

Assortative mating and rapid adaptation shape genetic variation in admixed Cape Verdeans

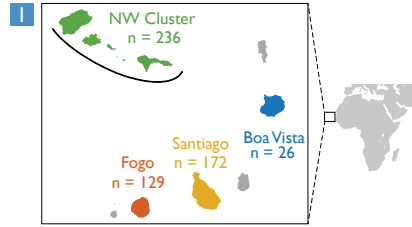


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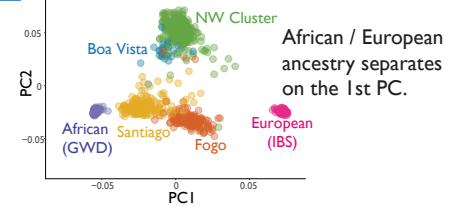
Background

- Recent admixture allows us to leverage patterns in genetic ancestry to understand short-term evolution.
- On very short timescales, changes in allele frequencies may be difficult to observe and biased by factors like nonrandom mating. In contrast, changes in ancestry patterns in admixed populations can be observed within tens of generations.
- Cape Verdeans** are admixed descendants of Portuguese colonizers and enslaved West African people who settled the islands during the 1400s.
- Here, we use ancestry patterns to infer the demographic and selective history of the last ~20 generations since founding.**



Data: genome-wide SNP data from 563 Cape Verdean individuals¹.

2 PRINCIPAL COMPONENTS ANALYSIS²

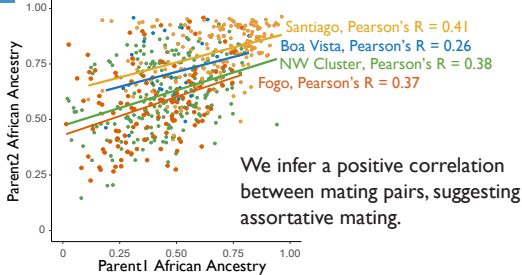


- This mixed ancestry provides the basis for examining mating patterns and the genetic consequences of admixture.

Ancestry and mating patterns

Inference of population history relies on assumptions about mating patterns

3 PARENTAL ANCESTRY ESTIMATES³



We infer a positive correlation between mating pairs, suggesting assortative mating.

- Assumptions about mating patterns can skew demographic inference.
- Multiple population genetic tools that assume random mating **underestimate the timing of admixture.**

TIMING OF ADMIXTURE INFERRED WITH ALDER⁴

	ISLAND	ESTIMATE
Known order of settlement beginning in the 1400s	Santiago	1779
	Fogo	1762
	Boa Vista	1788
	NW Cluster	1742

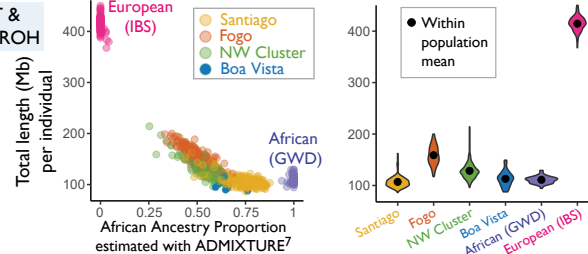
TIMING OF ADMIXTURE ALLOWING ASSORTATIVE MATING⁵

	ISLAND	ESTIMATE
Known order of settlement beginning in the 1400s	Santiago	1565
	Fogo	1515
	Boa Vista	1559
	NW Cluster	1515

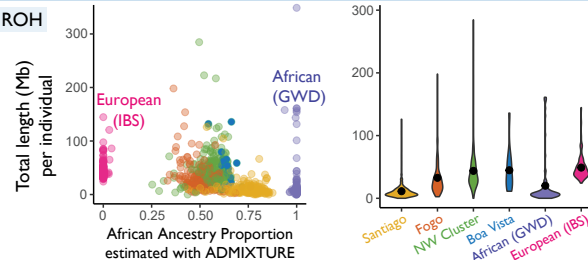
ROH reflect historical differences between the islands in their contributions from the source populations and mating patterns

- ROH** (runs of homozygosity) arise when identical haplotypes are inherited from a common ancestor.
 - Distant common ancestors contribute **shorter ROH**, where haplotypes have been broken up over generations.
 - In contrast, recent common ancestors generate **longer ROH**.
- Here, we identify ROH with a model-based approach⁶ and examine it in the context of the genetic contributions of the source populations.
- Despite recent colonization, some admixed individuals have **lower ROH** than African reference populations.

4 SHORT & MEDIUM ROH



5 LONG ROH

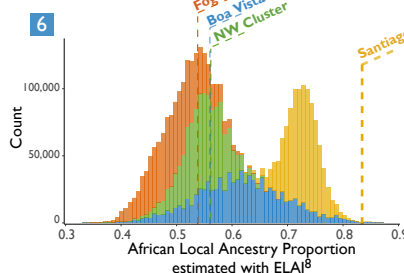


- Classifying ROH by length, we find that **shorter length ROH drive low overall levels of ROH** through mismatched local ancestry.
- Many admixed individuals still present **excess long ROH**, likely reflecting post-admixture processes.
- ROH content is **consistent with colonization history, recent population sizes, and mating patterns.**
 - The NW Cluster was founded most recently, likely involving a bottleneck; Fogo has higher incidence of first-cousin marriages; and Santiago is the oldest and largest population.

Local ancestry differences among the islands suggest adaptation to malaria

- We find elevated African ancestry and long African ancestry tracts in Santiago individuals surrounding the Duffy-negative allele.
- The Duffy-negative allele at the DARC locus protects against a malaria parasite.
- Notably, Santiago is the only Cape Verdean island with substantial history of malaria transmission.
- We explore this **preliminary evidence of adaptation** further in Oral Presentation #256 presented by Amy Goldberg.

Genome-wide distribution of local ancestry at autosomal SNPs (histogram) vs ancestry at the DARC locus (dashed lines)



Conclusions

- Ancestry patterns in Cape Verde reveal patterns of assortative mating over the last ~20 generations.
- Understanding these patterns is critical to demographic inference, as seen with biases in estimates of admixture timing.
- Runs of homozygosity (ROH) reflect the contributions of the source populations and patterns of assortative mating.
- Together, these results provide insight into the population history of Cape Verde. More generally, we show how admixed populations provide powerful test cases for understanding evolutionary processes within tens of generations.