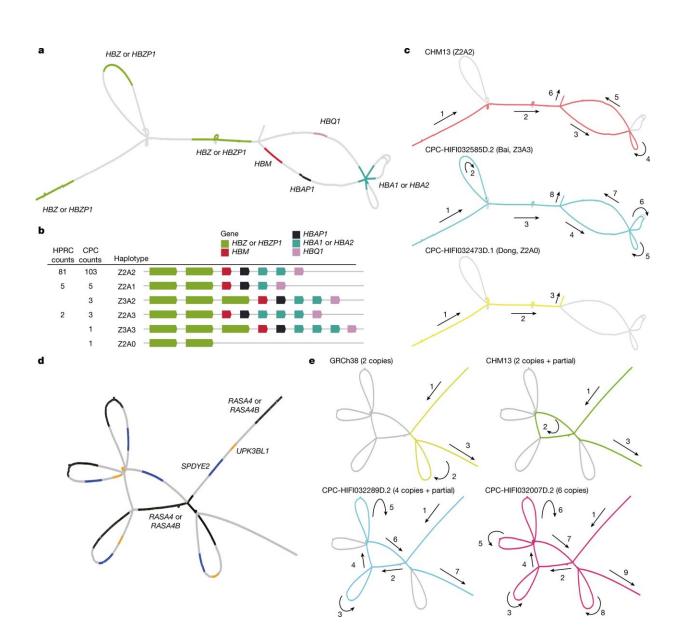


Consortium analyzes genomic sequences for 36 minority ethnic groups in China

June 15 2023, by Bob Yirka



Visualization of novel and complex SVs in the CPC pangenome graph. a, The



locations of α -globin genes on the CPC pangenome subgraph. b, Allele counts and linear structural visualization of all structural haplotypes from the Minigraph-Cactus graph among 116 CPC haploid assemblies and 94 HPRC haploid assemblies. The size and spacing of genes on the diagram do not represent the actual size of the chromosome. c, Paths of different α -globin gene haplotypes through the joint subgraph. The arrows indicate the direction of the paths. d, The locations of genes in the RASA4 region on the CPC subgraph. e, Paths of different structural haplotypes with diverse copy numbers of RASA4B. "partial" represents a 14.9-kb fragment of RASA4B. Credit: *Nature* (2023). DOI: 10.1038/s41586-023-06173-7

A large team of genetic engineers affiliated with institutions across China has completed the first phase of its initiative to create a pangenome reference of genome data for people of different ancestries in China. In their project, reported in the journal *Nature*, the group, which is part of The Chinese Pangenome Consortium (CPC), sought to sequence the genomes of minority groups across China. The editors at *Nature* have also published a Research Briefing outlining the work in the same journal issue.

The CPC was created in China to address a concern that has arisen in the global medical community—most genomic sequencing has involved people from the major population groups. For example, in China, Hans are the dominant ethnic group, so most genetic research in China has been conducted using Han genomes.

This is a problem because slight differences in genomes can result in treatments that fail to work or cause unexpected problems among people from minority <u>ethnic groups</u>. The <u>consortium</u> has divided its effort into several phases, the first of which was to choose a list of minority ethnic groups to study and then to conduct sequencing and <u>genetic analysis</u> on tissues collected from members of each group in that list.



In its first phase, the team created a list of 36 minority ethnic groups (there are currently 55 known minority ethnic groups in China) and collected tissues samples from 58 people who were represented on the list. They then conducted sequencing on all the samples, resulting in 116 assemblies (two from each volunteer). They conducted haplotype phasing to sort the assemblies by parentage. The team added 189 million base pairs of packaged sequences that varied between individuals. They also found 1,367 protein-coding duplications.

The research team also found what they describe as considerable variation between groups in certain sequences, which they suggest confirms the need for the CPC to continue its work.

More information: Yang Gao et al, A pangenome reference of 36 Chinese populations, *Nature* (2023). <u>DOI: 10.1038/s41586-023-06173-7</u>

A pangenome reference representative of 36 minority Chinese ethnic groups, *Nature* (2023). DOI: 10.1038/d41586-023-01675-w

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