

Emerging Capabilities for Forensic Genetic Analysis - Novel Investigative Leads and Improved Profiling Data from DNA

Christopher Phillips

Forensic Genetics Unit,
University of Santiago de Compostela



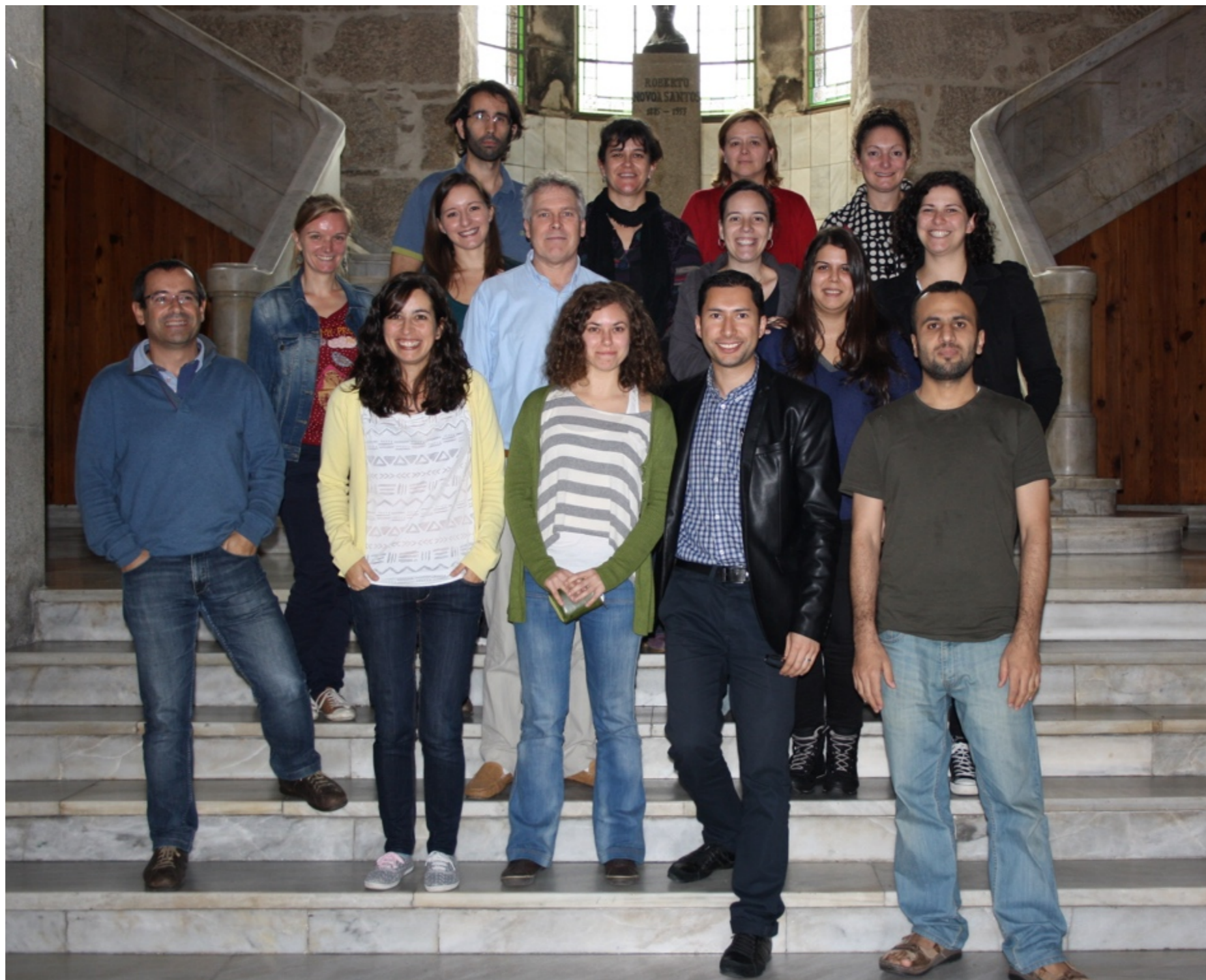
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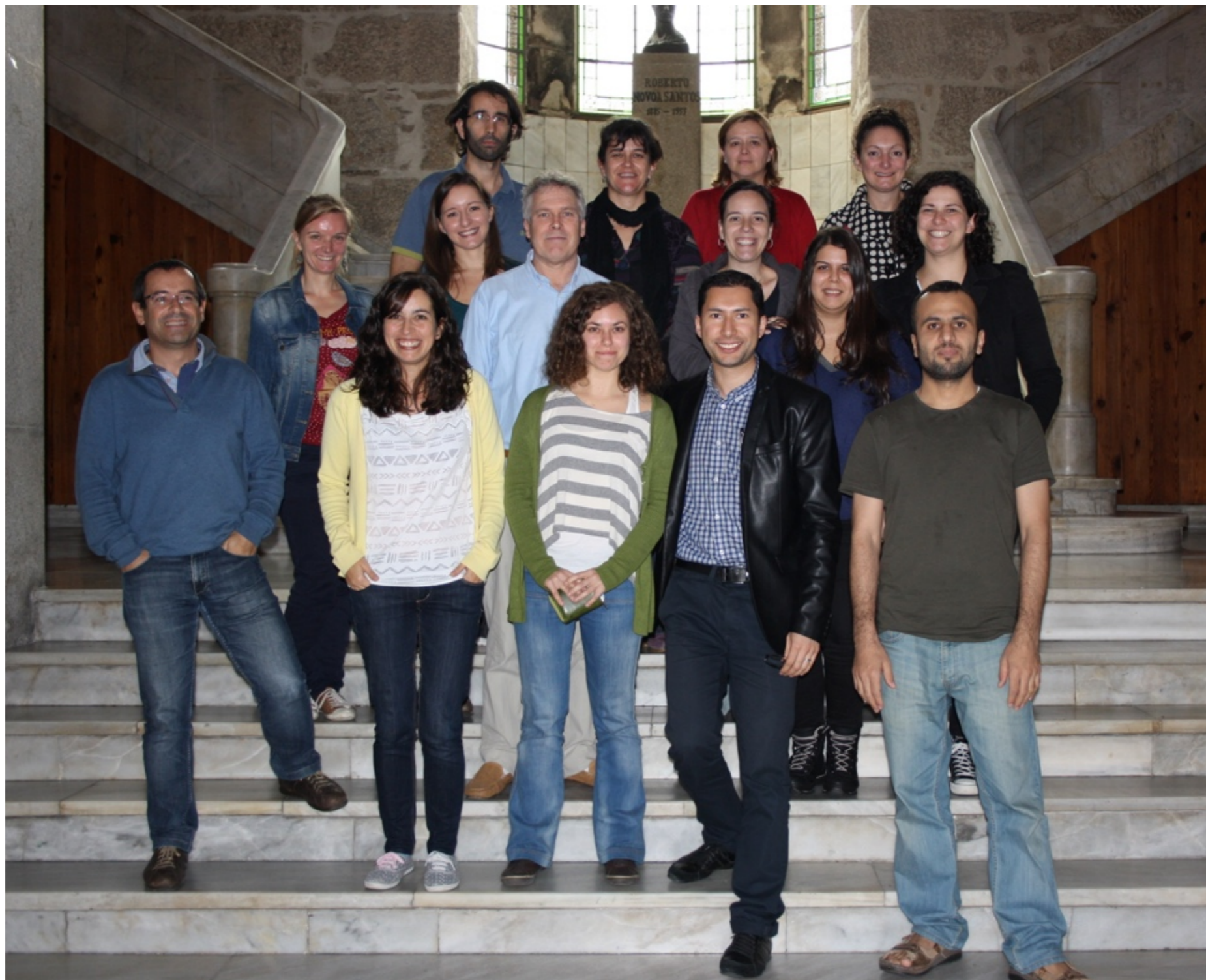




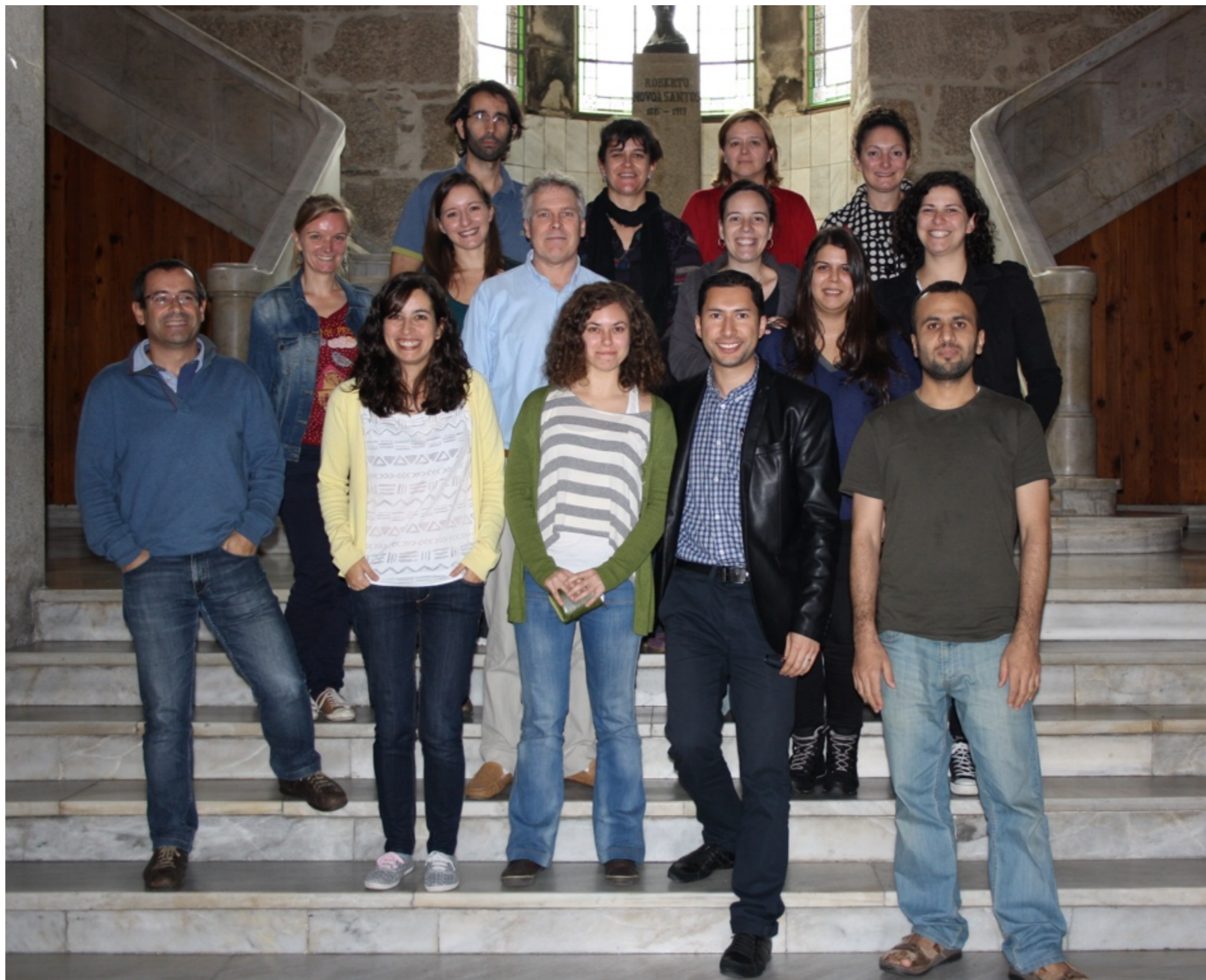
Markers

Data

Detection



- Markers** How many genetic markers can we type from contact traces?
- Data** What information can we give investigators beyond identity?
- Detection** Can the marker detection systems minimize false negatives?



MPS

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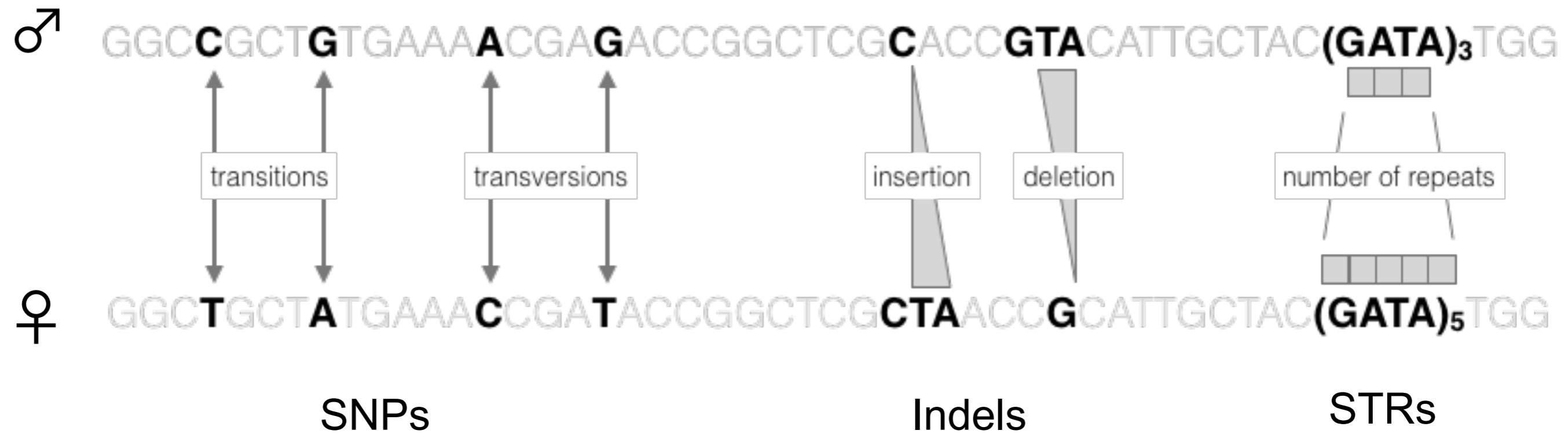
SNPs 'R' Us



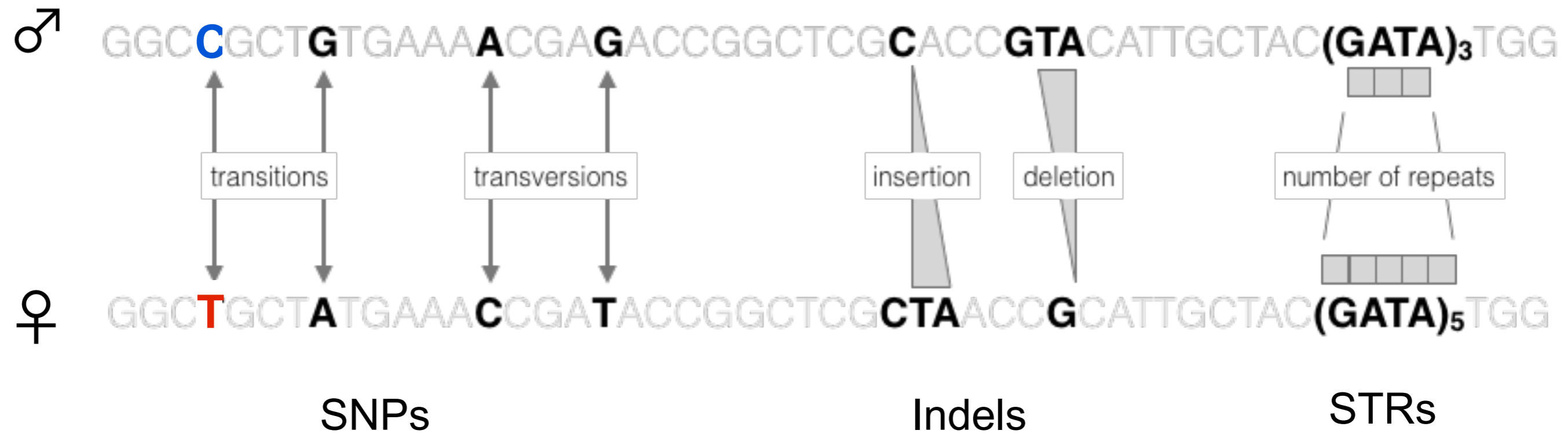
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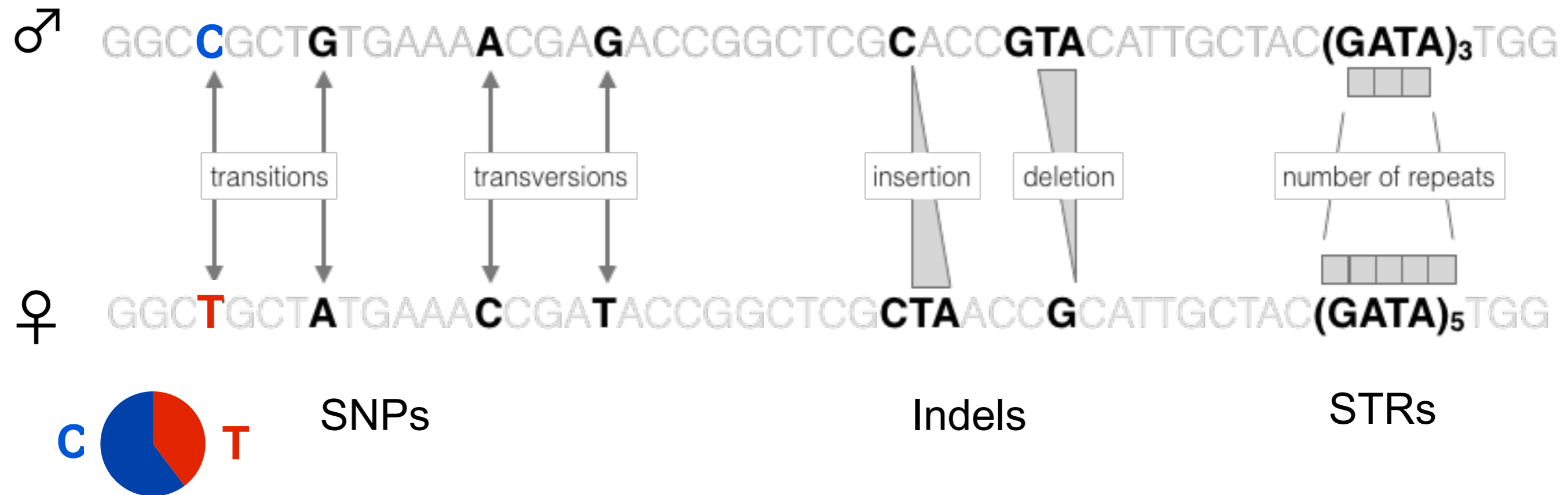
Types of genetic marker



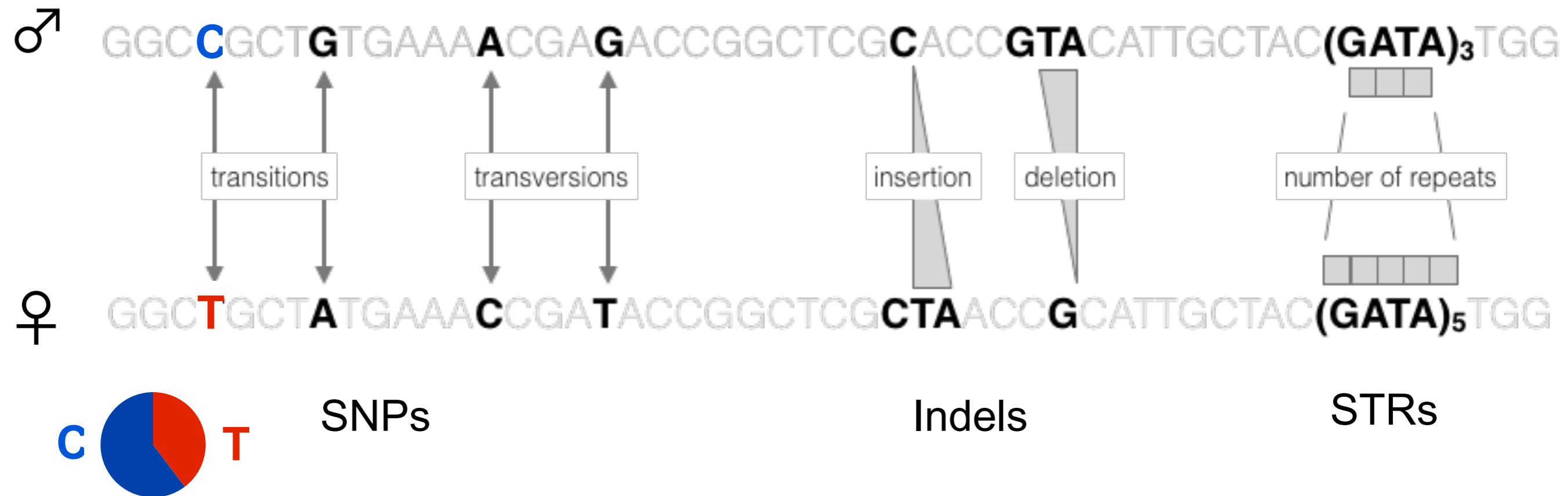
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Two forensic genetic analyses 'before' DNA - *Identification*

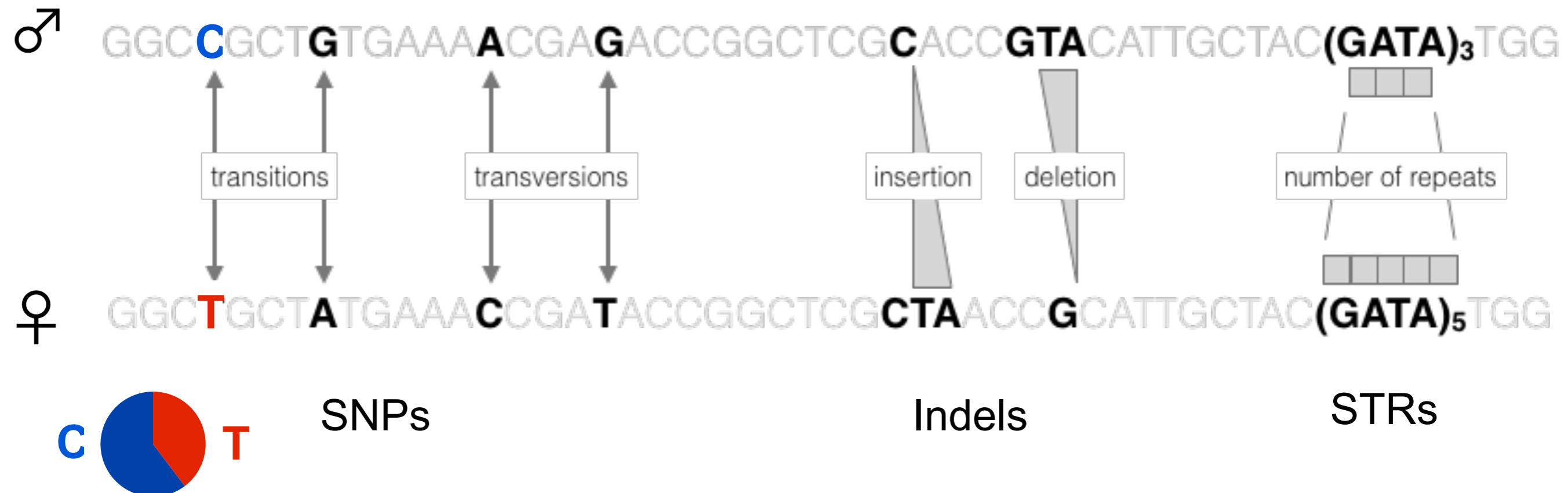
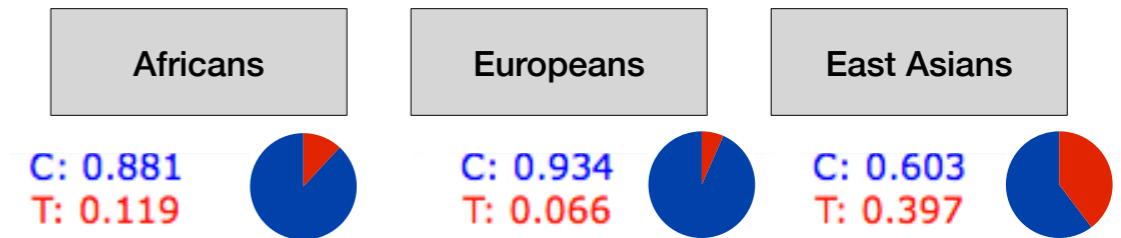


Two forensic genetic analyses 'before' DNA - *Identification*

Aggravated burglary in Essex (double murder): 1981

- Bled on glass and was *EsD 2-1* (CT) - details held on the *Blood Group Index*
- Five burglars active in Essex region with *EsD 2-1* (CT) type

System name	Gene	SNP identifier	% informative genotypes in Africans	% informative genotypes in Europeans	% informative genotypes in East Asians
Esterase D	ESD	rs9778	22.4	12.32	63.5

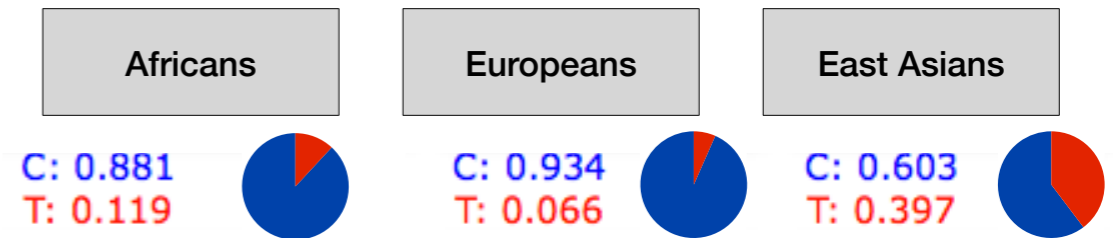


Two forensic genetic analyses 'before' DNA - *Ancestry analysis*

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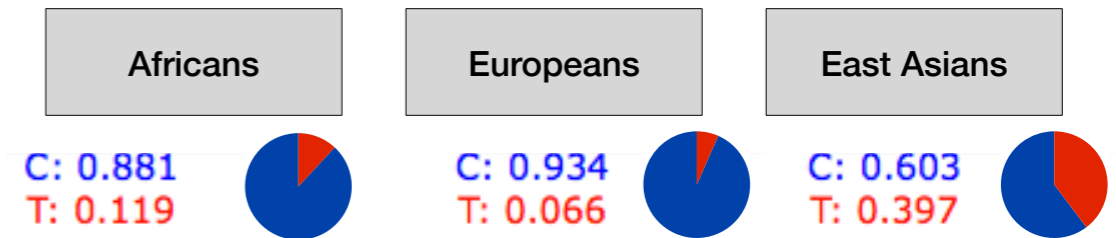


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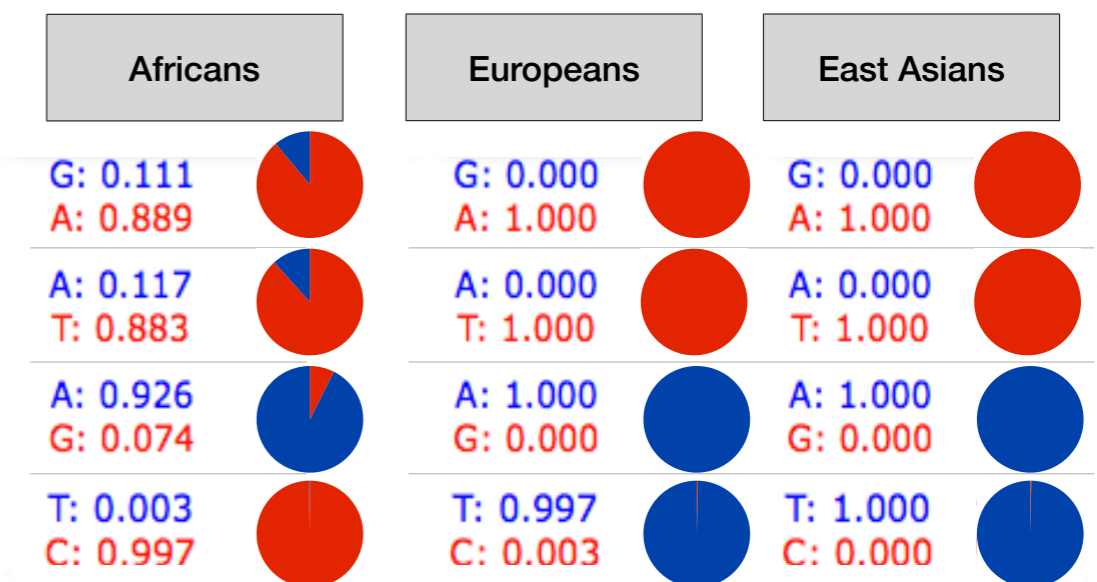
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South London serial rapist: 1981-2

- Used *Pep A* - a red cell membrane protein present in semen
- Detecting *2-1* (AG) linked attacks and indicated African ancestry

System name	Gene	SNP identifier	% informative genotypes	cumulative % informative	cumulative % uninformative
Pep A	CNDP2	rs733062	20.97	20.97	79.03
Hb S	HBB	rs334	22.03	38.38	61.62
CA II	CA2	rs2228063	15.54	47.96	52.04
Fy	DARC	rs2814778	99.66	99.77	0.23

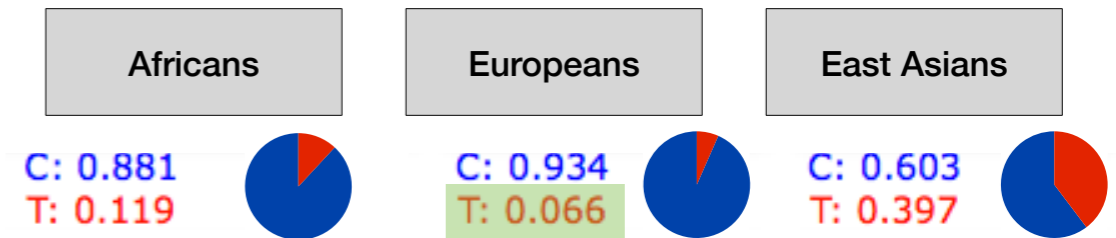


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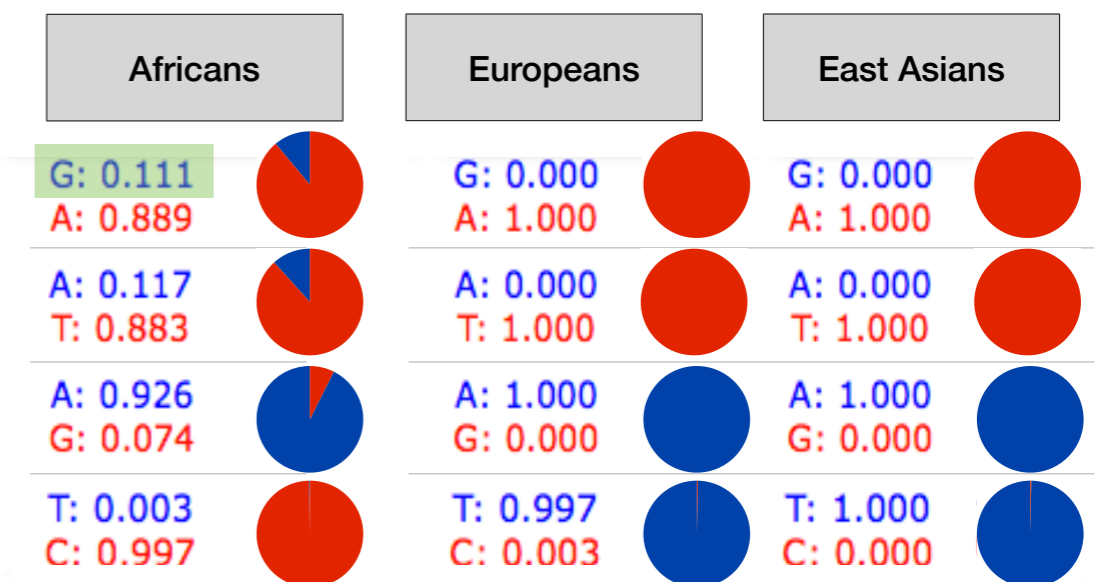


The rare

South London serial rapist: 1981-2

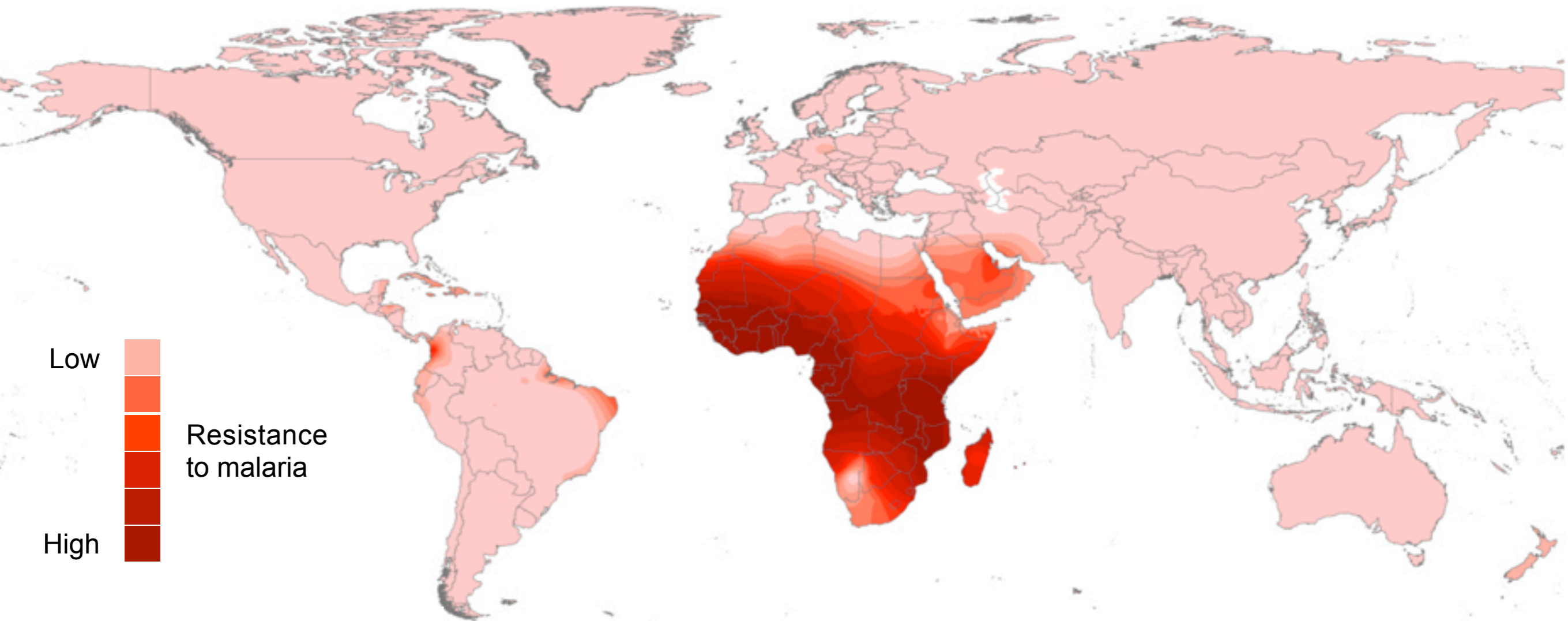
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The population specific

The Duffy SNP is African-informative due to a key evolutionary factor

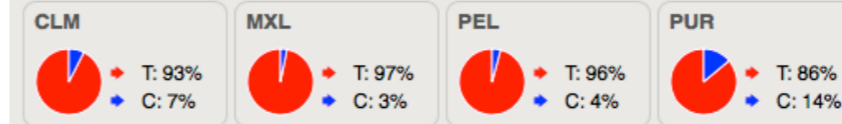


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AFR sub-populations

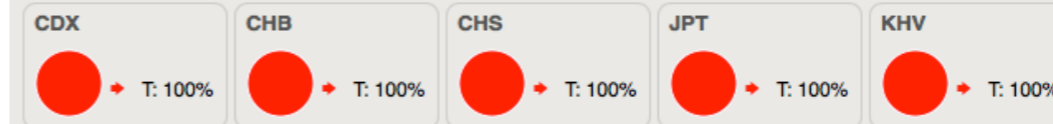


AMR sub-populations



1000 Genomes
A Deep Catalog of Human Genetic Variation

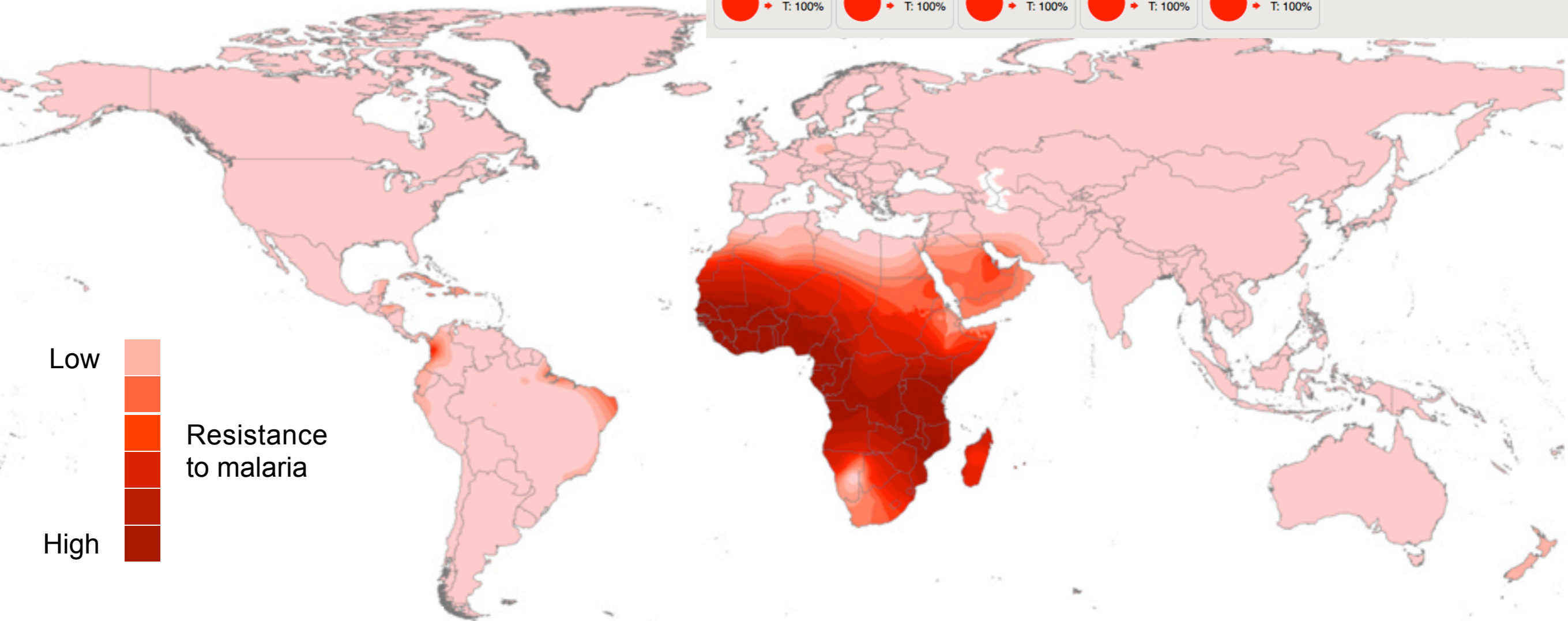
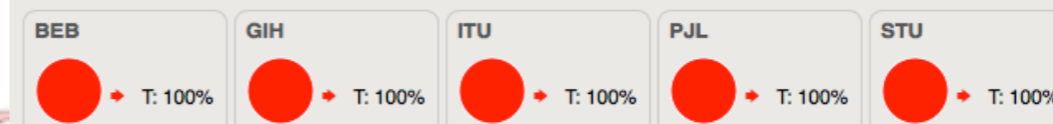
ASN sub-populations



EUR sub-populations

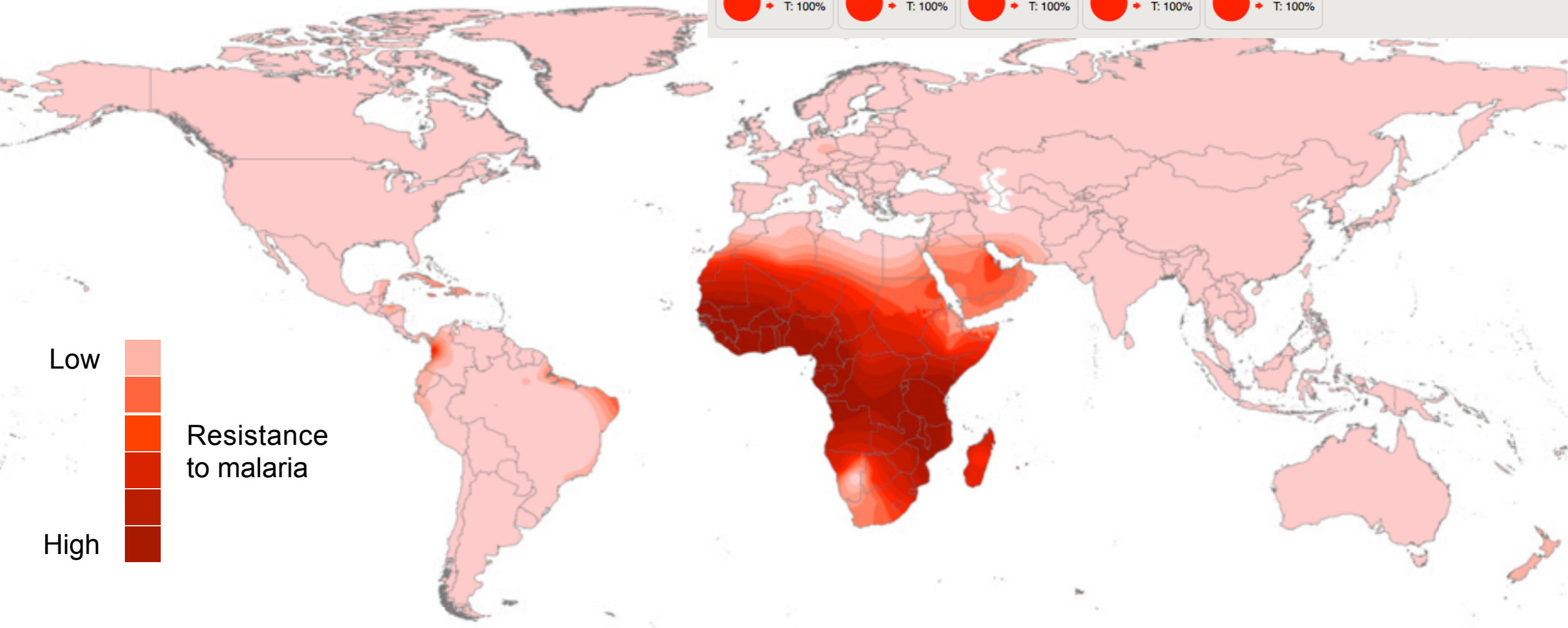
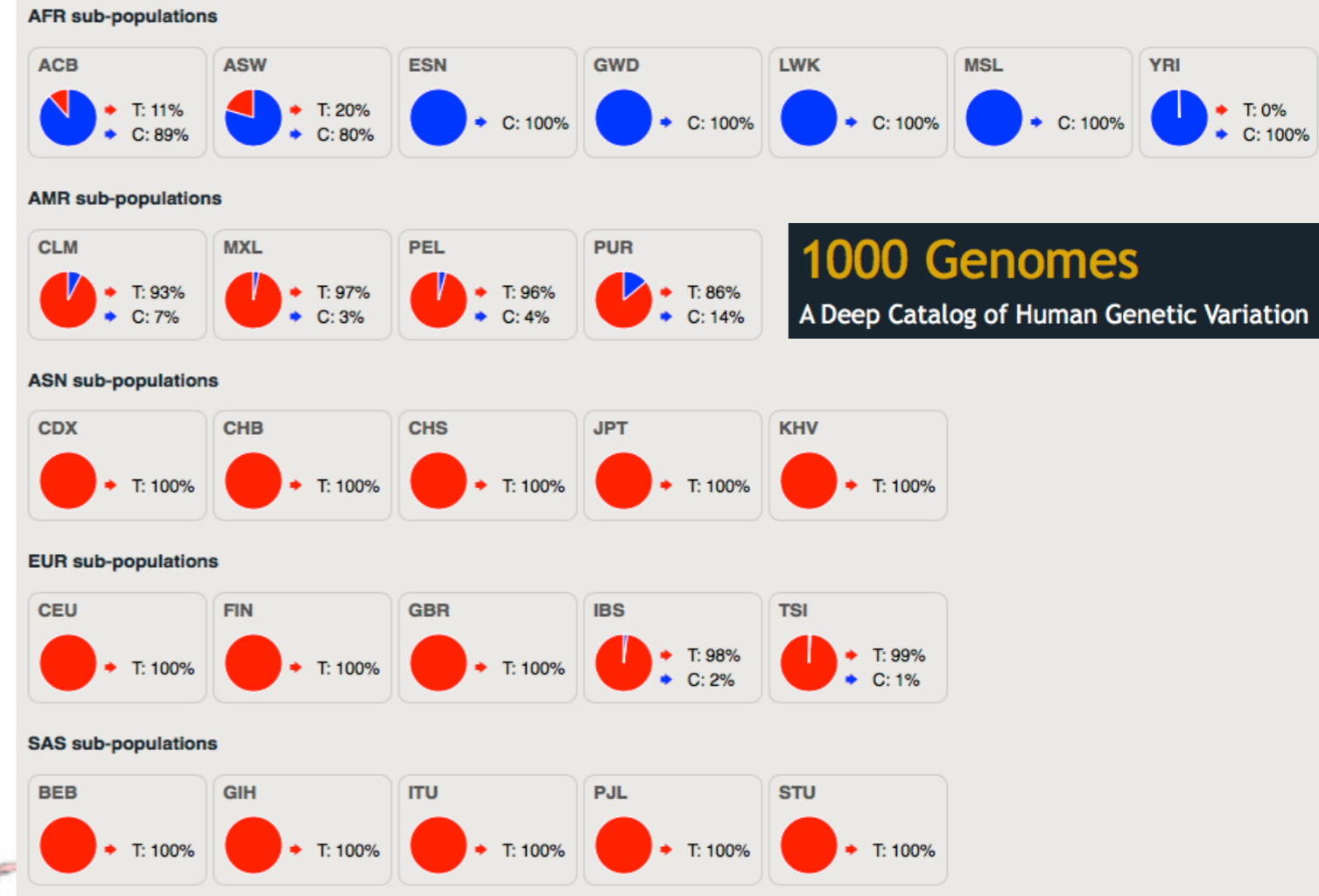


SAS sub-populations



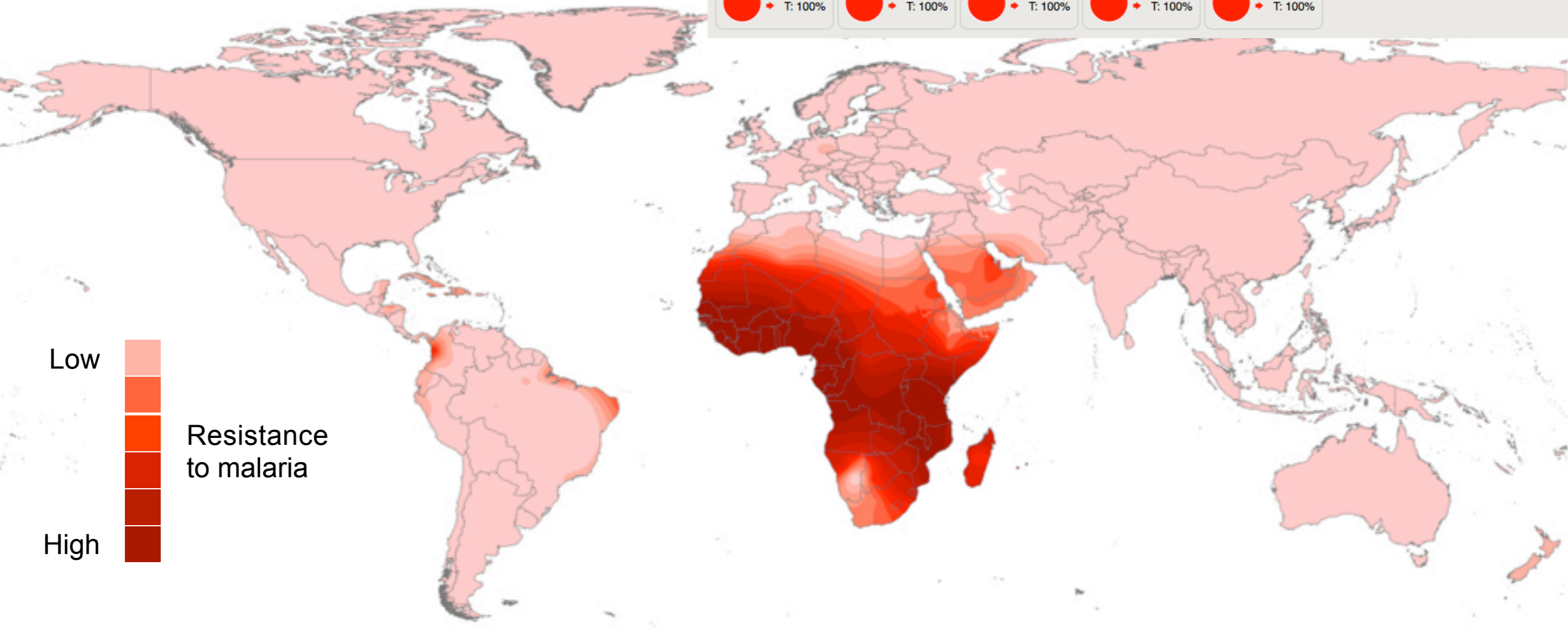
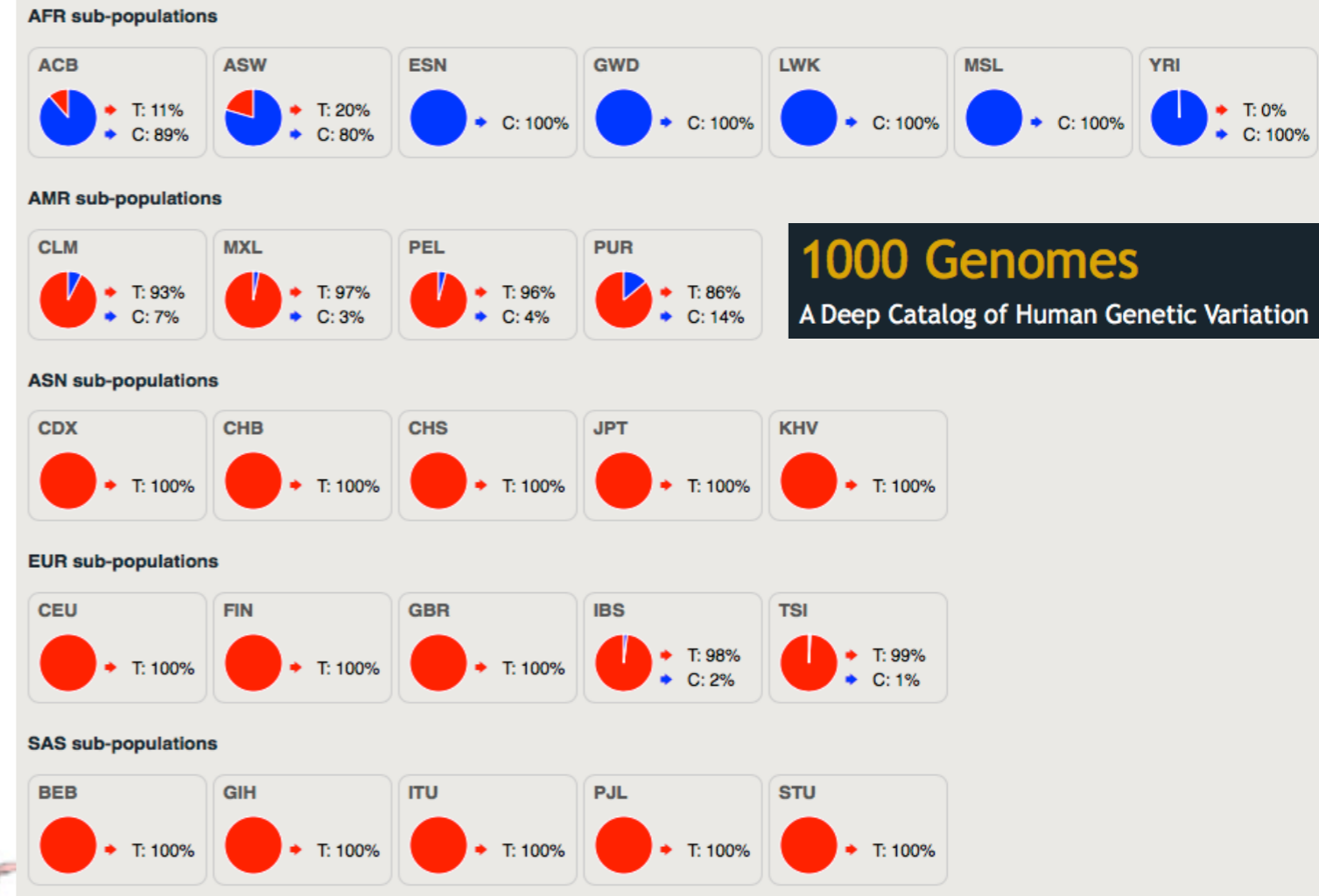
The Duffy SNP is African-informative due to a key evolutionary factor

- Sources of error:
Admixture



The Duffy SNP is African-informative due to a key evolutionary factor

- Sources of error:
 - Admixture
 - Atypical variation

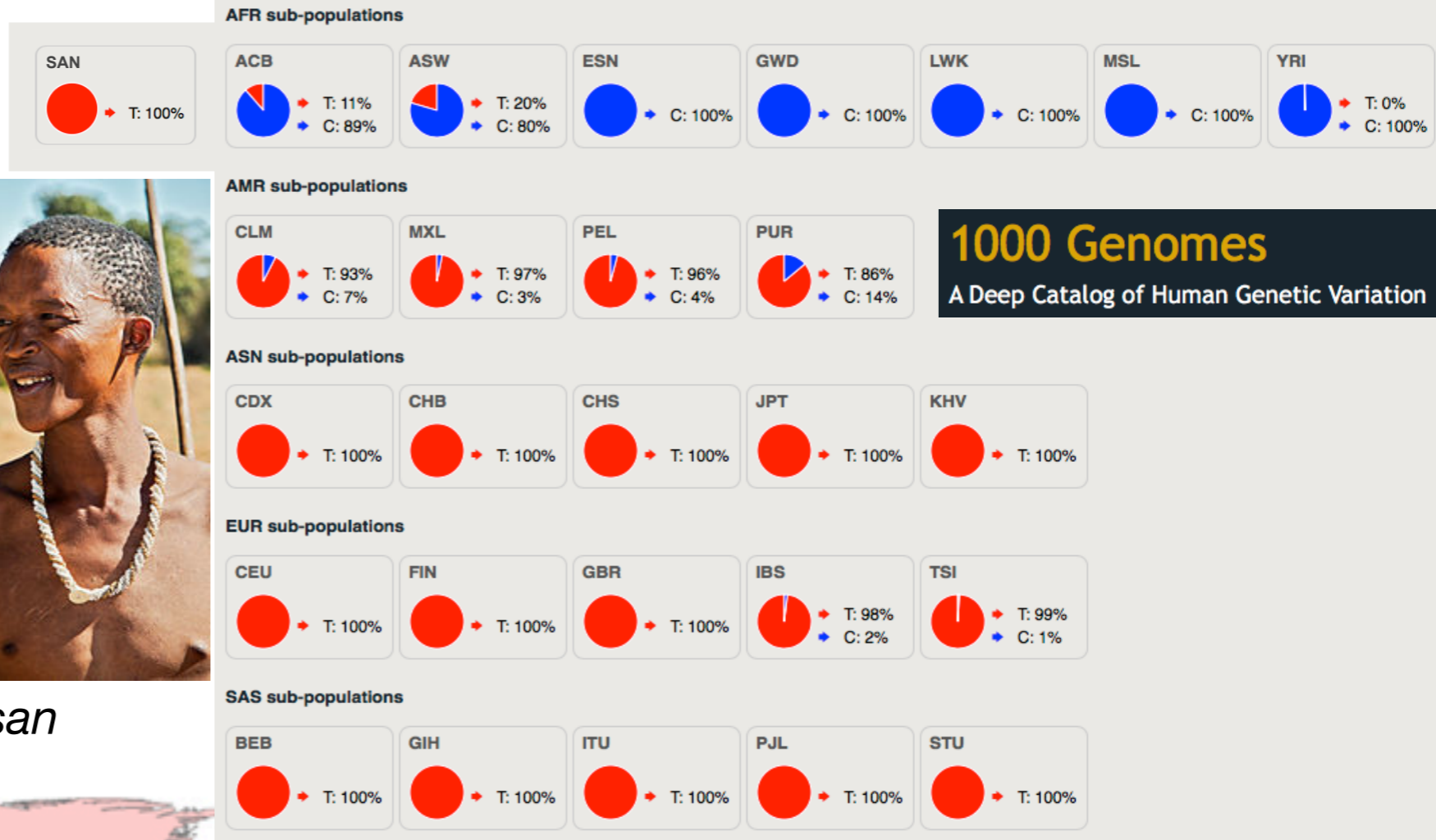


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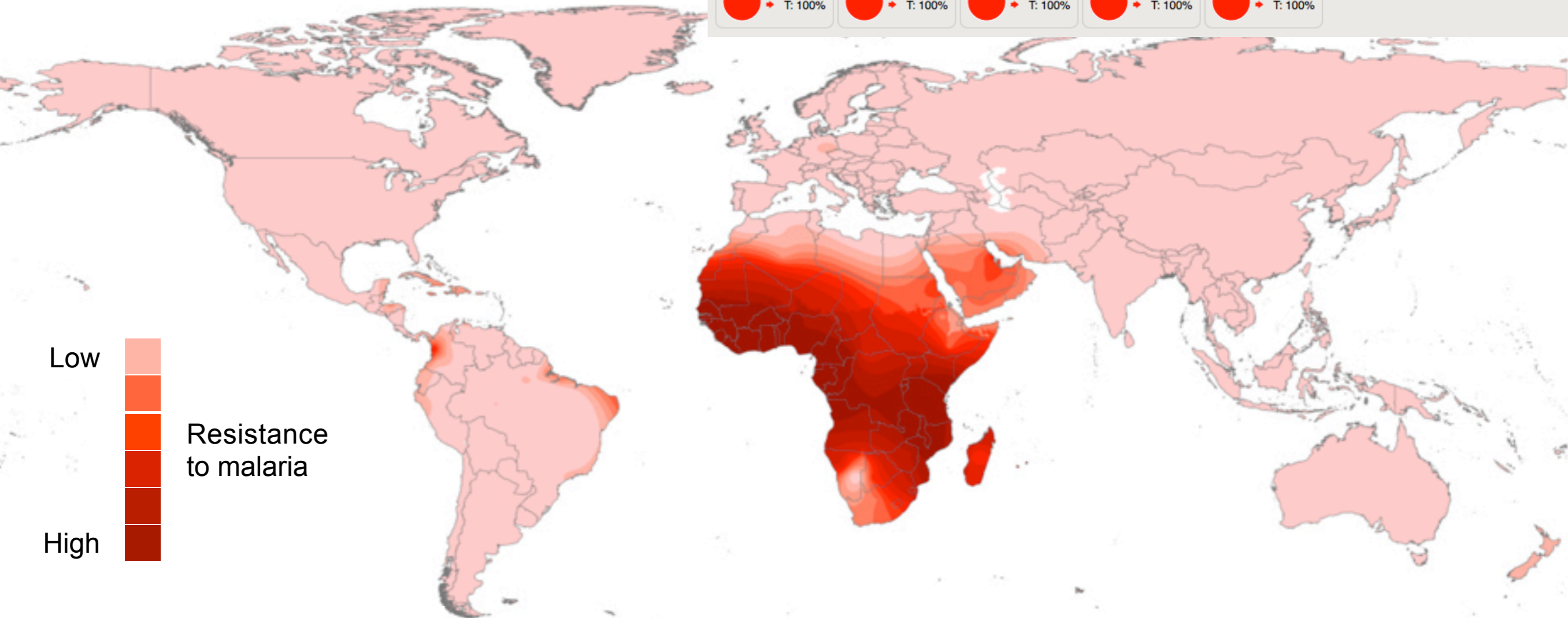
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Khoisan

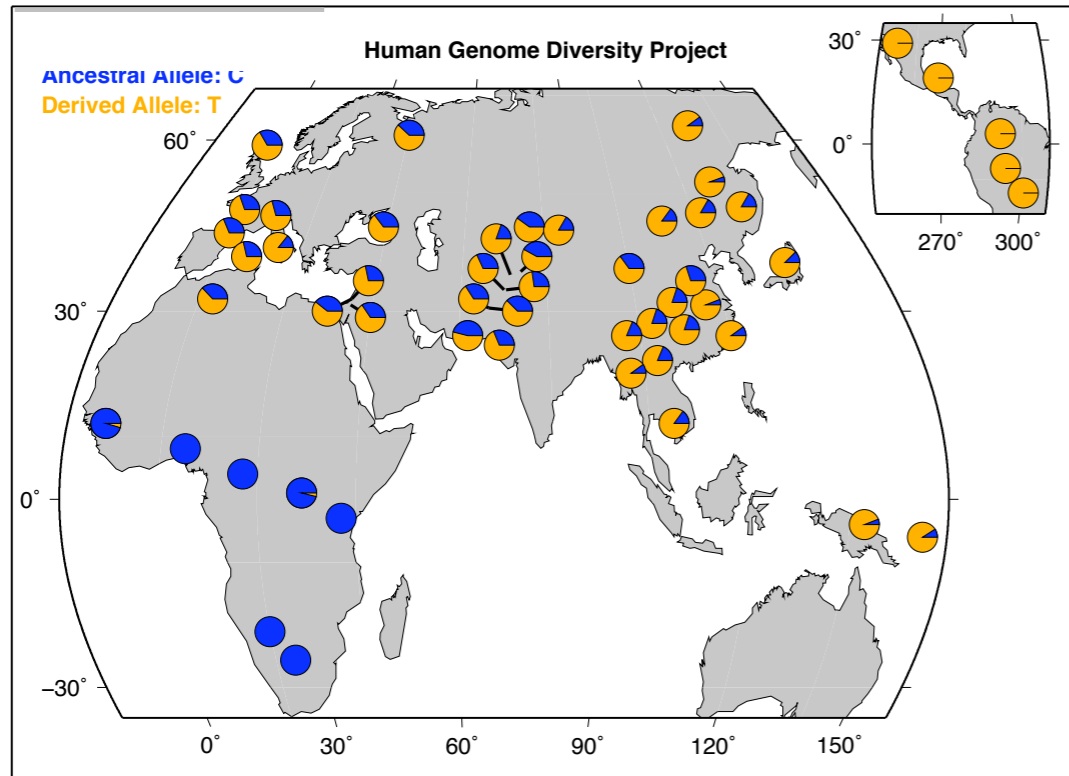


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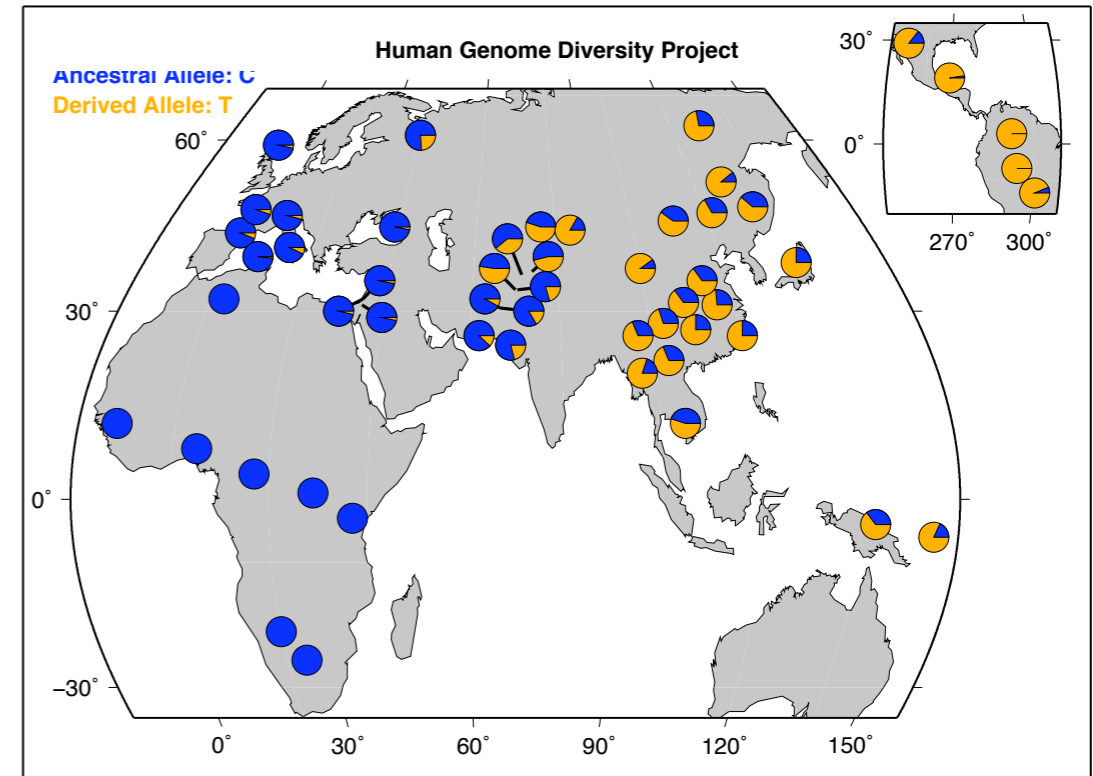


Have since identified more 'fixed' SNPs - frequencies near 1 or 0

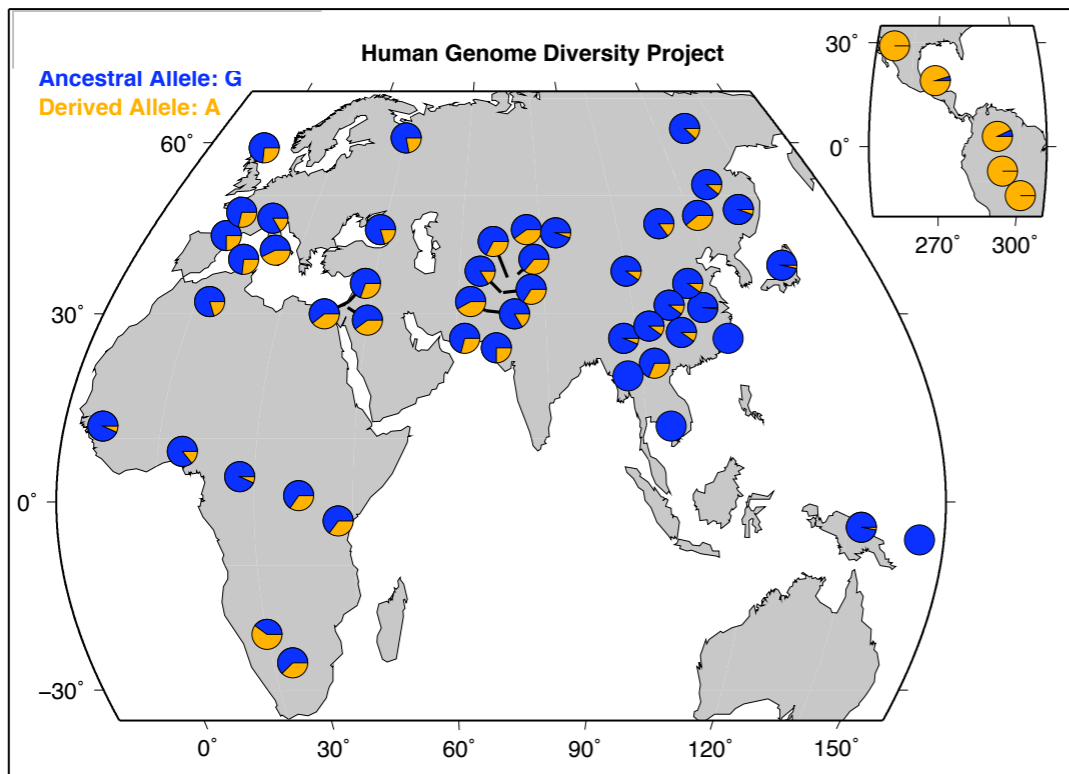
Best Native American-African



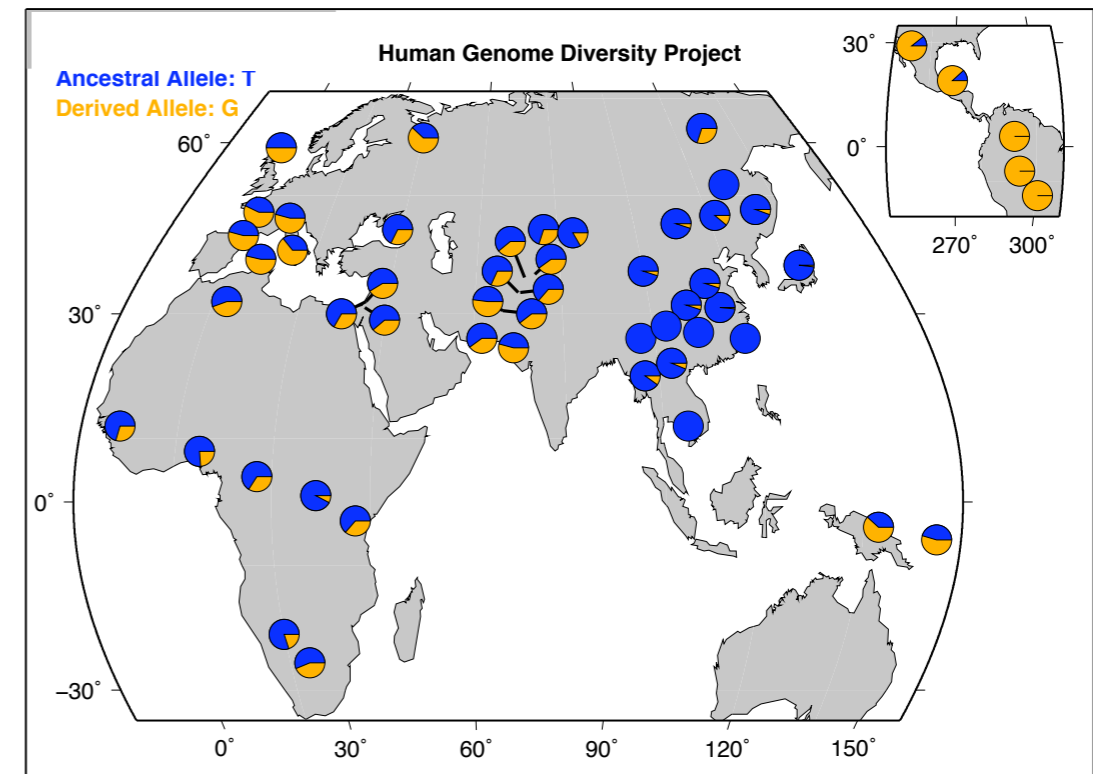
Best Native American-European



Best Native American-Oceanian



Best Native American-East Asian

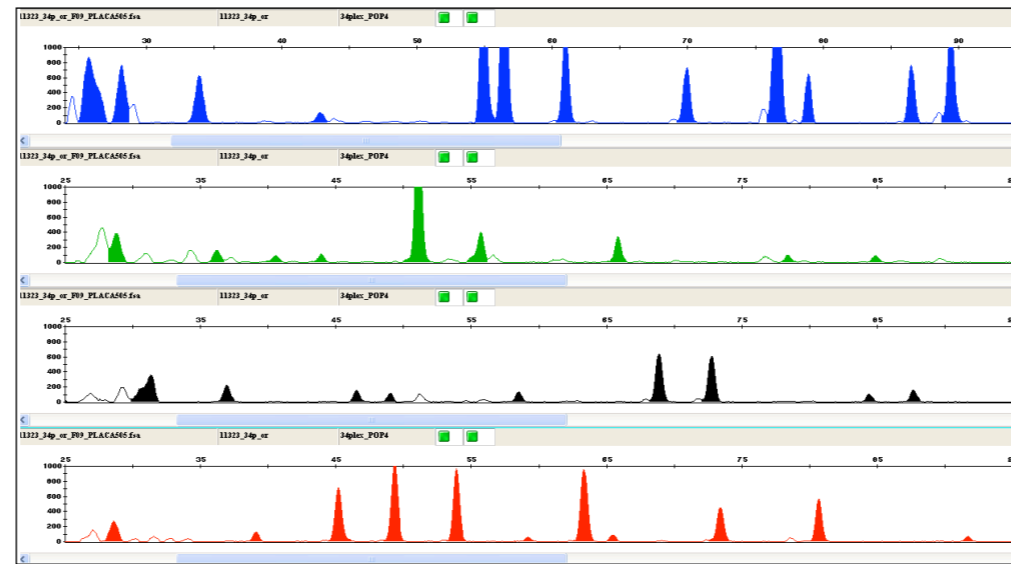


Apply validated Bayesian analysis regimes to the genetic data

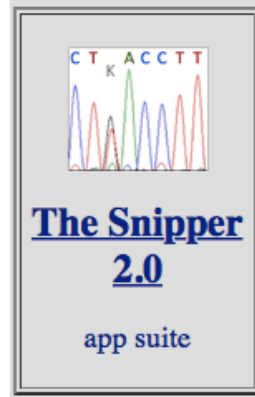
WWII soldier ID program



34-SNP profile of sample A (30/34 SNPs)



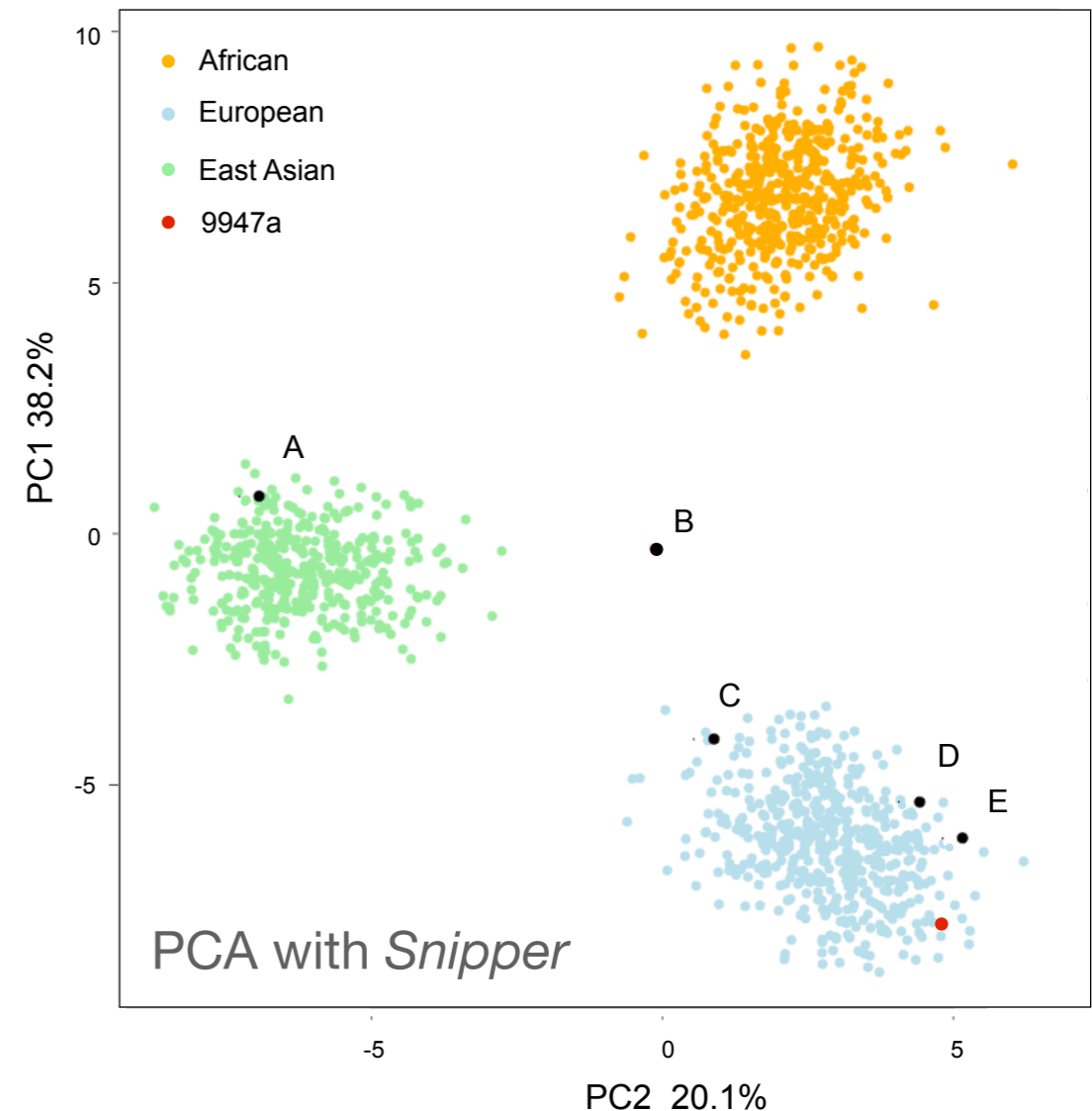
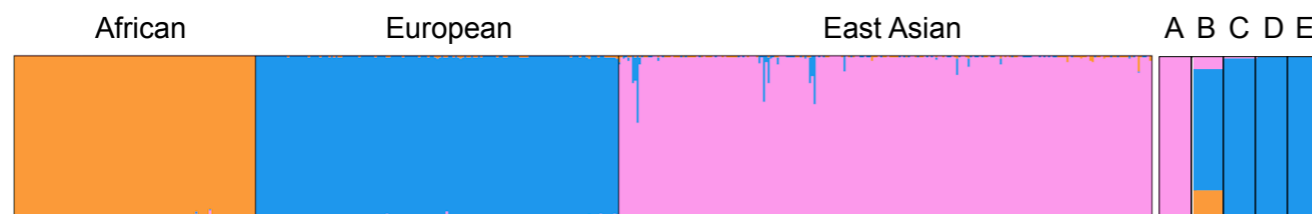
Binary AIM classification of multiple individuals
with an Excel file of populations (.xlsx format)



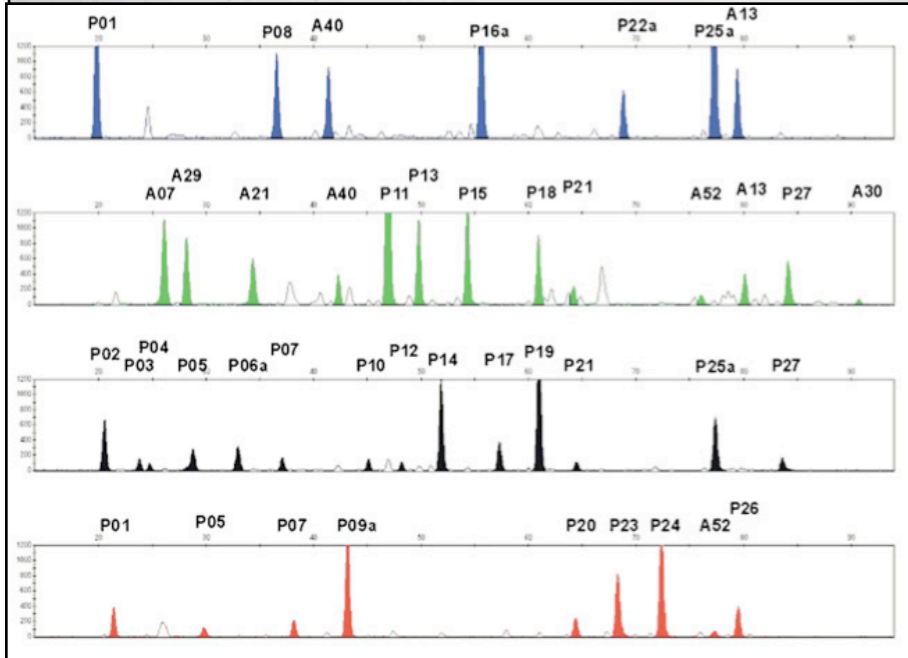
Bayes analysis with *Snipper*

- A:** 88% completeness 9,355,471,189,361,381,376 times more likely E ASN than EUR
- B:** 12% completeness 47 times more likely EUR than AFR
- C:** 56% completeness 69,184,156,498,170 times more likely EUR than E ASN
- D:** 88% completeness 237,256,787,732,999,681,605,632 times more likely EUR than E ASN
- E:** 97% completeness 2,795,277,788,041,023,852,877,185,024 times more likely EUR than E ASN

Identifying genetic clusters with STRUCTURE



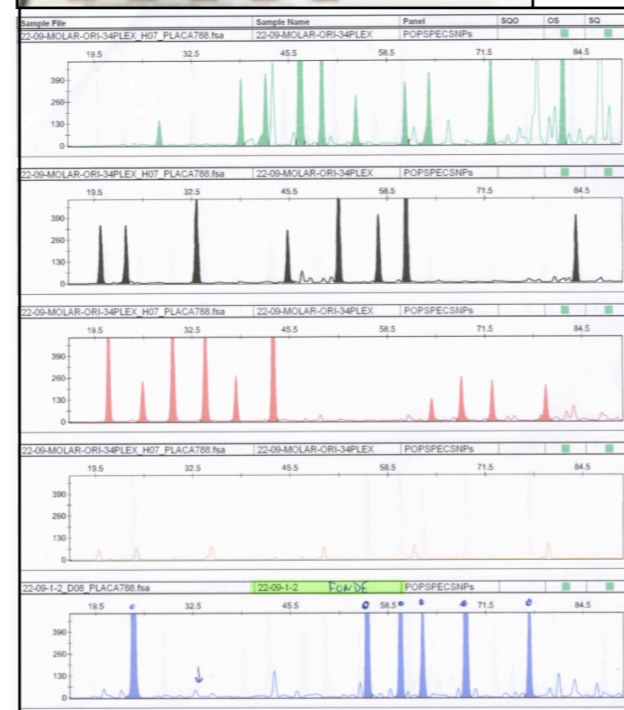
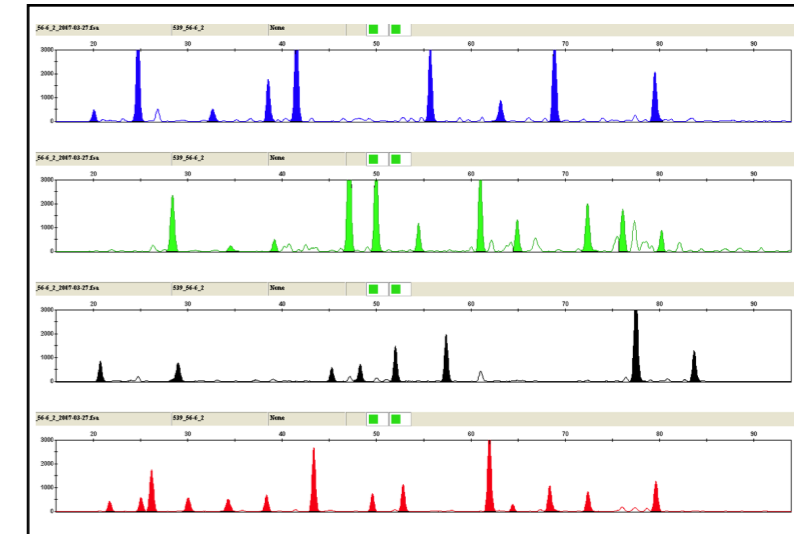
SNP-based tests work well with highly degraded DNA



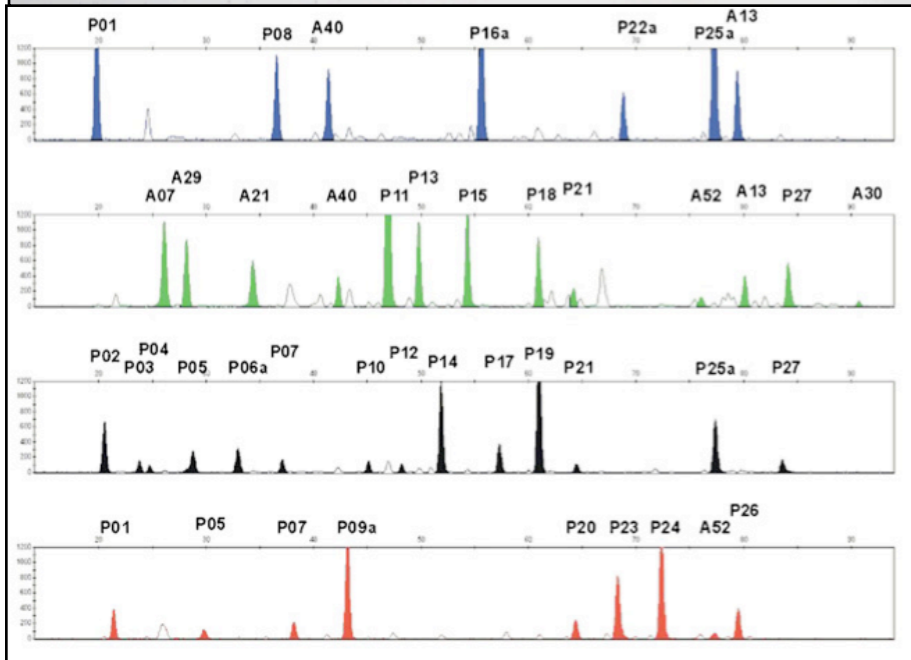
Femur



Both dentine



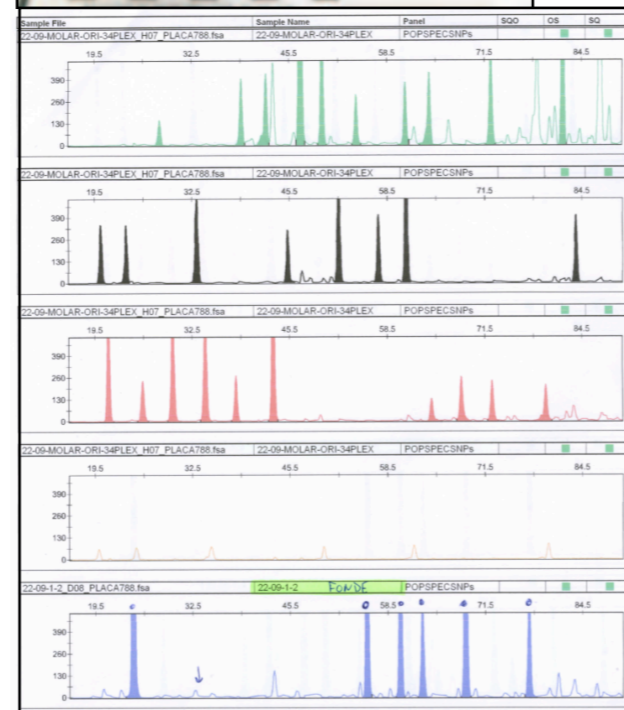
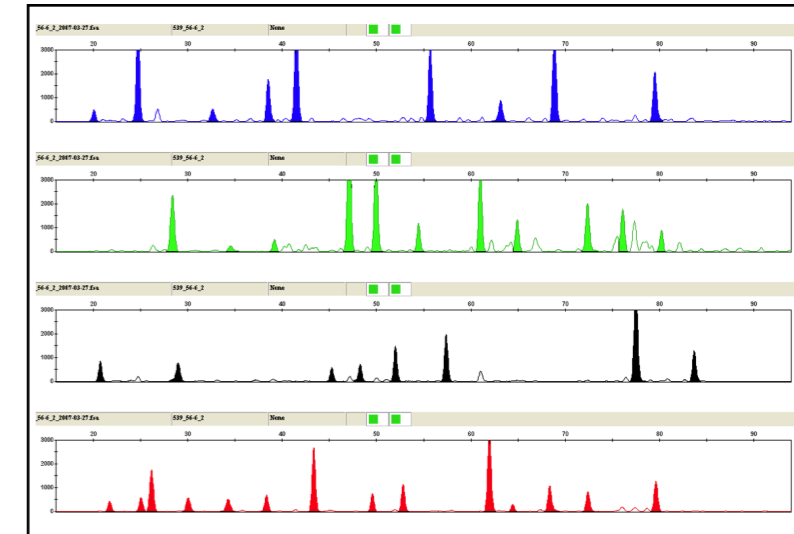
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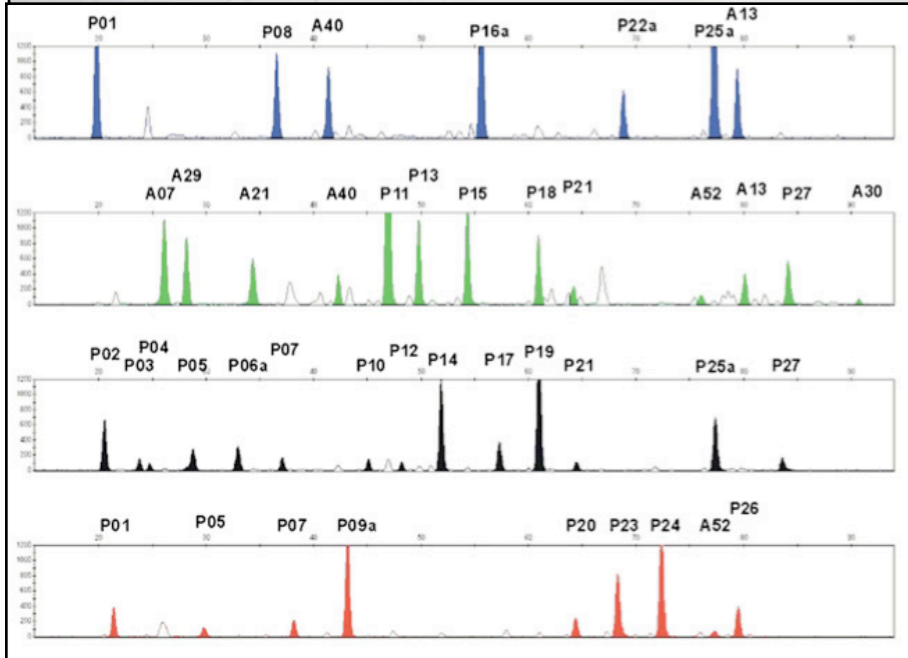
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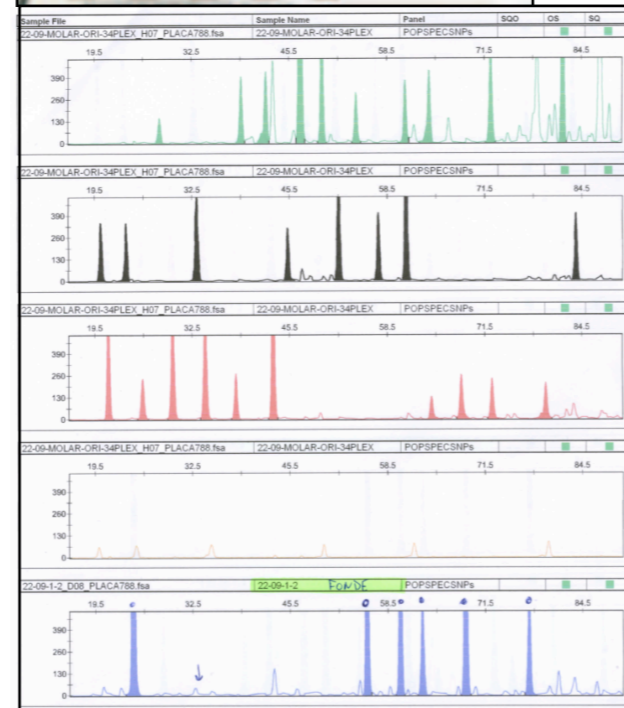
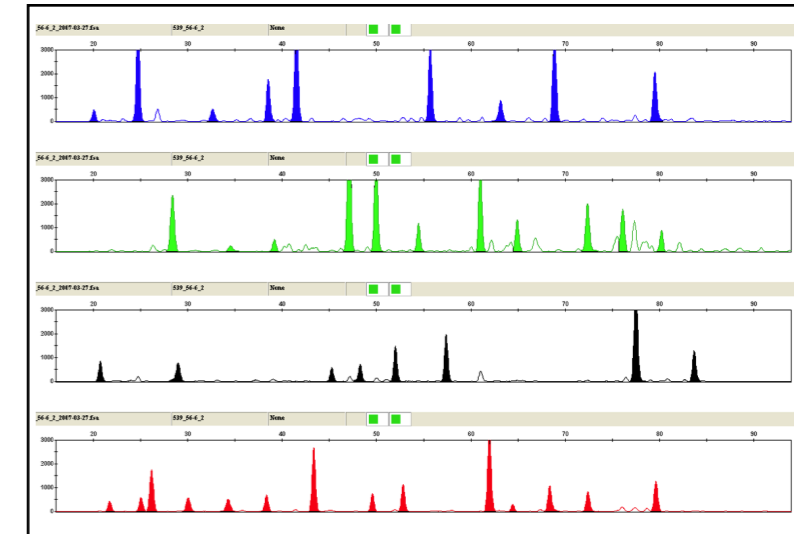
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Femur



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- Charred skeleton found after forest fire
- Adult male reported missing 10Y earlier
- Surviving daughter used to confirm identity
- Normal profiling failed, few MiniFiler STRs detected, SNPs gave near-complete profiles



A typical cold case review - the murder of Eva Blanco

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- Heavy rain obscured tyre marks and footprints at scene
- Small semen stain on underwear



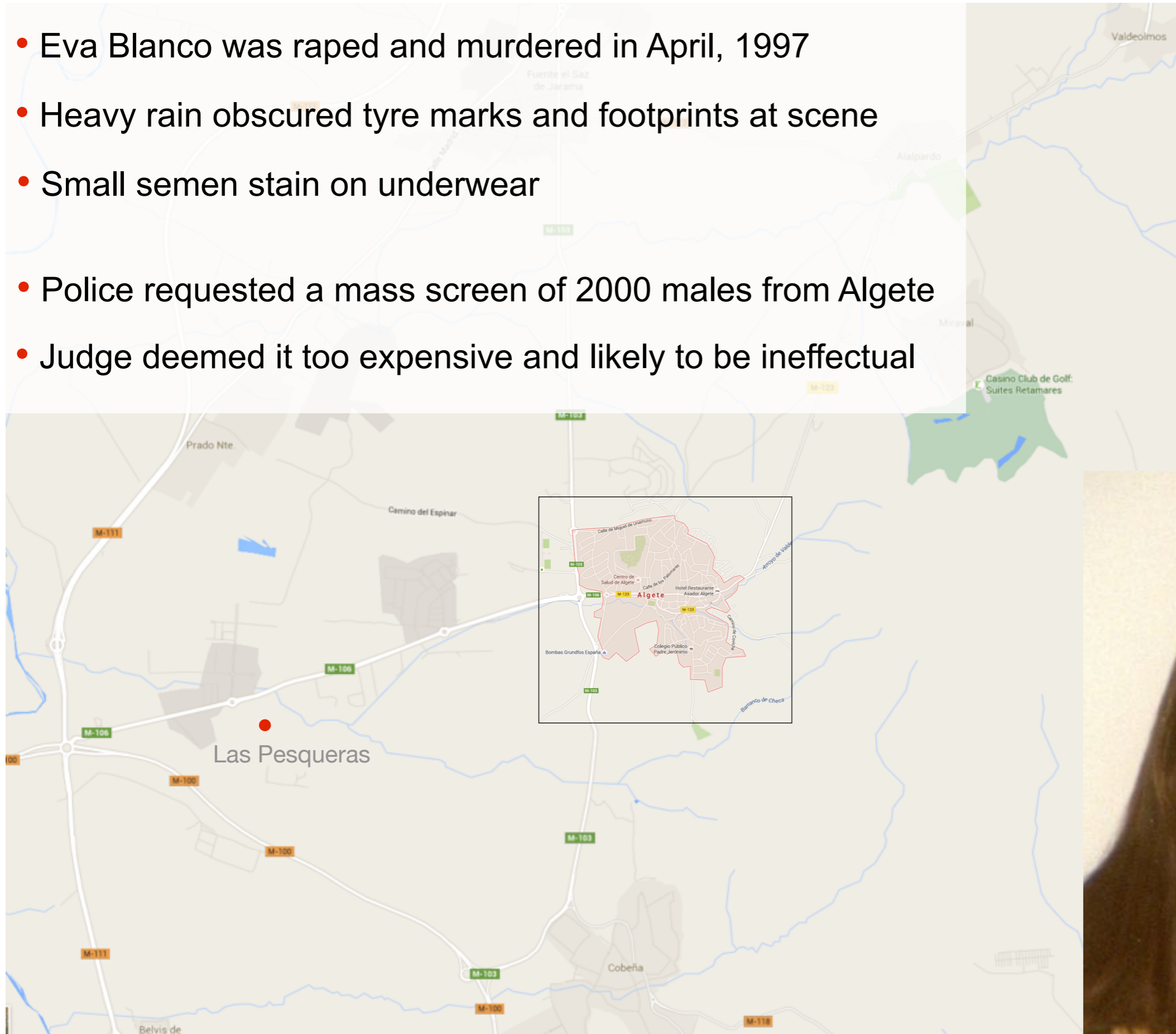
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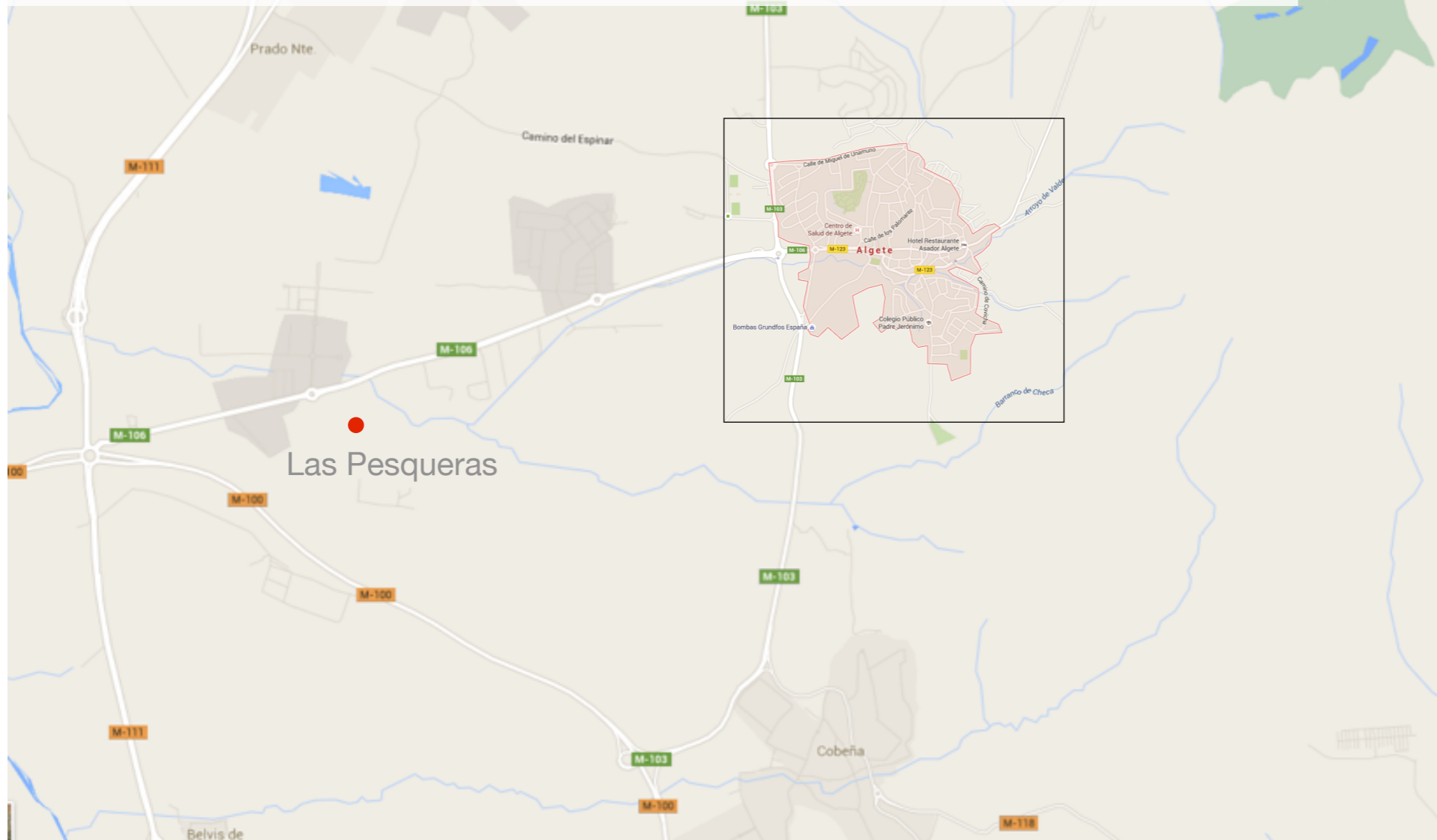
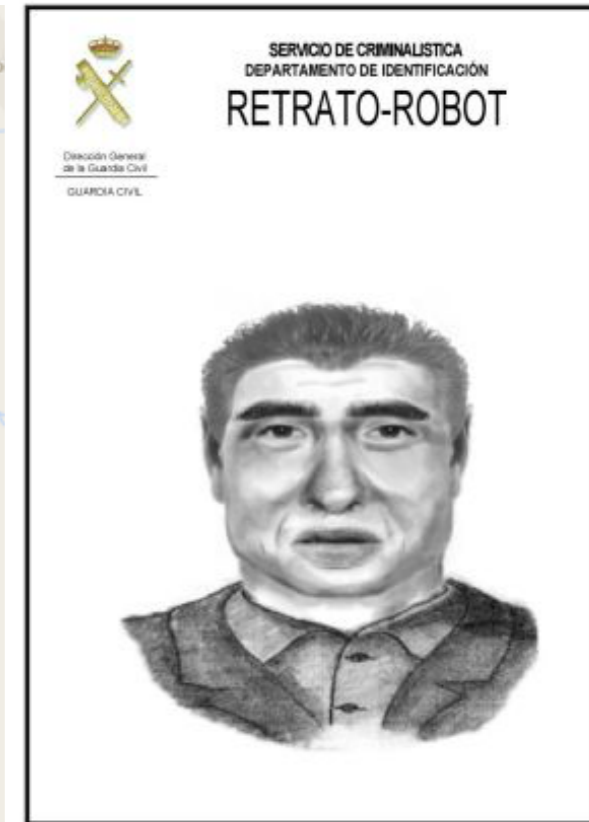
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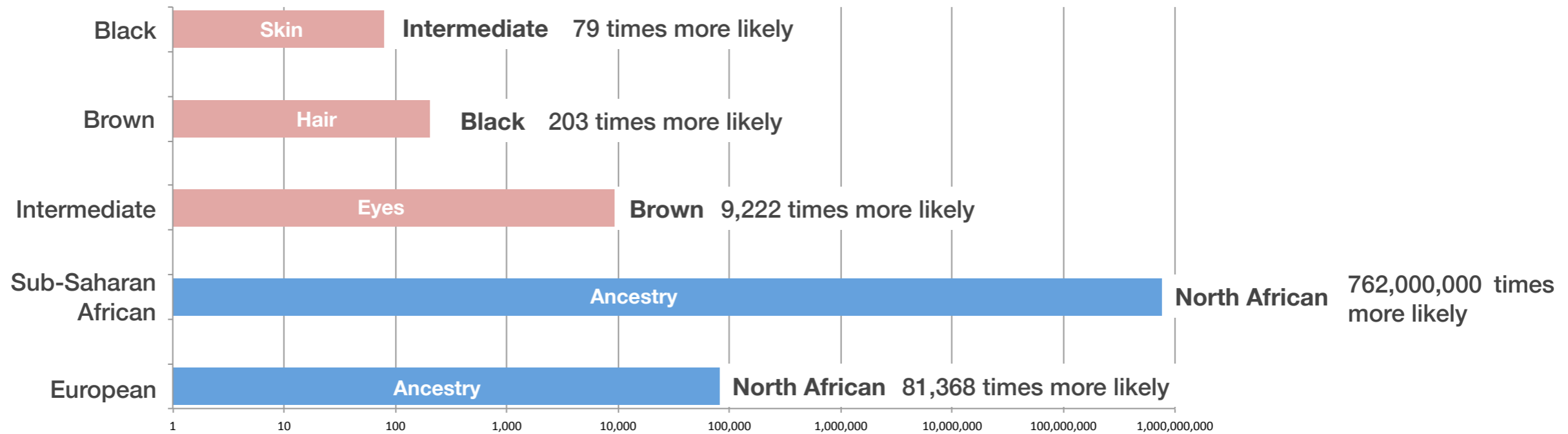
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- 63 pigmentation-predictive 'SHEP' SNPs typed

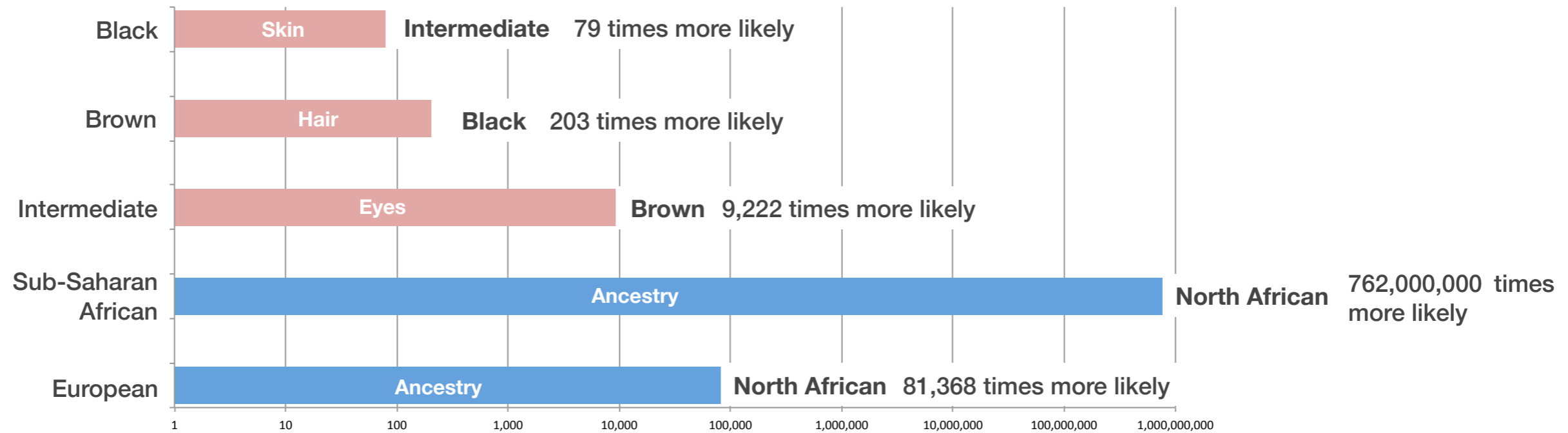
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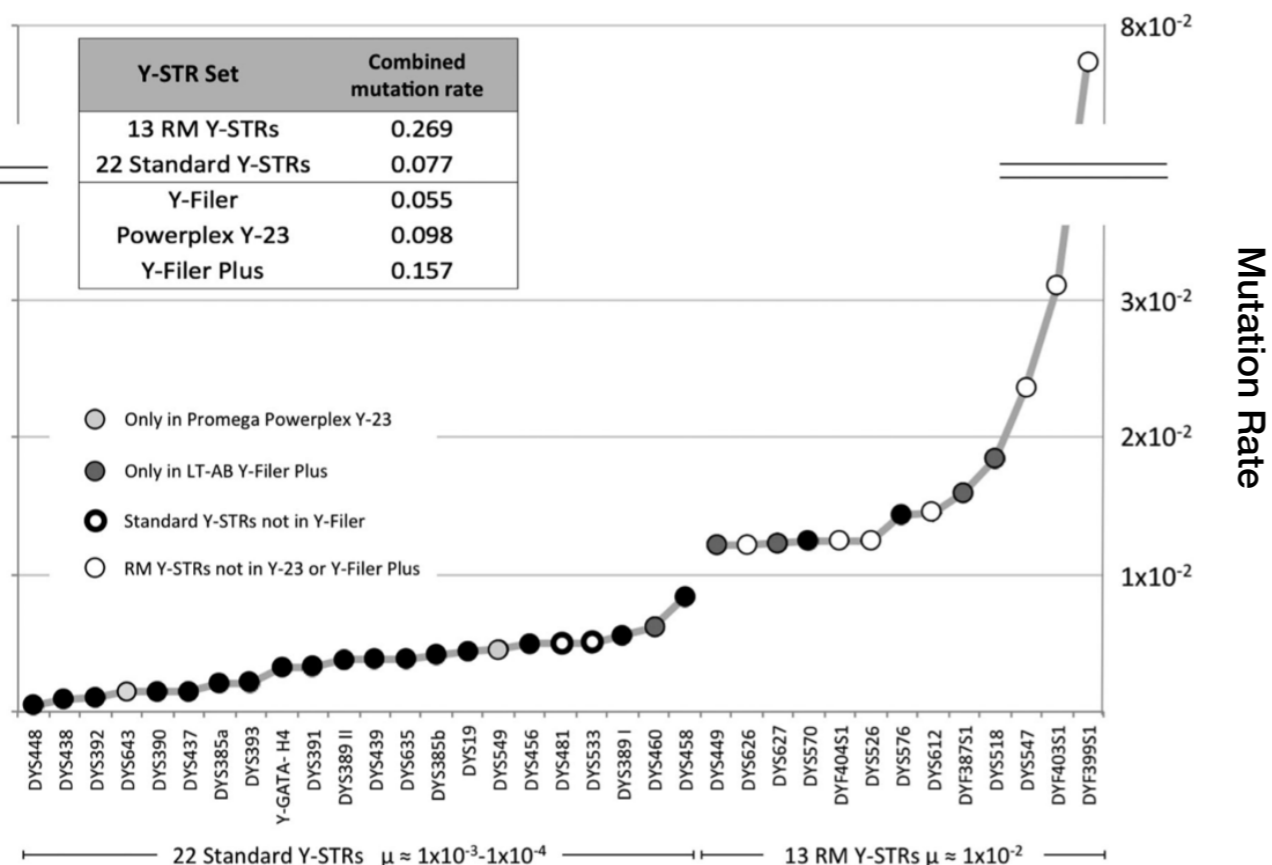
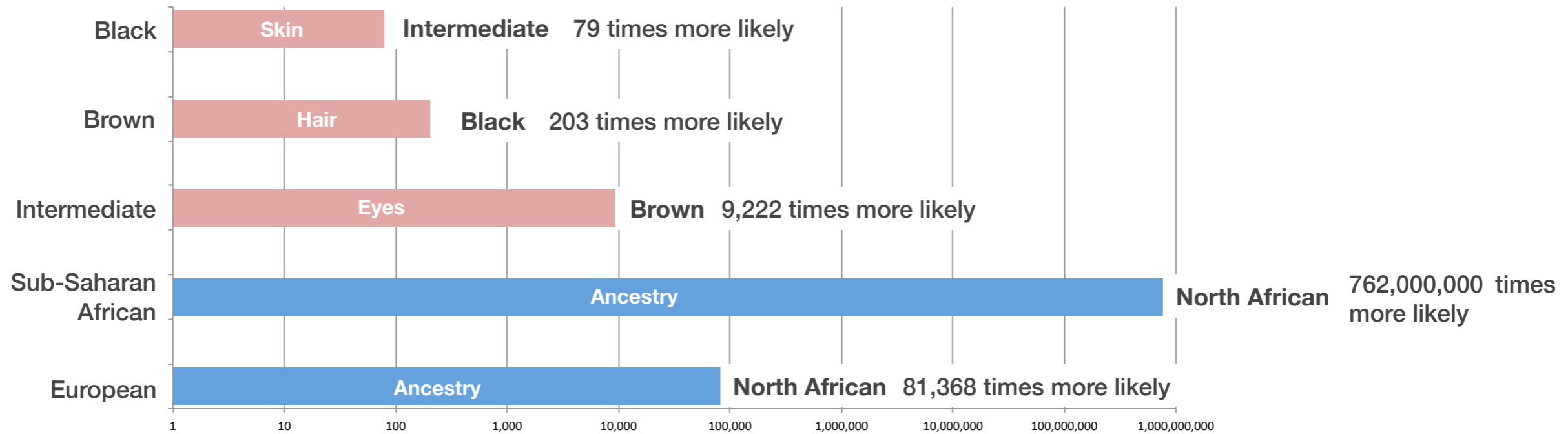
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- One sample gave a full Y-STR match apart from a single rapidly mutating Y-STR genotype - led to identification of a brother who had left Spain in 1999 and lived in France

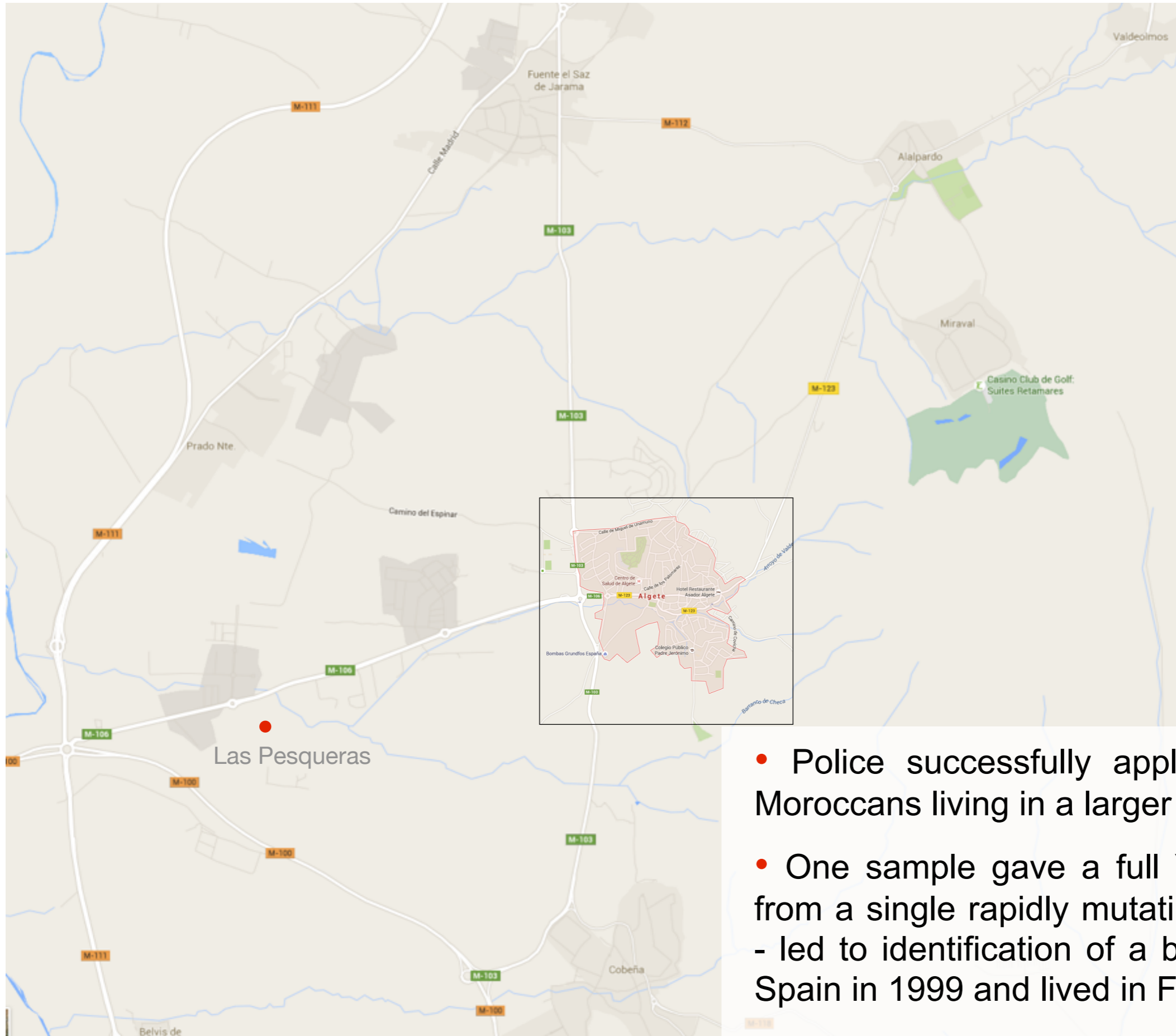
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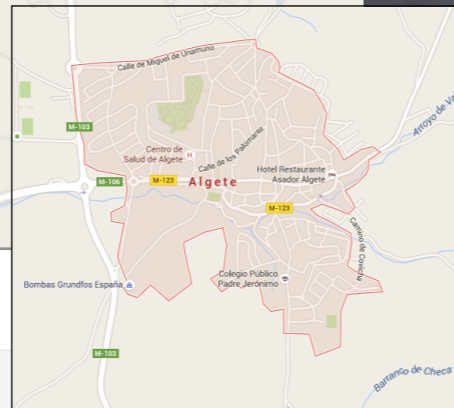


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Suspect arrested in France, October 2015



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EL PAÍS

CRIME

Madrid teen's suspected murderer arrested in France 18 years after crime

• Man who investigators believe murdered 17-year-old Eva Blanco was living in French village

PATRICIA ORTEGA DOLZ / F. JAVIER BARROSO | Madrid | 2 OCT 2015 - 17:38 CEST

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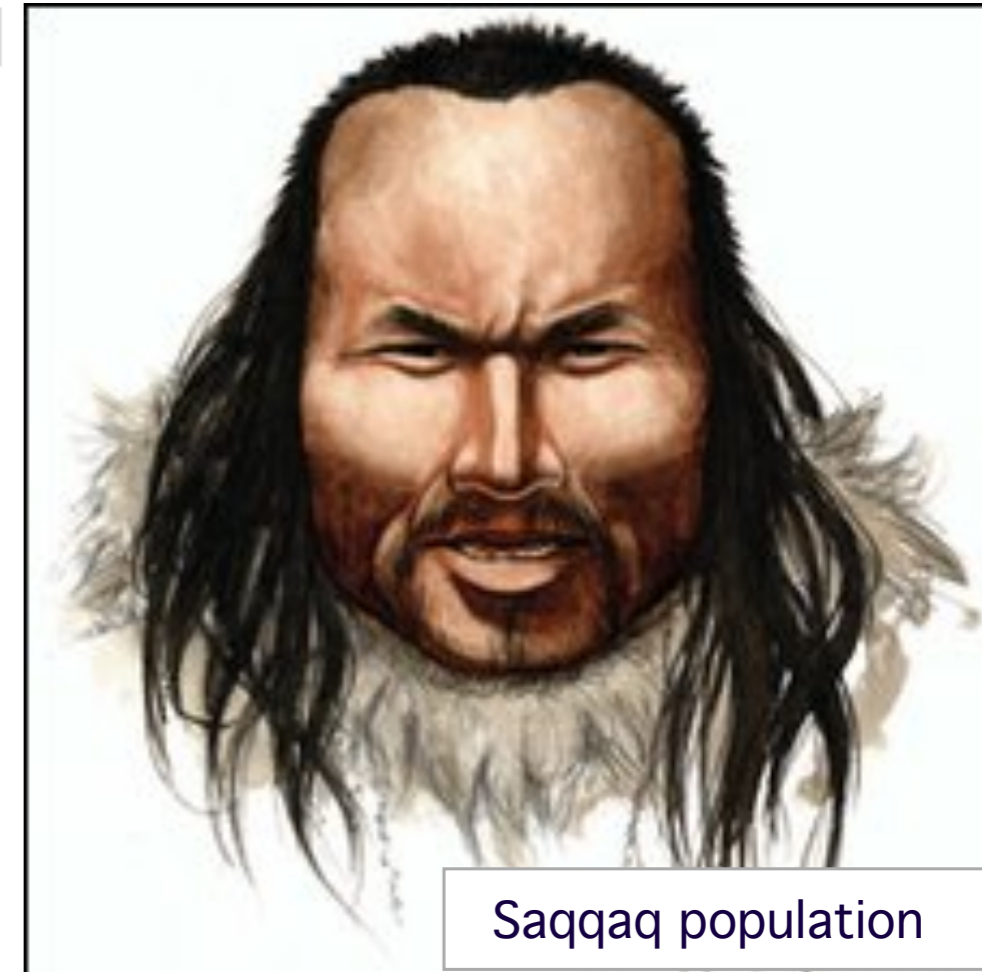
Palaeo-genomics researchers now provide photo-fits !

nature

ARTICLES

Ancient human genome sequence of an extinct Palaeo-Eskimo

We report here the genome sequence of an ancient human. Obtained from ~4,000-year-old permafrost-preserved hair, the genome represents a male individual from the first known culture to settle in Greenland. Sequenced to an average depth of 20×, we recover 79% of the diploid genome, an amount close to the practical limit of current sequencing technologies. We identify 353,151 high-confidence single-nucleotide polymorphisms (SNPs), of which 6.8% have not been reported previously. We estimate raw read contamination to be no higher than 0.8%. We use functional SNP assessment to assign possible phenotypic characteristics of the individual that belonged to a culture whose location has yielded only trace human remains. We compare the high-confidence SNPs to those of contemporary populations to find the populations most closely related to the individual. This provides evidence for a migration from Siberia into the New World some 5,500 years ago, independent of that giving rise to the modern Native Americans and Inuit.



Saqqaq population



SLC24A2/SLC45A2 *Hair colour/skin tone*

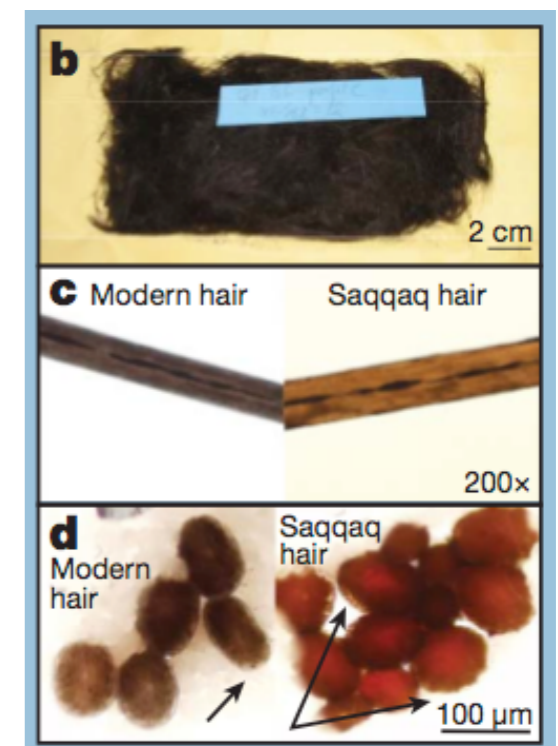
OCA2/HERC2 *Eye colour*

AR/EDA2R *MPBaldness*

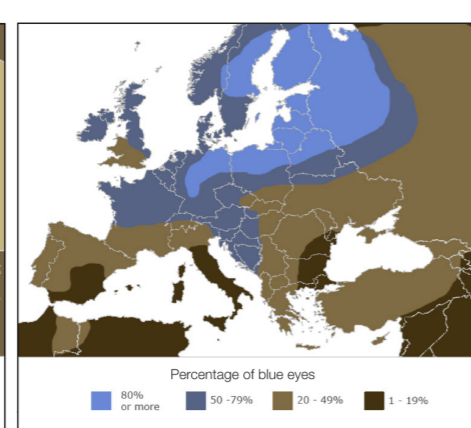
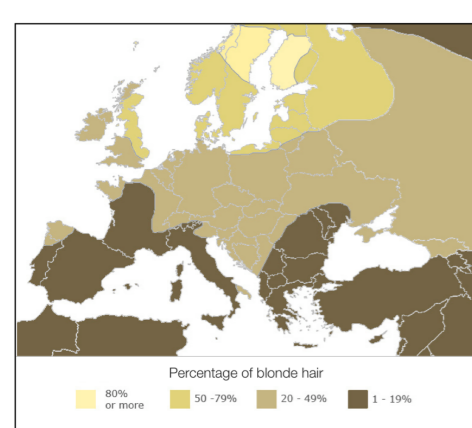
ABCC11 *Earwax viscosity*

EDAR *Shovel-shaped incisors*

ABO *Blood group*







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IrisPlex: A sensitive DNA tool for accurate prediction of blue and brown eye colour in the absence of ancestry information

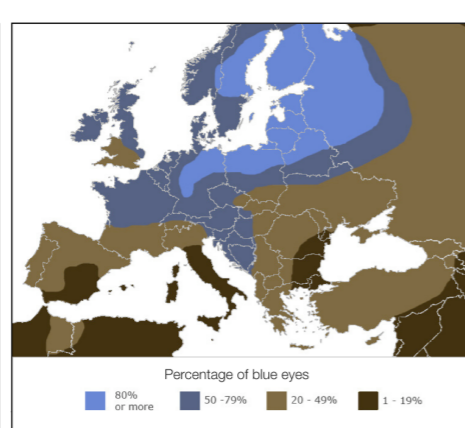
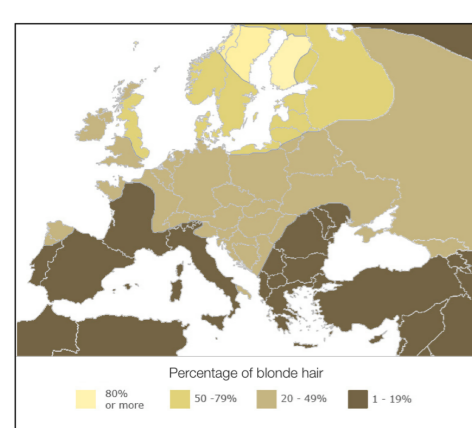
Susan Walsh, Fan Liu, Kaye N. Ballantyne, Mannis van Oven, Oscar Lao, Manfred Kayser*

The HirisPlex system for simultaneous prediction of hair and eye colour from DNA

Susan Walsh^a, Fan Liu^a, Andreas Wollstein^a, Leda Kovatsi^b, Arwin Ralf^a, Agnieszka Kosiniak-Kamysz^c, Wojciech Branicki^{d,e}, Manfred Kayser^{a,*}

IrisPlex
HirisPlex
HirisPlex-S





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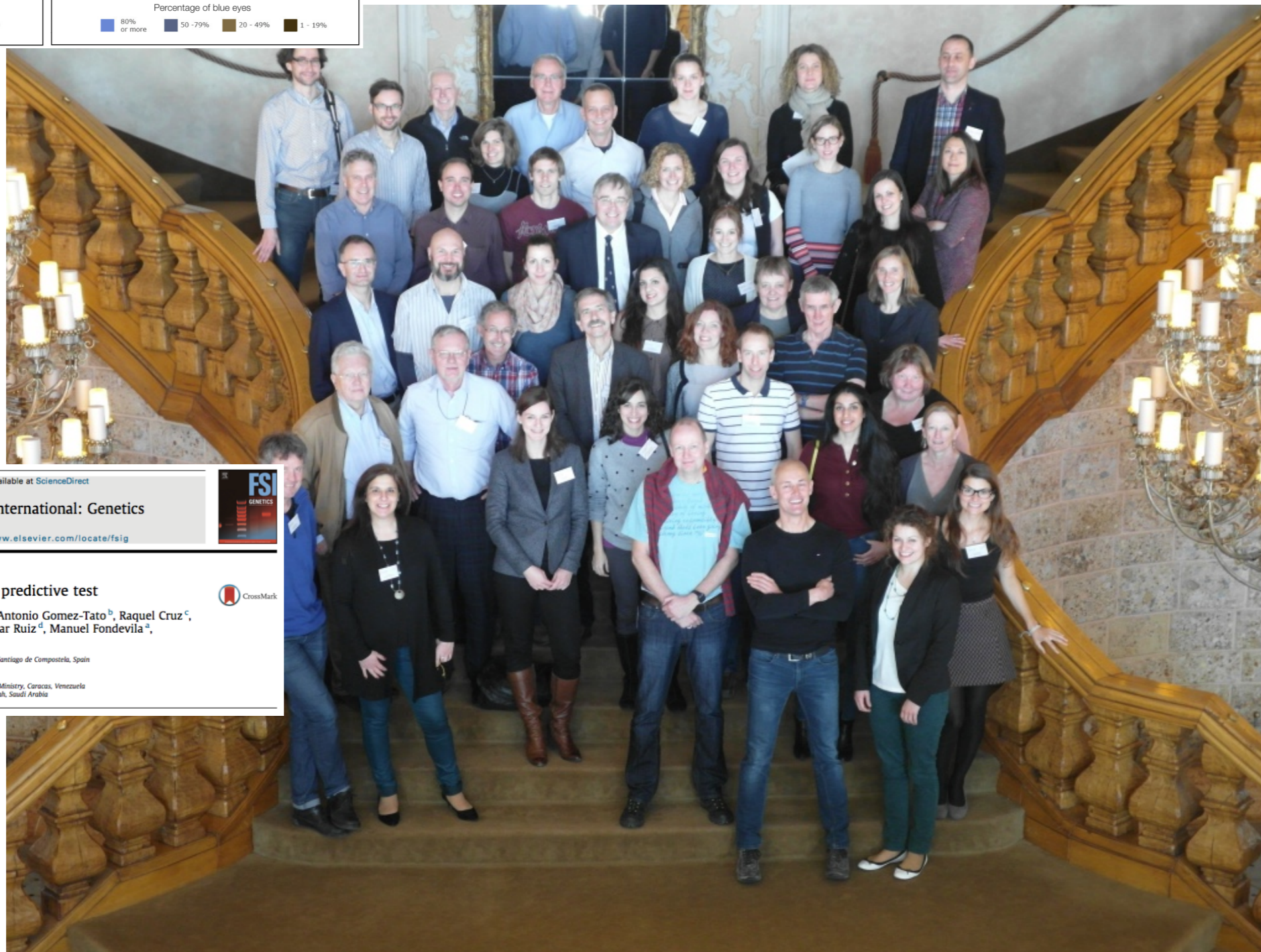
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IrisPlex

HirisPlex

HirisPlex-S

Skin colour in individuals with African co-ancestry



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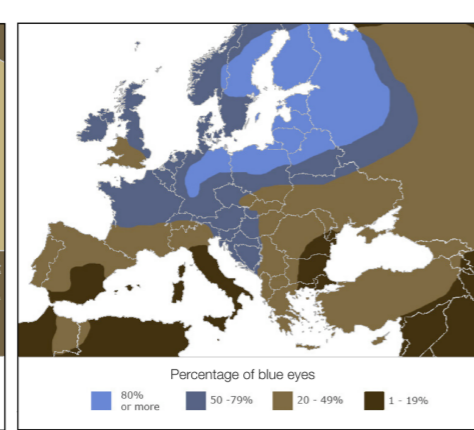
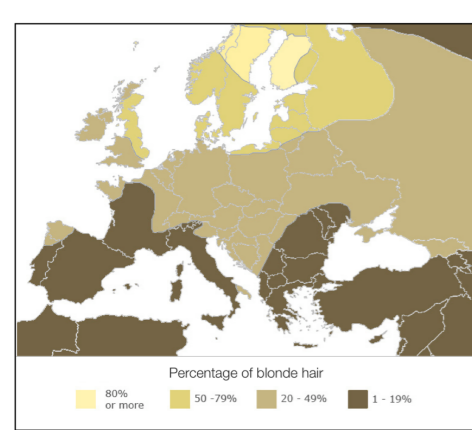
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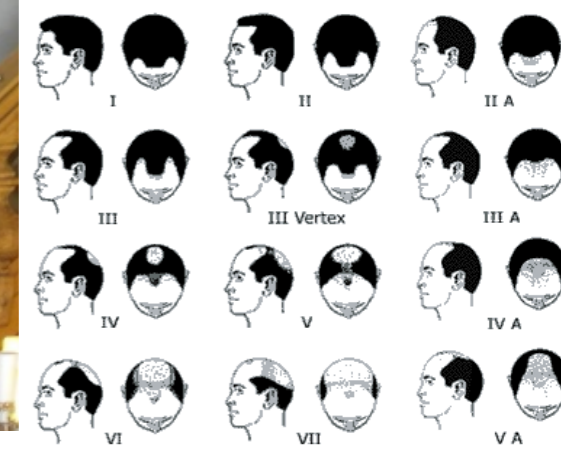
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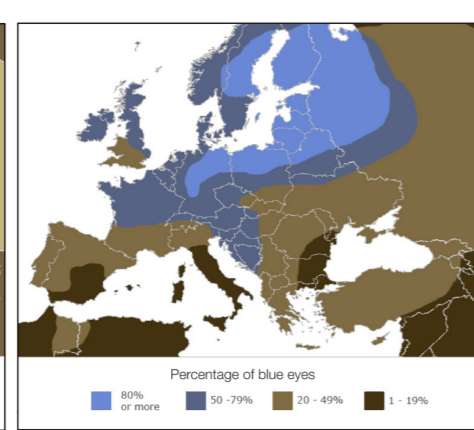
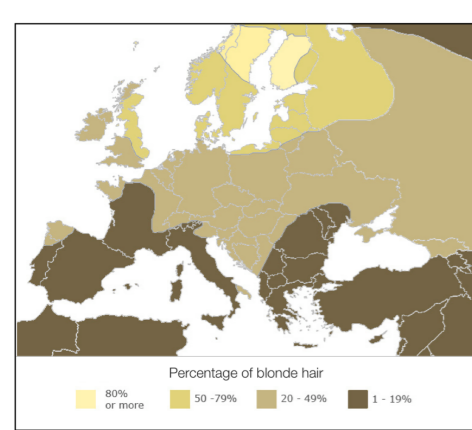
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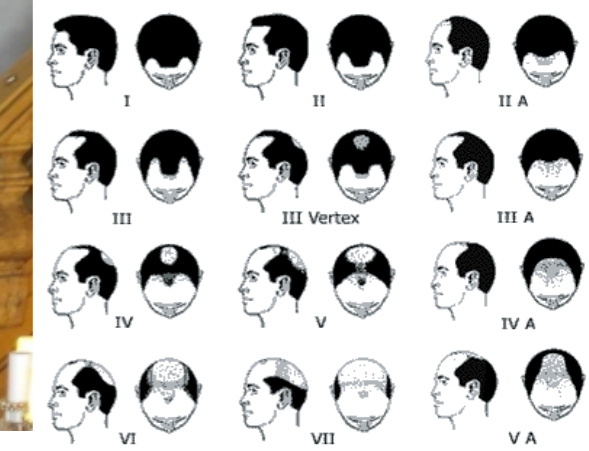




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PLOS ONE

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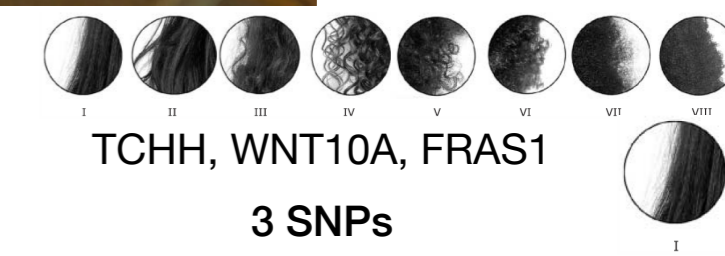
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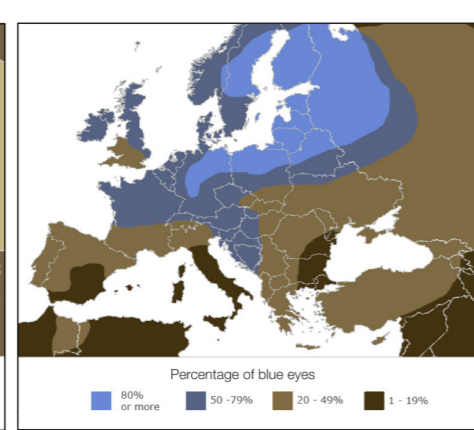
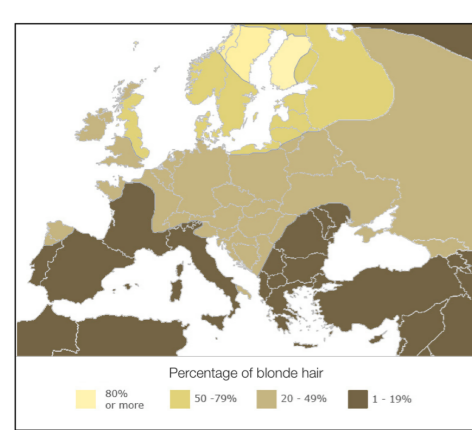
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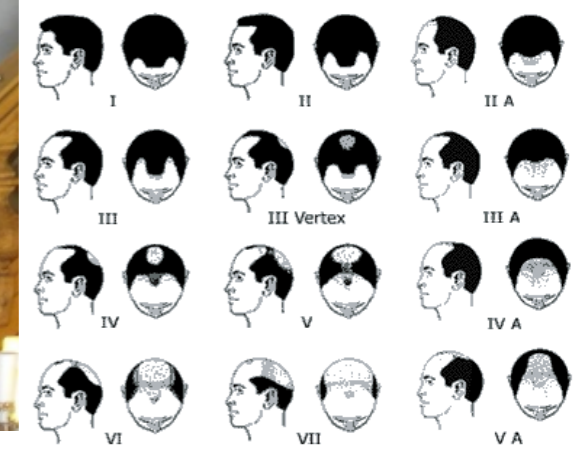




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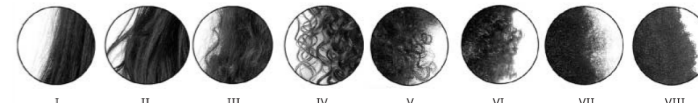


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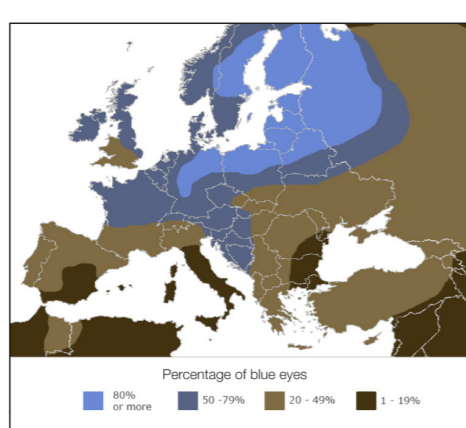
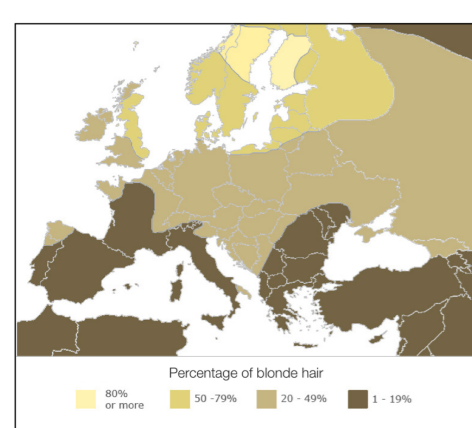
'Hair type'



TCHH, WNT10A, FRAS1

3 SNPs





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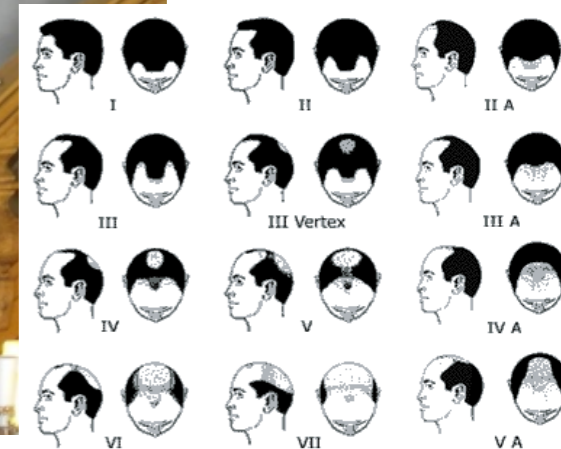
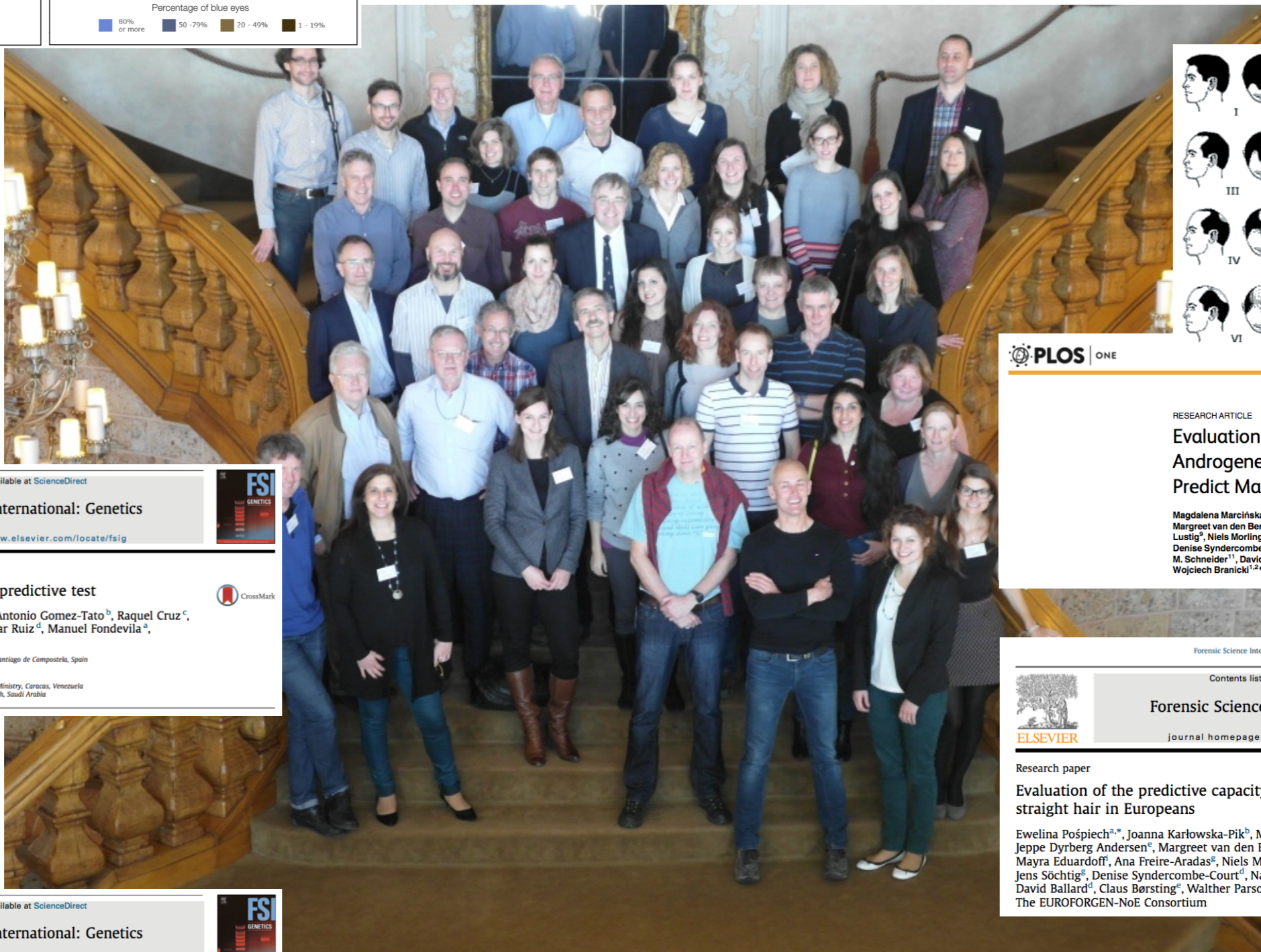
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Ancestry panel for MPS



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Building a forensic ancestry panel from the ground up: The EUROFORGEN Global AIM-SNP set

C. Phillips^{a,*}, W. Parson^{b,c}, B. Lundsberg^d, C. Santos^a, A. Freire-Aradas^a, M. Torres^e, M. Eduardoff^b, C. Børsting^d, P. Johansen^d, M. Fondevila^a, N. Morling^d, P. Schneider^f
the EUROFORGEN-NoE Consortium, Á. Carracedo^{a,e,g}, M.V. Lareu^a



'Hair type'



TCHH, WNT10A, FRAS1

3 SNPs



Where are we now?

Using Genome-wide SNPs for DNA Phenotyping and Kinship Inference



Ellen McRae Greytak, PhD

Parabon NanoLabs, Inc., 11260 Roger Bacon Dr. Suite 406, Reston VA 20190
ellen@parabon.com | parabon-nanolabs.com/snapshot

Introduction

DNA Phenotyping is the prediction of physical appearance from DNA. It can be used to generate leads in cases where there are no suspects or database hits, or to help identify remains. Using in-depth data mining and advanced machine learning, and with support from the US Department of Defense, we have built the Snapshot™ Forensic DNA Phenotyping System, which accurately **predicts genetic ancestry, eye color, hair color, skin color, freckling, and face shape** in individuals from any ethnic background, even admixed individuals. Each prediction is presented with a measure of confidence, and highly unlikely trait values are excluded. We have also developed a novel kinship inference algorithm that can accurately predict relatedness between any two individuals out to 6th-degree relatives using only their DNA.

SNP Technology

- Traditional DNA forensics uses short tandem repeats (STRs), which can only be used for matching and cannot generate leads in the absence of a suspect or CODIS hit
- Snapshot uses single nucleotide polymorphisms (SNPs), which code for the actual differences between people
- Millions of SNPs are found across the genome and provide the information needed to predict an individual's appearance

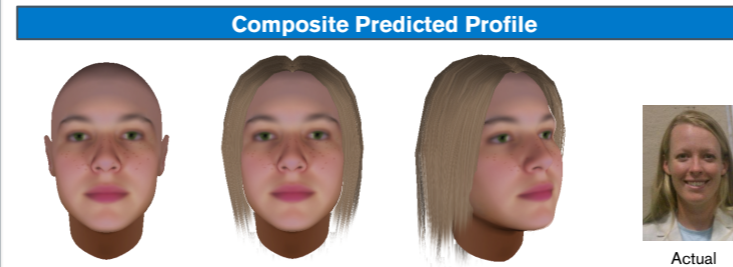
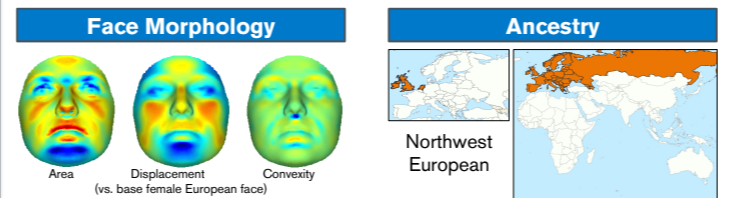
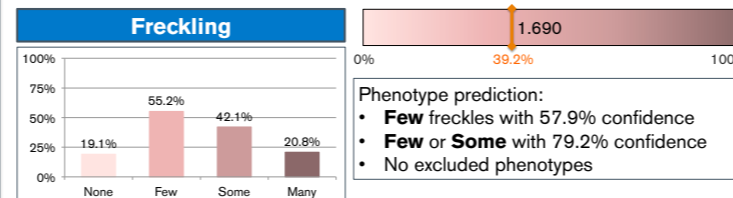
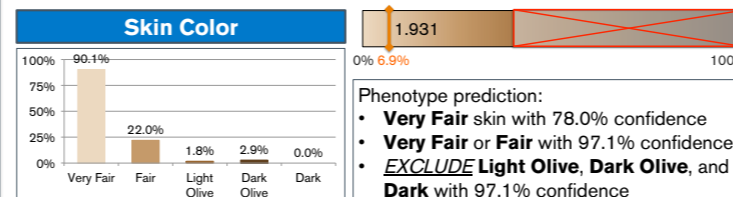
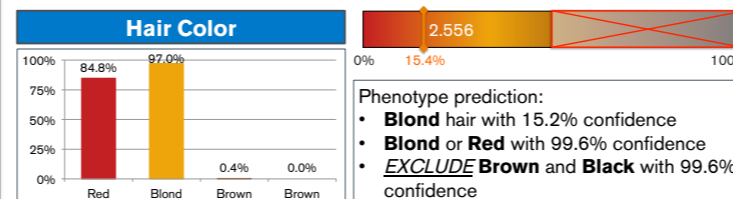
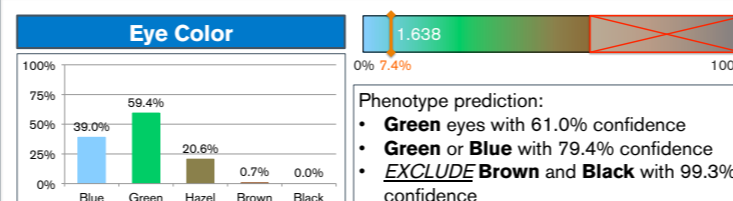
DNA Requirements

- Snapshot can accept data from any off-the-shelf Illumina genome-wide SNP genotyping array or next-generation sequencing machine
- Parabon has partnered with forensic laboratories to generate this data from casework samples
- Call rates sufficient for prediction can be obtained from as little as **50 pg** of extracted DNA, which can potentially be found in a fingerprint
- Prediction accuracy degrades smoothly with loss of SNPs from small or degraded samples, which is reported via wider confidence intervals

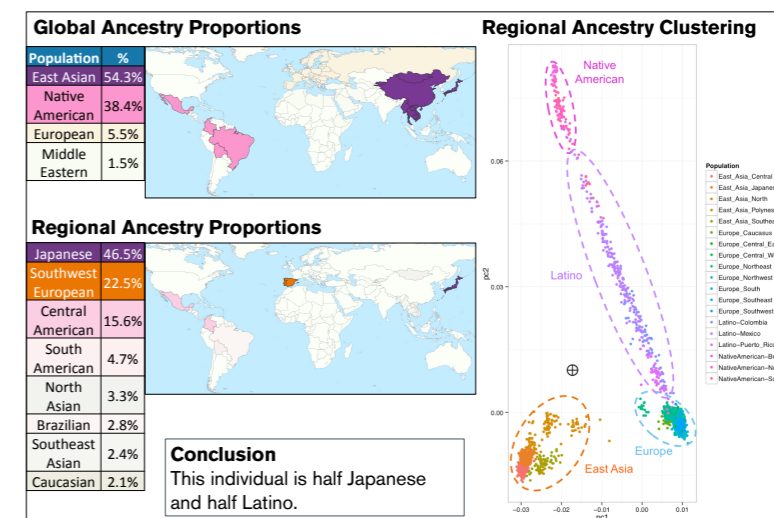
Genetic Ancestry

- Snapshot compares new, unknown subjects against background data from nearly 8,000 individuals with clearly defined ancestry
- Infers percent membership in each of 7 global populations (Africa, the Middle East, Europe, Central/South Asia, East Asia, Oceania, America)
- Also calculates percent ancestry from subcontinental populations (e.g., Northwest vs. Northeast Europe)
- Robust to admixture and can distinguish among closely-related populations

Example Prediction



Complex Ancestry Example



Distant Kinship Inference

- Uses genome-wide SNP data to compare two genomes and predict the degree of relatedness between the two individuals
- 100% accuracy for parent-offspring, full siblings, and 2nd-degree relatives
- >95% accurate at distinguishing 6th-degree relatives (2nd cousins once-removed) from unrelated pairs
- Can be used for identifying remains, validating kinship claims, and constructing relationship networks among subjects of interest

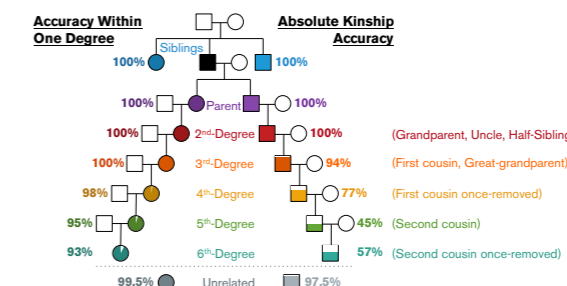


Figure 1: Snapshot Kinship accuracy, measured as the frequency of correct predictions of the exact degree of relatedness (absolute accuracy) and the frequency of predictions within one degree of relatedness (n = 1,282 relationships). All relationships are defined relative to the black square.



Extending the scope of data from conventional STR analysis

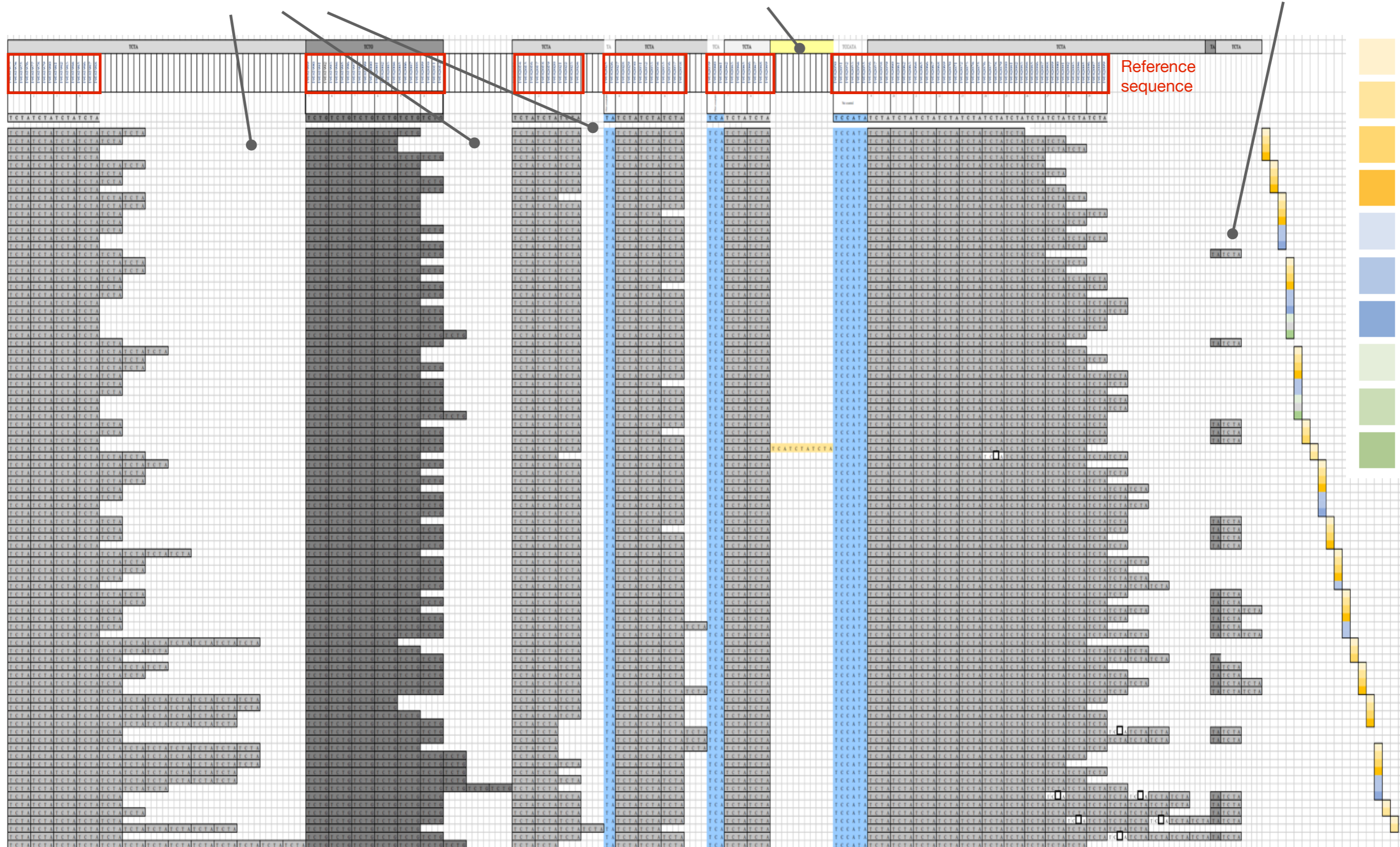
- More STRs/variation (MPS) aids mixture analysis and familial searching
- Enhanced Y-STR analysis can identify close male relatives - sometimes

MPS analysis of D21S11 reveals 5-6 STRs in a row

Nucleotides additional
to reference sequence

Ad-hoc 11 nucleotide
segmental duplication

TA [TCTA]_N motif not
in reference sequence



Where are we now?

Extending the scope of data from conventional STR analysis

- More STRs/variation (MPS) aids mixture analysis and familial searching
- Enhanced Y-STR analysis can identify close male relatives - sometimes

Adding more trait predictive tests and improving geographic resolution

- Hair morphology / early-onset male baldness. More ancestry SNPs
- The most promising new tests are age estimation and facial modeling

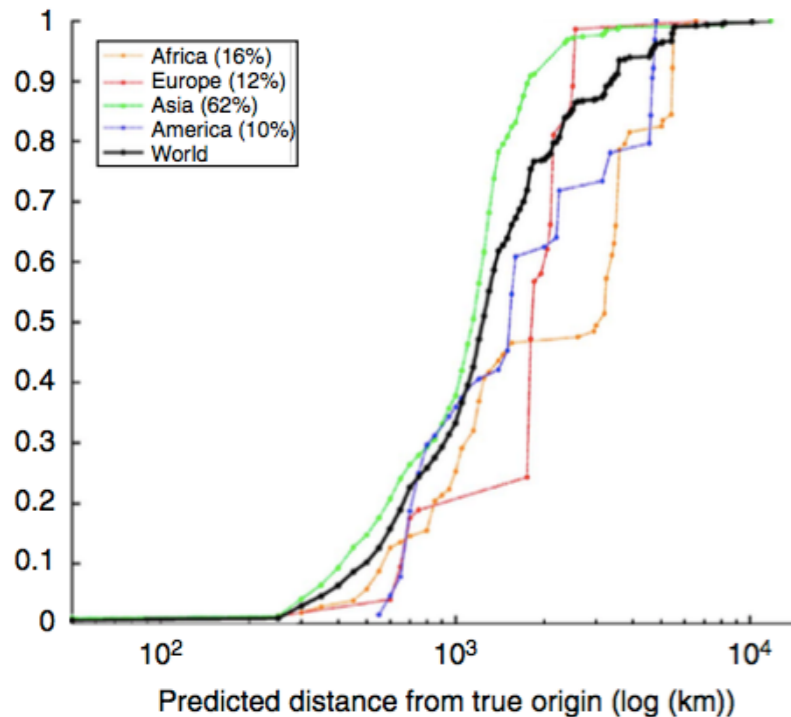
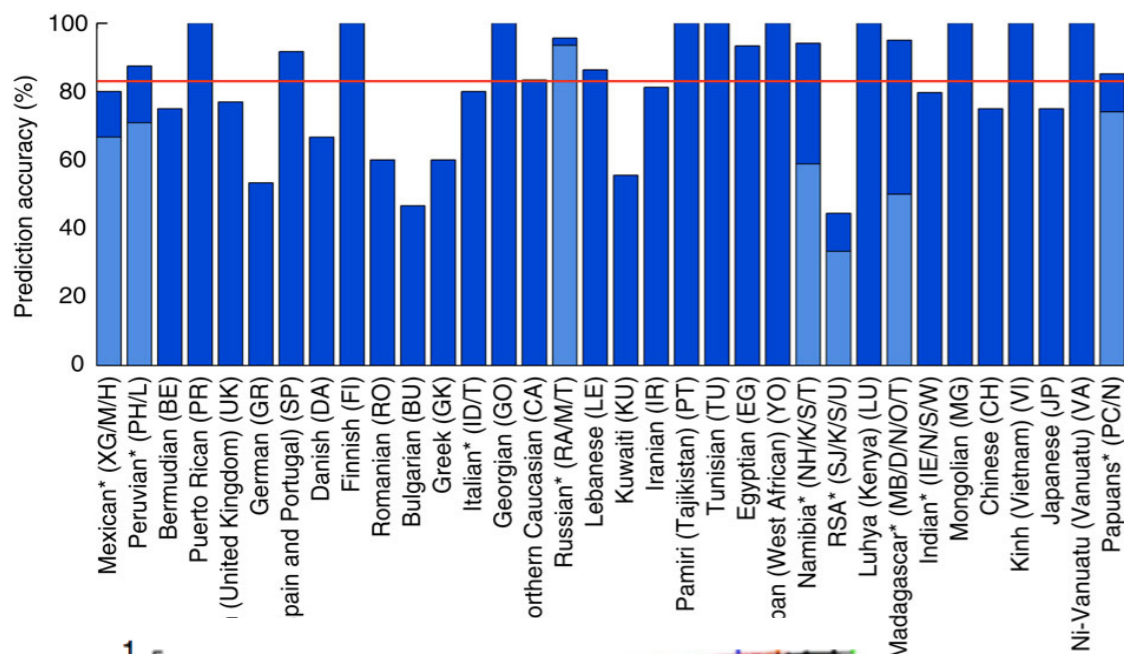
Possible to improve geographic precision with high density SNP sets

NATURE COMMUNICATIONS | ARTICLE

GPS 130,000 SNPs

Geographic population structure analysis of worldwide human populations infers their biogeographical origins

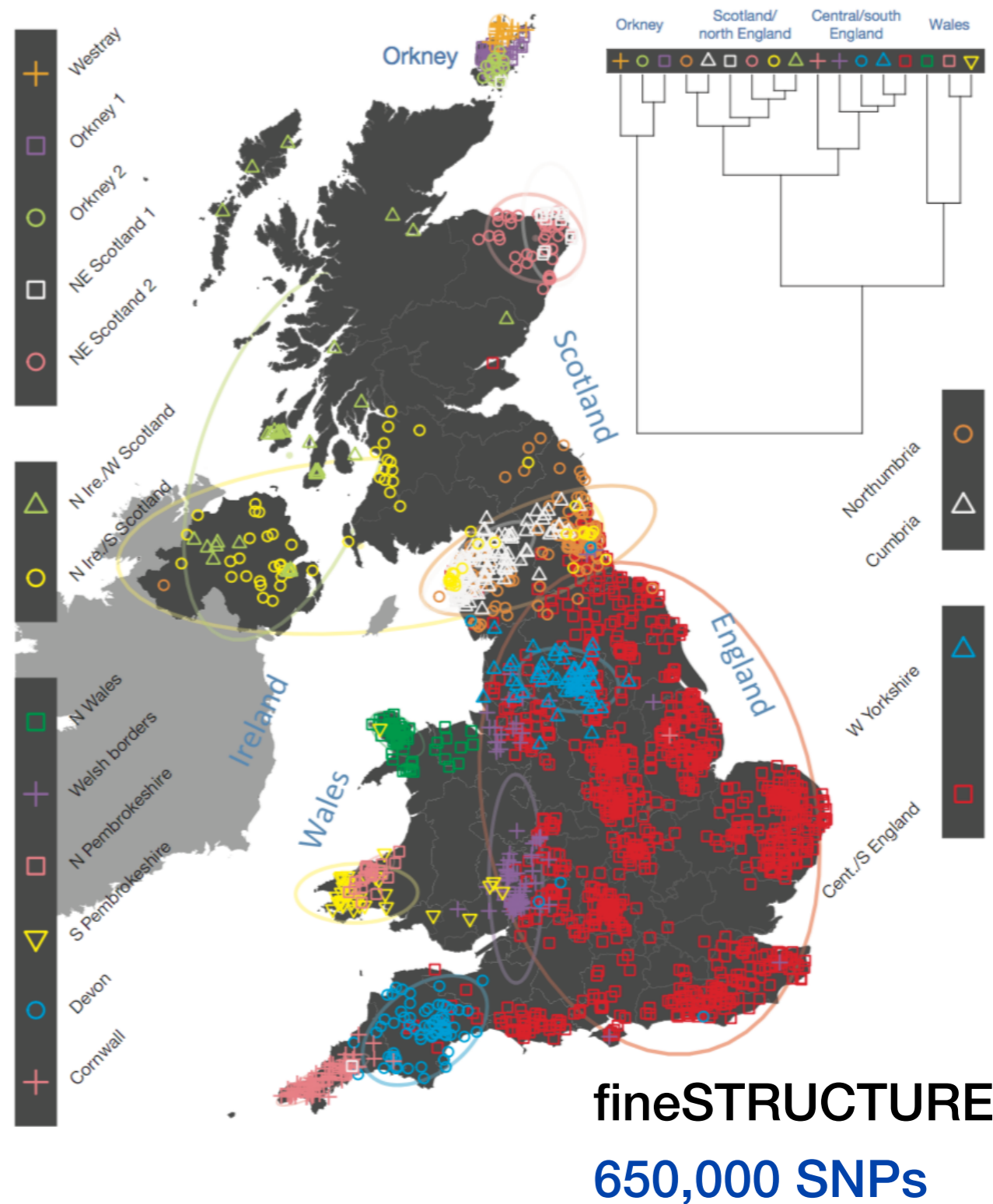
Eran Elhaik, Tatiana Tatarinova, Dmitri Chebotarev, Ignazio S. Piras, Carla Maria Calò, Antonella De Montis, Manuela Atzori, Monica Marini, Sergio Tofanelli, Paolo Francalacci, Luca Pagani,



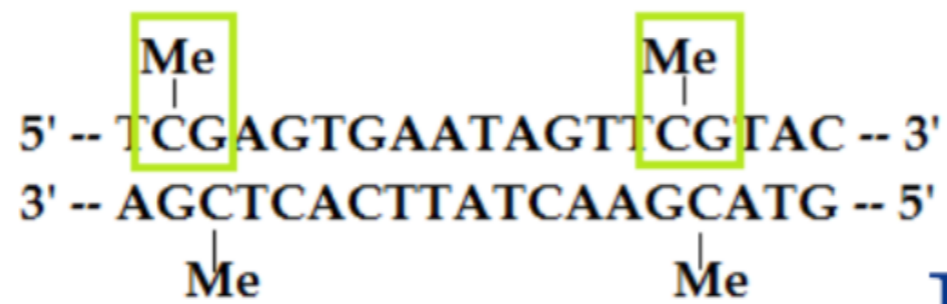
ARTICLE

The fine-scale genetic structure of the British population

Stephen Leslie^{1,2,3*}, Bruce Winney^{3*}, Garrett Hellenthal^{4*}, Dan Davison⁵, Abdelhamid Boumertit³, Tammy Day³, Katarzyna Hutnik³, Ellen C. Rovrvik³, Barry Cunliffe⁶, Wellcome Trust Case Control Consortium 2†.

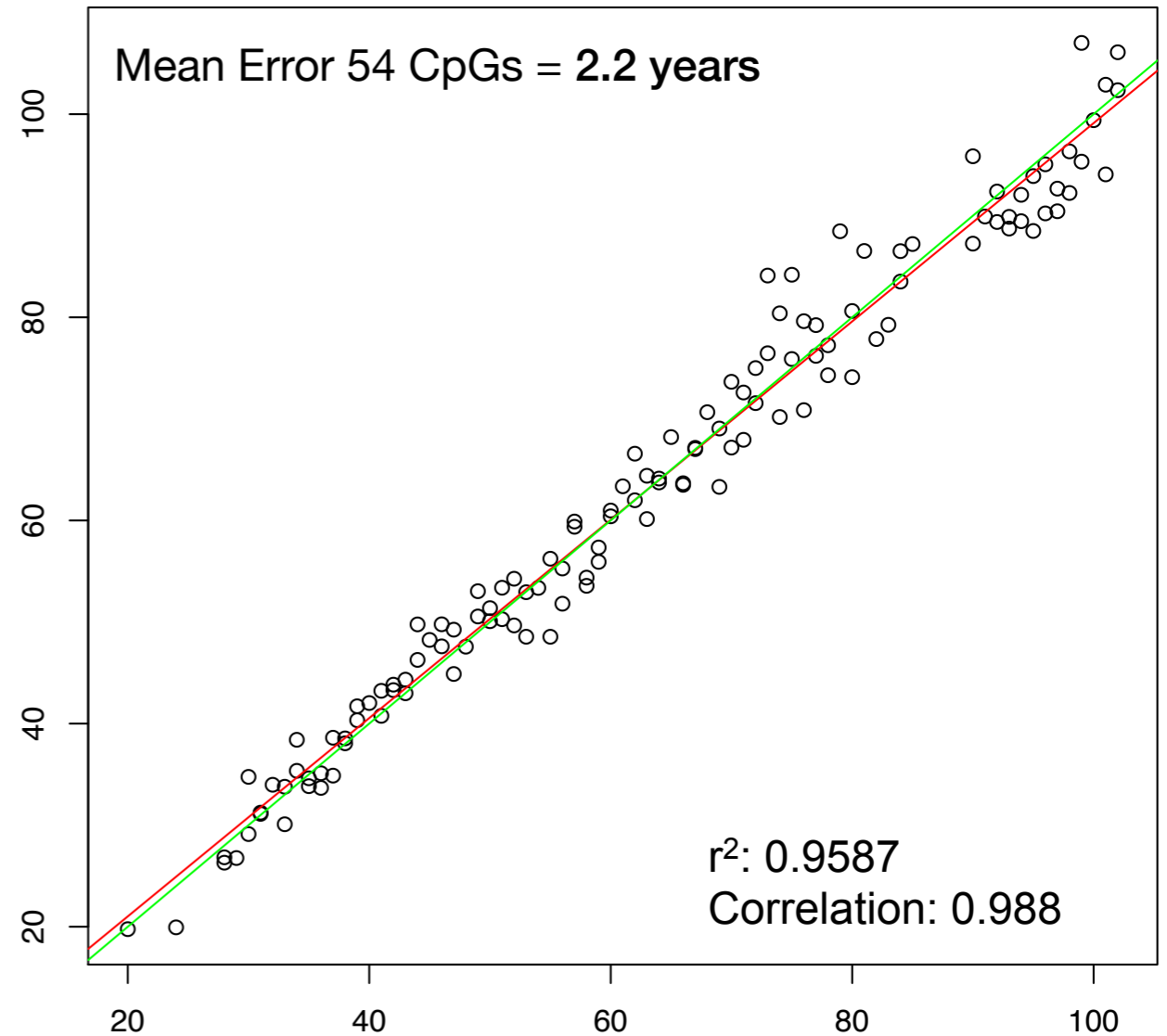
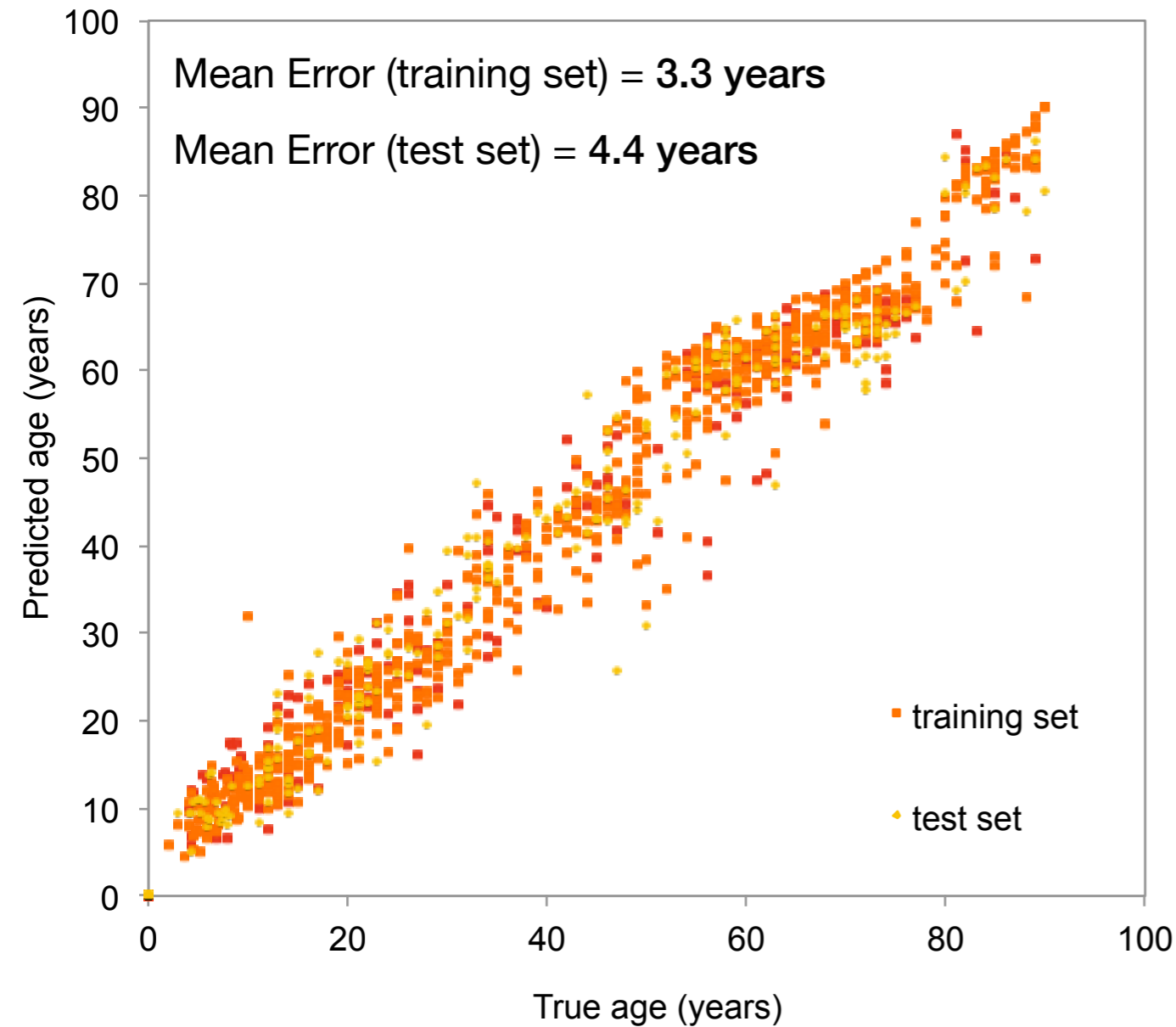


Forensic meth-age tests



ELOVL2

FHL2



- High variance in methylation levels between tissues and individuals
- Bisulphite treatment of methylated DNA is variable, often incomplete - creates noise

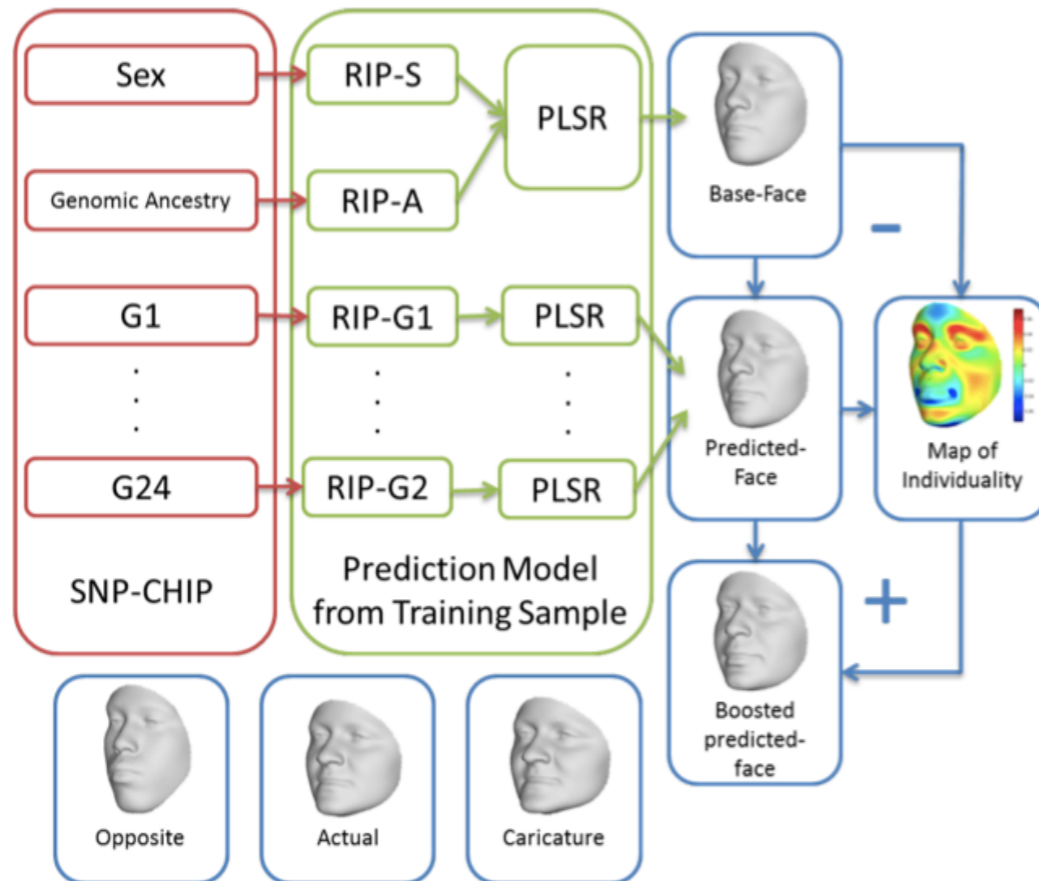
Facial modeling with SNP data



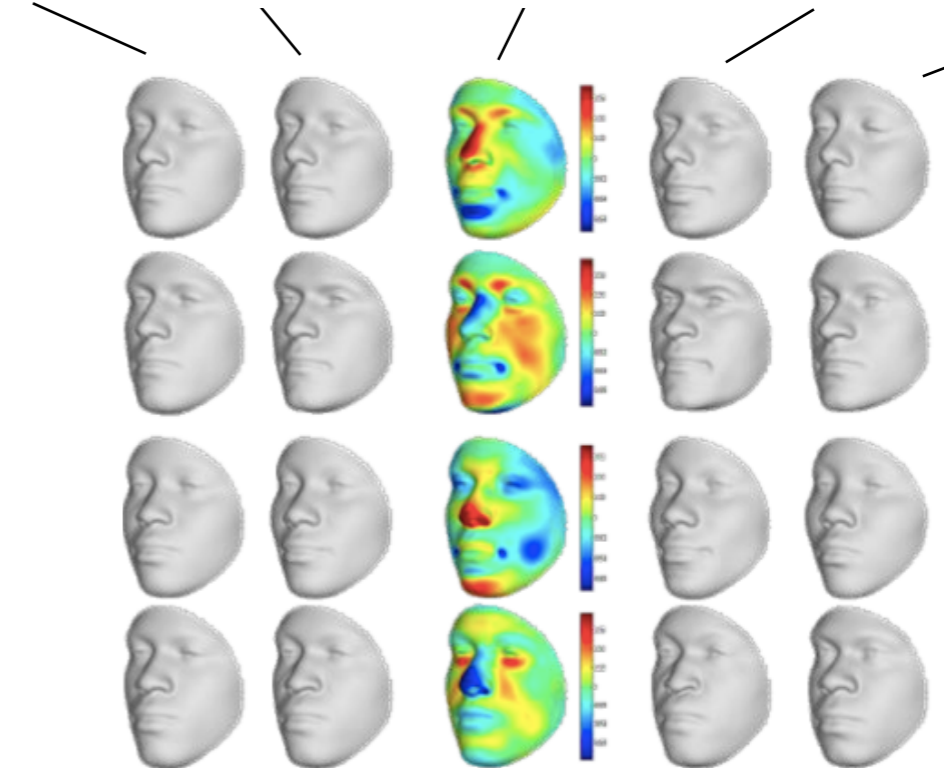
Modeling 3D Facial Shape from DNA

Peter Claes¹, Denise K. Liberton², Katleen Daniels¹, Kerri Matthes Rosana², Ellen E. Quillen², Laurel N. Pearson², Brian McEvoy³, Marc Bauchet², Arslan A. Zaidi², Wei Yao², Hua Tang⁴, Gregory S. Barsh^{4,5}, Devin M. Absher⁵, David A. Puts², Jorge Rocha^{6,7}, Sandra Beleza^{4,8}, Rinaldo W. Pereira⁹, Gareth Baynam^{10,11,12}, Paul Suetens¹, Dirk Vandermeulen¹, Jennifer K. Wagner¹³, James S. Boster¹⁴, Mark D. Shriver^{2*}

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Base Predicted Individuality Map Boosted Actual

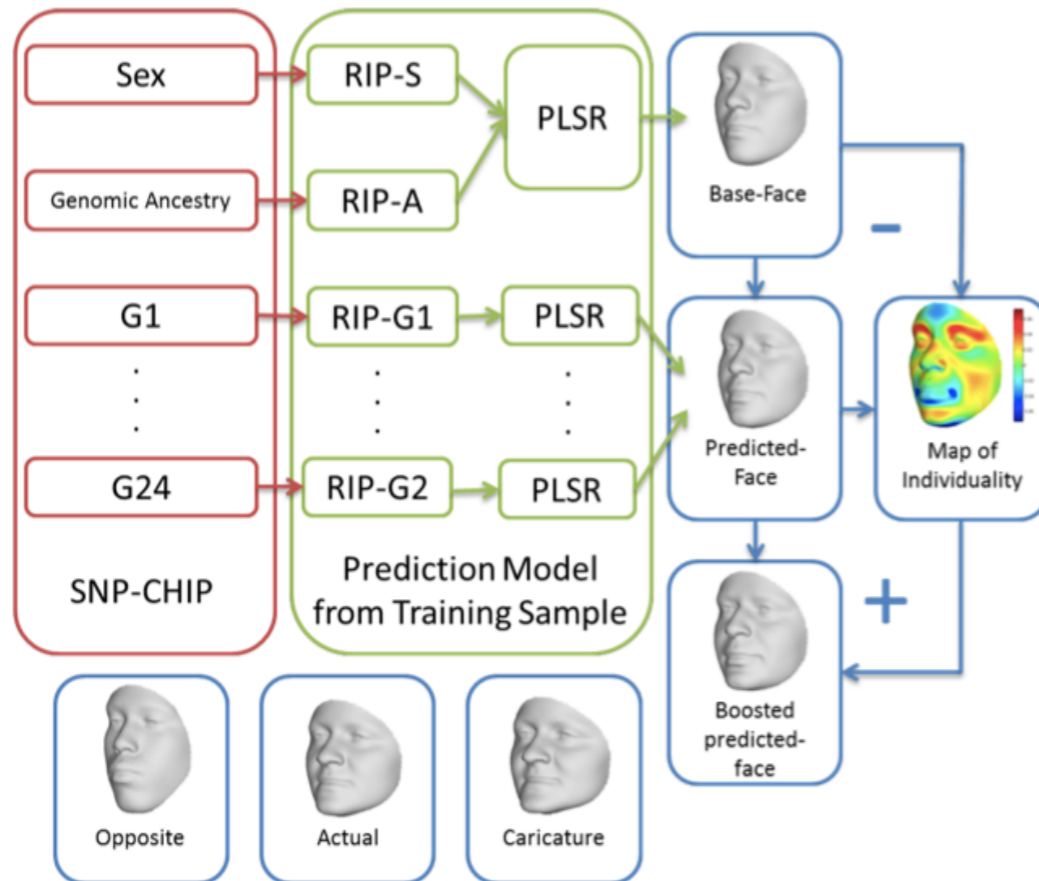


Facial modeling with SNP data

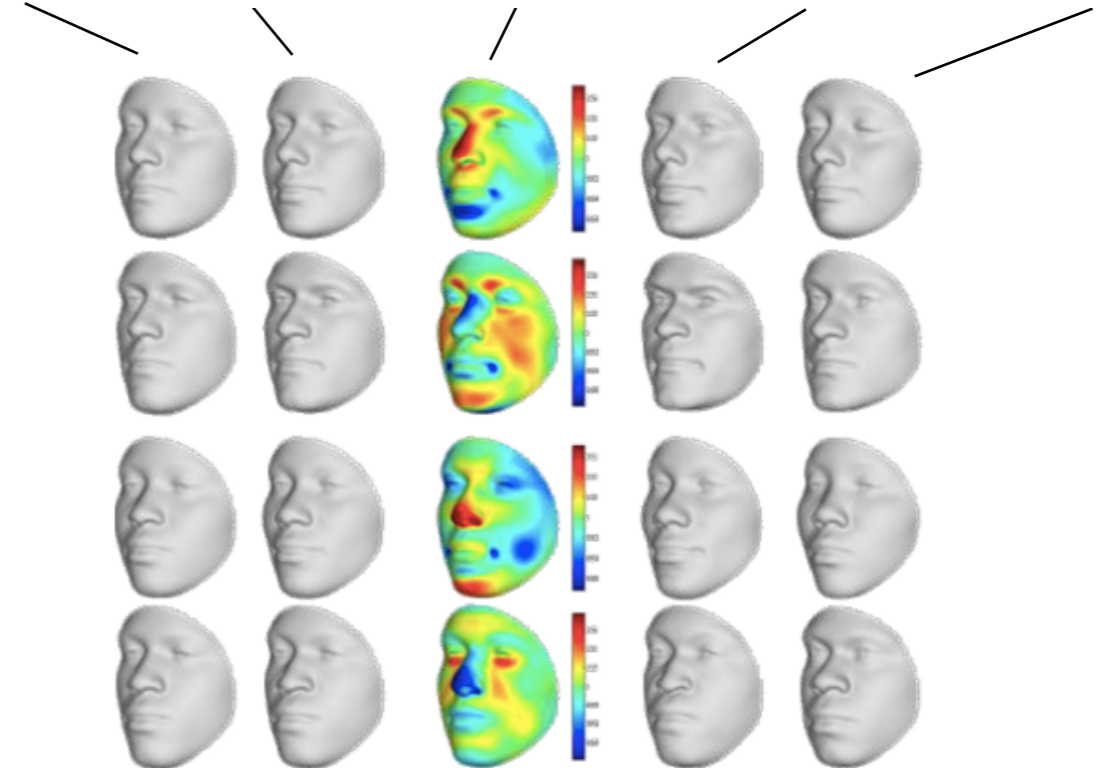


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Base Predicted Individuality Map Boosted Actual



Where are we now?

Extending the scope of data from conventional STR analysis

- More STRs/variation (MPS) aids mixture analysis and familial searching
- Enhanced Y-STR analysis can identify close male relatives - sometimes

Adding more trait predictive tests and improving geographic resolution

- Hair morphology / early-onset male baldness. More ancestry SNPs
- The most promising new tests are age estimation and facial modeling

Combining genetic tests can bring ‘synergy’ to appearance prediction

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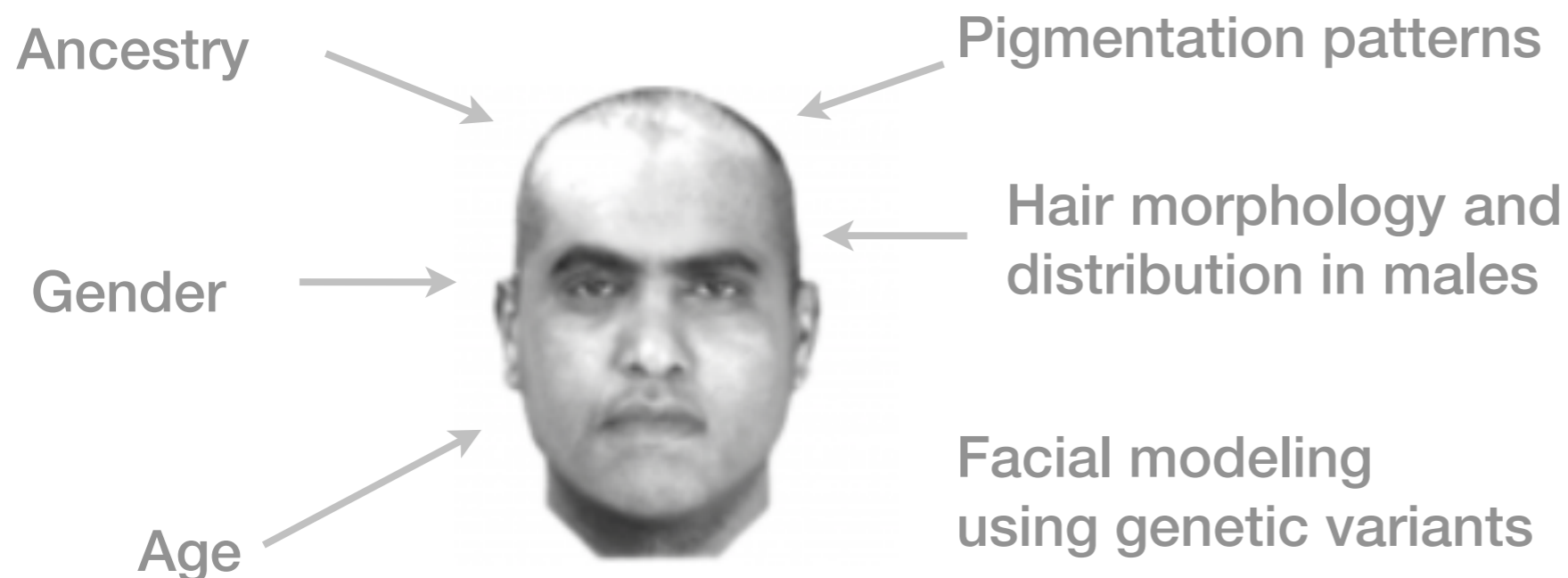
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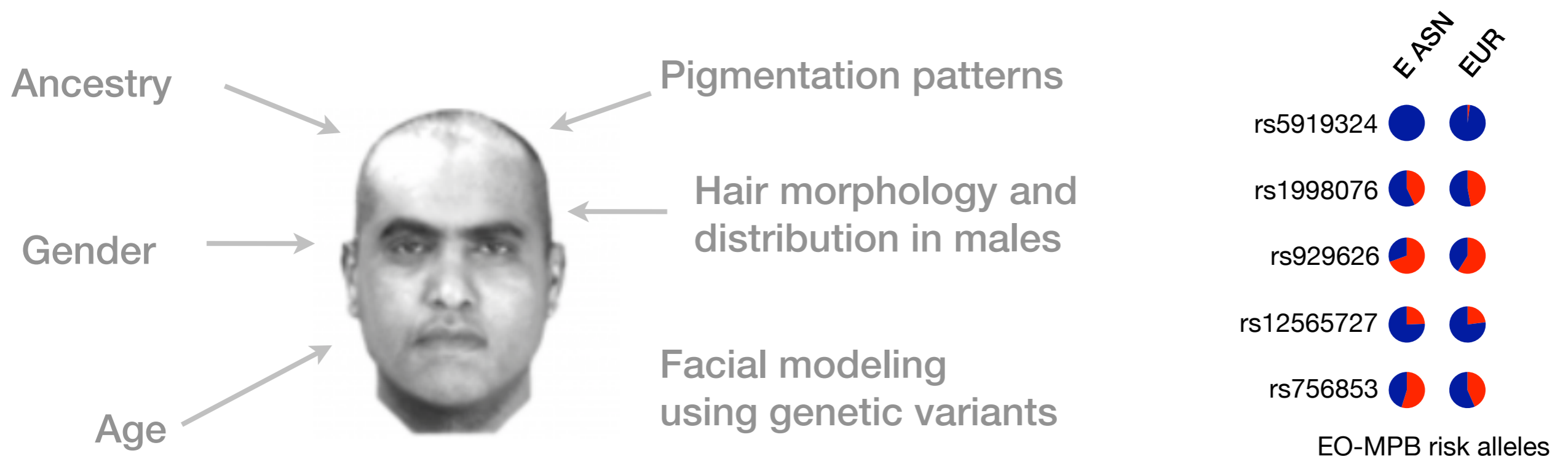
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Thank You



The Professor of anatomy's room, USC