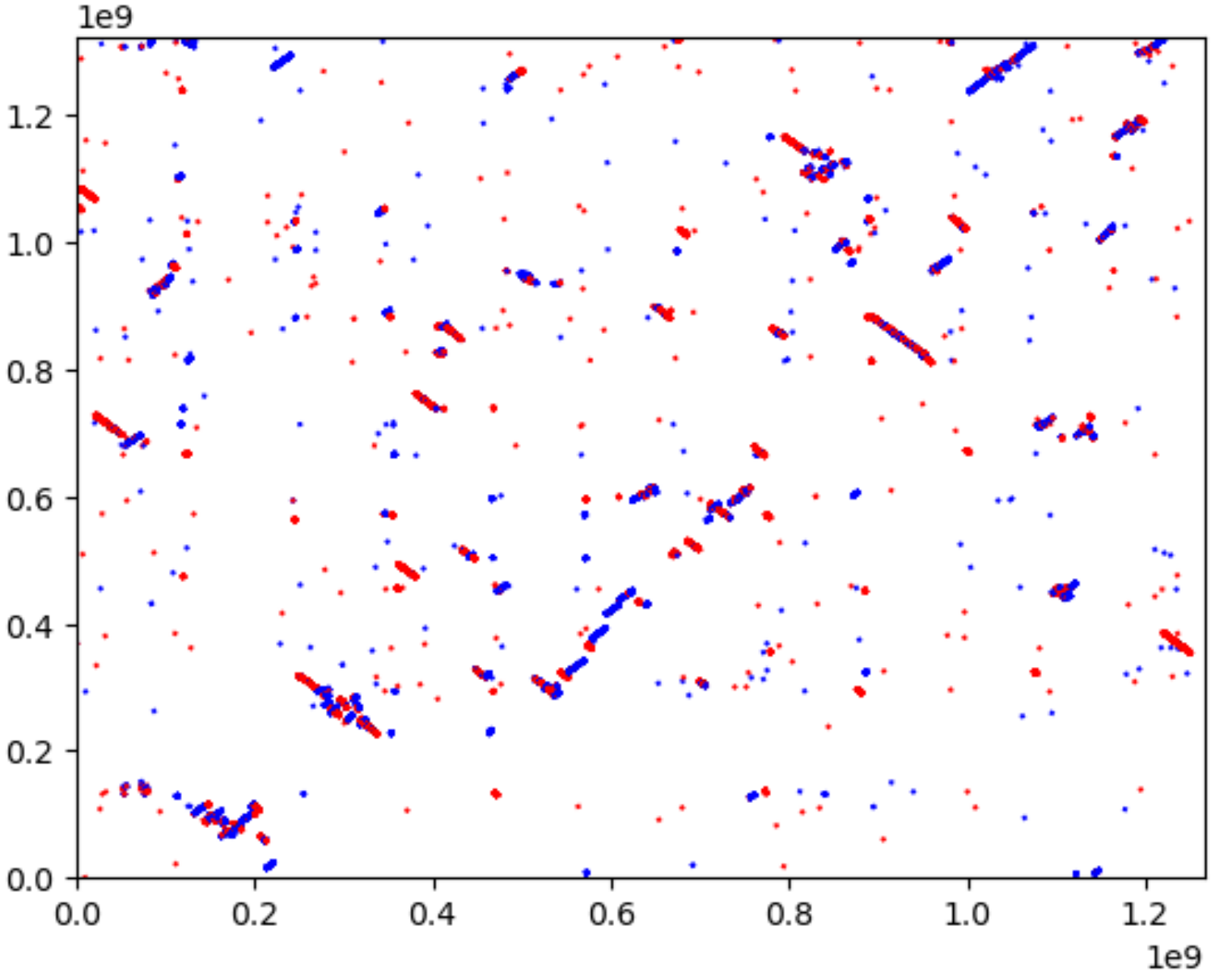
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