

Studying historical movements of populations using DNA

Garrett Hellenthal

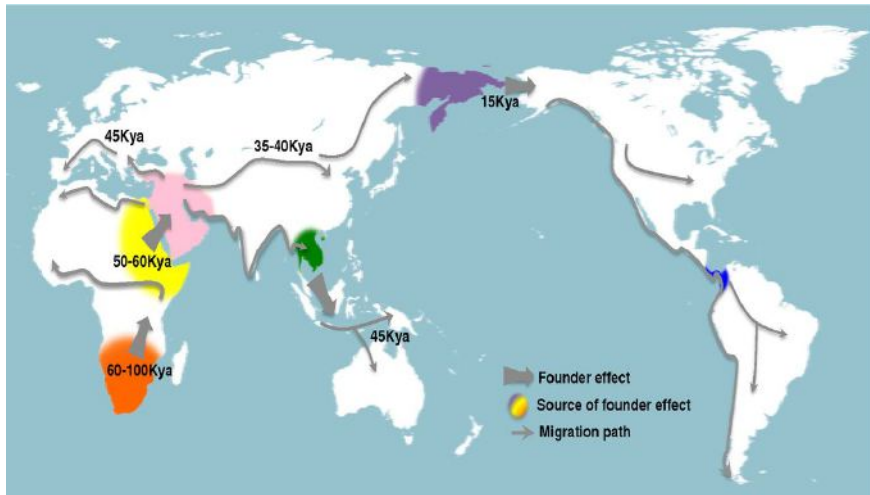
University College London

Adelphi Genetics Forum Teacher's Conference
Manchester
June 28, 2024

Outline

- 1 Background
- 2 Genetic “clusters”
- 3 Detecting intermixing (“admixture”) among groups
 - examples of recent (<4,500 years ago) intermixing
 - recent intermixing in African populations
 - examples of older (>4,500 years ago) intermixing
- 4 Spreading adaptive genetic variants through intermixing
- 5 Conclusions

Ancestral history of modern humans



(Henn et al 2012, *PNAS* 109:17758)

Genetic Variation data

- you inherit a sequence of ≈ 3 billion A,G,C,Ts from each parent

A A G G T C A A G T A T C
A A G G T C A A G T A T C

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G

T

G

T

G

T

T

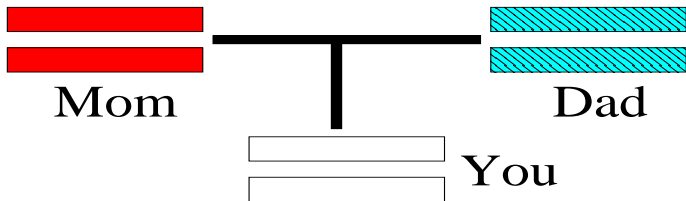
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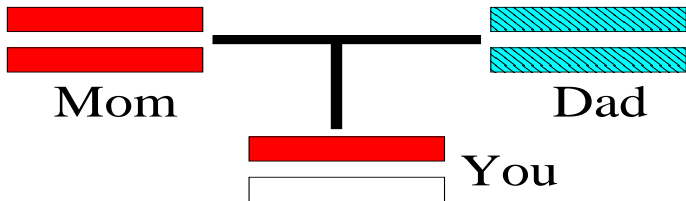
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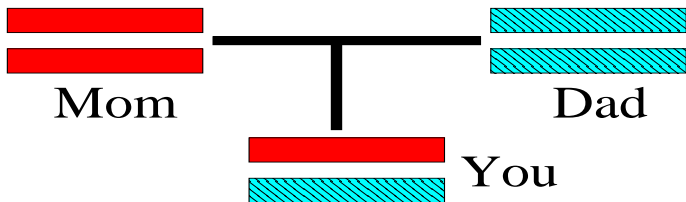
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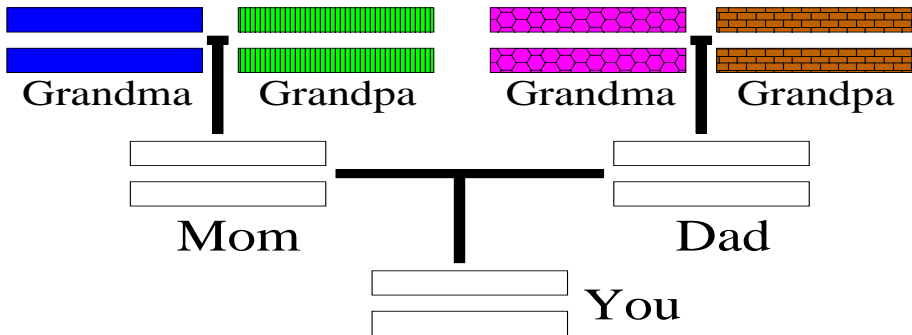
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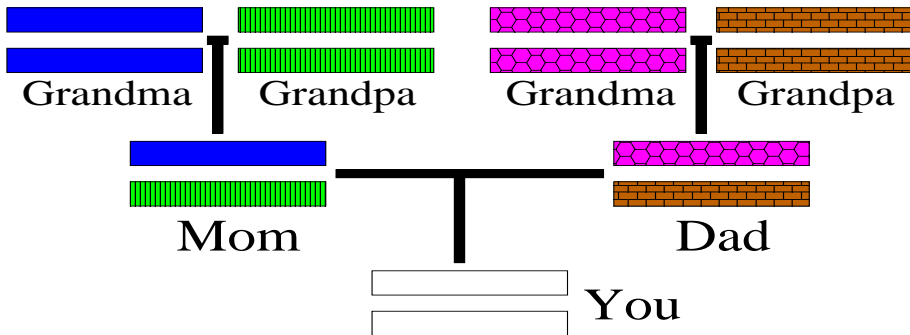
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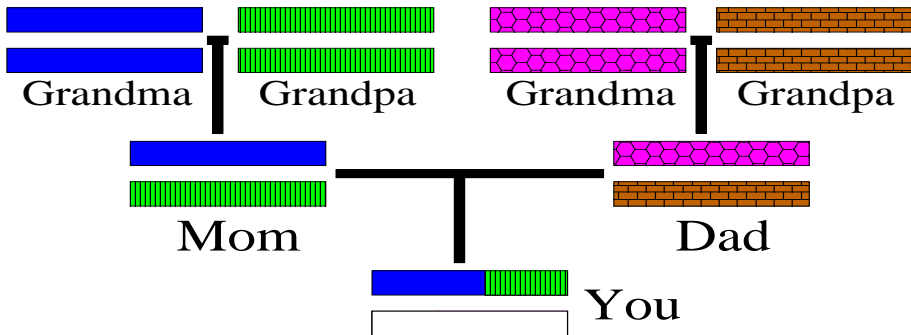


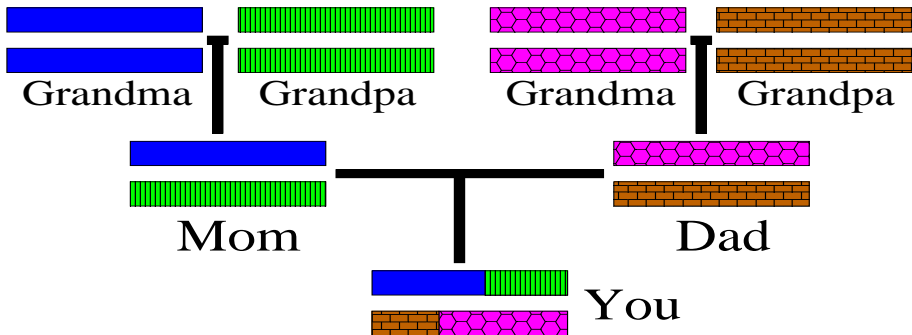


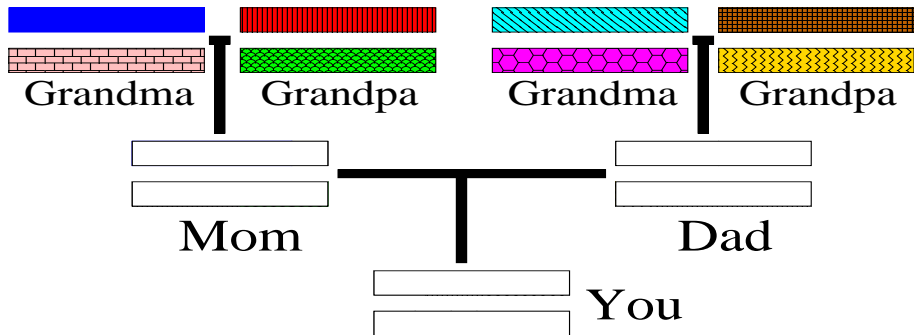


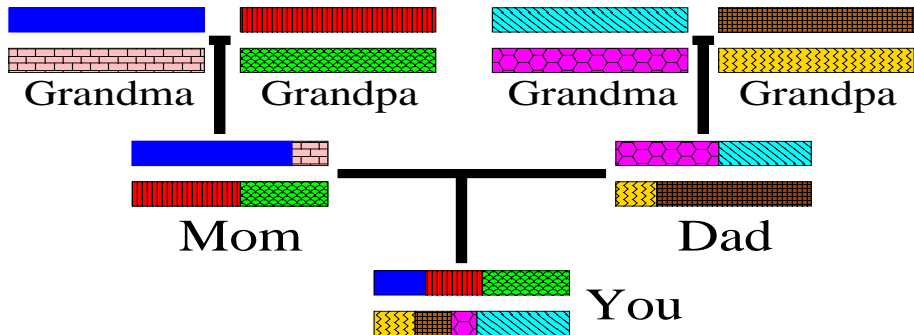


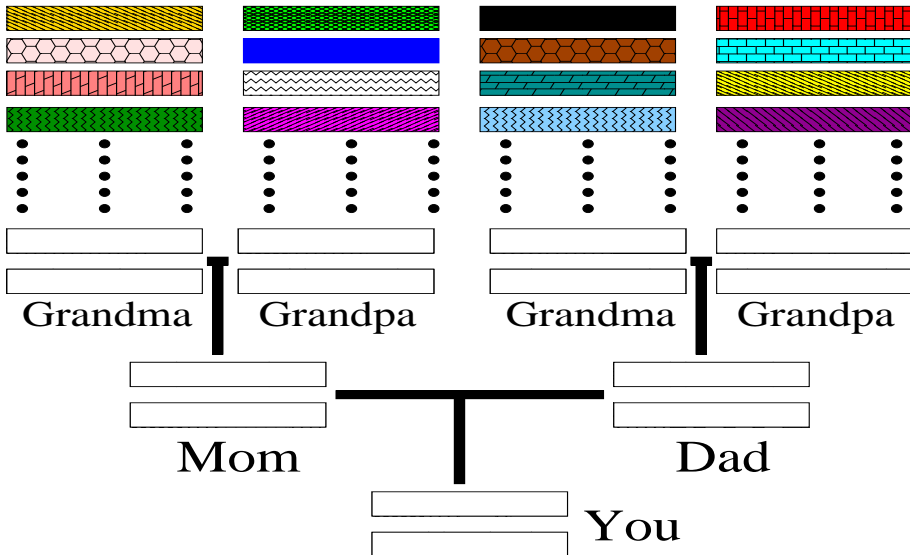


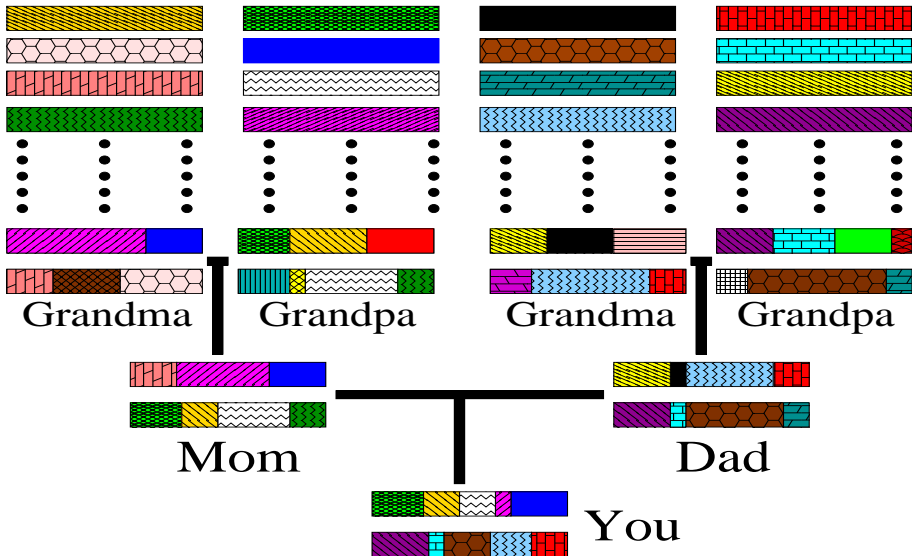


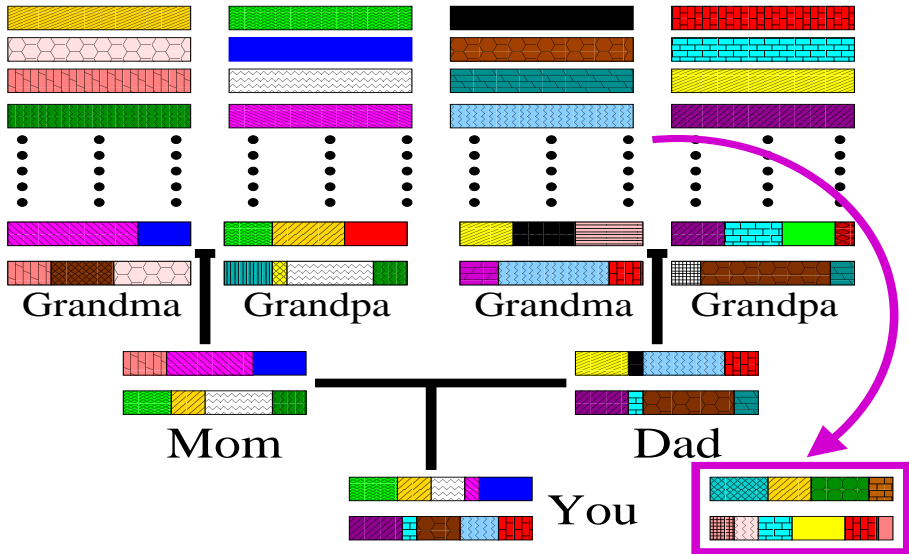


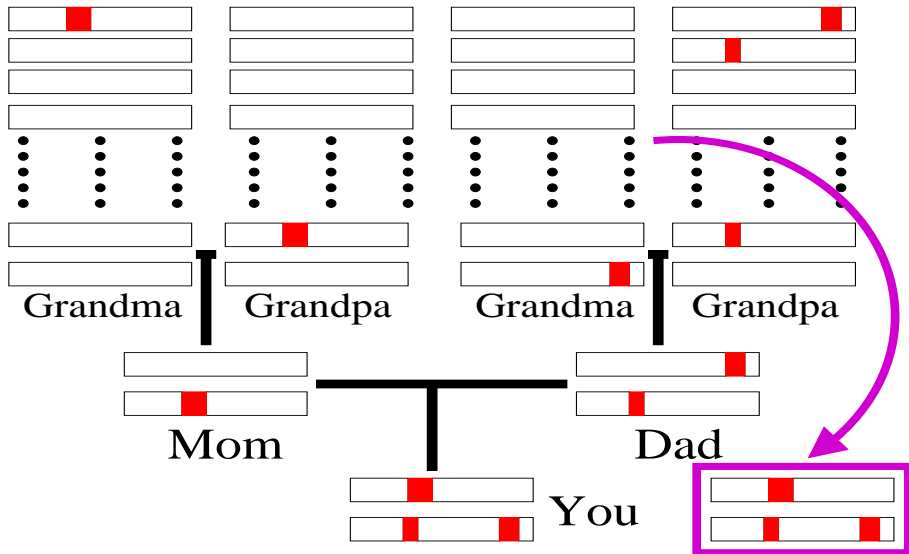




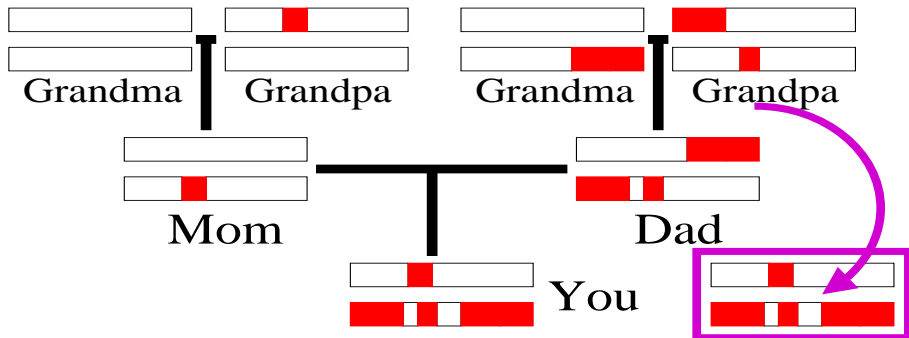




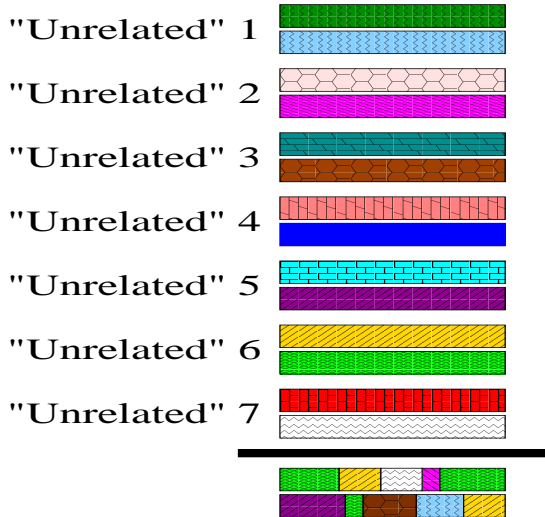




you and a stranger share **DNA segments**, inherited from the same ancestors that lived (e.g.) 10 generations ago



shared **DNA segments** are longer if inherited from a more recent ancestor (e.g. same grandparents)



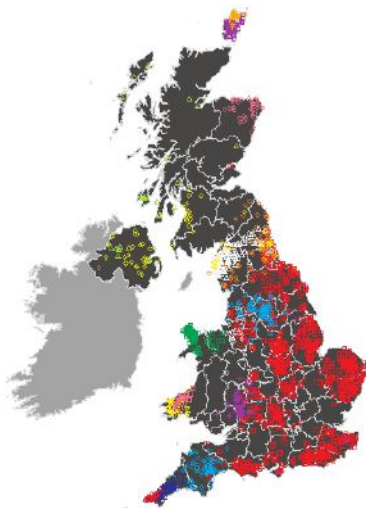
- compare genetic data of "unrelated" people
- for person at bottom, infer who they share long DNA sequence matches (and hence **more recent ancestors**) with

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Genetic "clusters" in the United Kingdom

- 2,039 people collected across rural areas of UK
- for each person, all four of their grandparents were born within 80km of one another
- **colors** reflect genetically similar people



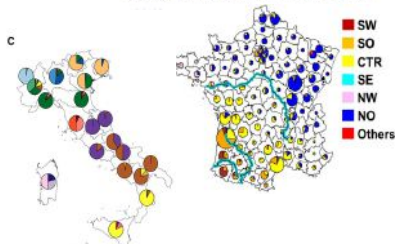
(Leslie et al 2015, *Nature* 519:309-314)

Article | Open Access | Published: 11 September 2020

Dutch population structure across space, time and GWAS designRita F. Byrne¹, Victor van Heerden, Project M-E ACS GWAS Consortium, Leonard H. van den Berg, Jan H. Vekke & Russel L. M. Maughan¹

Nature Communications 11, Article number 4556 (2020) | Cite this article

5820 Accesses | 2 Citations | 49 Altmetrics | Metrics

**The genetic history of France**Aude Saint-Pierre¹, Joana Garcia^{2,3}, Isabel Alves^{4,5}, Valérie Karstchhoff², Marina Gaultier², Philippe Anagnostis³, Jean-François Garavito⁴, Christophe Zalcou⁴, Martial Morlet⁵, Pilar Galan⁶, Serge Hercowitz⁶, Joaquin Mathieson⁷, Richard Peltre⁸, Emmanuel Colin^{1,9} and Christian Ginès^{1,2}

Article | Open Access | Published: 01 February 2019

Patterns of genetic differentiation and the footprints of historical migrations in the Iberian PeninsulaDerek Byrnfelt, Grego Fernández-Rodríguez, Clara-Pau Fonto, Inés Quintela, Angel Garracido, Peter Donnelly & Simon Myers¹

Nature Communications 10, Article number 151 (2019) | Cite this article

188 Accesses | 26 Citations | 886 Altmetrics | Metrics

**Genetic history of the population of Crete**

Petros Drineas, Fotis Tzietsos, Anna Plantinga, Iosif Lazaridis, Evangelia Yannaki, Anna Riazou, Katerina Kanaki, Marolets Michalodimitrakis, Francisco Perez-Jimenez, Giustina De Silvestro ... See all authors >

RESEARCH ARTICLE**Population structure of modern-day Italians reveals patterns of ancient and archaic ancestries in Southern Europe**A. Barone^{1,2,3}, S. Anzi^{1,2,3}, F. Barbieri^{1,2,3}, G. Altanbonini¹, S. Balari¹, G. Bolla¹, & S. Sarno¹,
* See all authors and affiliations

Article | Published: 17 September 2018

Genomic history of the Sardinian populationChangshou W. A. Chiang^{1,2}, Joseph H. Martin, Carlo Sidoti, Agnès Bordinado, Marwan Al-Aadi, Margherita Zolotarewska, Mariastella Pittalis, Ines Boonman, Andrea Marchio, Sergio Palla, M. Stari, Andrew Angelis, Kim E. Lohmeyer, Daniela R. Ribeiro, David Schlesselman, Francesco Ciullo, John Novembre^{1,2}

Nature Genetics 50, 1428–1434 (2018) | Cite this article

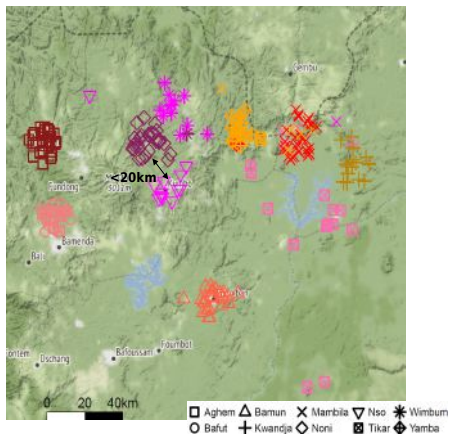
1819 Accesses | 52 Citations | 514 Altmetrics | Metrics



Fine-scale genetic structure within Grassfields of Cameroon



Colour=Genetic cluster
Shape= Ethnic group

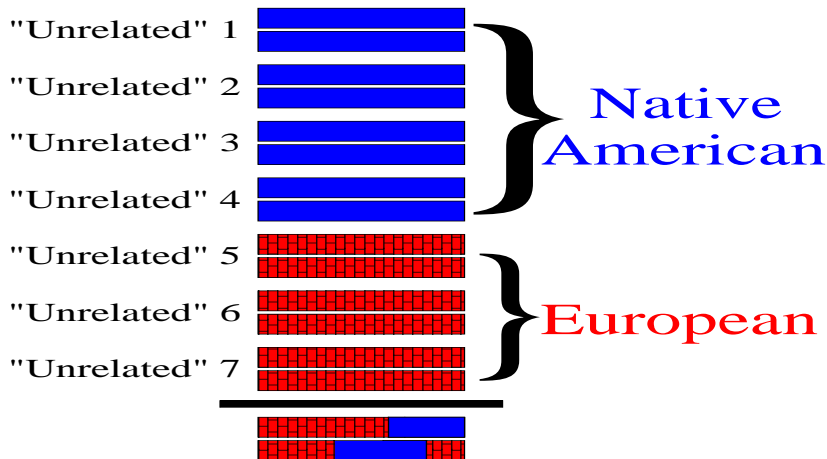


(Bird et al 2023, *Sci Adv* 9:eabq2616)

Outline

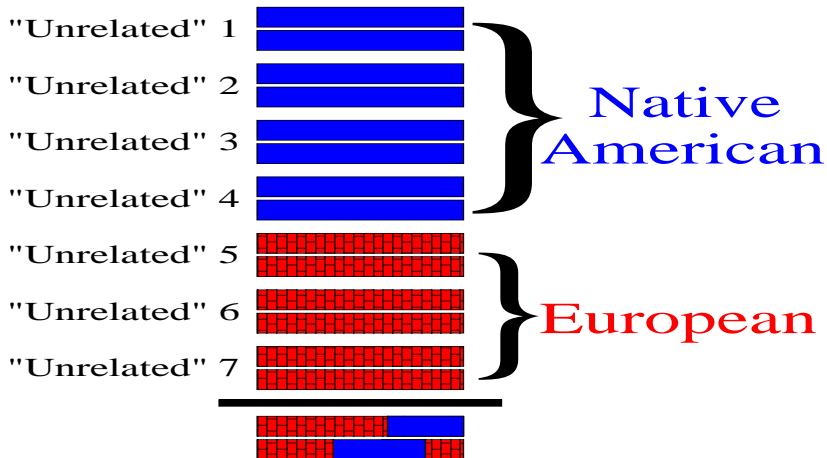
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Detecting when groups intermixed



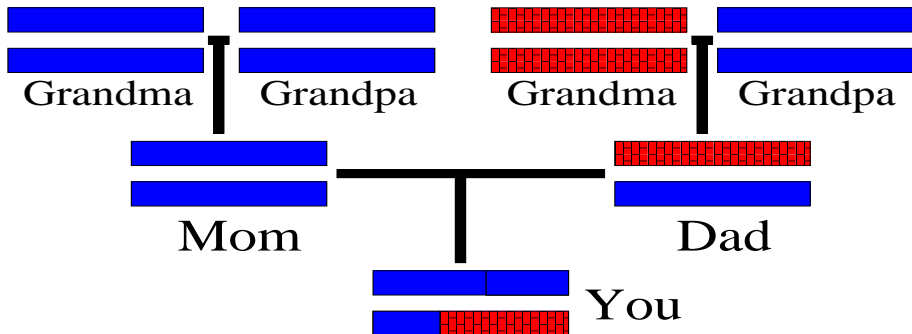
Example: Latin Americans

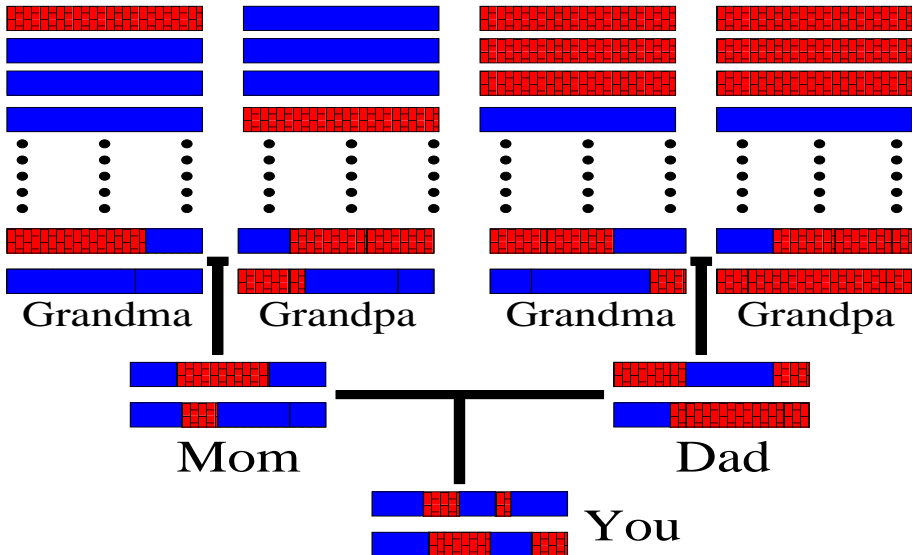
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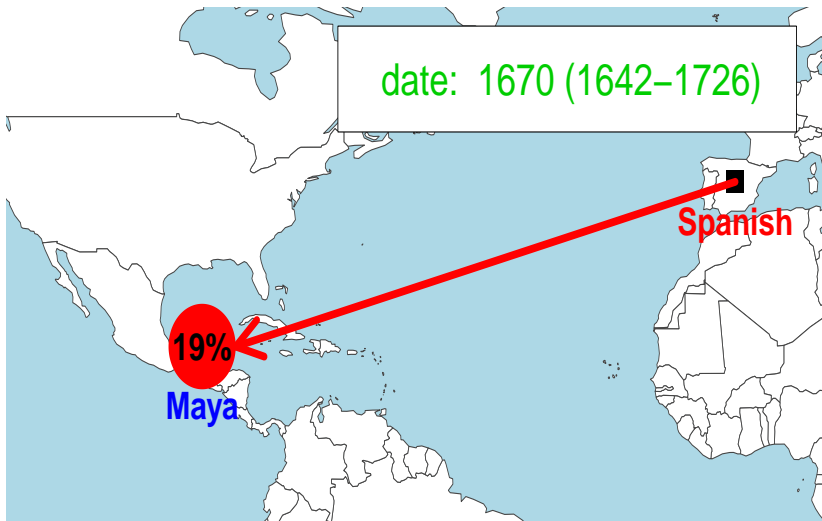
Example: Latin Americans

→ can date when the groups intermixed by **sizes** of **blue** and **red** segments



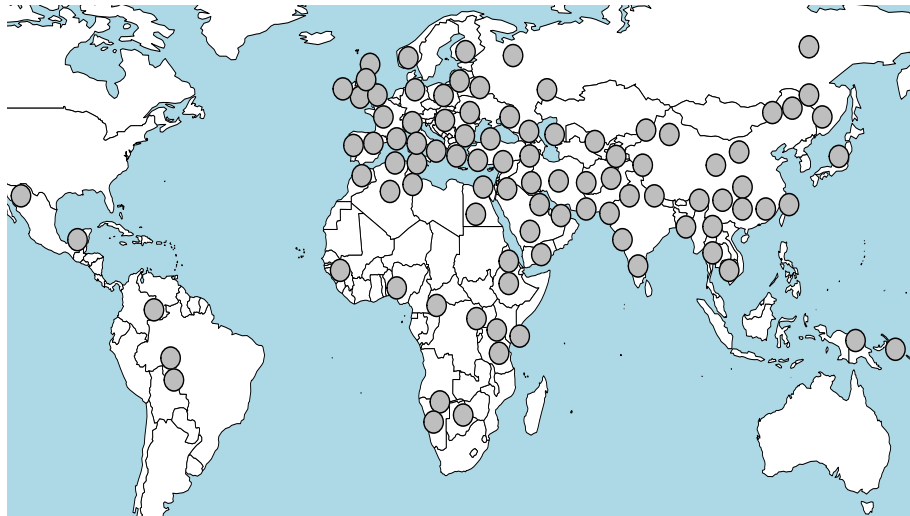


Maya – DNA inherited from colonial-era **Europeans**

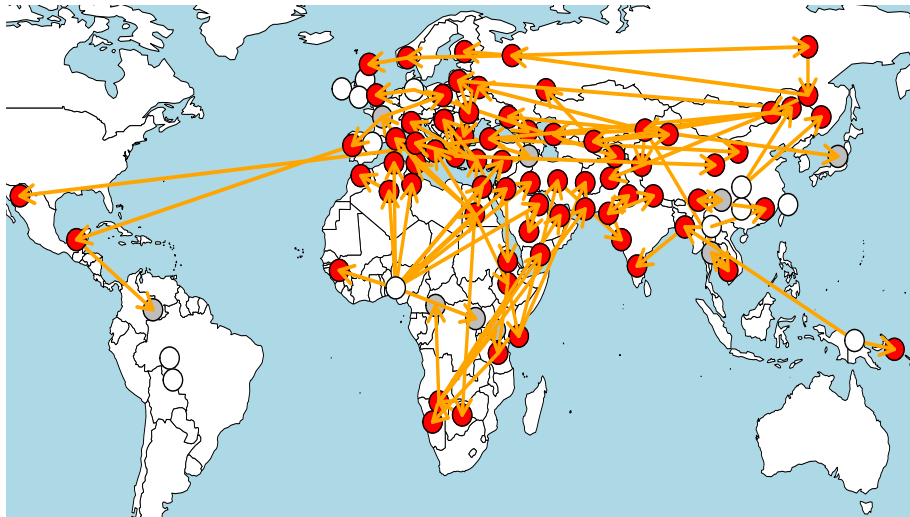


(Hellenthal et al 2014, *Science* **343**:747)

Inferred recent (<4,500 years ago) intermixing events



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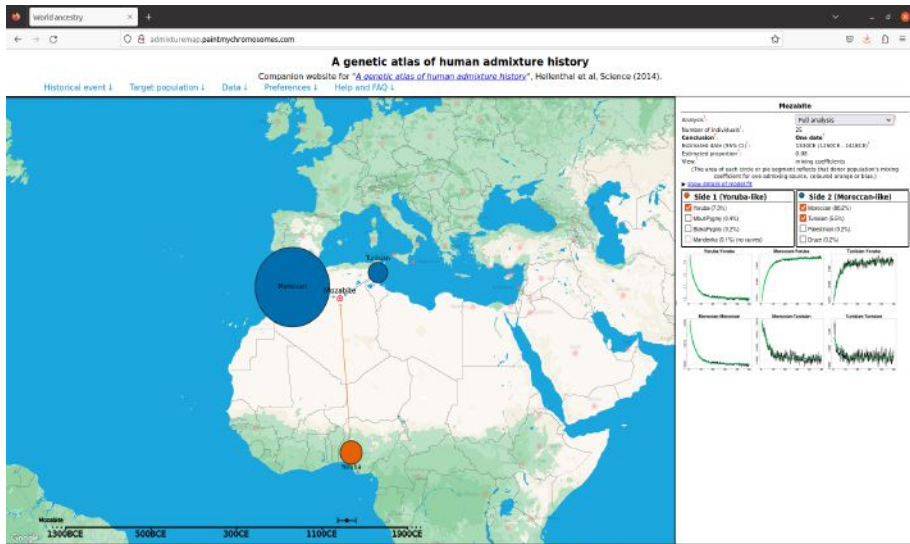
www.admixturemap.paintmychromosomes.com

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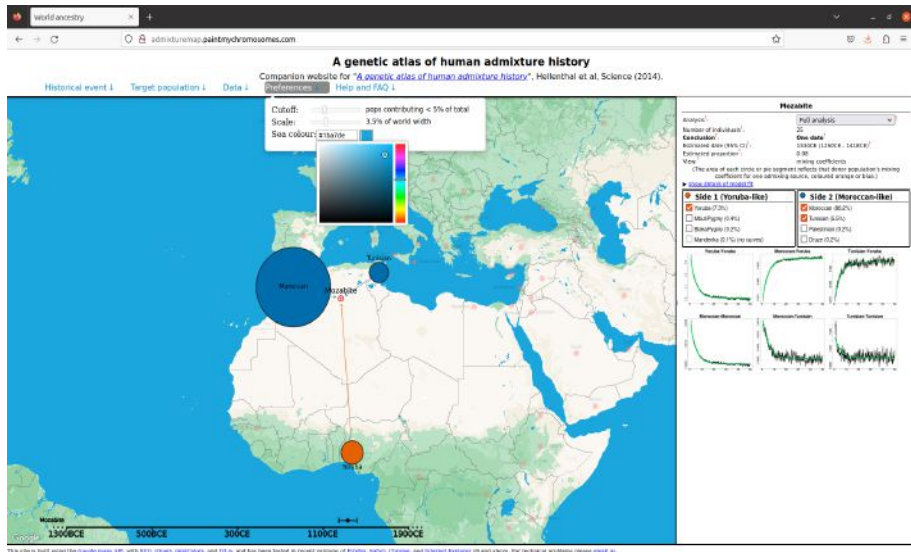
(Gavin Band)

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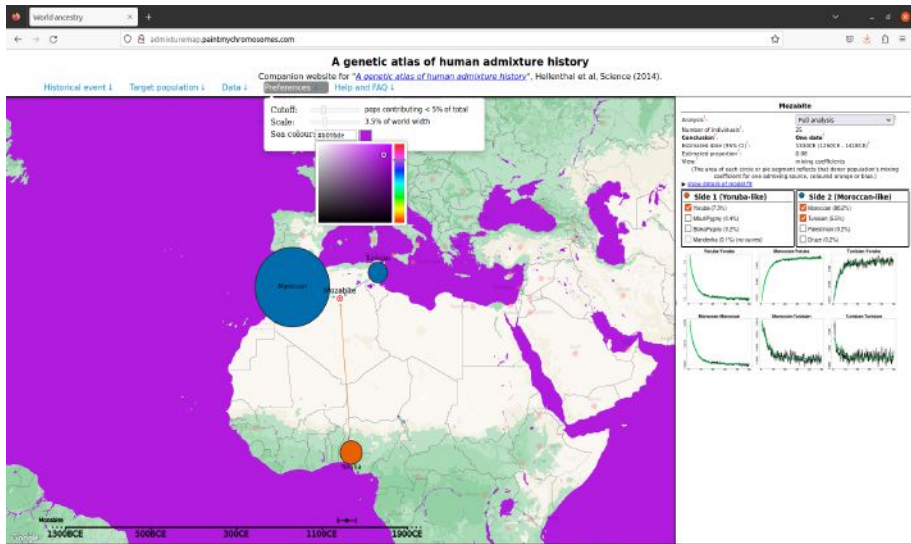
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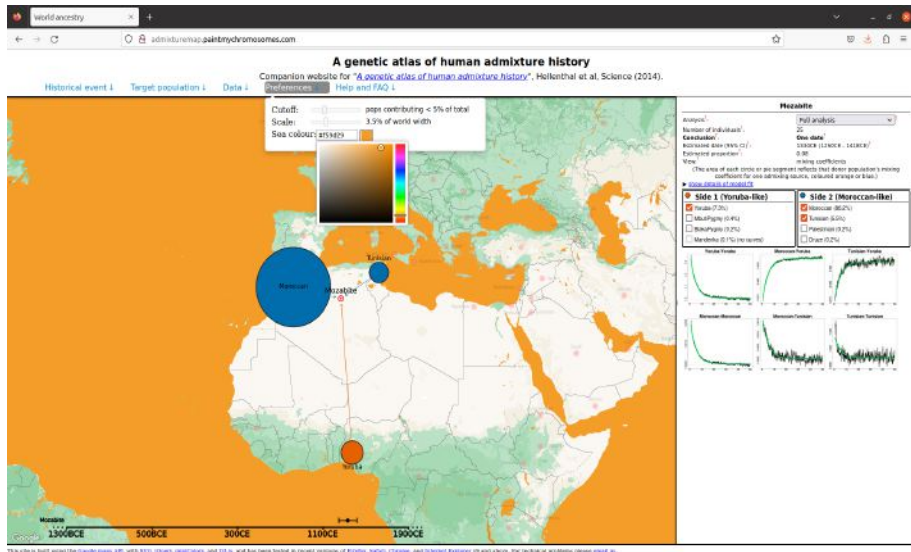
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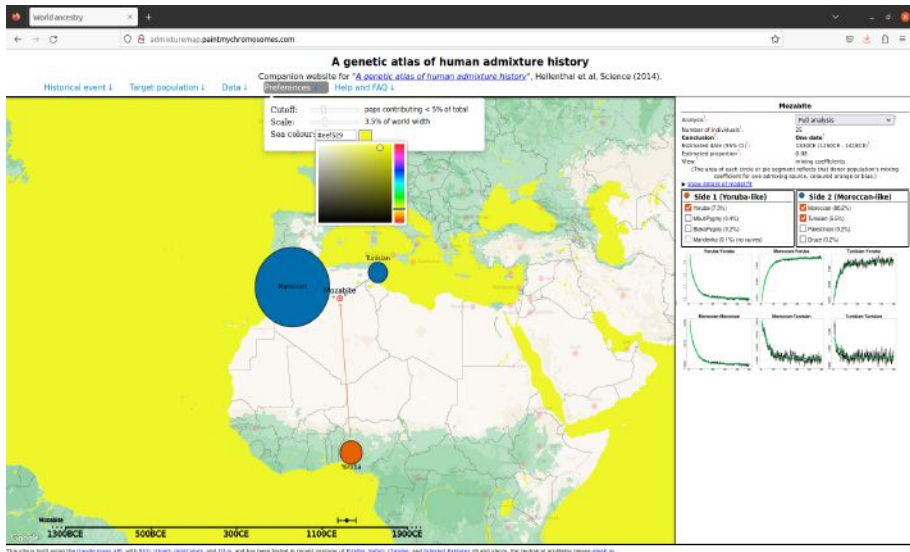
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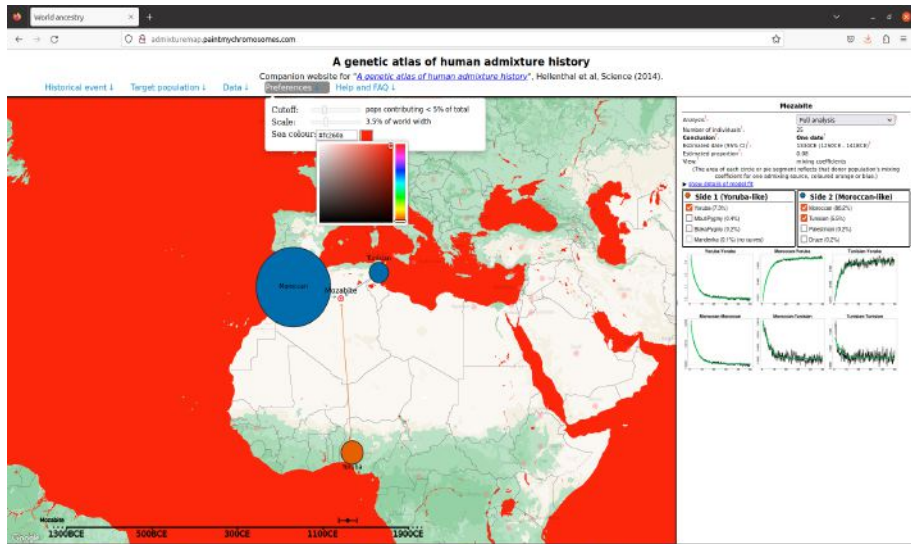
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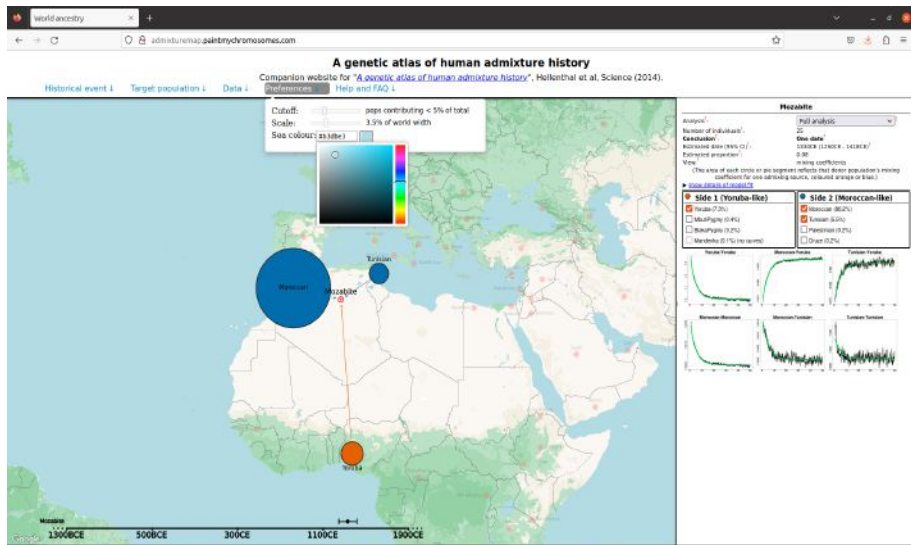
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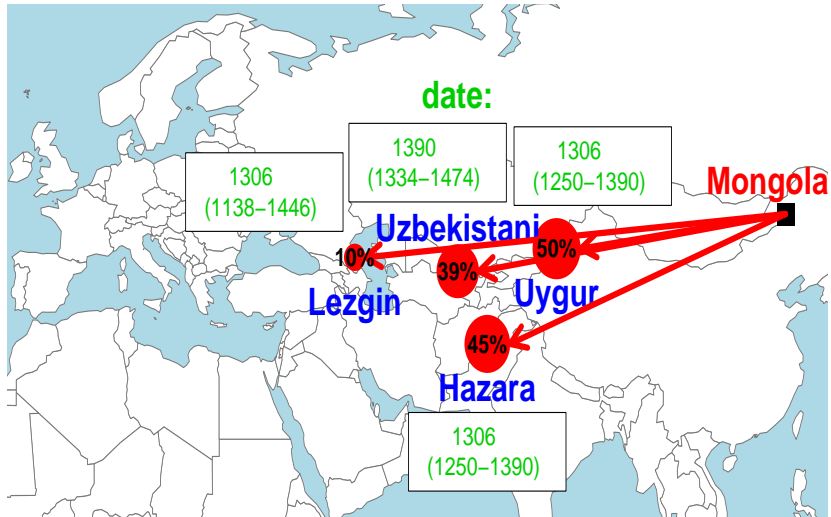


(Gavin Band)

Outline

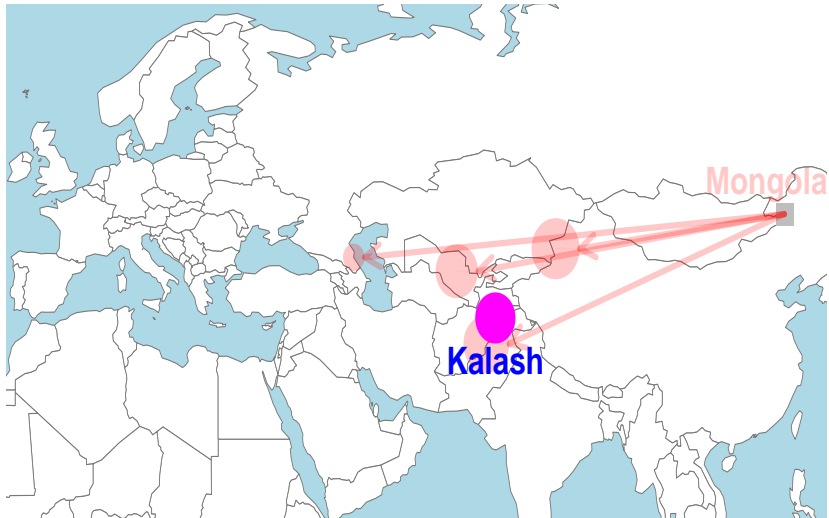
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1. Mongol Empire (1206-1368) of Genghis Khan



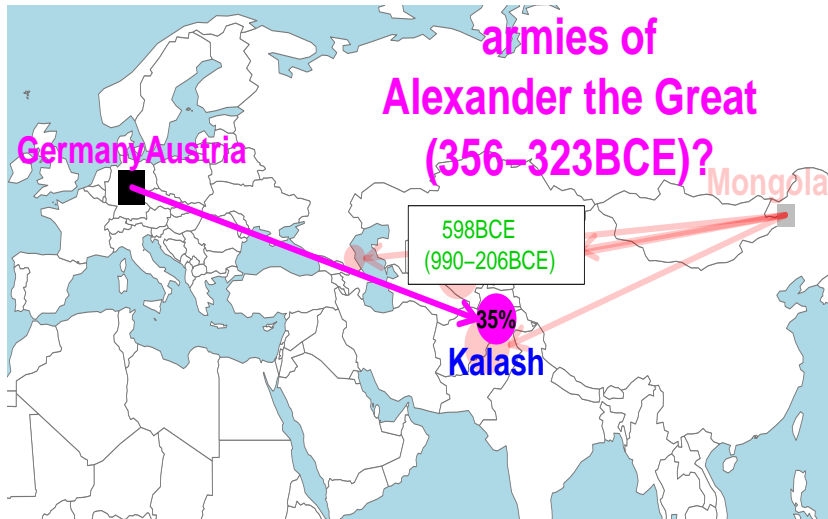
(Hellenthal et al 2014, *Science* **343**:747)

2. Kalash of Pakistan



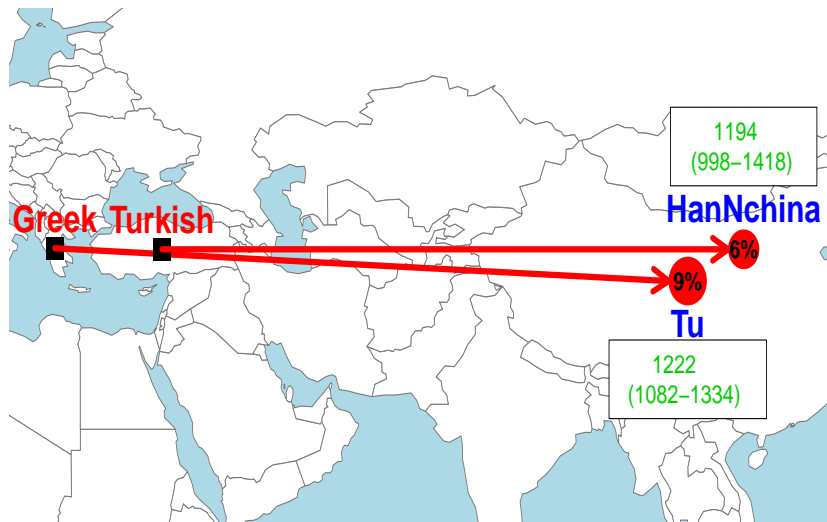
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2. Kalash of Pakistan



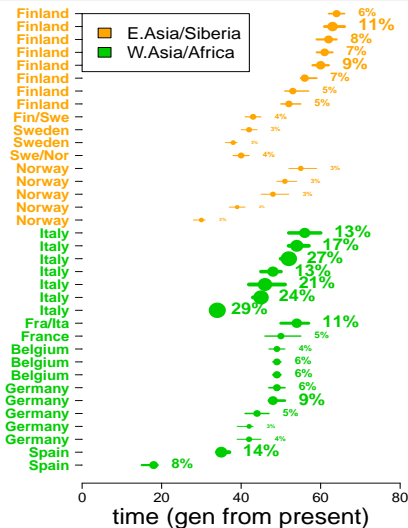
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3. Silk Road traders?



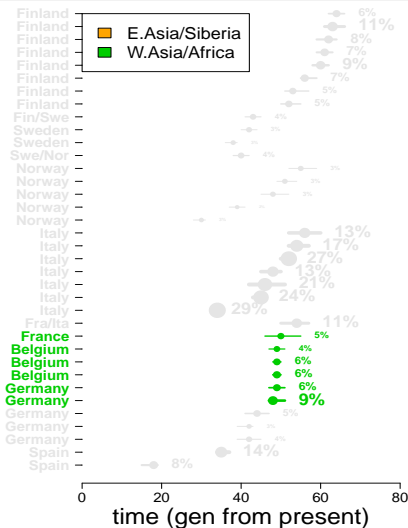
(Hellenthal et al 2014, *Science* **343**:747)

4. intermixing in Europe



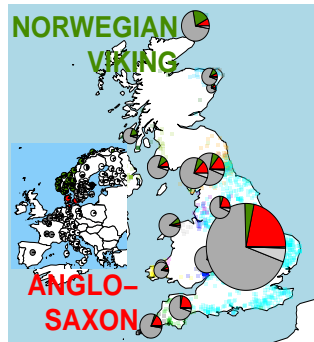
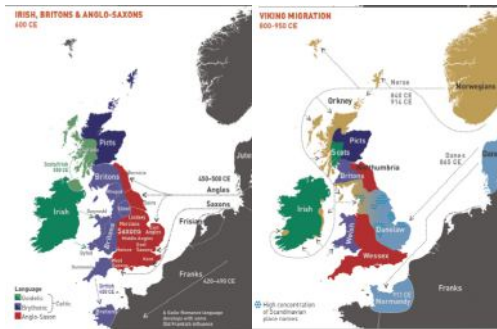
(Wangkumhang et al 2022, *Genome Res* 32:1553-64)

4. intermixing in Europe → Roman Empire?



(Wangkumhang et al 2022, *Genome Res* 32:1553-64)

5. Anglo-Saxon, Norse Viking migrations into UK



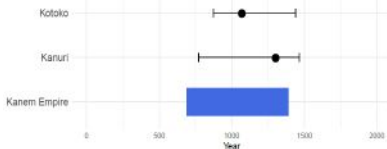
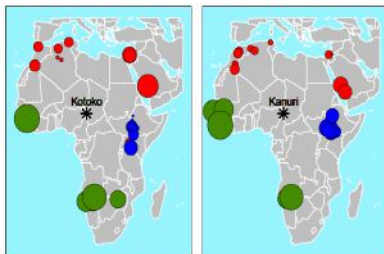
(Leslie et al 2015, *Nature* 519:309-14)

- **5-6th century Anglo-Saxon** migrations into England (contributed $\approx 10-40\%$ of DNA)
- **9-10th century Norse Viking** migrations into Scotland (contributed $\approx 25\%$ of DNA)

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Admixture in Cameroonian Kotoko/Kanuri



**Kanem
Empire
(700-
1380AD)**

'Because of its location, it served as a point of contact in **trade between North Africa, the Nile Valley, and the sub-Saharan region**'

Encyclopaedia Britannica

(see also Shriner & Rotimi 2018, *Am J Phy Anthropol* **167**:804)

(Bird et al 2023, *Sci Adv* **9**:eabq2616)



Kingdom of Aksum (\approx 150BC-960)



(Bird et al 2023, *Sci Adv* **9**:eabq2616)

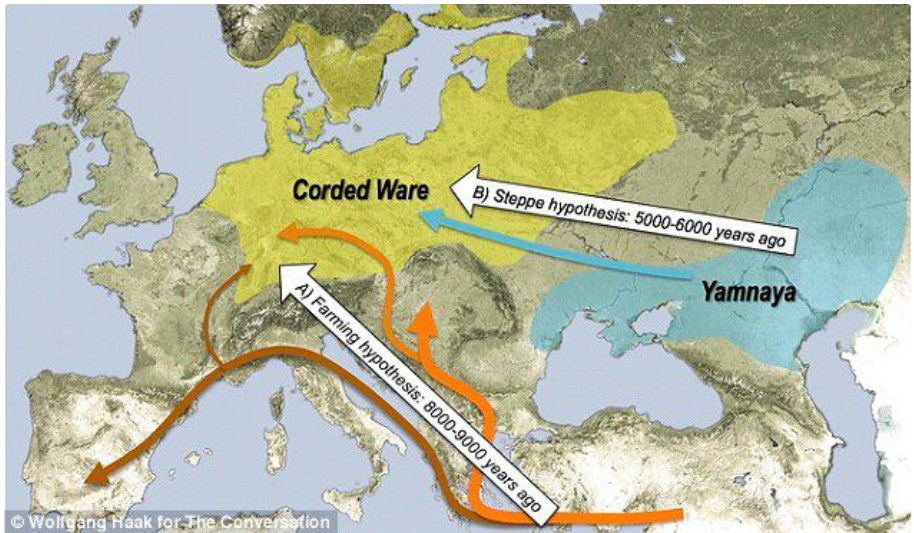
(**right:** by Aldan-2, commons.wikimedia.org/w/index.php?curid=75096897)

Sudan: intermixing between **Near East** and **Sub-Saharan Africa**
dated to \approx 700-1300

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Intermixing >4,500 years ago



(Haak et al 2015, *Nature* 522:207-11)



Intermixing >40,000 years ago

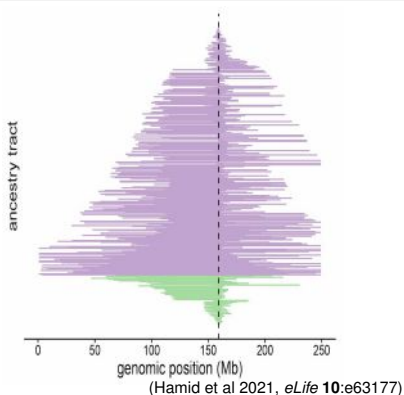
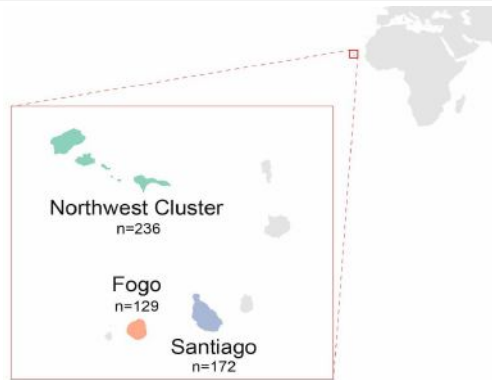


Nature Reviews | Genetics
 (Stoneking & Krause 2011, *Nat Rev Genet* 12:603)
 (Green et al 2010, *Science* 328:710)
 (Reich et al 2010, *Nature* 468:1053)

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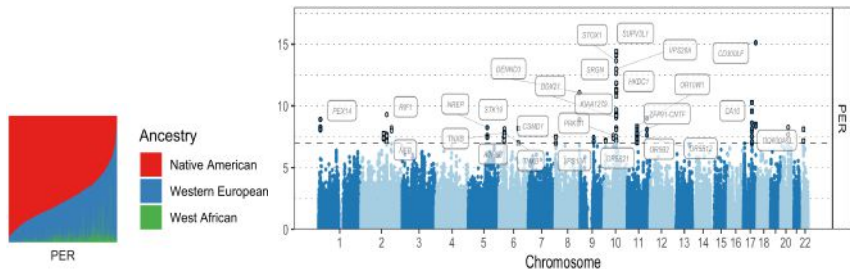
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Spreading adaptive genetic variants through intermixing



- **The Republic of Cabo Verde** settled in ≈ 1460 by **Portuguese** and **W.Africans**
- mixed people today carry more **W.African** ancestry at *DARC* gene, to protect against malaria

Spreading adaptive genetic variants through intermixing



(Mendoza-Revilla et al 2022, *Mol Biol Evol* **39**:msac076)

Peruvians: mixture of **Native American**, **W.European** and **W.African** ancestries

- **Native American** ancestors adapted to living at high elevations?
- since arrival of **Europeans**, adaptation linked to **immunity genes**

Have Neanderthals/Denisovans given us adaptive genetic variants?



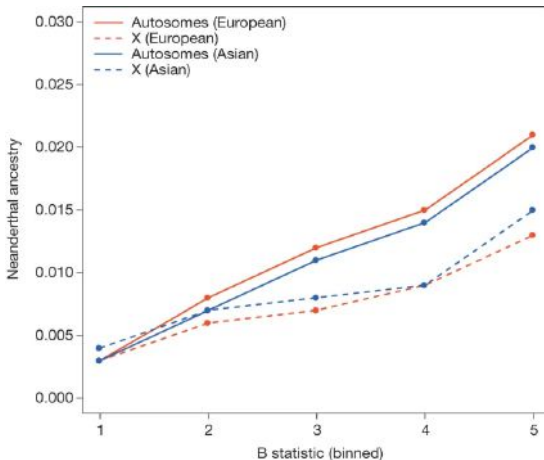
(Huerta-Sanchez et al 2014, *Nature* 512:194)



- **Denisovans** gave to Tibetans a version of gene (*EPAS1*) associated with efficient oxygen uptake
- **Neanderthal** regions associated with risk of COVID-19

(Zeberg & Paabo 2020 *Nature* 587:610 / Zeberg & Paabo 2021 *PNAS* 118:e2026309118)

Have Neanderthals/Denisovans given us adaptive genetic variants?



(Sankararaman et al 2014, *Nature* 507:354)

- overall: less **Neanderthal** DNA in functionally important regions

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- anatomically modern humans have been intermixing throughout 1,000s to 10,000s of years
 - including with other archaic human groups (Neanderthals, Denisovans)

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- can also use DNA to infer **when past groups intermixed**
 - often correlated with known migrations (but not always)
- anatomically modern humans have been intermixing throughout 1,000s to 10,000s of years
 - including with other archaic human groups (Neanderthals, Denisovans)
- evidence that intermixing sometimes spreads adaptive genetic variants into new populations/environments

Acknowledgements

- S.Myers, D.Falush, D.Lawson, P.Wangkumhang
- N.Bird, S.Lopez, L.van Dorp, S.Morris, P.Awah, A.Tarekegn, M.Fomine, T.Oljira, D.Zeitlyn, E.Bekele, N.Bradman, M.Thomas
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