



# CURSO DE FÉRIAS 2020

## GENÉTICA E EVOLUÇÃO

10 a 14 de fevereiro

Instituto de Biociências • USP

# Minicurso: Biologia Evolutiva

## Aula: Genômica Populacional

Palestrante: Kelly Nunes

Laboratório de Genética Evolutiva

<http://www.genevol.ib.usp.br>

# O Laboratório de Genética Evolutiva - IBUSP



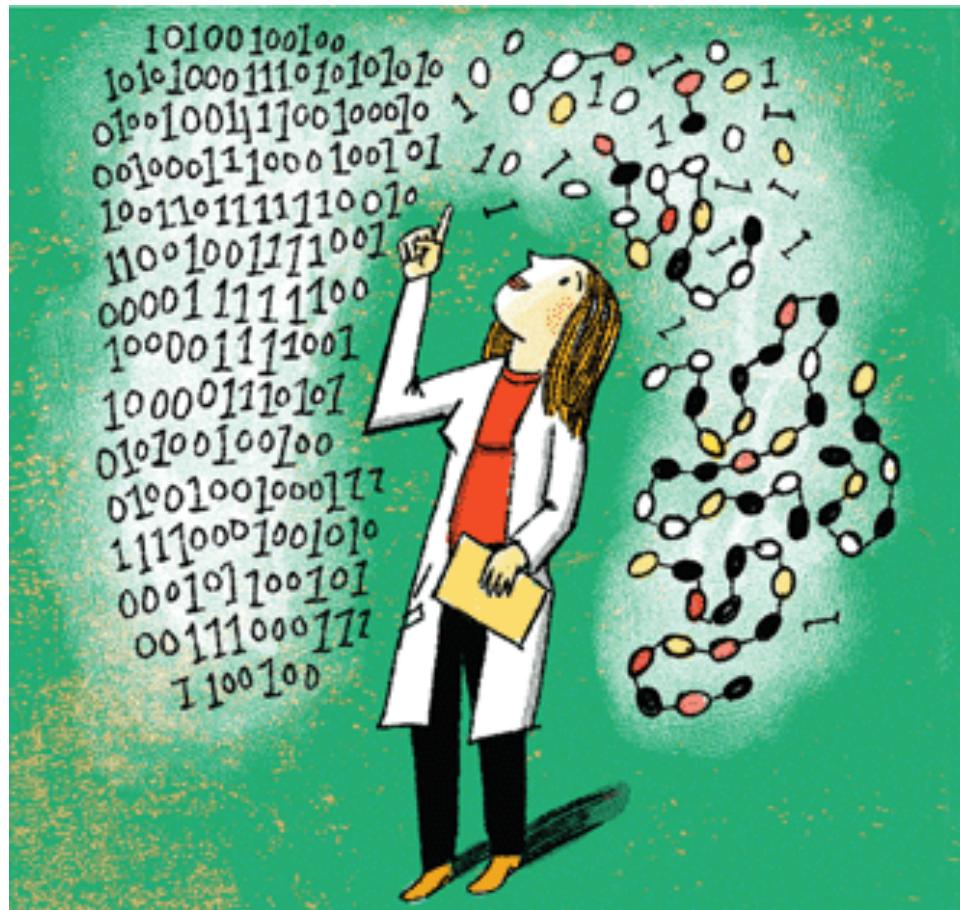
**Professor Responsável:** Diogo Meyer ([diogo@ib.usp.br](mailto:diogo@ib.usp.br))

## Linhas de Pesquisa:

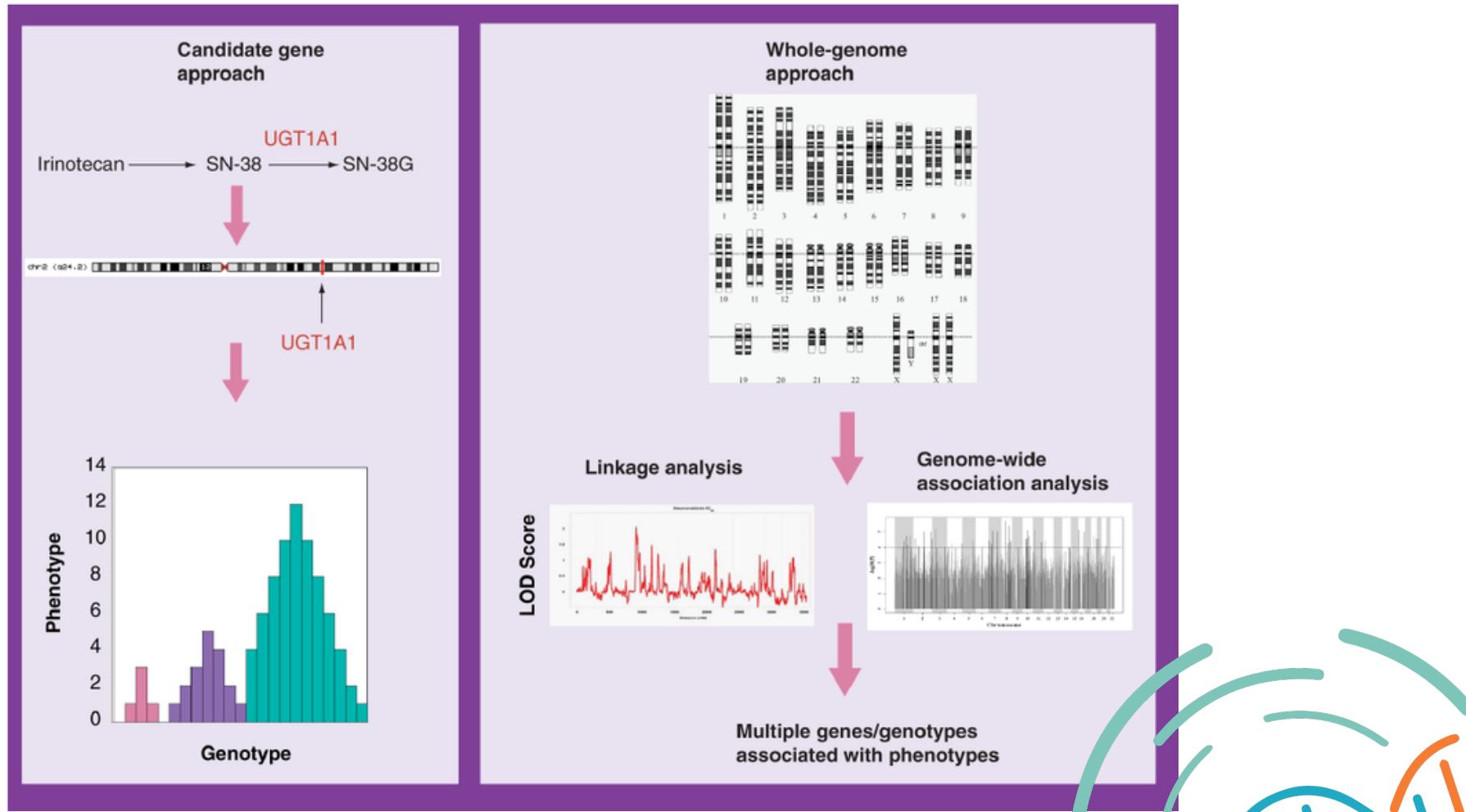
- Genômica Populacional:
  - História das populações miscigenadas
  - Padrões de Seleção Natural em populações miscigenadas
- Genômica Evolutiva:
  - Evolução dos genes HLA
- Transcriptôma
  - Expressão dos genes HLA
  - Mapeamento de eQTLs



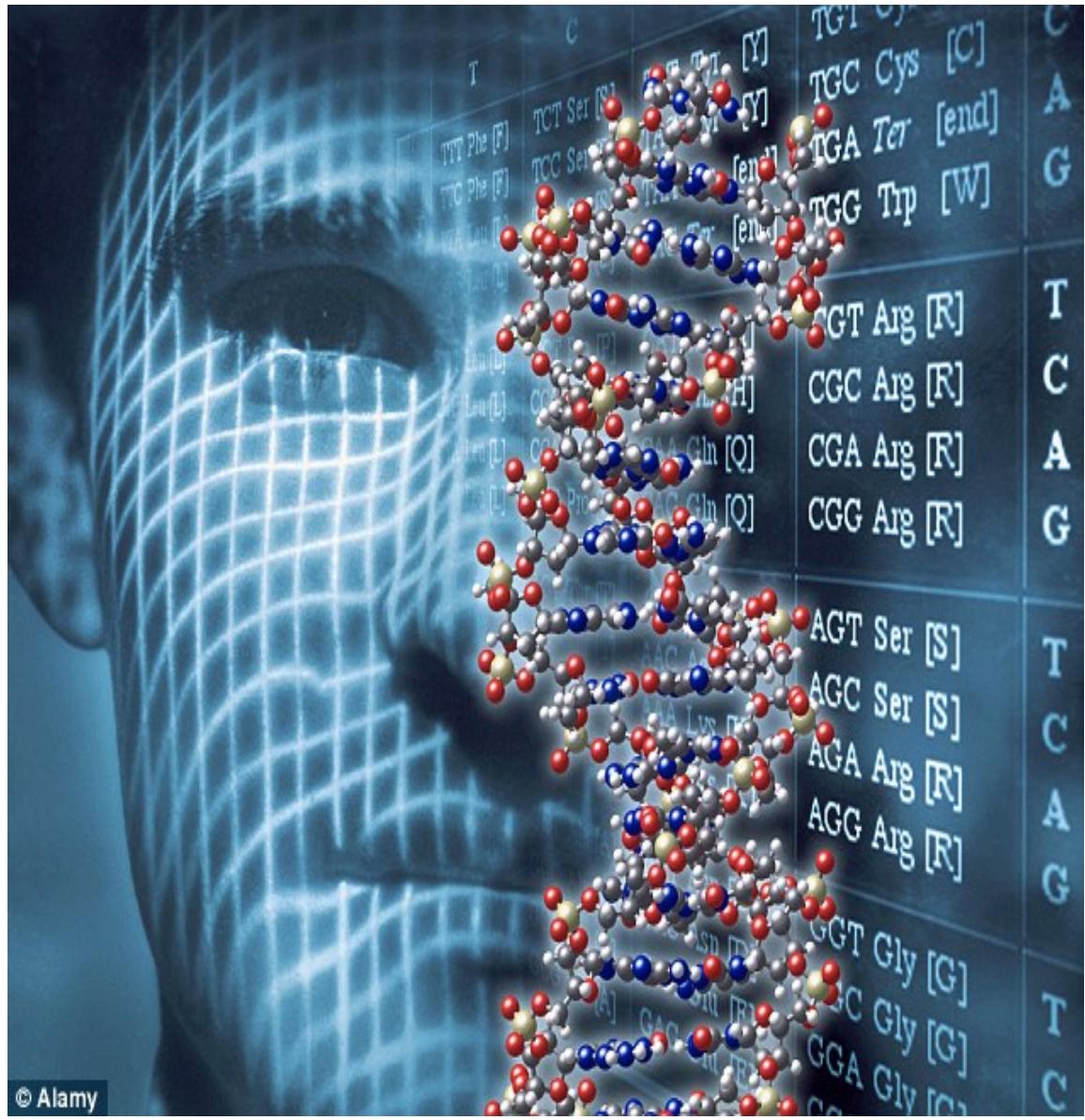
# A Genômica Populacional



# Genes candidatos x Scans ao longo do Genoma



# O Genoma Humano

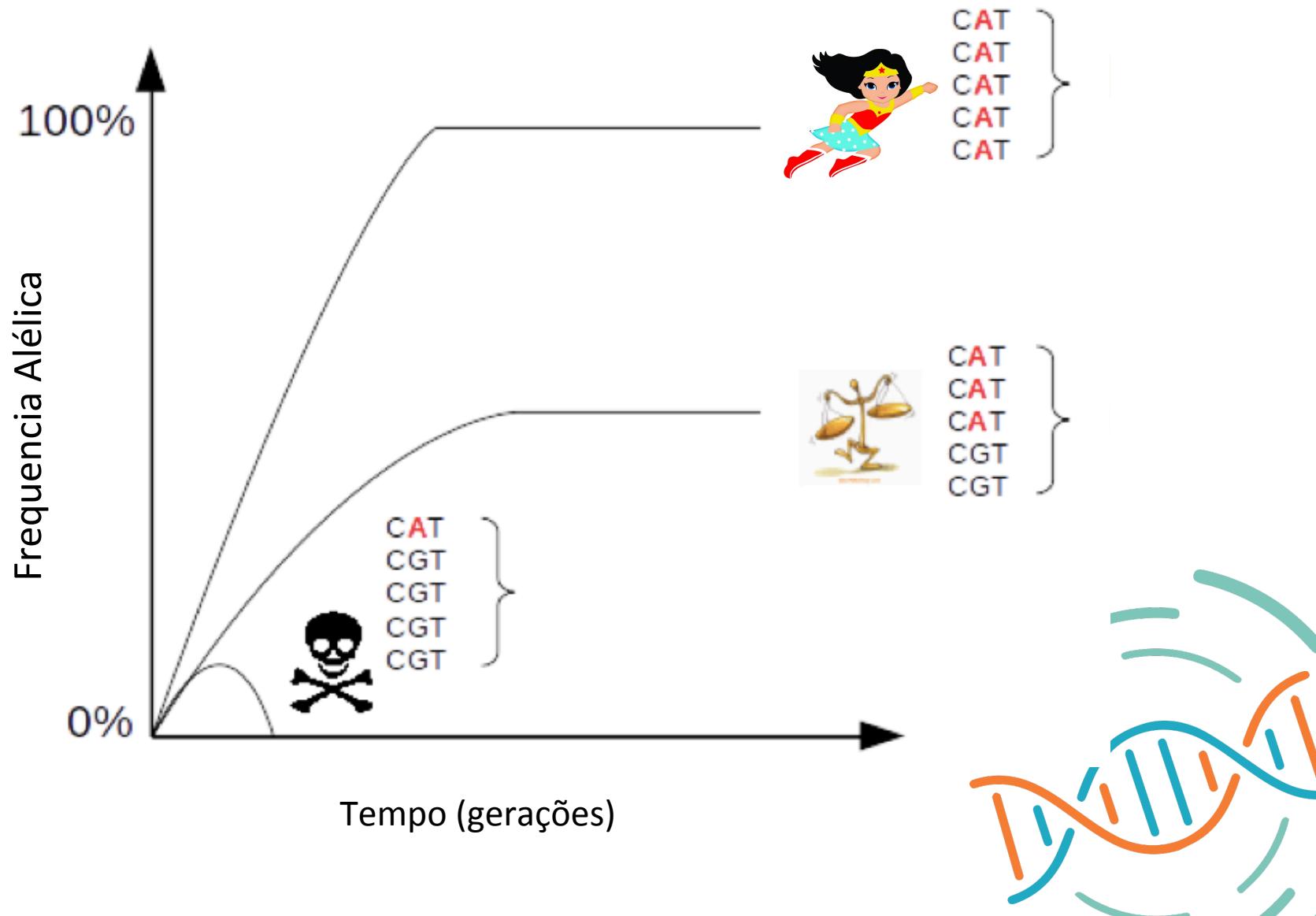


# Por que as mutações são mantidas?

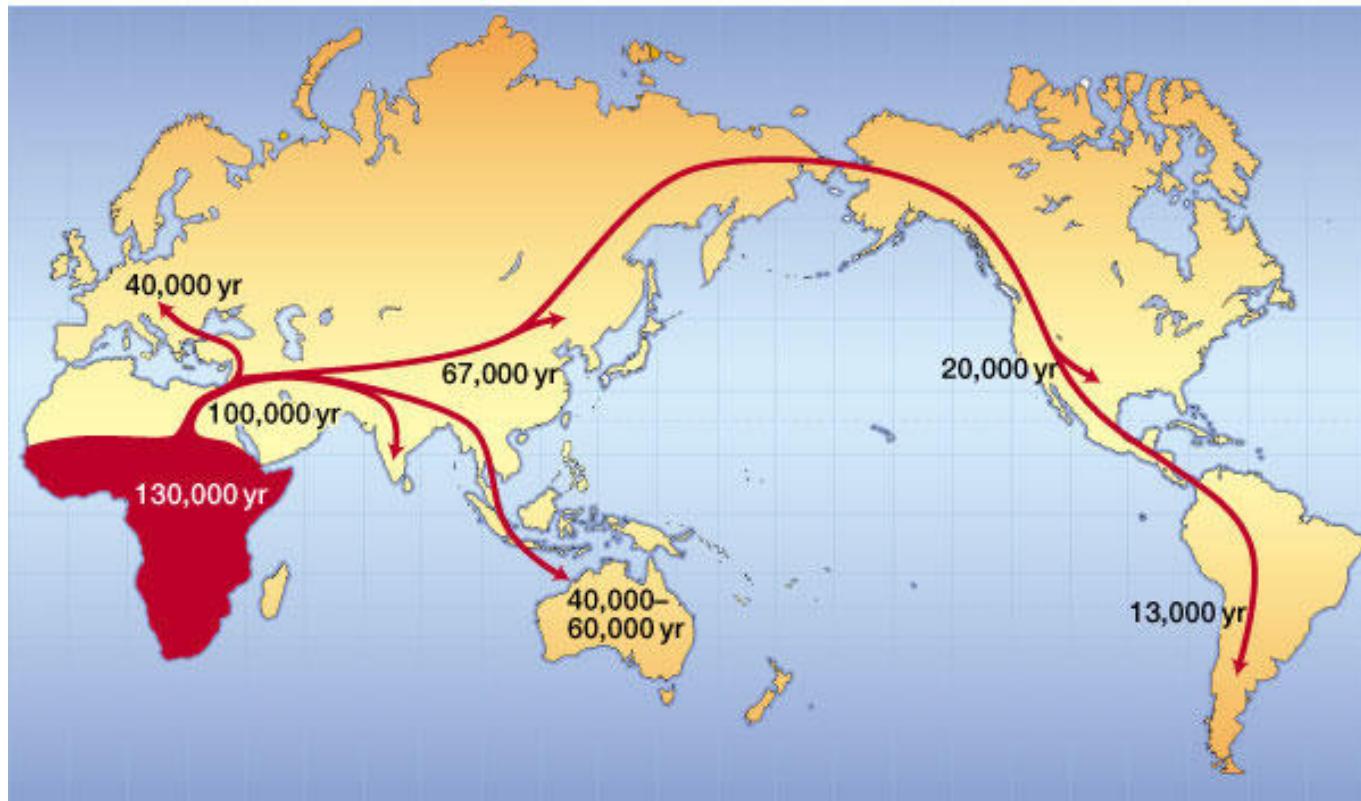
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CGTGTAGGGGTCTGCA  
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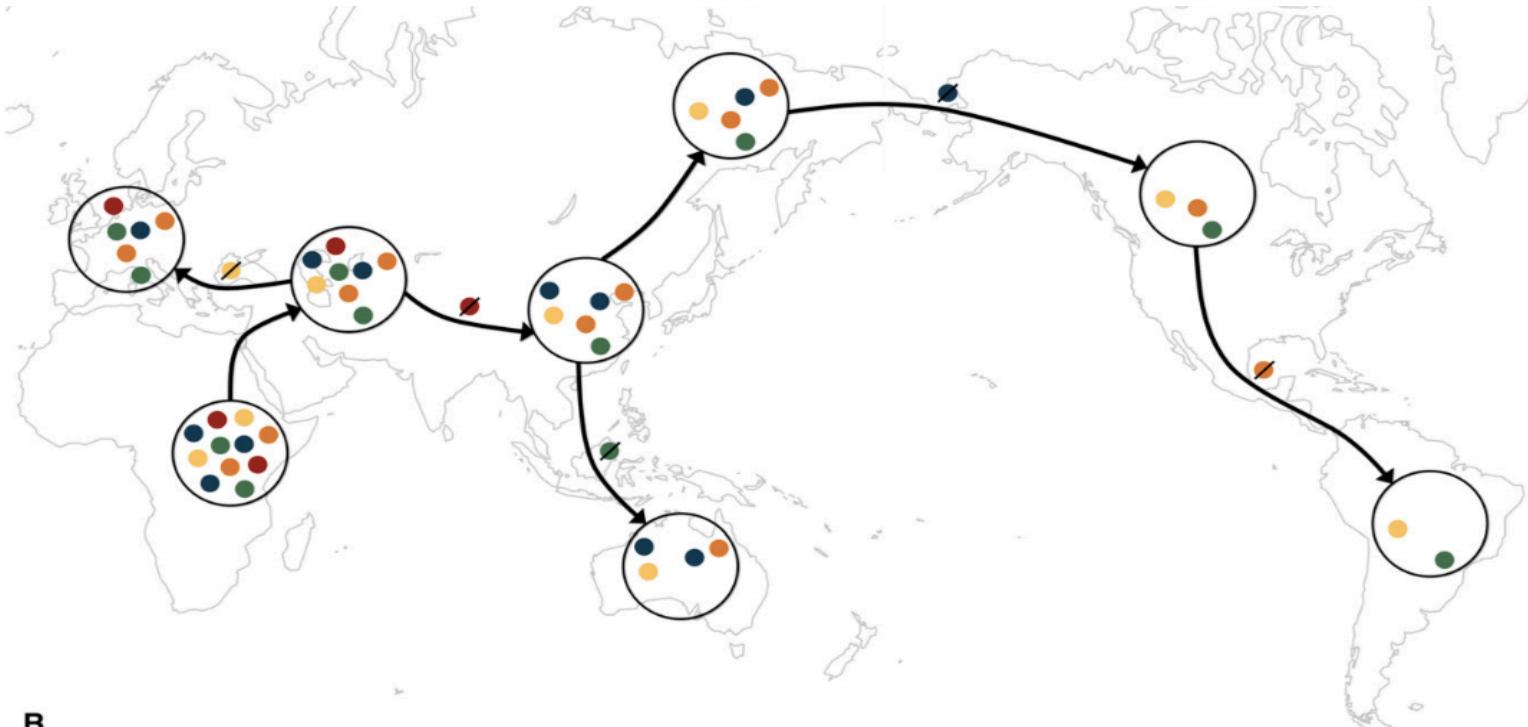
# Qual o destino de uma mutação?



# A Saída do Homem Moderno da África



# A Saída do Homem Moderno da África: Deriva Genética



**Fonte:** Rosenberg NA, Kang JT. Genetic Diversity and Societally Important Disparities. *Genetics*. 2015 Sep;201(1):1-12.

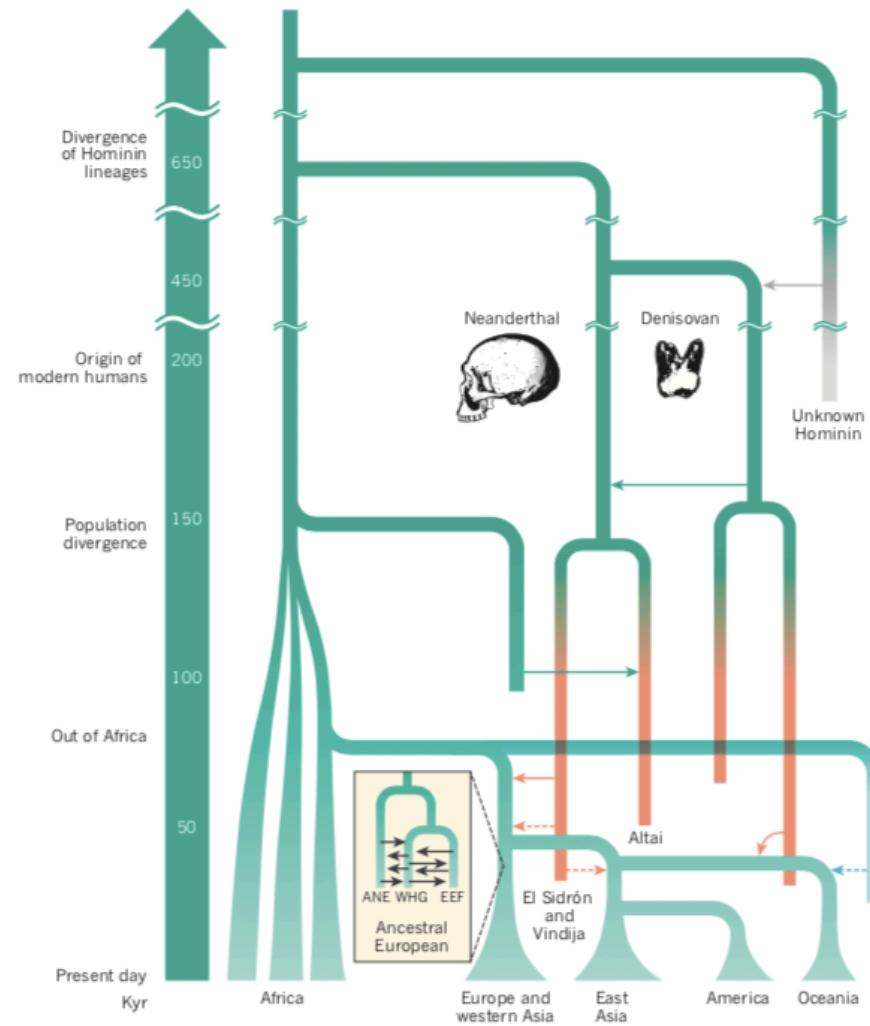


# A Saída do Homem Moderno da África: Seleção Natural



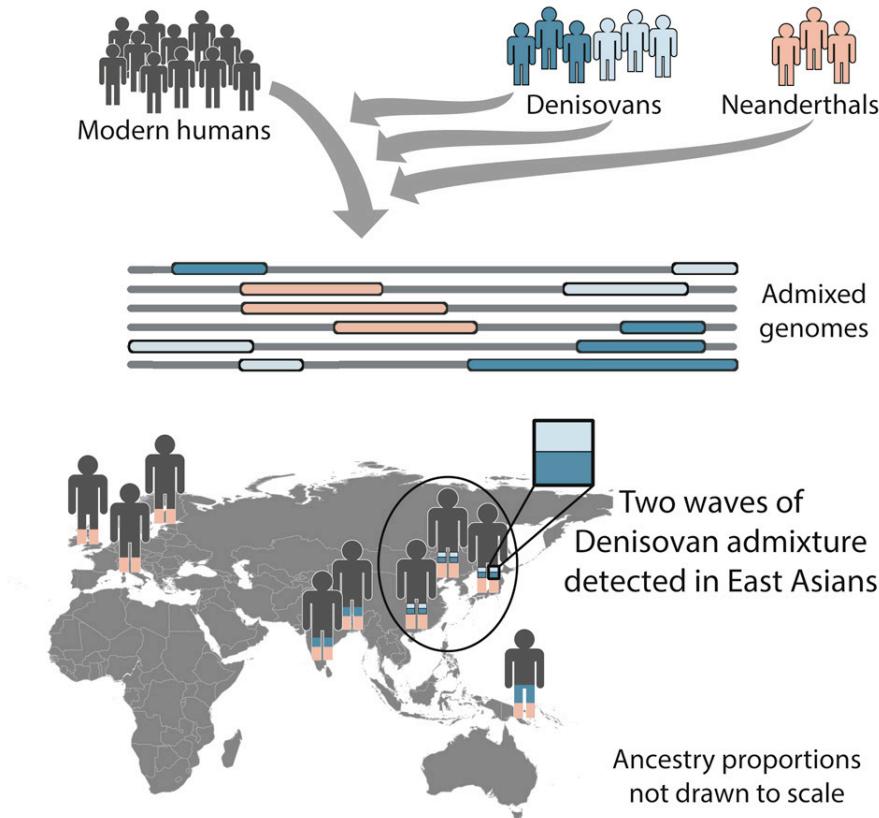
**Fonte:** Tishkoff S. GENETICS. Strength in small numbers. Science. 2015 Sep 18;349(6254):1282-3. doi: 10.1126/science.aad0584.

# A Saída do Homem Moderno da África: O encontro com outros homídeos



**Fonte:** Nielsen R, Akey JM, Jakobsson M, Pritchard JK, Tishkoff S, et al. Tracing the peopling of the world through genomics. *Nature*. 2017 Jan 18;541(7637):302-310

# A introgressão do genoma de outros homíndeos em humanos modernos



**Fonte:** Browning SR, et al. Two Pulses of Archaic Denisovan Admixture. *Cell*. 2018 Mar 22;173(1):53-61.e9.

# Cada população apresenta sua história evolutiva



# O quão diferenciada são as populações humanas?

Sample	Number of regions	Number of populations	Variance components and 95% confidence intervals (%)		
			Within populations	Among populations within regions	Among regions
World	1	52	94.6 (94.3, 94.8)	5.4 (5.2, 5.7)	
World	5	52	93.2 (92.9, 93.5)	2.5 (2.4, 2.6)	4.3 (4.0, 4.7)
World	7	52	94.1 (93.8, 94.3)	2.4 (2.3, 2.5)	3.6 (3.3, 3.9)
World-B97	5	14	89.8 (89.3, 90.2)	5.0 (4.8, 5.3)	5.2 (4.7, 5.7)
Africa	1	6	96.9 (96.7, 97.1)	3.1 (2.9, 3.3)	
Eurasia	1	21	98.5 (98.4, 98.6)	1.5 (1.4, 1.6)	
Eurasia	3	21	98.3 (98.2, 98.4)	1.2 (1.1, 1.3)	0.5 (0.4, 0.6)
Europe	1	8	99.3 (99.1, 99.4)	0.7 (0.6, 0.9)	
Middle East	1	4	98.7 (98.6, 98.8)	1.3 (1.2, 1.4)	
Central/South Asia	1	9	98.6 (98.5, 98.8)	1.4 (1.2, 1.5)	
East Asia	1	18	98.7 (98.6, 98.9)	1.3 (1.1, 1.4)	
Oceania	1	2	93.6 (92.8, 94.3)	6.4 (5.7, 7.2)	
America	1	5	88.4 (87.7, 89.0)	11.6 (11.0, 12.3)	

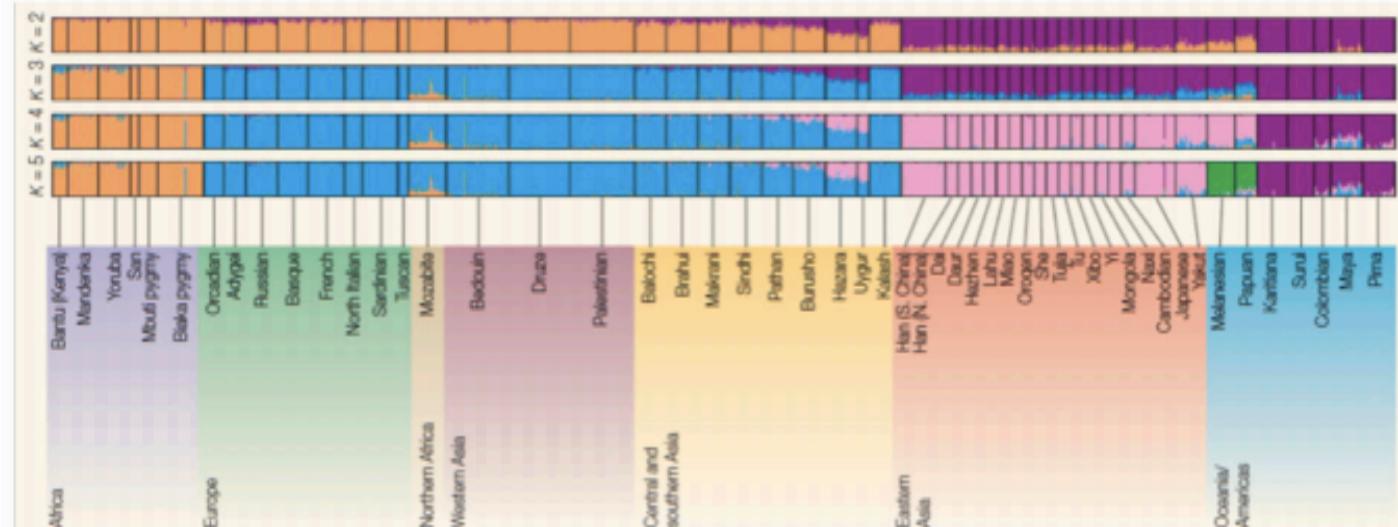
Fonte: Rosenberg NA, Pritchard JK, Weber JL, Cann HM, Kidd KK, Zhivotovsky LA, Feldman MW. Genetic structure of human populations. Science. 2002 Dec 20;298(5602):2381-5.



# O quanto diferenciada são as populações humanas?

## Genetic Structure of Human Populations

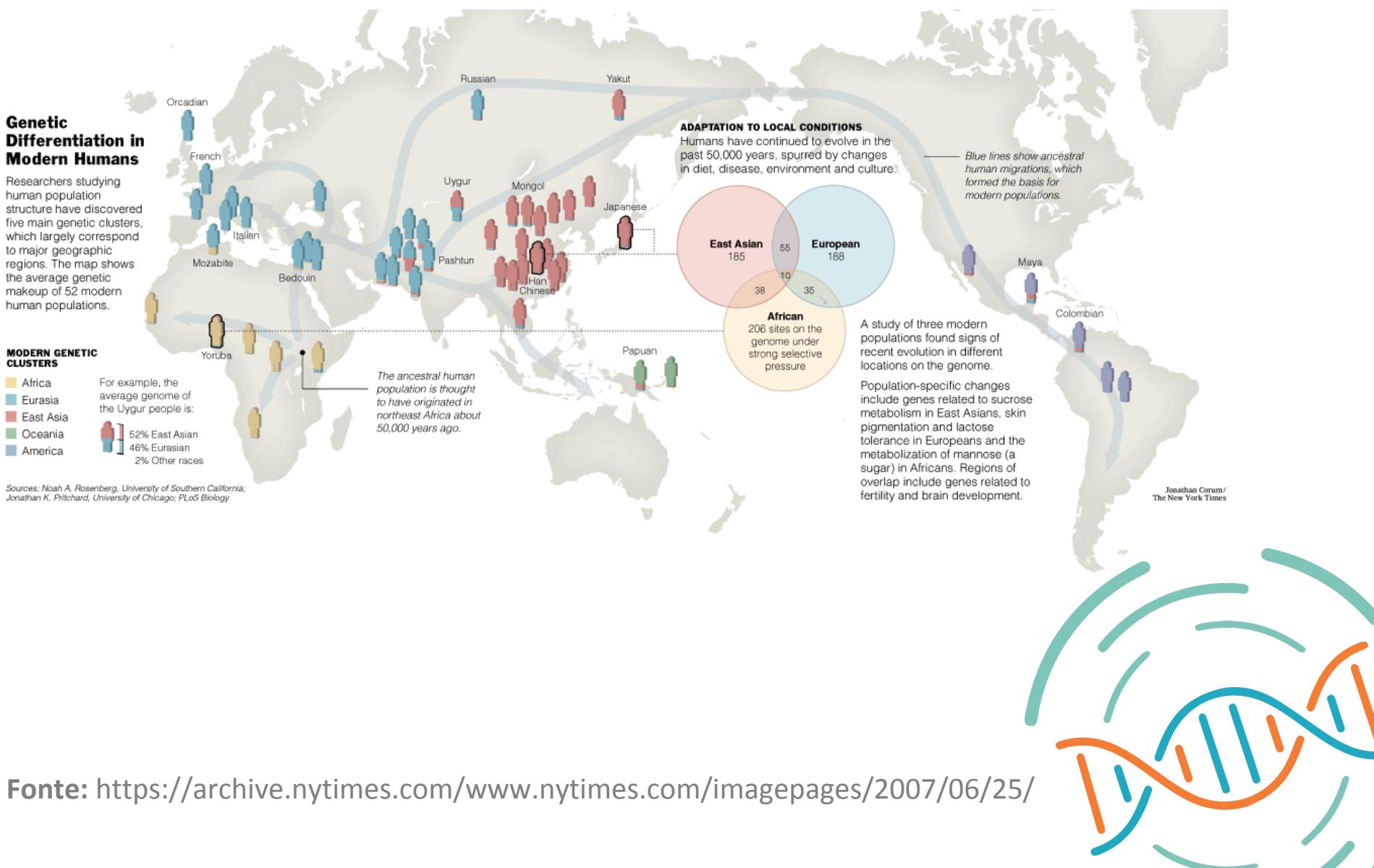
Noah A. Rosenberg,<sup>1\*</sup> Jonathan K. Pritchard,<sup>2</sup> James L. Weber,<sup>3</sup>  
Howard M. Cann,<sup>4</sup> Kenneth K. Kidd,<sup>5</sup> Lev A. Zhivotovsky,<sup>6</sup>  
Marcus W. Feldman<sup>7</sup>



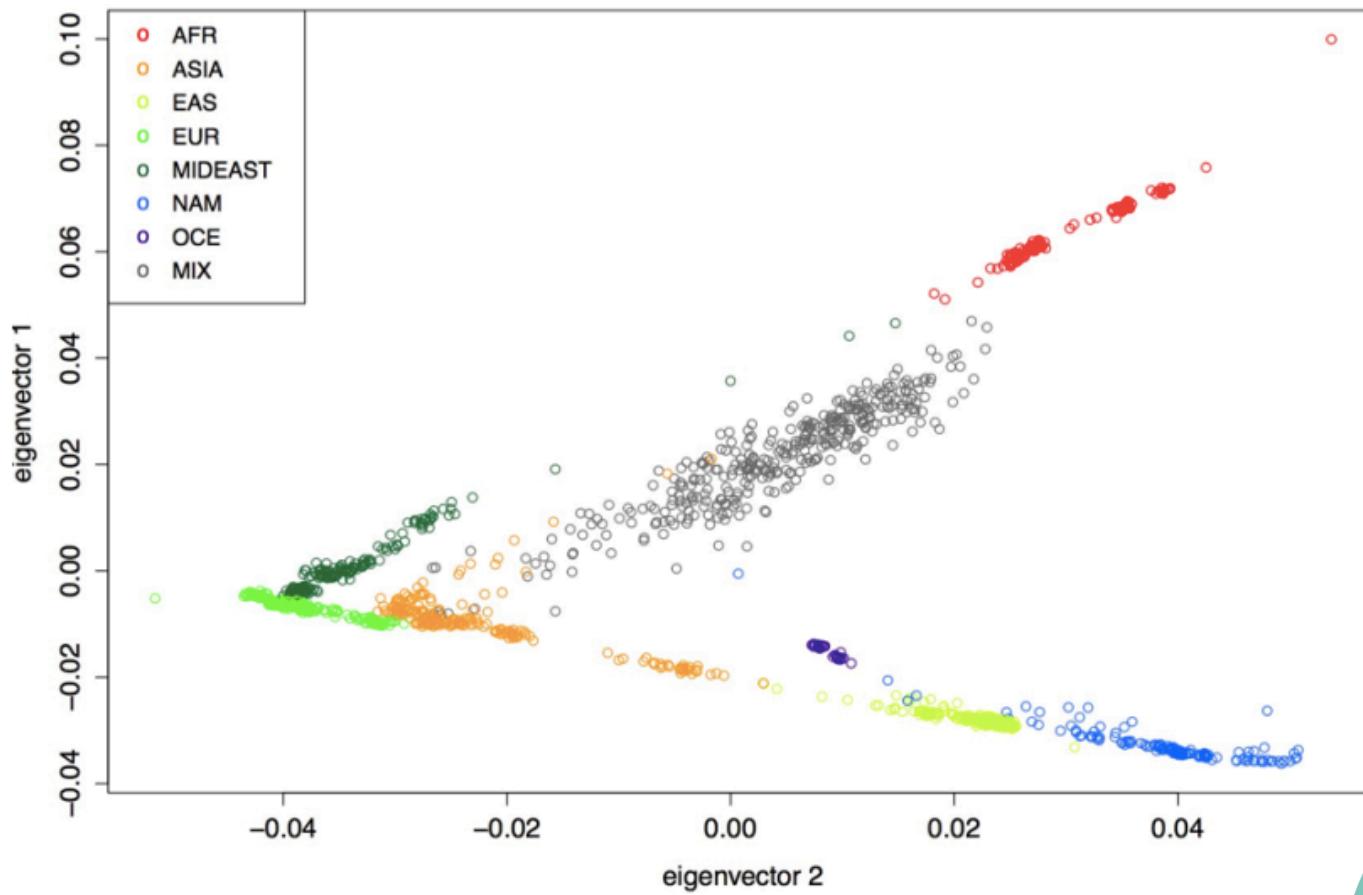
Fonte: Rosenberg NA, Pritchard JK, Weber JL, Cann HM, Kidd KK, Zhivotovsky LA, Feldman MW. Genetic structure of human populations. Science. 2002 Dec 20;298(5602):2381-5.



# O quanto diferenciadas são as populações humanas?



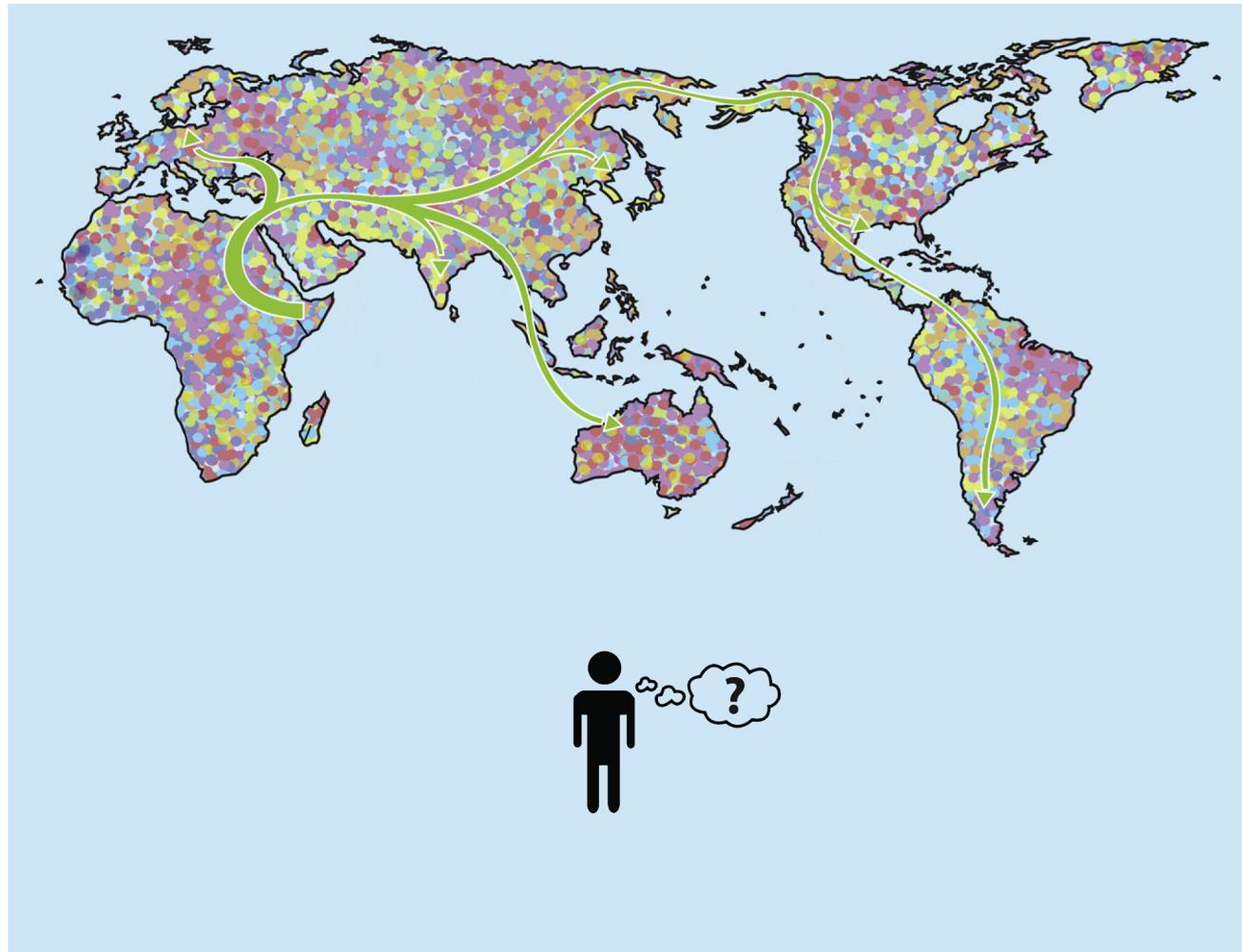
# O quanto diferenciada são as populações humanas?



Fonte: Dados do LABORATÓRIO GENEVOL - IBUSP



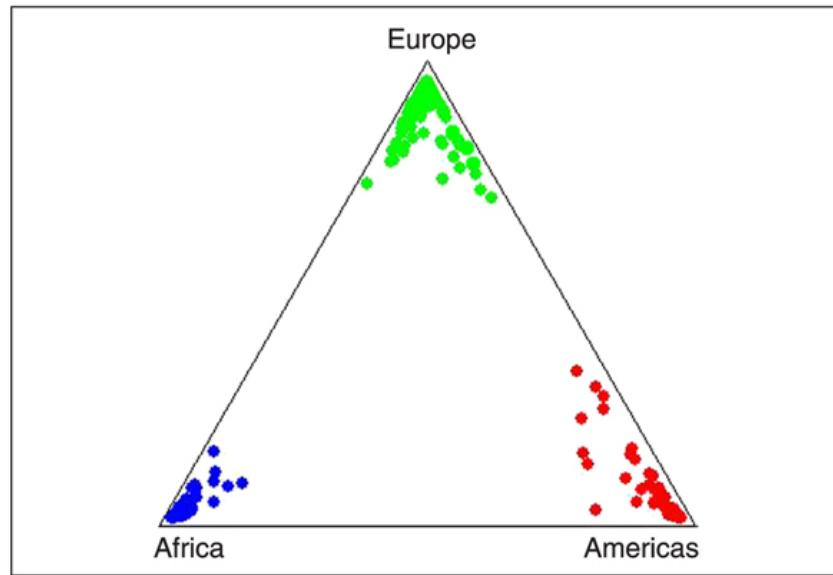
# Como é possível atribuir a ancestralidade genética?



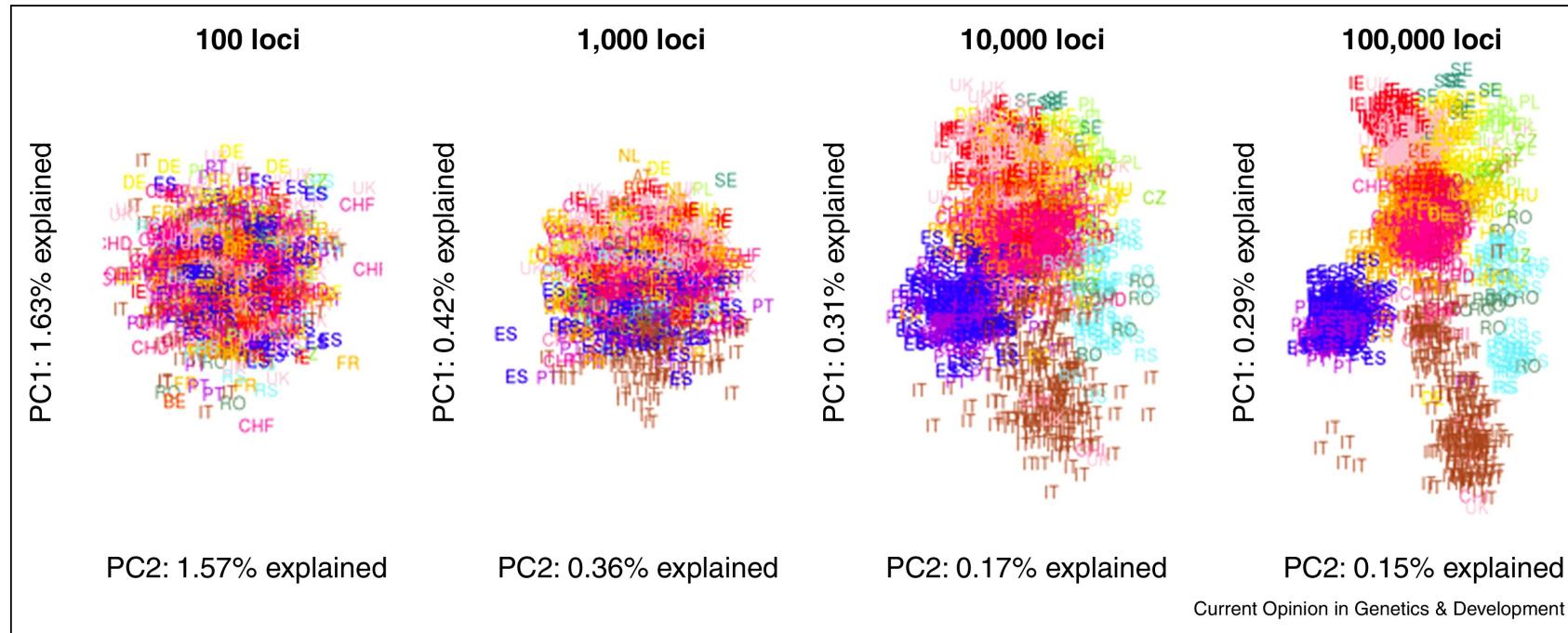
# Ancestralidade genética: Era pré-genômica

Marcadores Informativos de Ancestralidade (AIMs): dezena de marcadores

FREQ	AFR	EUR	NAM
Marcador 1	92%	5%	1%
Marcador 2	0%	95%	3%
Marcador 3	3%	2%	100%



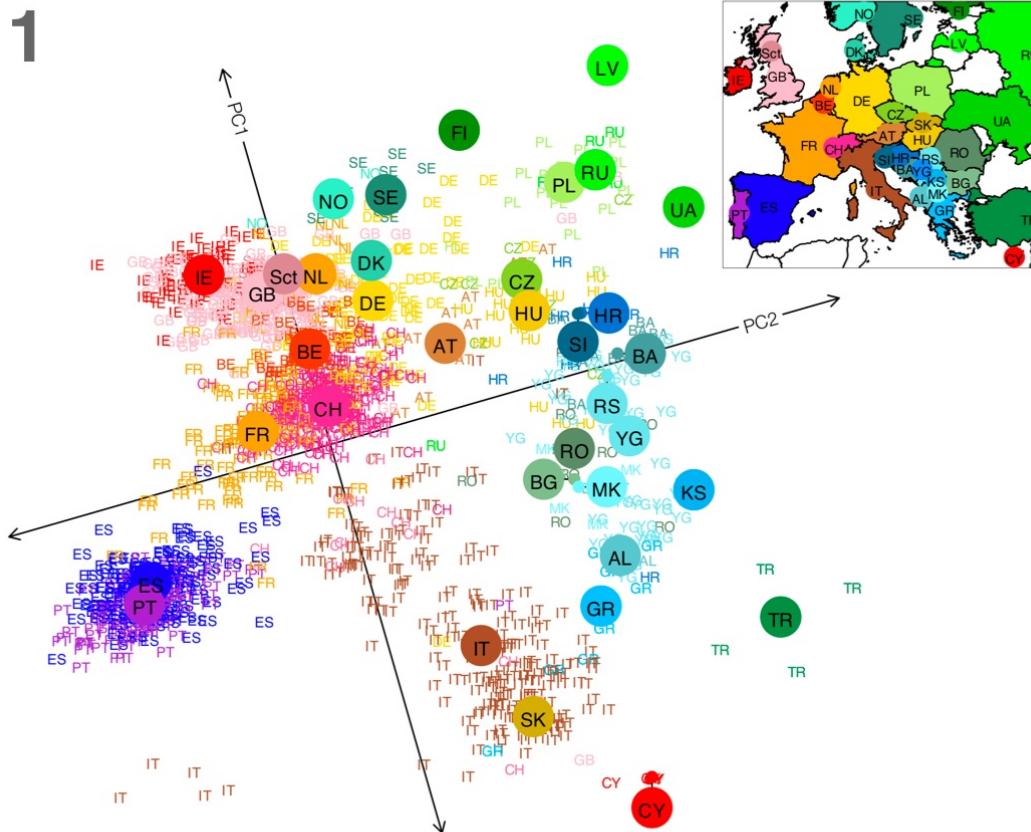
# Ancestralidade genética: Era Genômica



**Fonte:** John Novembre, Benjamin M Peter (2016) Recent advances in the study of fine-scale population structure in humans. Current Opinion in Genetics & Development



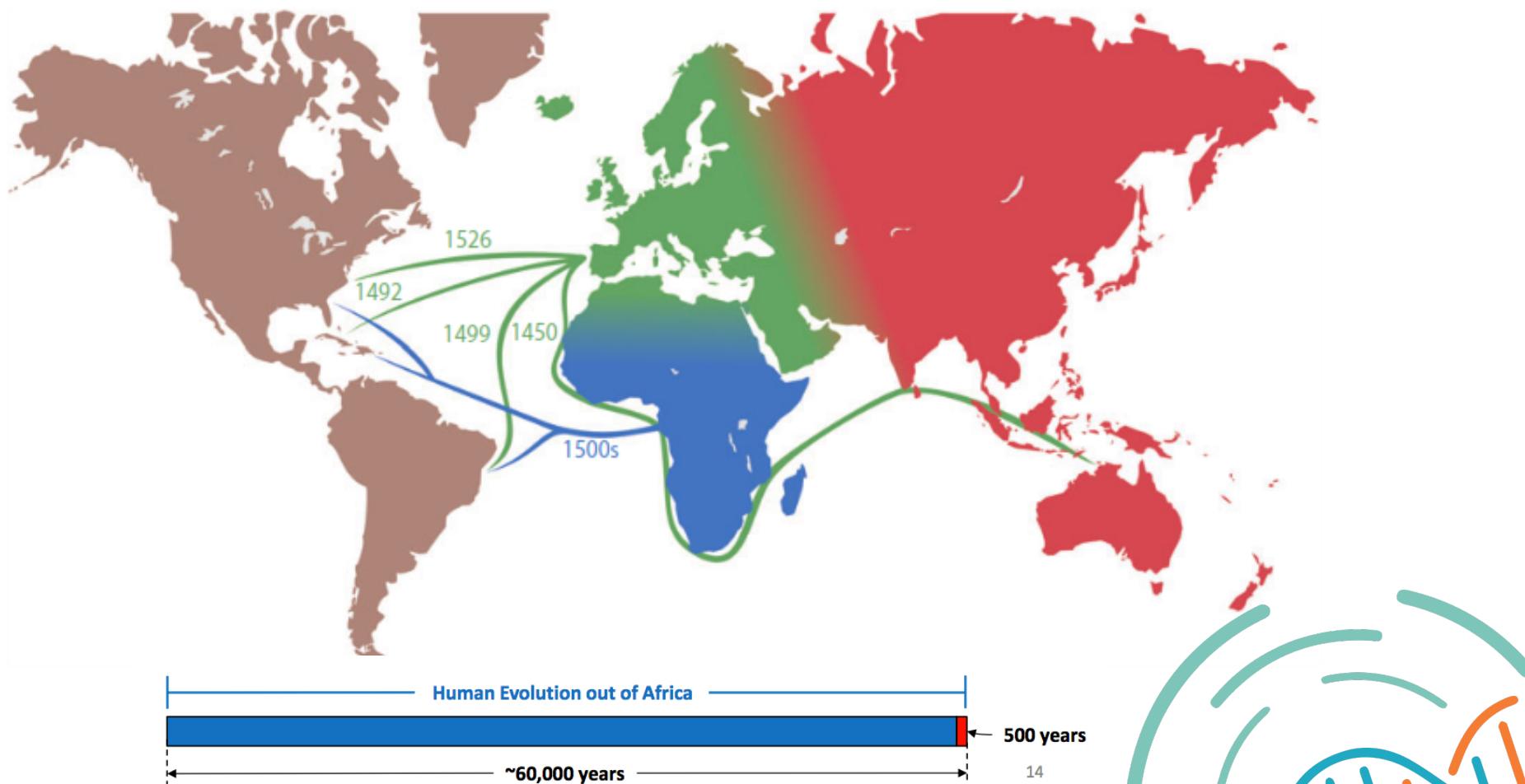
# Ancestralidade genética: Era Genômica



**Fonte:** Novembre J, et al . Genes mirror geography within Europe. Nature. 2008 Nov 6;456(7218):98-101.

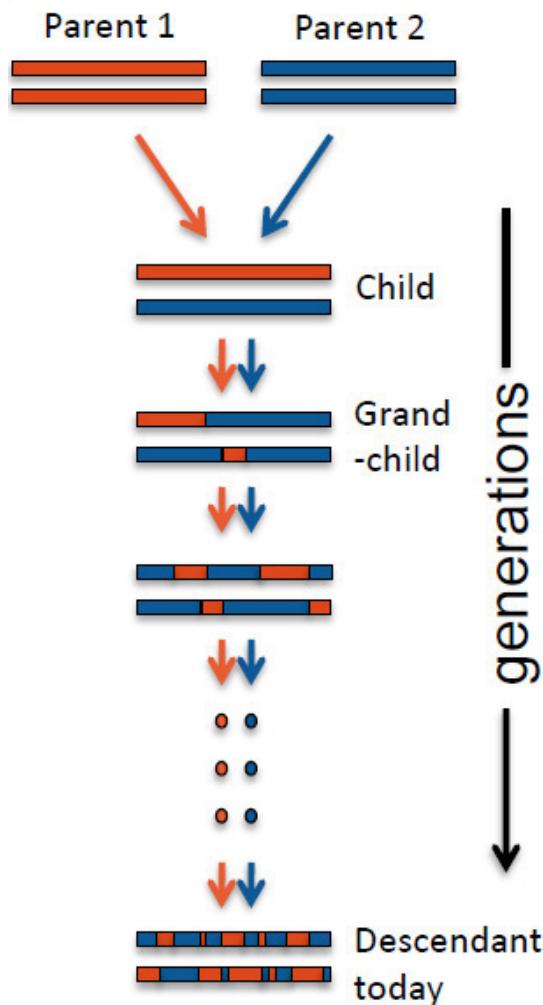


# As grandes navegações e o processo de miscigenação



Fonte: Winkler CA, Nelson GW, Smith MW. Admixture mapping comes of age. Annu Rev Genomics Hum Genet. 2010;11:65-89.

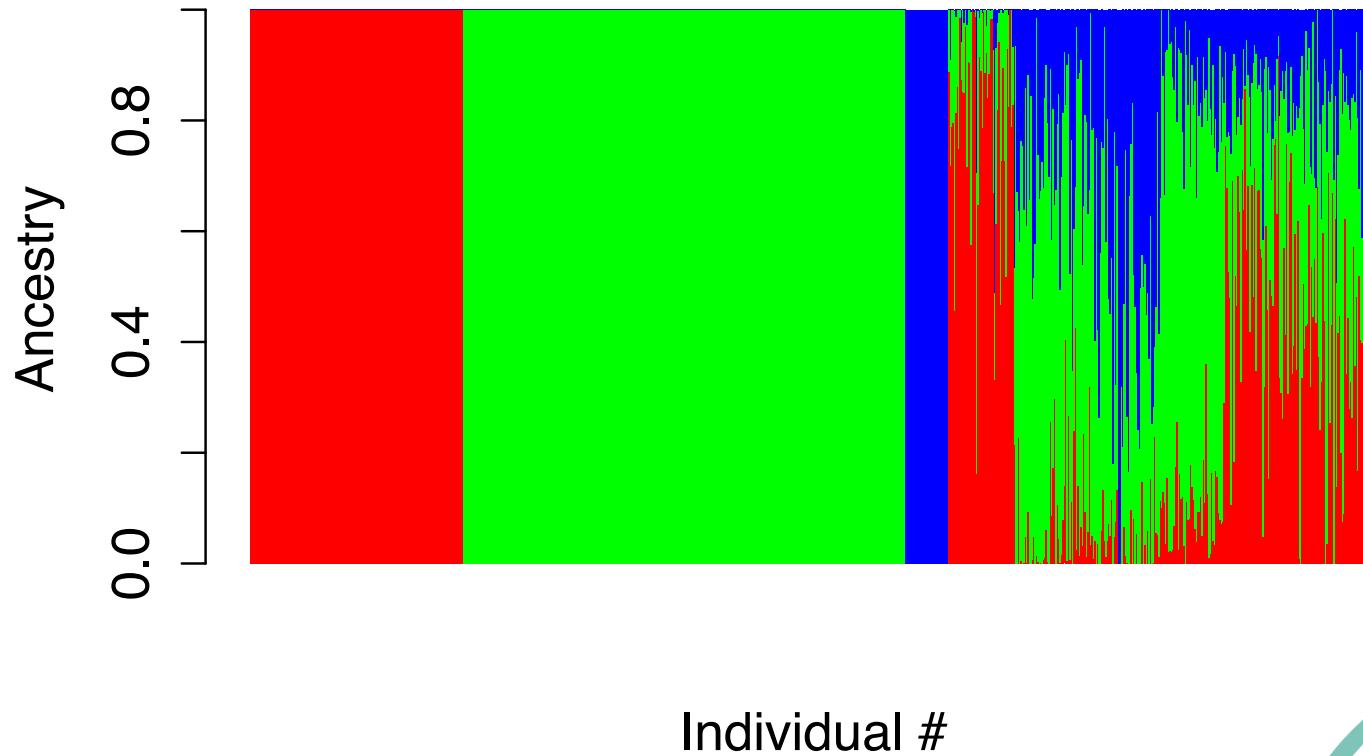
# Como é o Genoma de um indivíduo miscigenado?



Fonte: Winkler CA, Nelson GW, Smith MW. Admixture mapping comes of age. Annu Rev Genomics Hum Genet. 2010;11:65-89.



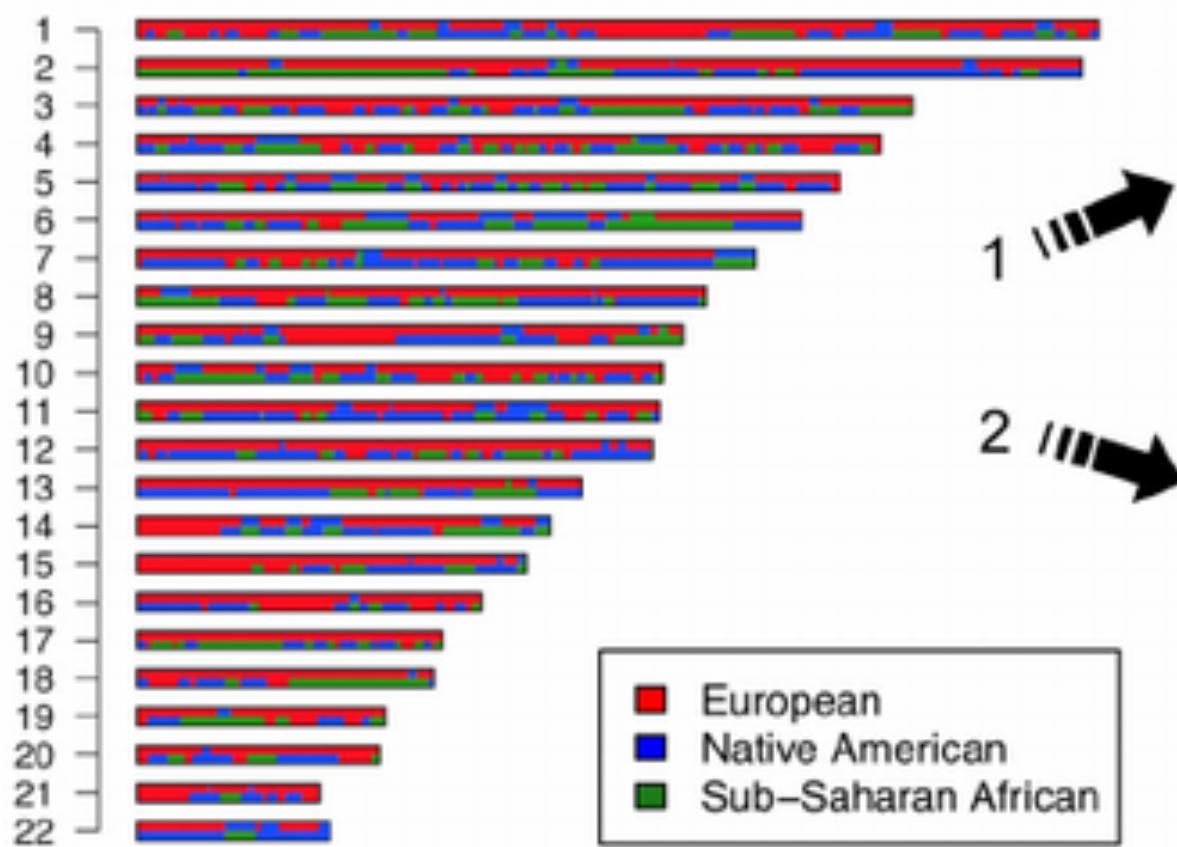
# Ancestralidade Global



Fonte: Dados do LABORATÓRIO GENEVOL - IBUSP



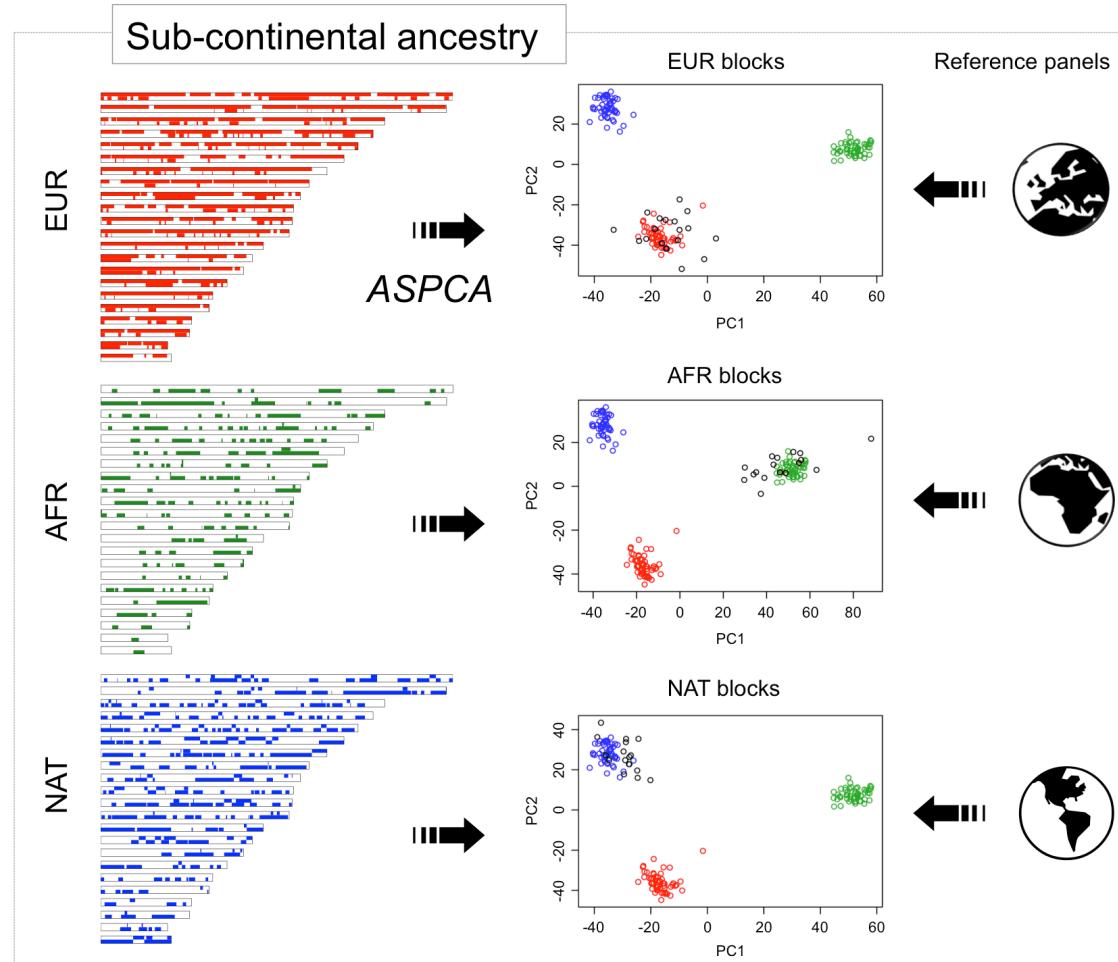
# Ancestralidade Local



Fonte: Moreno-Estrada A et al. Reconstructing the population genetic history of the Caribbean. PLoS Genet. 2013 Nov; 9(11):e1003925.



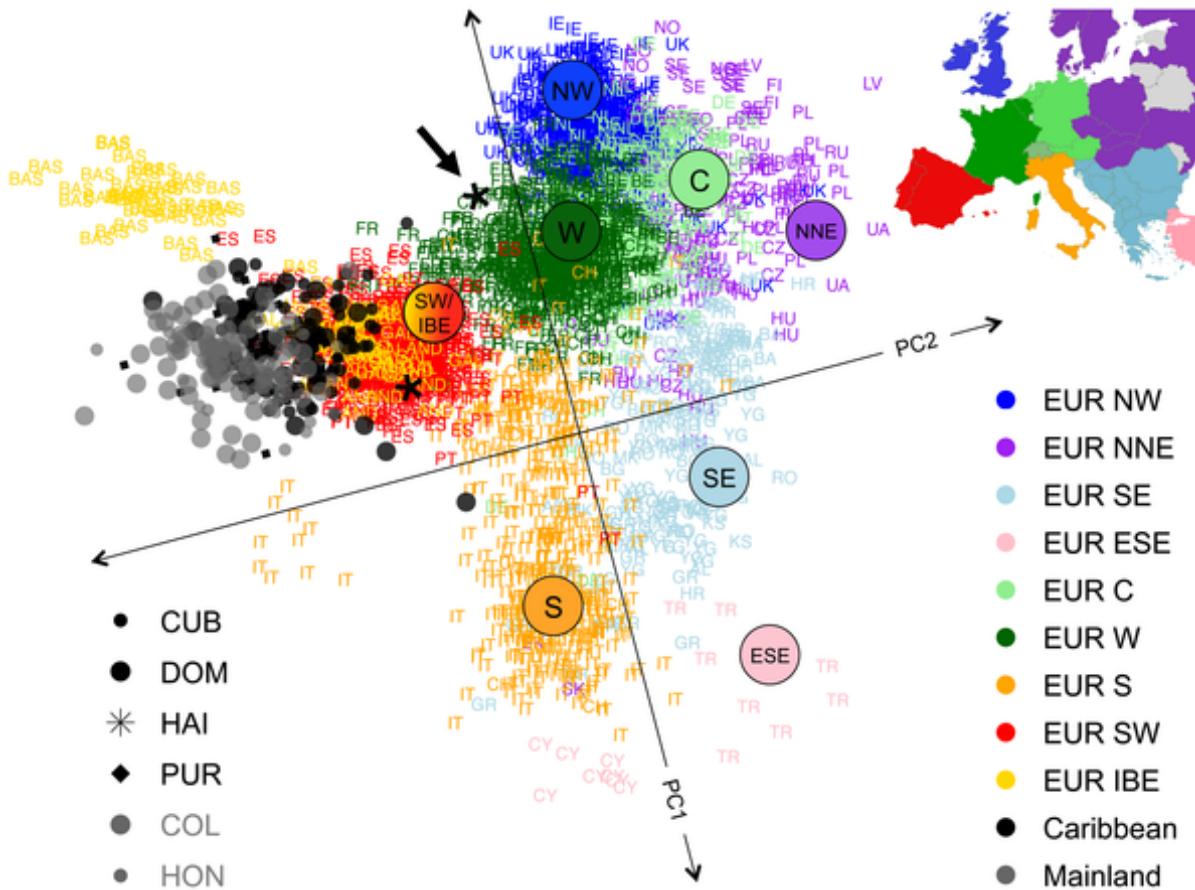
# Ancestralidade Subcontinental



Fonte: Moreno-Estrada A et al. Reconstructing the population genetic history of the Caribbean. PLoS Genet. 2013 Nov; 9(11):e1003925.



# Ancestralidade Subcontinental



**Fonte:** Moreno-Estrada A et al. Reconstructing the population genetic history of the Caribbean. PLoS Genet. 2013 Nov; 9(11):e1003925.

