Recombination between heterologous human acrocentric chromosomes

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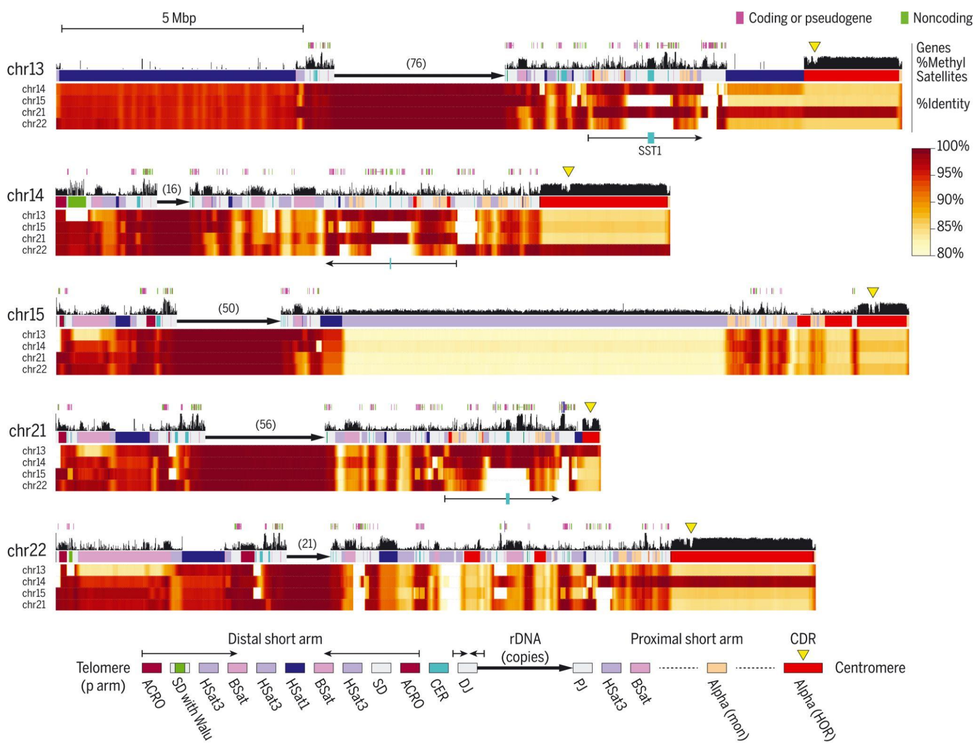
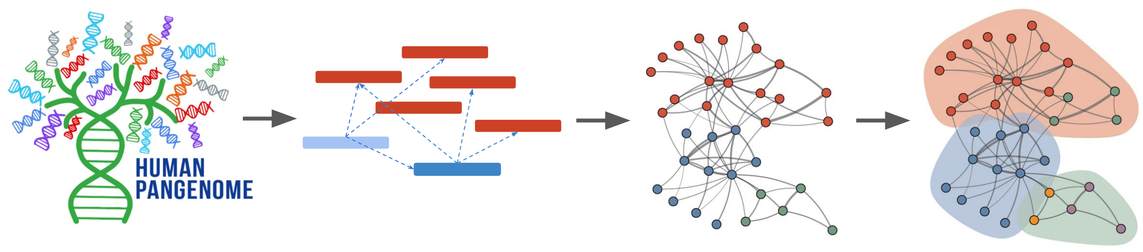
1) Filling 8% of the reference which was incomplete

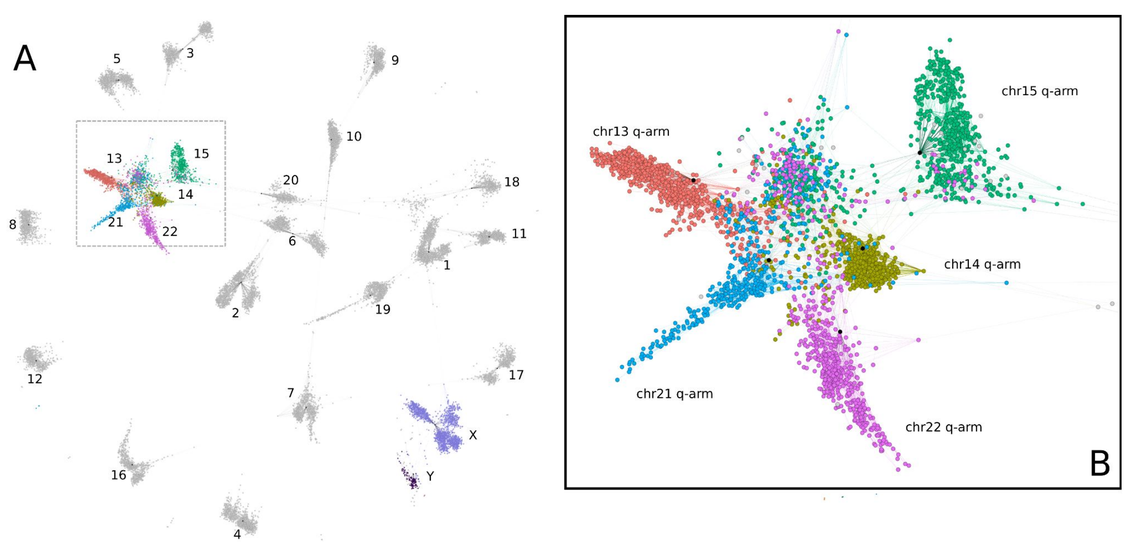
- The acrocentric p-arms were assembled for the **first time**.
- **High similarity** between p-arms of the acrocentric chromosomes seen in **one genome**.



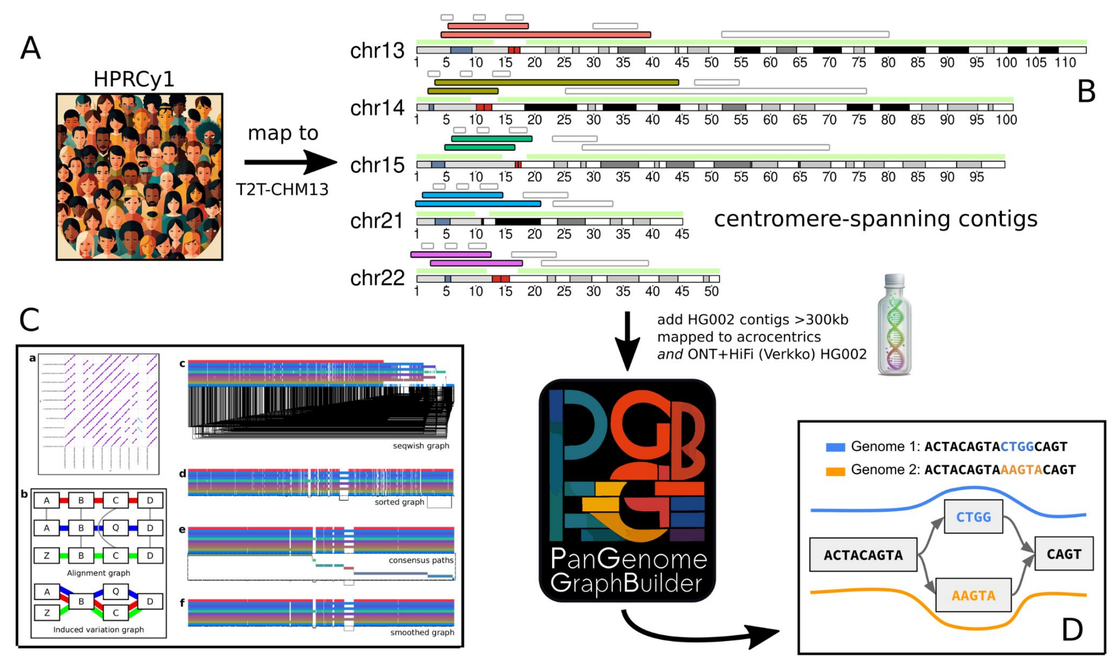
2) Homology-based community detection

47 near-complete assemblies → Homology mapping → Mapping graph → Community detection

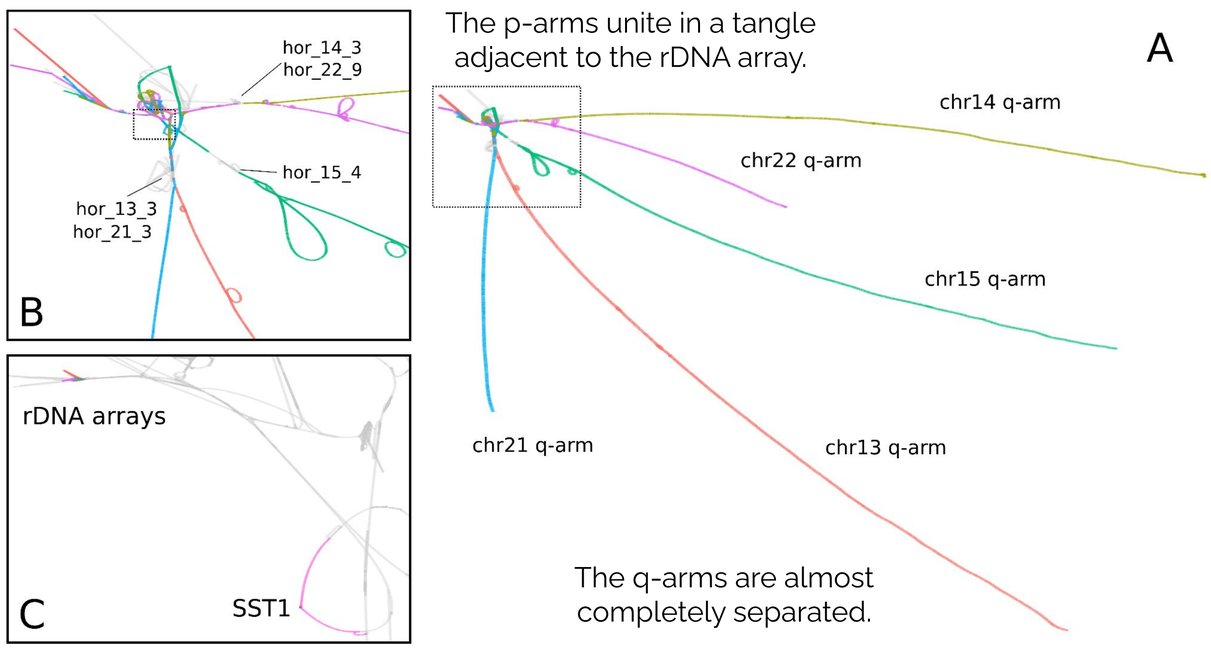




3) Building an acrocentric pangenome variation graph



4) Acrocentric pangenome variation graph



5) Pangenome untangling

Identify cut points (⊕) in the graph

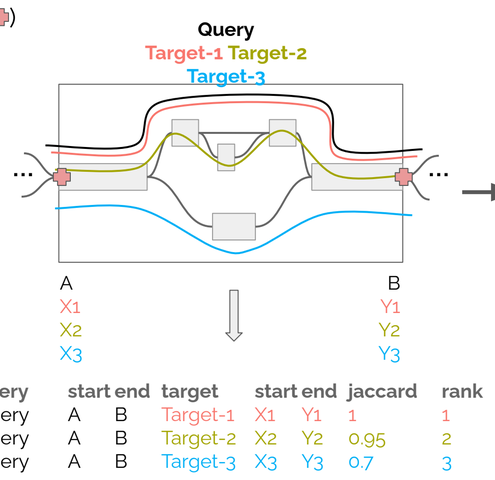
⇩

Define segment boundaries:

[A,B)
[X1, Y1)
[X2, Y2)
[X3, Y3)

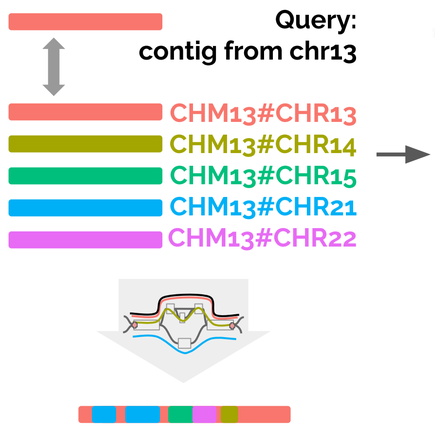
⇩

Compare segments



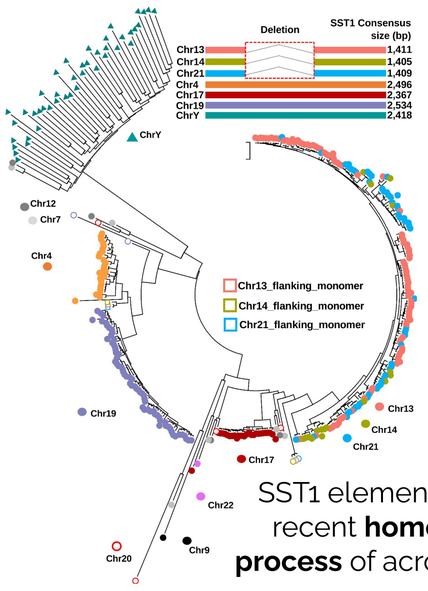
query	start	end	target	start	end	jaccard	rank
Query	A	B	Target-1	X1	Y1	1	1
Query	A	B	Target-2	X2	Y2	0.95	2
Query	A	B	Target-3	X3	Y3	0.7	3

6) Resolving homology mosaics



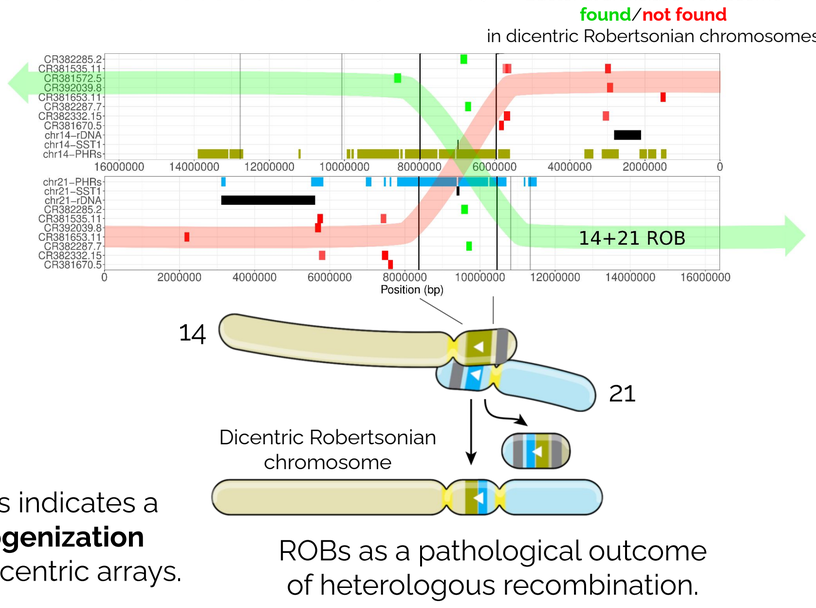
7) Pseudo-homologous regions (PHRs)

9) Robertsonian translocations (ROBs)



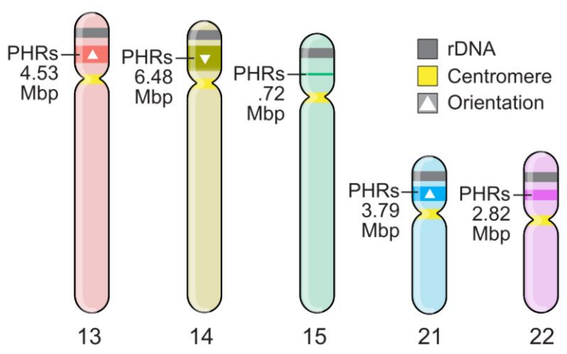
SST1 elements indicates a recent **homogenization process** of acrocentric arrays.

Bacterial artificial chromosome
found/**not found**
in dicentric Robertsonian chromosomes

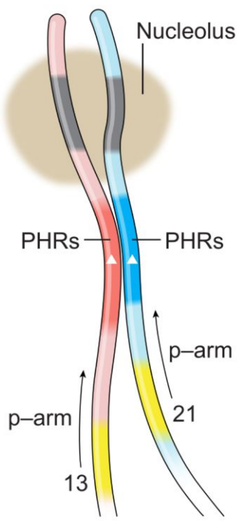


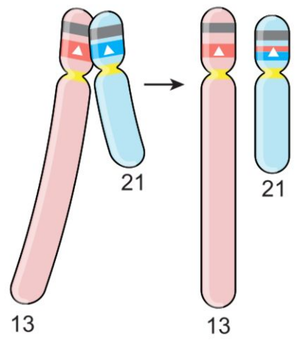
ROBs as a pathological outcome of heterologous recombination.

8) Heterologous recombination



A PHR on 13, 14 and 21 is nearly identical on all three chromosomes, but is typically **inverted** on 14 relative to 13 and 21.





Recombination largely depends on **sequence homology** and **physical proximity**.

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