

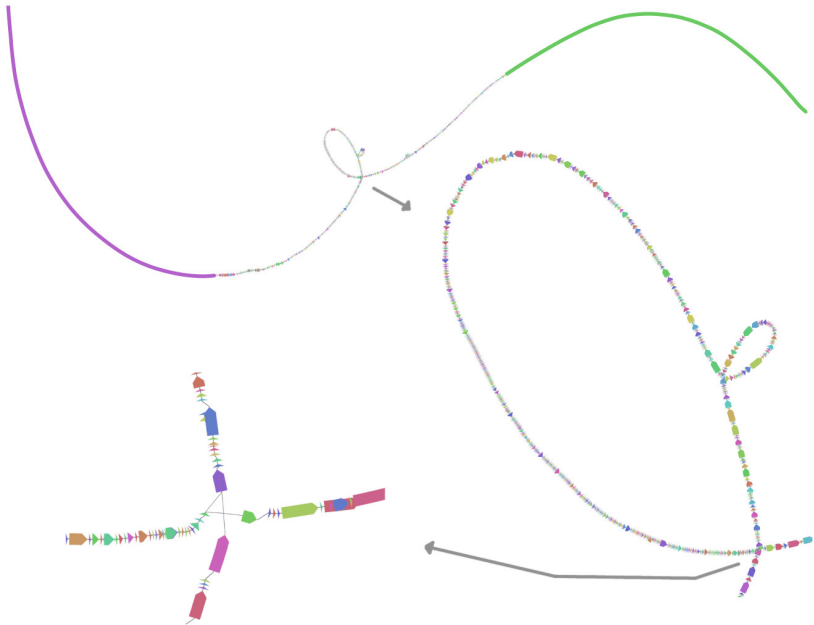
Guessing RCCX modules from a collapsed pangenome

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Pangenome with collapsed RCCX region

- ▶ A pangenome was built from the HPRC haplotypes using PGGB.
- ▶ The global structure of the pangenome looks good
 - ▶ The modules were collapsed nicely
 - ▶ Haplotypes loop back, traversing the collapsed RCCX part of the pangenome multiple times.
 - ▶ The large transposable element polymorphism at the beginning of the RCCX module is visible too.
 - ▶ No other major rearrangement.

Pangenome with collapsed RCCX region



Question

Can we guess the location of module 1 (with CY21A1P) and module 2 (with CYP21A2) in the haplotypes based on how they traverse the pangenome?

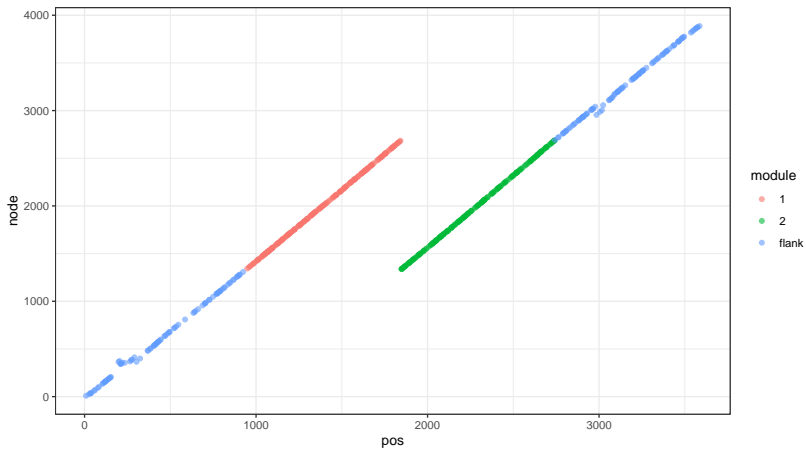
- ▶ Each module 1 should tend to traverse similar nodes across different haplotypes.
- ▶ A PCA-like might analysis might pick up the two types of modules.
- ▶ If we can guess the modules, we won't have to rely on annotation (and filter out haplotypes with suspicious annotations).

Find the looping edge from the reference path

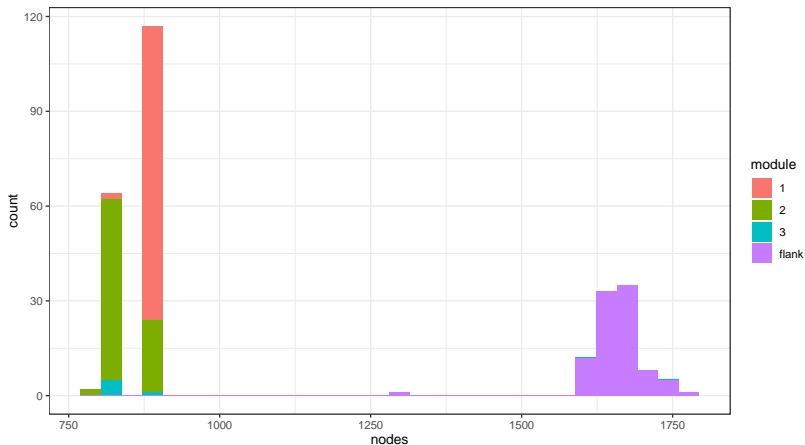
As the edge where the node IDs differ the most

path	pos	node	strand	next.node	jump
ref	1846	2689	+	1336	1353

Split each haplotype at the looping edges

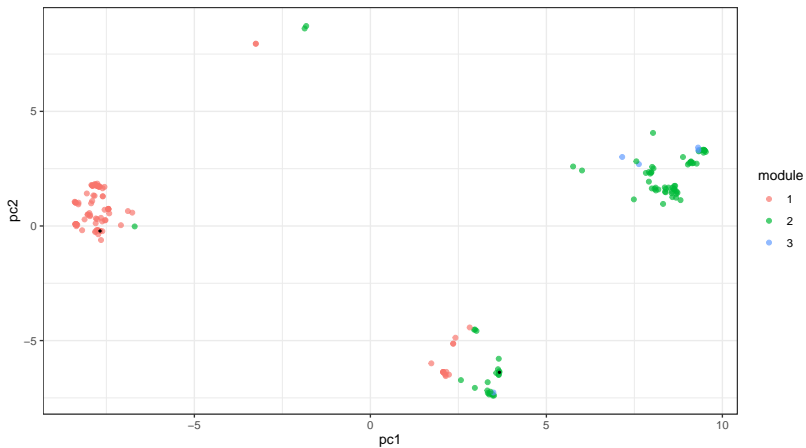


Any small sub-path to remove?



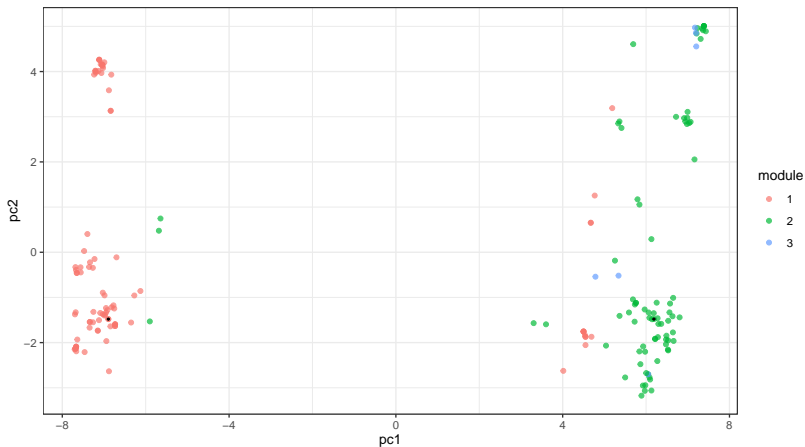
No, they seem to have consistent size.

PCA with all the module nodes



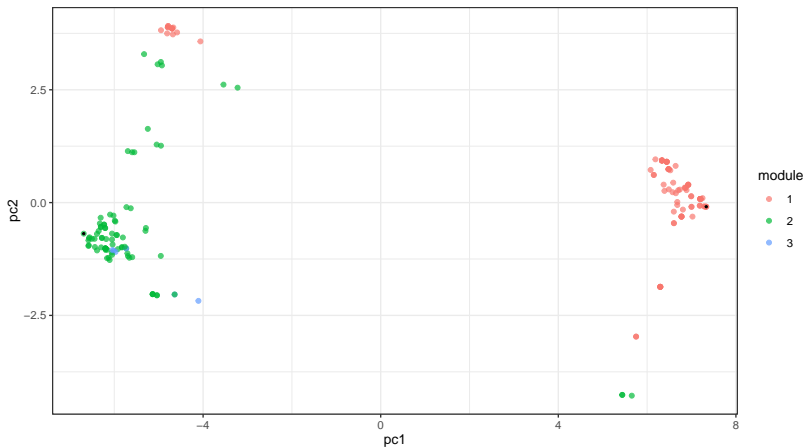
Black points highlights modules in the reference haplotype.

Removing the TE polymorphism



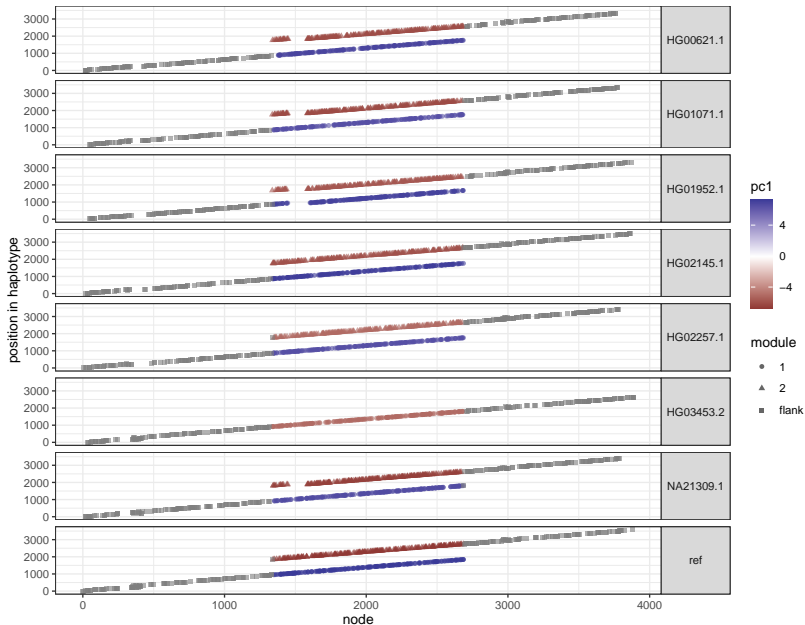
Black points highlights modules in the reference haplotype.

Using module-spec nodes in the reference haplotype

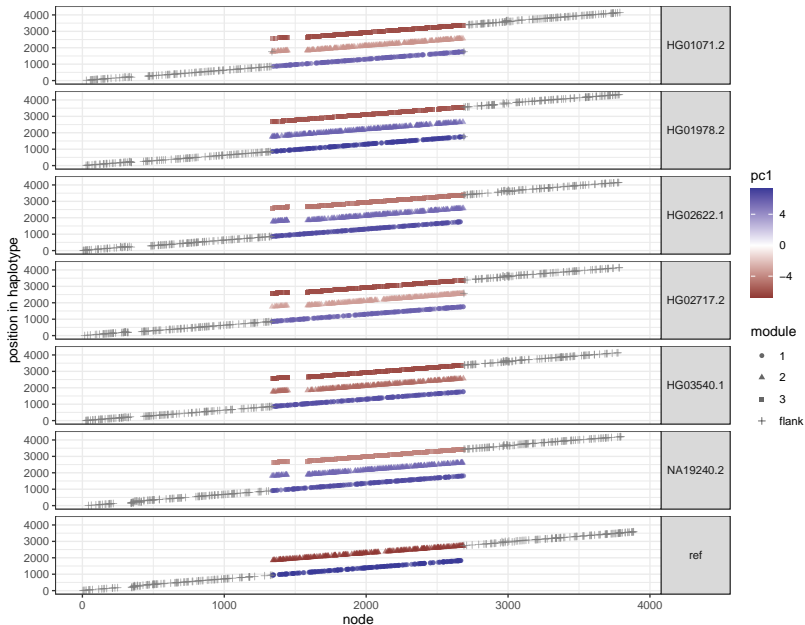


Black points highlights modules in the reference haplotype.

Random haplotypes colored by PC1



Tri-modular haplotypes



Uni-modular haplotypes

