6th DNA polymorphisms in human populations

Musée de l'Homme, Paris, 7–9 December 2016

NEW FINAL PROGRAM AND ABSTRACT BOOK

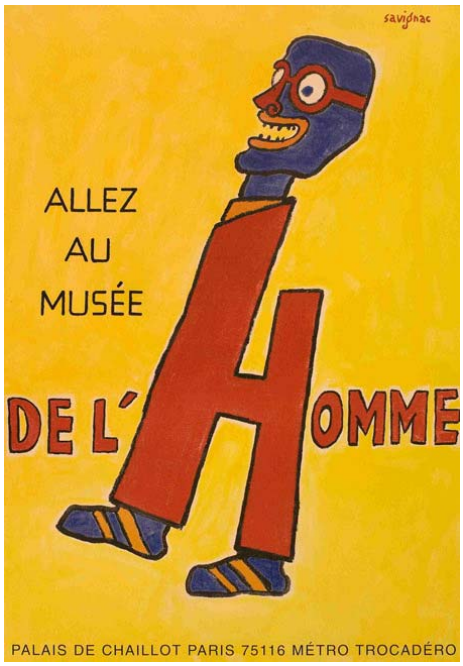
35 TALKS AND 24 POSTERS

VS. 2.3 (DECEMBER 4, 2016) – 56 PAGES

INDEX AT PAGE 3

The pagination of this document is set to be fine when printing recto/verso. Save resources.

http://ecoanthropologie.mnhn.fr/DPHP2016/DPHP2016.htm



6TH INTERNATIONAL CONFERENCE OF THE SERIES
DNA Polymorphisms in Human Populations

Convenor	Franz Manni
Conference Assistant	Nancy Wise
Assistants	Bérénice Alard Nina Marchi
Scientific Committee	Frederic Austerlitz, Celine Bon, Raphaelle Chaix, Pierre Darlu, Jean-Marc Elalouf, Evelyne Heyer, Laure Segurel, Paul Verdu.
Venue	*Musée de l'Homme* 17, place du Trocadéro, 75116 Paris France
Dates	From the 7th to the 9th of December, 2016
Website	http://ecoanthropologie.mnhn.fr/DPHP2016/DPHP2016.htm
Contact	dnaparis2016@gmail.com

INDEX

NB *Pages of abstracts can be found in the detailed schedule (pages 7 – 13)*

REGISTRATION AND COSTS

To register please connect to the webpage of the conference:
http://ecoanthropologie.mnhn.fr/DPHP2016/DPHP2016.htm
and download the REGISTRATION FORM, fill it in and send it back, by Email to:
dnaparis2016@gmail.com

PRICES APPLY AS IT FOLLOWS

Please add an extra-cost of 50 Euros to all payments made later than November 19, 2016

FULL CONFERENCE	*Full access to all plenary sessions and to the workshop you decide to attend.* 200 Euros for students 250 Euros for professionals *This price includes 1 cocktail reception, 2 lunches, coffee breaks and the conference package, but NOT the conference dinner (cost = 60 Euros).*
ONLY PLENARY SESSIONS	*Access to the plenary sessions of the 8th and 9th December but* NOT *to the workshops of the 9th of December* 120 Euros for students 150 Euros for professionals *This price includes 1 cocktail reception, 1 lunch, coffee breaks and the conference package, but NOT the conference dinner (cost = 60 Euros).*
ONLY WORKSHOPS	*Access to one of the three workshops held the 9th of December but* NOT *to the plenary sessions of the 8th and 9th of December.* 100 Euros for students 120 Euros for professionals Workshops: "Measuring Culture" "Genomic Demography" "Ancient DNA" *You have to choose the one you want to attend.* *This price includes 1 proper lunch, coffee breaks and the conference package.*

Free entrance:
The employees and students of the National Museum of Natural History of Paris (MNHN) can freely access the conference by showing their Multipass card *(No cocktail, lunches or conference package offered)*.

ALL PAYMENTS SHOULD BE MADE BE WIRING THE MONEY TO THE BANK ACCOUNT SPECIFIED IN THE REGISTRATION FORM AND BY SCANNING AND SENDING THE PROOF OF PAYMENT TO dnaparis2016@gmail.com
(No Paypal accepted, all wiring costs at your expenses)
French scientists can pay with a *BON DE COMMANDE*

Foreword

Our ambition is to provide a representative overview of what human population geneticists are doing at the moment. There is no constrained topic, but it has to be related to human diversity and, possibly, cross-disciplinary.

Since fifteen years our disciplines have experienced an increasing specialization. This is why we would like our conference to be a bit old-fashioned, like when you could go to a population genetics meeting, learn and understand almost everything. But, there is room for more in-depth discussions in the frame of three workshops:

1. Measuring Culture
Measuring cultural differences and evolution.
(Organized by Pierre Darlu and Franz Manni)

2. Genomic Demography
Inferring demography, gene flow and admixture from genome-wide data
(Organized by Frederic Austerlitz and Paul Verdu)

3. Ancient DNA
Ancient DNA: from archaeological sites to genome analyses
(Organized by Celine Bon and Jean-Marc Elalouf)

While the first (December, 8) and the last day (December, 10) will consist in classical plenary lectures and poster sessions, the workshops will take place the second day (December, 9). You will have to choose the one you would like to attend. The workshops will allow large round-table discussion and the presentations can be less formal.

GENERAL SCHEDULE AT A GLANCE

December, 7 ■	*7:00 pm – 9:00 pm*	Registrations and cocktail reception *(included in the registration fee)*. *Venue*: *Grande Galerie de l'Evolution du* *Muséum National d'Histoire Naturelle* 36, rue Geoffroy Saint-Hilaire 75005 Paris http://www.mnhn.fr/fr/visitez/lieux/grande-galerie-evolution *N.B. You can register also the next day (8th) or directly the 9t if you decide to attend only a workshop.*
December, 8 ■	*9:00 am – 9:30 am*	*Registrations* *Venue*: Musée de l'Homme 17, Place du Trocadéro 75116 Paris – France
	9:30 am – 1:35 pm	PLENARY SESSIONS (+ 1 coffee break)
	1:45 pm – 2:30 pm	*Lunch (included in the registration fee)*
	2:30 pm – 3:30 pm	POSTER SESSION (coffee available)
	3:30 pm – 5:45 pm	PLENARY SESSIONS
	8:15 pm	*Conference dinner at the restaurant* La Coupole NOT included in the registration fee. Cost: 60 Euros, with wine.
December, 9 ■	*9:30 am – 10:00 am*	*Registrations* *Venue*: Musée de l'Homme 17, Place du Trocadéro 75116 Paris – France
	10.00 am – 1:30 pm	WORKSHOPS
	(There is no constraint timing, participants and organizers will agree on a timetable).	*1 Measuring Culture* *2 Genomic Demography* *3 Ancient DNA* *You have to choose the one you want to attend.* *Coffee available from 11:15am to 12:00pm.*
	1:30 pm – 2:30 pm	*Lunch (included in the registration fee)*
	2:45 pm – 3:30 pm	WORKSHOPS (Flash presentations)
	(There is no constraint timing, participants and organizers will agree on a timetable).	*1 Measuring Culture* *2 Genomic Demography* *3 Ancient DNA* *You have to attend the one you started with.* *11:15am to 12:00pm*.
	3:30 pm – 4:15 pm	PLENARY SESSIONS (+ 1 coffee break at 4:45 pm)
	4:15 pm – 4:30 pm	*Award „Loredana Castrí" attributed to the best poster presentation*
	4:30 pm – 5:30 pm	*Conference dispersal and free visit of the* Musée de l'Homme *Sandwhiches available*

DETAILED SCHEDULE: *PLENARY SESSIONS*

December, 7 ■	*6:00 pm – 8:00 pm*	Registrations and cocktail reception *(included in the registration fee)*. *Venue*: *Grande Galerie de l'Evolution du* *Muséum National d'Histoire Naturelle* 36 rue Geoffroy Saint-Hilaire 75005 Paris http://www.mnhn.fr/fr/visitez/lieux/grande-galerie-evolution
December, 8 *Morning* ■	*9:00 am – 9:30 am*	*Registrations* *Venue*: Musée de l'Homme 17, Place du Trocadéro 75116 Paris – France http://www.museedelhomme.fr/
	9:30 am – 9:40 am	Evelyne HEYER and Franz MANNI *Welcome message*
	9:40 am – 9:50 am	Bruno DAVID, *President of the National Museum of Natural History* *Welcome message*
	9:50 am – 10:25 am	*(35' talk)* Kenneth K. KIDD *Page 19* *Using ancestry informative SNPs (AISNPs) to infer ancestry*
	10:25 am – 10:45 am	*(20' talk)* Francisco ÚBEDA DE TORRES *Page 24* *The non-paradoxical paradox of recombination hotspots*
	10:45 am – 11:05 am	*(20' talk)* Etienne PATIN *Page 22* *Reconstructing the genetic and adaptive history of Bantu-speaking populations in Africa and North America*
	11:05 am - 11:35 am	*Coffee Break*
	11:35 am – 11:55 am	*(20' talk)* Guido Alberto GNECCHI RUSCONE *Page 18* *Dissecting the role of demography and natural selection in shaping the genomic background of low- and high-altitude Nepali populations from the Gaurishankar region*
	11:55 am – 12:15 pm	*(20' talk)* Chiara BARBIERI *Page 14* *Reconstructing human diffusion and collapse in the Americas*
	12:15 pm – 12:35 pm	*(20' talk)* César FORTES-LIMA *Page 18* *Reconstruction of African links in African-American populations and new genomic insights into the Atlantic world*
	12:35 pm – 12:55 pm	*(20' talk)* Nina MARCHI *Page 22* *Human dispersal and inbreeding avoidance in inner Asia*
	12:55 pm – 1:15 pm	*(20' talk)* Phillip ENDICOTT *Page 16* *Polynesian origins, a perspective from the Society Islands*
	1:15 pm – 1:35 pm	*(20' talk)* Francesc CALAFELL *Page 15* *A Bronze Age lineage dominates the Y-chromosome landscape in the Iberian Peninsula*
	1:45 pm – 2:30 pm	*Lunch at the Trocadéro Business Centre* *(5 minutes walk -- included in the registration fee)*

DETAILED SCHEDULE: *PLENARY SESSIONS*

December, 8
Afternoon
■

2:30 pm – 3:30 pm — POSTER SESSION (see next pages)→→→

Sandwhiches available

3:30 pm – 3:50 pm — *(20' talk)* Franz MANNI *Page 21*
The Bantu expansion and the peopling of Gabon, a genetic and linguistic overview

3:50 pm – 4:10 pm — *(20' talk)* Pavel FLEGONTOV *Page 17*
Rare variants and autosomal haplotypes reveal the Siberian roots of North American Na-Dene populations

4:10 pm – 4:30 pm — *(20' talk)* Pradiptajati KUSUMA *Page 20*
The sea-nomad Bajo: Origin, dispersal, and adaptation

4:30 pm – 4:50 pm — *(20' talk)* Mohan REDDY BATTINI *Page 23*
Origins of Austro-Asiatic Populations and their Status in the Peopling of India

4:50 pm – 5:10 pm — *(20' talk)* Eveline ALTENA *Page 14*
Genetic history of the Dutch population

5:10 pm – 5:45 pm — *(35' talk)* Laurent EXCOFFIER *Page 16*
Genomic insights into the settlement of Australia by modern humans

8:15 pm — *Conference dinner NOT included in the registration fee; Cost: 60 Euros, with wine.*

Restaurant :
La Coupole,
102 Bd du Montparnasse,
75014 Paris
http://www.lacoupole-paris.com

DETAILED SCHEDULE: *POSTER SESSION*

December, 8
■
2:30 pm – 3:30 pm

1 Frédéric AUSTERLITZ *Page 25*
Comparing population history inferred from genetic and linguistic data in Central Asia

2 Chiara BARBIERI *Page 26*
The co-evolution of languages and genes tracking down matches and mismatches

3 Jeremy GARDENT *Page 30*
A simple overview of music throughout Gabon

4 Joaquim PÉREZ-LOSADA *Page 36*
Support for a linguistic serial founder effect originating in Africa

5 Francesco MONTINARO *Page 35*
Complex ancient genetic structure and cultural transitions in Southern-Africa populations

6 André FLORES-BELLO *Page 30*
Genetic analysis and evolution of Rh blood group system in Basques

7 Florence PETIT *Page 36*
Confrontation of Red Cell Blood Groups distribution to environmental data

8 João C. TEIXEIRA *Page 38*
Comparative genomics of innate immunity in human and non-human primates

9 Antony HERZIG *Page 31*
Human Population Isolates: Challenges in Phasing and Imputation

10 Samantha BRUNEL *Page 28*
Genetic diversity during the Neolithic: what about France?

11 Neus ISERN *Page 32*
Ancient DNA and the spread of the Neolithic in Europe

12 Aurore MONNEREAU *Page 34*
Palaeogenetic analysis of Bronze Age/Iron Age transition in Southern Central Asia

13 Patrizia SERVENTI *Page 38*
Iron Age Italic population genetics: the Piceni from Novilara (8th-7th century BC)

14 Bérénice ALARD *Page 25*
Dynamics of clans in human unilineal populations: a genetic approach

15 Goki LY *Page 32*
Reassessing the influence of social organization on genomic diversity: the case of Austroasiatic populations of South-East Asia

16 Valentina DOMINICI *Page 29*
Overcoming the Dichotomy: New Insights into the Genomic Diversity of Open and Isolated European Populations

17 Athanasios KOUSATHANAS *Page 33*
Demographic history and deleterious variation in Hunter-Gatherer and Farmer African Populations

18 Vladimir BAJIĆ *Page 26*
Genetic history of southern African Khoisan populations reveals time dependent intensity of sex-biased gene flow

19 Gwenna BRETON *Page 27*
Genetic diversity and demographic history of Sub-Saharan human populations based on genome-wide markers

20 Nicolas BRUCATO *Page 30*
Genetic legacy of the Indian Ocean trading network

21 Enrico MACHOLDT *Page 33*
Exploring the population history of Vietnam

22 Teresa NUTILE *Page 35*
Analysis of exome-sequencing variants in Cilento isolates

23 Stefania SARNO *Page 37*
Genomic ancestry of southern Italy, insights into a complex history of admixture

24 Levon YEPISKOPOSYAN and Anahit HOVHANNISYAN *Page 39*
Genetic mapping of historical Armenia

DETAILED SCHEDULE: *WORKSHOPS*

December, 9 ■	09:30 am – 10:00 am	*Registrations*
	10:00 am – 1:30 pm	WORKSHOPS
	1:30 pm – 2:30 pm	*Lunch at the Trocadéro Business Centre* *(5 minutes walk -- included in the registration fee)*
	2:45 pm – 3:30 pm	WORKSHOPS (flash presentations)
	3:30 pm – 4:30 pm	PLENARY SESSION
	3:30 pm – 4:15 pm	*(45' talk)* Mattias JAKOBSSON *Page 22* *Genes mirror migrations and cultures in prehistoric Europe*
	4:15 pm – 4:30 pm	*Award „Loredana Castrí"* *Attributed to the best poster presentation*
	4:30 pm – 5:30 pm	*Conference dispersal and free visit of the* Musée de l'Homme *(Coffee and sandwiches available)*

WORKSHOP 1 *Measuring Culture* *(see next pages)*

Measuring cultural differences and evolution

(Organized by Pierre Darlu and Franz Manni)

Since about twenty years, computational methods and approaches have been applied to cultural differences. The purpose was to measure and quantify traits that, previously, were seen as computationally intractable by the specialists of the disciplines involved (cultural anthropology, linguistics, musicology, etc.).
Artefacts, languages and music are now coded as vectors, multistate matrices, distance matrices that allow an unprecedented degree of historical and geographical inference. But…
What are we inferring? From black boxes to new hypotheses, is the signal robust enough?
The purpose of this workshop is to attract a panel of specialists, as diverse as possible, in order to address the weaknesses and strengths of computational methods that go beyond the understanding of cultural anthropologists, linguists, musicologists, etc.

WORKSHOP 2 *Genomic Demography* *(see next pages)*

Inferring demography, gene flow and admixture from genome-wide data

(Organized by Frederic Austerlitz and Paul Verdu)

Population genetics has enabled a reconstruction of the past demographic history of human populations. The increasing availability of genome-wide datasets at regional or global scales allows, in principle, to investigate in a much greater detail the demographic history of populations. Classical population genetics has to be adapted to genome-wide data and the development of new analytical tools has become crucial.
In this workshop, we aim at discussing the strengths and limitations of existing and forthcoming approaches to real genomic data, with a special focus on demographic changes over the time, complex gene-flow and admixture processes.

WORKSHOP 3 *Ancient DNA* *(see next pages)*

From archaeological sites to genome analysis

(Organized by Celine Bon and Jean-Marc Elalouf)

Ancient genomes provide a direct insight into the evolution of populations and species. In this workshop, we will review the different sources of genetic material that are available in archaeological sites, concerning vegetal and animal genomes, humans being included under the latter rubric.
During the workshop, classical aspects of ancient-DNA studies will be discussed, from the development of on-site genetic analyses to specific laboratory protocols and the estimation of population genetics statistics using ancient, low-coverage genomes. A variety of data sources will be reviewed, including ancient microbiome data. Thus, this workshop will cover up-to-date protocols in palaeogenetic analyses and their contributions to archaeological or historical investigations.

WORKSHOP 1 *Measuring Culture**

Only the order is provided, the exact schedule depends on the length of the scientific discussion after each presentation.

December, 9

■

WORKSHOP *Measuring Culture*
Measuring cultural differences and evolution.
Organized by Pierre Darlu and Franz Manni

10:00 am – 1:30 pm *Talks of the morning in the following order:**

Pierre DARLU and Franz MANNI
General overview of the aims and themes of the workshop

Franz MANNI *Page 44*
The descendants of the Middle-Ages Spanish population do not speak Spanish and live by the coast

Leonardo ARIAS *Page 41*
Flowing genes and languages, the role of rivers in structuring genetic variation in native American populations from Nort-West Amazonia

Kristina TAMBETS *Page 44*
The genetic structure of the Uralic-speaking populations - Do genetic and linguistic data tell the same story?

Daniel CORACH *Page 42*
Genetic Relationships of Southern South-American Tribal Groups Inhabiting Argentinean Territory Assessed by Uni and Biparentally Transmitted Polymorphic Markers Correlates with Linguistic Affinities.

Eugenio BORTOLINI *Page 41*
Isolation by distance, demic diffusion, and cultural transmission in the genomic era: A case study based on folktale distribution over Eurasia and Africa

Valentin THOUZEAU *Page 45*
Reconstructing demographic and cultural history of human populations from genetic and linguistic polymorphism data

Franz MANNI *Page 43*
The cultural package? Comparing the diversity of music and languages in Gabon (Africa)

Pierre DARLU *Page 43*
Cladistics, Networks or Distance methods? Controversial representations of the geographical distribution of linguistic traits

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS
(upon organizers' invitation)

(10' talk) Jeremy GARDENT *Page 30*
A simple overview of music throughout Gabon

(10' talk) Goki LY *Page 32*
Reassessing the influence of social organization on genomic diversity: the case of Austroasiatic populations of South-East Asia

(10' talk) Joaquim PEREZ-LOSADA *Page 36*
Support for a linguistic serial founder effect originating in Africa

(10' talk) Antony HERZIG *Page 31*
Human Population Isolates: Challenges in Phasing and Imputation

3:30 pm – 4:30 pm PLENARY SESSION (see page 10)

WORKSHOP 2 *Genomic Demography*

Only the order is provided, the exact schedule depends on the length of the scientific discussion after each presentation.

December, 9

■

WORKSHOP *Genomic Demography*
Inferring demography, gene flow and admixture
from genome-wide data
Organized by Frederic Austerlitz and Paul Verdu

10:00 am – 1:30 pm *Talks of the morning in the following order:**

Frederic AUSTERLITZ and Paul VERDU
General overview of the aims and themes of the workshop
Simone Andrea BIAGINI *Page 47*
Human population genomics of the western Mediterranean
Ashot MARGARYAN *Page 48*
7800 years of mitochondrial genetic continuity in Armenia
Gerard SERRA-VIDAL *Page 50*
Study of the human north African genome landscape through the analysis of complete genomes
Lara R. ARAUNA *Page 47*
Recent historical migrations have shaped the gene pool of Arabs and Berbers in North Africa
Sandra OLIVEIRA *Page 49*
The maternal history of south-west Angola
Alex MAS-SANDOVAL *Page 49*
Native American pre-colonization genetic history through admixed Brazilians

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS
(upon organizers' invitation)

(10' talk) Nicolas BRUCATO *Page 28*
Genetic legacy of the Indian Ocean trading network
(10' talk) Enrico MACHOLDT *Page 33*
Exploring the population history of Vietnam
(10' talk) Stefania SARNO *Page 37*
Genomic ancestry of southern Italy, insights into a complex history of admixture

3:30 pm – 4:30 pm PLENARY SESSION (see page 10)

WORKSHOP 3 *Ancient DNA*

Only the order is provided, the exact schedule depends on the length of the scientific discussion after each presentation.

December, 9

■

WORKSHOP *Ancient DNA*
Ancient DNA: from archaeological sites to genome analyses
Organized by Celine Bon and Jean-Marc Elalouf

10:00 am – 1:30 pm *Talks of the morning in the following order:**

Celine BON
General overview of the aims and themes of the workshop
Jean-Marc ELALOUF *Page 53*
Validation of a portable system for ancient DNA analysis on archeological sites
Lasse VINNER *Page 55*
Comparison of target enrichment methods for ancient human samples
Andrew E. BENNET *Page 53*
Ancient DNA bares its teeth: What ancient dental calculus can tell us about past populations
E.A.MATISOO-SMITH *Page 54*
Mitochondrial Genomes of Ancient Phoenicians
PAGANI Luca *Page 55*
Genome-wide allele frequency estimates from population level ultra-low coverage aDNA samples.
Etienne GUICHARD *Page 54*
Impact of non-LTR retrotransposons in the differentiation and evolution of the Homo genus

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS
(upon organizers' invitation)

(5' talk) Samantha BRUNEL *Page 28*
Genetic diversity during the Neolithic: what about France?
(5' talk) Neus ISERN *Page 32*
Ancient DNA and the spread of the Neolithic in Europe
(5' talk) Aurore MONNEREAU *Page 34*
Palaeogenetic analysis of Bronze Age/Iron Age transition in Southern
Central Asia
(5' talk) Patrizia SERVENTI *Page 38*
Iron Age Italic population genetics: the Piceni from Novilara (8th-7th century BC)

3:30 pm – 4:30 pm PLENARY SESSION (see page 10)

.

PLENARY SESSION - TALKS
Abstracts in alphabetical order

TALK PLENARY SESSIONS

GENETIC HISTORY OF THE DUTCH POPULATION

ALTENA Eveline1, Risha Smeding1, Thirsa Kraaijenbrink1, Kristiaan van der Gaag1,2, Eileen Vaske1, Paul Reusink1, Anna Friedler1, Yoan Diekmann3, Mark G. Thomas3, Peter de Knijff1.

1 Forensic Laboratory for DNA Research, Dept. of Human Genetics, Leiden University Medical Center, Leiden, the Netherlands.
2 Current address: Dept. biologische sporen, Netherlands Forensic Institute, Den Haag, the Netherlands.
3 Dept. of Genetics, Evolution and Environment, University College London, London, UK.

Although the Netherlands is a small country, previous studies indicated complex geographic patterns of genetic variation in the modern Dutch population. Based on archaeological and historical evidence, and the dynamic Dutch landscape, it is most likely that patterns of genetic variation in the modern population were primarily shaped since medieval time. A clear understanding of when and how the modern patterns emerged, however, requires genetic data from historic Dutch populations.
We analyzed several Dutch archaeological population samples from different locations and from a time range between the early medieval period and the post-medieval period. This collection of nearly 800 skeletons was examined for autosomal, Y-chromosomal and mitochondrial variation. The resulting dataset, together with comparable data from more than 2000 modern Dutch individuals, allows us to review our historical genetic past in detail, both in time and space. Finally, we test for population continuity by statistical modeling of frequency changes in mitochondrial and Y-chromosomal haplogroups.

TALK PLENARY SESSIONS

RECONSTRUCTING HUMAN DIFFUSION AND COLLAPSE IN THE AMERICAS

BARBIERI Chiara1, Ghirotto Silvia2, Arias Leonardo3, Sandoval Jose4, Sevini Federica5, De Fanti Sara6, Franceschi Zelda7, Franceschi Claudio5, Luiselli Donata6, Stoneking Mark3, Fujita Ricardo4, Heggarty Paul1, Powell Adam1

1 Department of Linguistic and Cultural Evolution, Max Planck Institute for the Science of Human History, Jena, Germany
2 Department of Life Sciences and Biotechnologies, University of Ferrara,
Ferrara, Italy
3 Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany
4 Centro de Genetica y Biologia Molecular, Universidad San Martin de Porres (USMP), Lima, Peru
5 C.I.G. Interdepartmental Centre L. Galvani for Integrated Studies on Bioinformatics, Biophysics and Biocomplexity, University of Bologna, Bologna, Italy
6 Department of Biological, Geological and Environmental Sciences, University of Bologna, Bologna, Italy
7 Dipartimento di Storia Culture Civiltà, University of Bologna, Bologna, Italy

The Americas, with their relatively recent history of population diffusion and collapse after European contact, represent an ideal case-study for testing the genetic footprints left by demographic changes. Native American mtDNA genomes have been successfully employed to reconstruct the timing and magnitude of the initial population expansion into the continent. However, previous studies were based on collections of individual lineages and were missing actual population samples. This limited perspective lacked the resolution to explore regional population spread and diversification. Furthermore, the recent collapse was not adequately tested in populations with different prehistories and in different regions.

In this study we focus on Meso- and South-America to explore 1 the diversification of populations who crossed the Isthmus of Panama into South America and 2 the traces of a recent demographic collapse in various sets of population mitogenomes. Our dataset comprises 320 full mtDNA genomes from 13 populations from Mesoamerica and from different ecogeographic regions of South America: the Andes, Amazonia and the Gran Chaco. A Bayesian approach is employed to reconstruct population demographies and to test different scenarios of expansion and collapse.

Our results suggest different prehistories for the populations studied, irrespective of their geographic location. Similarities between Mesoamerican and Andean populations indicate a possible connection on the Pacific coast, which is tested with spatially-informed simulations. Coalescent simulations support the recent collapse in most of our populations, helping us to understand the impact of recent events on population mitogenomes.
In conclusion, populations in the Americas experienced different demographic trajectories and strong diversification, which possibly drove the high cultural and linguistic diversity reported today. Including mtDNA genomes from modern populations along with ancient samples promises to shed light into the past of the Americas and into the demographic dynamics behind population divergence.

TALK PLENARY SESSIONS

A BRONZE AGE LINEAGE DOMINATES THE Y-CHROMOSOME LANDSCAPE IN THE IBERIAN PENINSULA

CALAFELL F1, P. Villaescusa2, N. Solé-Morata1, A. Carracedo3, K. Rouault4, C. Férec4, O. Hardiman5, A. Santurtun6, S. Jiménez7, M. F. Pinheiro8, B. M. Jarreta9, M. M. De Pancorbo2

1 Institute of Evolutionary Biology (CSIC-Universitat Pompeu Fabra), CEXS-UPF-PRBB, Barcelona, Catalonia, Spain
2 BIOMICs Research Group, Lascaray Research Center, University of Basque Country UPV/EHU, Vitoria-Gasteiz, Spain
3 Forensic Genetics Unit, Institute of Legal Medicine, University of Santiago de Compostela, Spain; Galician Foundation of Genomic Medicine (SERGAS), CIBERER (University of Santiago de Compostela), Santiago de Compostela, Spain.
4 Inserm UMR1078, Génétique, Génomique fonctionnelle et Biotechnologies, Brest 15 Cedex 2, France.
5 National Neuroscience Centre, Beaumont Hospital, Dublin, Ireland.
6 Unit of Legal Medicine, Department of Physiology and Pharmacology, University of Cantabria, Santander, Spain
7 Institute of Legal Medicine of Alicante, Spain.
8 National Institute of Legal Medicine and Forensic Sciences, Portugal.
9 Laboratory of Genetics and Genetic Identification, University of Zaragoza, Spain.

The genetic landscape of the Iberian Peninsula is dominated (as in the rest of Western Europe) by haplogroup R1b, which comprises two thirds of the Y chromosomes; the rest is divided roughly equally between E-M35, G, I, and J. Within R1b, R1b-S116 (also known as P312 dominates, with ~60% in Spain;it further trifurcates into three major branches having distinct geographical distributions: M529 (L21 radiating from the British Isles, U152 in France, Switzerland and N. Italy, and DF27 in the Iberian Peninsula. DF27 is poorly known, and we have sought to characterize its distribution and diversity, with the aim of reconstructing its history. We have typed DF27 and six of its derived SNPs, as well as 16 Y-STRs in 2,993 males from 32 populations located in Spain, Portugal, France and Ireland; SNP allele frequencies were also gathered from the reference populations in the 1000 Genomes Project. We confirmed that DF27 is the most frequent haplogroup in Iberia, with an average frequency ~45%, while it dropped to <15% right across the Pyrenees. Within Iberia, it ranged from 40% in most populations to ~75% in Basques. Elsewhere, it showed high frequencies in Colombia and Puerto Rico, which implies it can be used to trace Iberian male migrations into the Americas.

However, our most striking result is how young DF27 is. We estimated from STR variation that DF27 originated 4,000±150 years ago (ya); it took it just 120 generations to grow to ~12 million carriers in Iberia and ~75 million in Central and South America (assuming just 1/3 paternal Iberian ancestry). This places the origin of DF27 in the early Bronze Age, and at least 2,000 years after the arrival of the Neolithic, which was supposed to be the last major event that shaped the European genetic landscape. The DF27 expansion may be part of a global trend, in which bursts of male lineages have been observed at different periods, and in different geographical regions (Poznik et al. 2016.

Poznik et al. 2016. Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nat Genet. 486:593-9

TALK PLENARY SESSIONS

POLYNESIAN ORIGINS, A PERSPECTIVE FROM THE SOCIETY ISLANDS

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The origin of the Polynesian speaking peoples is frequently associated with the pioneering settlement of the Pacific east and south of the Solomons, otherwise known as Remote Oceania. This commenced 3,200 years ago, reaching Tonga and Samoa by 2,900 BP, and is clearly marked by the Lapita cultural complex in the archaeological record of the region. Lapita is widely considered to be the outcome of cultural and genetic admixture of Austronesian speaking peoples from Southeast Asia with coastal groups in the Bismarck archipelago and New Guinea, prior to settling Remote Oceania.
According to the dominant cultural-historical paradigm, Polynesian culture, language and people, evolved out of Lapita in the Tonga and Samoa region (western Polynesia) within a few hundred years of first settlement. However, most of the hundreds of islands and atolls contained within the Polynesian triangle--a vast area of the Pacific defined by Hawai'i, Easter Island and New Zealand--were settled during a second wave of dispersal, by peoples ancestral to today's eastern Polynesians.
The exploration and settlement of eastern Polynesia was previously considered to occur early in the Polynesian archaeological record, but recent improvements in radiometric dating hygiene indicate that it did not take place until 2000 years after the first arrival of people in Tonga. The first detectable landfall during this last major period of human colonisation is currently dated to 950 years BP on the leeward Society islands, located at the geographical and cultural centre of the Polynesian triangle, after which the remainder of eastern Polynesia was settled in as little as 150-200 years. This significant temporal discontinuity with Lapita demonstrates the need to incorporate a wider range of hypotheses into model testing of Polynesian origins in general, and to reconsider the origins of eastern Polynesians in particular.
Due to the importance of the Societies in the settlement history of this vast region of the Pacific, we produced high-resolution, genome-wide, genetic data from three leeward islands, including Borabora and Rai'atea, historically the two most important cultural centres. Using the enhanced ability provided by haplotype based approaches to capture subtle population structure and detect multiple admixture events in the recent past, we identified multiple donor populations in both Near Oceania and Southeast Asia, indicating greater complexity than existing models account for. We also detect two significant phases of admixture during the past 2000 years; the first involves people of European ancestry, dating to the period bracketed by the three voyages of Captain Cook during the late 18th century, whilst the most recent date for the second is significantly after the Lapita settlement of western Polynesia but before the first colonisation of eastern Polynesia. Genomic sequencing of key mitochondrial and Y-chromosomal DNA lineages provides important complementary information on the prehistory of Polynesians as a whole.

TALK PLENARY SESSIONS

GENOMIC INSIGHTS INTO THE SETTLEMENT OF AUSTRALIA BY MODERN HUMANS

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The exact settlement history of Sahul (Australia, New Guinea and Tasmania) remains largely unknown. Indeed, there is still an intense discussion on whether the ancestors of Australo-Papuans left Africa earlier than Eurasians, and the exact divergence time and levels of gene flow between Australian Aborigines and Papuans are uncharacterized. A detailed analysis of high-coverage genomes of 83 Aboriginal Australians and 25 Papuans from the New Guinea Highlands reveals that Sahul was colonized by the ancestors of Australo-Papuan more than 50 kya and that these two groups diverged 25-40 thousand years ago (kya), with very little subsequent gene flow. The fact that this divergence occurred long before the separation of Australia and Papua (only some 10Kya), suggests the existence of an ancient population structure in Sahul. Moreover, all studied Pama-Nyungan speaking Aboriginal Australians seem to descend from a single founding population that differentiated as early as ~10-32 kya. At odds with a previous genomic study, we have no evidence for two exits out of Africa, but we infer an early divergence of Australian and Papuan ancestors from Eurasians, about 57 kya. Interestingly, the evidence for a single exit out of Africa is only found if we explicitly take into account archaic admixture in Australo-Papuans.

TALK PLENARY SESSIONS

RARE VARIANTS AND AUTOSOMAL HAPLOTYPES REVEAL THE SIBERIAN ROOTS OF NORTH AMERICAN NA-DENE POPULATIONS

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Pre-history of Native Americans of the Na-Dene language family remains controversial. Continuity of Na-Dene and Paleo-Eskimos, associated with the Saqqaq and Dorset archaeological cultures that appeared in America after ~4,800 YBP, was proposed under the three-wave model of America's settlement. However, recent genetic studies produced conflicting results (Reich. et al. 2012, Raghavan et al. 2014, 2015). We performed reconstruction and dating of Na-Dene population history, using public high-coverage sequencing data (1,206 individuals from 94 populations) and a recently developed coalescent method relying on rare alleles (Rarecoal). Since the initial report in Schiffels et al. (2016), we have improved the software and added pulse-like admixture events as a new feature. We also applied model-free approaches for analysis of rare allele and autosomal haplotype sharing, investigated with the ChromoPainter and fineSTRUCTURE tools on two independent SNP array datasets of 634 and 1,283 individuals. All methods detected Central and West Siberian ancestry exclusively in a fraction of Na-Dene individuals, but not in other Native Americans. Using Rarecoal, we have modelled a gene flow from a Siberian population that split 6,500-7,000 years ago into the ancestors of Na-Dene. Our results are consistent with a Paleo-Eskimo admixture into the First American ancestors of Na-Dene, and a much later and less extensive bidirectional admixture was detected between Na-Dene and Neo-Eskimos. The recent gene flow from Siberia to Na-Dene is in agreement with the Dene-Yeniseian language macrofamily proposal and with succession of archaeological cultures in Siberia.

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Raghavan, M. *et al*. Genomic evidence for the Pleistocene and recent population history of Native Americans. *Science* 349, 1–20 (2015).
Reich, D. *et al*. Reconstructing Native American population history. *Nature* 488, 370–374 (2012).
Schiffels, S. *et al*. Iron Age and Anglo-Saxon genomes from East England reveal British migration history. *Nat. Commun*. 7, 10408 (2016).

TALK PLENARY SESSIONS

RECONSTRUCTION OF AFRICAN LINKS IN AFRICAN-AMERICAN POPULATIONS AND NEW GENOMIC INSIGHTS INTO THE ATLANTIC WORLD

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The transatlantic slave trade enacted as the most traumatic long-distance migration in human history. It changed the genetic composition of the entire New World, for instance, in French Guiana and Suriname most of enslaved Africans escaped and established new independent African settlements since then, called Noir Marron communities. Currently, descendants of enslaved Africans through the Americas present uncertain African source population, complex patterns of admixture with non-African groups, and different colonial past during the slave trade and after its abolition. To uncover long-standing questions on their demographic history, we analyzed two sets of uniparental markers and genome-wide data (4.5 million variants) for eight African-American populations (four Noir Marron communities, African-Brazilian, African-Colombian, African-Barbadian and African-American in the United States), and their putative Sub-Saharan African source population. Moreover, we obtain a fine-scale genetic perspective of the European and Native American gene-flow across each chromosome. Both, the Afro-Brazilian and the African-American in the United States present a strong influence of a European pulse of migration. Besides the Afro-Colombian population presents the highest Native American proportion and different admixture timing in good agreement with history records. MtDNA genomes and sexual chromosomes unravel sexual asymmetric admixture patterns in admixed African-American populations, consistent with an excess of European male contribution and elevate African female ancestry. In the Noir Marron communities, all genetic systems revealed strong African ancestry (above 98%). Both, global and local ancestry inferences highlighted remarkably West-African genome-wide ancestry in those communities, and haplotype sharing linked to the populations residing today in the historical Gold Coast and Bight of Benin regions. This study provides new light into African-American legacy of their African founder groups, and presents a historical and genomic framework to reconstruct admixture migration events.

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TALK PLENARY SESSIONS

DISSECTING THE ROLE OF DEMOGRAPHY AND NATURAL SELECTION IN SHAPING THE GENOMIC BACKGROUND OF LOW- AND HIGH-ALTITUDE NEPALI POPULATIONS FROM THE GAURISHANKAR REGION

GNECCHI RUSCONE Guido Alberto 1, JEONG Choongwon 2, DE FANTI Sara 1, TRANCUCCI Michela 1, GENTILINI Davide 3, DI BLASIO Anna Maria 3, CHILDS Geoff 4, CRAIG Sienna R. 5, BASNYAT Buddha 6, SHERPA Mingma G. 7, SHERPA Phurba 7, MARINELLI Giorgio 8, NATALI Luca 8-9, PELUZZI Davide 8, BEALL Cynthia 10, DI RIENZO Anna 2, PETTENER Davide 1, LUISELLI Donata 1, SAZZINI Marco 1

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Adaptation to high-altitude has been extensively studied in Himalayan populations by searching for signatures of natural selection on the Tibetan and, to a lesser extent, Sherpa genomes. Recent studies have also pinpointed how demographic processes (e.g. archaic introgression and/or population admixture) might have played a role in the spread of genetic variants favourable at high-altitude. Nepali populations from the Gaurishankar mountain range represent an intriguing case study to test such interplay between demography and natural selection in shaping the distribution of these adaptive traits. In fact, this region hosts populations belonging to three main ethnic groups distributed along a wide altitudinal range (900-4,900 m). People speaking Indo-Aryan languages are culturally related to Indian populations and live at low altitudes. Groups speaking Tibeto-Burman languages (i.e. Tamangs and Sherpas) are instead supposed to descend from Tibetan populations and to have moved to Nepal in historical times. While Tamangs have largely spread across Gaurishankar valleys up to medium altitudes, local Sherpa communities mainly settled in few high-altitude villages in the Rolwaling valley.
Patterns of population structure and genomic relationships of these groups with 1,152 additional Sherpa, Tibetan, South Asian and East Asian subjects belonging to 72 populations were inferred from genome-wide data generated for more than 700,000 SNPs on 75 individuals collected during three field expeditions.
Evidence of extensive admixture was found in low-altitude Indo-Aryan speaking groups, with East-Asian ancestry components presumably introduced by Tibeto-Burman migrants. These latter groups have instead experienced limited (or null in the case of Sherpas) gene flow from local Nepali populations, with Tamangs showing closer affinity to Tibeto-Burman people settled in Northeastern India than to high-altitude Rolwaling Sherpas or Tibetans. This picture suggests a more complex distribution of East Asian ancestry components in Nepali populations with respect to what previously thought.
Searching for shared or unique selective events underlying high-altitude adaptation in the Gaurishankar Tibeto-Burman groups was finally performed to further disentangle the differential role played by demography and natural selection in shaping the genomic background of these Himalayan populations.

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TALK PLENARY SESSIONS

USING ANCESTRY INFORMATIVE SNPS (AISNPS) TO INFER ANCESTRY

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Ancestry informative SNPs (AISNPs) are being identified by many groups and several different panels have been published. But how to interpret the results for an individual is not always clear. Several principles need to be considered. First, inference can only be as good as the set of reference populations. Second, allele frequency variation must exist among the populations being considered. Finally, because most inference is based on the quantitative nature of allele frequencies, closely related populations will be difficult to discriminate among. Current AISNP panels differ greatly in the reference populations but the intersection of SNPs in most published panels only allows an individual to be assigned to one of five "continental" groups with little uncertainty. In general, the panels with large numbers of reference populations yield inferences with considerable uncertainty. We are using likelihood ratios to evaluate inference of an individual's possible population of origin. Likelihood ratios less than one or two orders of magnitude among the more likely populations of origin do not allow meaningful distinctions. A problem has been that the different panels have very few SNPs and reference populations in common; comparisons become problematic. We have been enhancing the number of reference populations for our panel of 55 AISNPs with data on 137 different population samples; the panel allows 10

regional clusters of populations to be distinguished and some finer distinctions within those clusters. We have also developed a panel that is the union of three different published panels and contains 192 AISNPs with data on 80 populations. These and other panels of AISNPs are freely available for use in FROG-kb <frog.med.yale.edu>.

TALK PLENARY SESSIONS

THE SEA-NOMAD BAJO: ORIGIN, DISPERSAL, AND ADAPTATION.

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The Indonesian archipelago has been a home for sea-farer experts and sea nomads since the Austronesian expansion era, as late as 4,000 years ago. Among them, the sea-nomad Bajo occupy widely the coastal areas in a multitude of different islands until nowadays. The Bajo are known to be a mobile boat-dwelling people to do trading in distant places, and diving in the deep sea to fish to make living. Although many Bajo communities live in far-distanced locations, they share a common culture, notably marked by a unique language: The Sama-Bajaw, a language that belongs to the Malayo-Polynesian branch of Austronesian language family. Their relative cultural homogeneity tends to balance their constant interactions with diverse ethnic groups with which they live closely geographically (e.g. the Dayak and Banjar in Borneo, the Bugis in Sulawesi, etc.). This appears to be often assumed as inter-cultural marriages and integration of non-Bajo individuals into the community ("maritime creolization"), leading to a presumption that, despite a common cultural identity, the Bajo genetic diversity is heterogeneous. In addition to the absence of any archaeological and/or written history of the Bajo, this apparent complexity is the reflection of the absence of consensus on the geographical origin, as well as the scenario of the Bajo dispersal. To provide insights on these questions, here we present the autosomal genome-wide SNPs study on three Bajo communities (n=73, living in coastal area of 1 Kotabaru island, Southeast Borneo, 2 Derawan archipelago, Northeast Borneo, and 3 Kendari, Southeast Sulawesi, separated with minimum straight line distance of 690 km. We compared the data to an exhaustive dataset from published data and our newly generated data from ethnic populations of interest known to interact with Bajo communities (i.e. the Samihim and Banjar in southeastern Borneo, the Bugis and Mandar in Sulawesi, and North Maluku people), composed in total of 2,890 individuals. Our results show that the genetic diversity of the sea nomad Bajo, similar to linguistic evidence, reveal a nearly identical identity across far-distant communities, revealing a recent gene flow and/or common ancestry. Moreover, our analyses indicate a clear scenario of dispersal within the Indonesian archipelago. Finally, we examined whether their particular way of life, on the frequent sea-dwelling and long-period sea-fishing, have left genetic traces by selective pressure.

TALK PLENARY SESSIONS

GENES MIRROR MIGRATIONS AND CULTURES IN PREHISTORIC EUROPE

JAKOBSSON Mattias

University of Uppsala, Sweden

Genomic information from ancient human remains is beginning to show its full potential for learning about human prehistory. I review the last few years' dramatic finds about European prehistory based on genomic data from humans that lived many millennia ago and relate it to modern-day patterns of genomic variation. The early times,

the upper Paleolithic, appears to contain several population turn-overs followed by more stable populations after the Last Glacial Maximum and during the Mesolithic. Some 11,000 years ago the migrations driving the Neolithic transition start from around Anatolia and reach the north and the west of Europe millennia later. This event is followed by major migrations during the Bronze age. These findings show that culture and lifestyle were major determinants of genomic differentiation and similarity in pre-historic Europe rather than geography as is the case today.

TALK PLENARY SESSIONS

THE BANTU EXPANSION AND THE PEOPLING OF GABON, A GENETIC AND LINGUISTIC OVERVIEW

MANNI Franz (1)(2) and John Nerbonne.(2)(1)

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To compare the genetic and linguistic diversity of the Gabon population and in order to contribute new elements to the scenarios concerning the early Bantu expansion, parental and autosomal markers have been typed for 17 Bantu populations living in Gabon to compute Fst distance matrices. Previously published results concerning the same data suggest that the population is very homogeneous and almost undifferentiated—with the exception of the Fang group that later entered from the North. We have tried to make the genetic dataset more representative of the 17 ethnic groups by filtering out all the DNA donors that were born outside the areas typically inhabited by their respective communities. The new results confirm the lack of differentiation, which is even smaller than previously observed.

Linguistic data have been treated in a similar way, which is by computing pairwise matrices of aggregate Levenshtein distances. Two independently-obtained datasets have been processed (Bastin et al. 1999; ALGAB) accounting for a total of 126 different varieties consisting in lists of words. They lead to similar results, showing that the languages cluster into the same number of groups (B10-30; B40; B50-60-70), with the exception of the group B20, whose unity is questionable. The linguistic distances we computed, based on the comparison of segments, word by word, are fully compatible with the classification of Grollemund et al. (2015) based, instead, on shared vocabulary, where sharing is operationalized as the percentage of words (not) having the same origin. This coding is unnecessary with the Levenshtein method, making it simpler to use and, for the larger amount of information it accounts for, more sensitive.

Besides the Fang group, two possible scenarios can be envisaged: *1)* Gabon has been peopled by genetically-similar Bantu immigrants that have remained homogeneous because the timeframe involved has been too short for them to "(èdifferentiate"; or *2)* separate migrations waves have entered Gabon (possibly at different times and from different directions), but the genetic differences characterizing them have been defaced, perhaps because of the frequent relocation of the ethnic groups typical of a largely mobile population. This second hypothesis is favoured by some linguists that see the languages of Gabon as having had distinct exogenous origins.

This study confirms well delimited linguistic areas, suggesting that the population of Gabon has remained quite local during the time-span necessary for the linguistic difference to arise, contradicting the claims of frequent and long-range internal migrations. Also, the geographic location of the varieties, as well as their classification into groups, is compatible with a differentiation in-situ (without the necessity to have been "imported" from the outside). For example the cluster B40 (located along the Atlantic coast) can be a linguistic group developed by Bantus that moved southward Cameroon, along the sea. By favouring the first scenario, we suggest that Gabon was not "circumnavigated" by the Bantu wave but was peopled more directly from the Benue river valley (Cameroon) where the Bantu originated, meaning that Gabon is one of the first regions occupied by the Bantu populations in their migration.

N.B. This talk is linked to a second presentation (Workshop "Measuring Culture") concerning the comparison of linguistic and musical variability in Gabon, as two aspects of cultural lineage.

ALGAB : Hombert, J.M., 1990, "Atlas linguistique du Gabon", Revue gabonaise des Sciences de l'homme, 2, pp. 37-42
Bastin Y., Coupez A., Mann M. 1999. *Continuity and Divergence in the Bantu Languages: Perspectives from a Lexicostatistic Study*. Tervuren, MRAC.

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TALK PLENARY SESSIONS

HUMAN DISPERSAL AND INBREEDING AVOIDANCE IN INNER ASIA

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Inbreeding is the biological consequence of reproduction between closely related individuals. It results in an increase in the number of homozygous sites within genomes and a decrease in genetic diversity. This can reveal recessive deleterious alleles associated with genetic diseases, decrease fertility and impede the adaptive response of individuals. In humans, two strategies can limit inbreeding. First, individuals can migrate out of their native group and mate inside a new group, which corresponds to geographic exogamy. Second, in the absence of dispersal, individuals can mate within their groups according to specific matrimonial rules.
In Inner Asia, multiple human populations with contrasted social organisations and different levels of geographic exogamy cohabit. This area therefore represents an interesting opportunity to test for the presence of inbreeding avoidance strategies. In this study, we collected both ethnological and genomic data for 369 men and 177 women in 18 populations from Inner Asia (Uzbekistan, Tajikistan, Kyrgyzstan, Siberia and Mongolia). This allowed us to detect the presence of geographical exogamy for each couple and to estimate the genetic inbreeding of each individual.
First, based on genetic estimates, all populations are less inbred than under random mating, suggesting they all have some strategies to avoid inbreeding. Second, we found that the proportion of exogamous couples was highly variable between populations, from 0% to 72%. Furthermore, we found that the endogamous populations are less inbred than the exogamous ones. Moreover, mostly or entirely endogamous populations are organized under a cognatic society while mainly exogamous populations are patrilineal. Social organization (patrilineal or cognatic), correlated to differences in dispersal behaviours, seems to lead to different patterns of genetic inbreeding.

TALK PLENARY SESSIONS

RECONSTRUCTING THE GENETIC AND ADAPTIVE HISTORY OF BANTU-SPEAKING POPULATIONS IN AFRICA AND NORTH AMERICA

PATIN Etienne1,2,3, Marie Lopez1,2,3, Rebecca Grollemund4, Guillaume Laval1,2,3, Christine Harmant1,2,3, Hélène Quach1,2,3, Laure Lémée5, Béatrice Régnault5, Nathaniel J. Dominy6, George H. Perry7, Luis B. Barreiro8, Alain Froment9, Paul Verdu10, Evelyne Heyer10, Jean-Michel Dugoujon11, Luis Pereira12,13,14, Lolke Van der Veen15, Patrick Mouguiama-Daouda15, Jean-Marie Hombert15, Lluís Quintana-Murci1,2,3

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The Bantu language family includes ~310 million speakers in Africa, yet the genetic and adaptive history of Bantu-speaking populations remains largely unexplored. We genome-wide genotyped 1,318 individuals from 35 populations of central Africa, covering the homeland of Bantu language expansions.
We demonstrate that Bantu-speaking populations of eastern and southern Africa originate from the south of central Africa, supporting a southward migration of early Bantu speakers across the equatorial rainforest. We show that rapid adaptation of Bantu-speaking populations to newly colonized environments has been facilitated by gene flow from autochthonous populations, particularly at the HLA region.
We estimate that Bantu-speaking populations contributed to 30% of the African ancestry of African Americans, whose genomes present no evidence of positive selection since admixture with European Americans. Our results broaden our knowledge of the complex evolutionary history of sub-Saharan Africa, which will help identifying genetic risk factors for disease in Africans and African Americans.

TALK *PLENARY SESSIONS*

ORIGINS OF AUSTRO-ASIATIC POPULATIONS AND THEIR STATUS IN THE PEOPLING OF INDIA

REDDY BATTINI Mohan

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Indian subcontinent is known for its enormous ethnic, linguistic and cultural heterogeneity, which is the result of historic and prehistoric processes of migration and peopling of India. Therefore, one finds Indian populations to speak large number of languages belonging to four major linguistic families, viz. Austro-Asiatic (AA), Dravidian, Indo-European and Tibeto-Burman. The Austro-Asiatic linguistic family, which is considered to be the oldest of all the families in South Asia, has substantial presence in Southeast Asia. Indian Austro-Asiatics are represented by some 25 Mundari tribes inhabiting Chotanagpur Plateau and its peripheral areas in Bihar, Bengal, Orissa and Maharashtra, 8 sub-groups of Khasi from Meghalaya (Khasi-Khmuic family) and the Mon-Khmer Nicobarese and Shompen from Andaman and Nicobar Islands.

The origin and expansion of the Austro-Asiatic populations has been a contentious issue and so far there has been no formidable archeological or linguistic evidence in favour of any of the hypotheses suggested. We have attempted comprehensive molecular genetic study of almost all the Austro-Asiatic tribes of India (25 tribes), encompassing the entire ethnic, geographic and linguistic heterogeneity among them, to assess the genetic unity, history and migration of these people. I shall examine the different hypotheses, based on both the genetic and non-genetic evidences. Northeast India, the only region which currently forms a land bridge between the Indian subcontinent and Southeast Asia, has been proposed as an important corridor for the initial peopling of East Asia. Given that the Austro-Asiatic linguistic family is considered to be the oldest and spoken by certain tribes in mainland India, Northeast India and entire Southeast Asia, we expect that populations of this family from Northeast India should provide the signatures of genetic link between Indian and Southeast Asian populations. In order to test this hypothesis, we analyzed mtDNA and Y-Chromosome SNP and STR data of the 8 groups of the Austro-Asiatic Khasi from Northeast India and the neighboring Garo and compared with 214 relevant other Austro-Asiatic and non-Austro-Asiatic populations of India and Asia.

Our results suggest strong paternal genetic link (based on Y-Chromosome evidence) not only among the subgroups of Indian AA populations but also with those of Southeast Asia. The results also indicate that the Y-haplogroup, O-M95, had originated in-situ in the Indian AA populations and particularly among the ancestors of present day Mundaris ~65,000 yrs BP and their ancestors carried it further to Southeast Asia via northeast Indian corridor. Subsequently, in the process of expansion, the Mon-Khmer populations of Southeast Asia migrated and colonized Andaman and Nicobar Islands at a much later point of time (~16,000 yrs BP). However, maternal link particularly between Mundaris and Southeast Asian AA populations is not evident, suggesting primarily male mediated migrations of AA populations from India to Southeast Asia. The results further suggest that the Austro-Asiatic Khasi tribes of Northeast India represent a genetic continuity between the populations of South and

Southeast Asia, thereby advocating that northeast India could have been a major corridor for the movement of populations from India to East/Southeast Asia. Our results are consistent with the linguistic evidence which suggest that the linguistic ancestors of the AA have originated in India and then migrated to Southeast Asia.

TALK PLENARY SESSIONS

THE NON-PARADOXICAL PARADOX OF RECOMBINATION HOTSPOTS

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Recombination hotspots are small chromosomal regions, where meiotic crossover events happen with high frequency. Due to the nature of the recombination process alleles that prevent recombination are over-transmitted rendering recombination hotspots transient and overall recombination difficult to explain. How is it possible that recombination hotspots (and recombination) persists over evolutionary time when recombination hotspots are self-destructive? This fundamental question is known as the recombination hotspot paradox and has attracted much attention in recent years. Yet, that attention has not translated into a fully satisfactory answer. No existing model adequately explains what maintains the allelic variability in recombination hotspots, while permitting over-transmission of the recombination resistant alleles. Here, we formulate a population genetics model for recombination hotspots finding a chaotic dynamic of allele frequency that maintains variability over time. This dynamic accounts for all empirical observations regarding the molecular mechanisms of recombination hotspots, thus providing a fully satisfactory answers.

POSTER SESSION
Abstracts in alphabetical order

POSTER

DYNAMICS OF CLANS IN HUMAN UNILINEAL POPULATIONS: A GENETIC APPROACH

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Societies are often structured into descent groups, such as clans. The descent group affiliation is transmitted either maternally (matrilineal populations) or paternally (patrilineal populations). People of a same descent groups define themselves through a common ancestry, on the male (patrilineal populations) or female (matrilineal populations) line. Consequently, women from the same matrilineal clan should be related through their maternal lines while men from the same patrilineal clan should be related through their paternal lines. However, if there is no recent common ancestry and/or if a clan incorporates individuals through horizontal processes, a lower relatedness is expected. In this study, we investigated clan dynamics in four matrilineal and four patrilineal South-East Asian populations using uniparental genetic data. Indeed, the maternally transmitted mitochondrial DNA and paternally transmitted Y-chromosome are powerful tools to explore fine scale sex-specific relatedness patterns. We sequenced the mitochondrial HVS-1 sequence (438 individuals), in addition to 17 Y-chromosome STRs loci (420 individuals). We show that the mitochondrial relatedness within matrilineal clans is higher than the Y-chromosome relatedness within patrilineal clans. This suggests that the descent rule is more strictly respected in matrilineal than in patrilineal populations or that patrilineal clans might be a conglomerate of men from diverse origins. Interestingly, ethnographic observations show that patrilineal clans from the studied populations tend to incorporate men from other villages and clans. Thus, genetic data unveil contrasted dynamics for matrilineal and patrilineal clans in South-East Asia.

POSTER

COMPARING POPULATION HISTORY INFERRED FROM GENETIC AND LINGUISTIC DATA IN CENTRAL ASIA

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Genetic and linguistic contribute to the understanding of the biological and cultural history of human populations. We compared the diversity of Central Asian populations as it is mirrored by this kind of data. These human groups belong to two distinct linguistic families: Indo-Iranian and Turkic. Concerning the linguistic data, we used a modified Swadesh lists of concepts concerning basic vocabulary. Words were classified into cognates, i.e. homologous words related by common ancestry. For genetic polymorphism data, we used mitochondrial DNA sequences, Y-chromosome and autosomal microsatellites. To infer the genealogical tree of the populations the

program starBeast has been used for both datasets. We compared the two trees obtained and found that the autosomal microsatellite tree had the best congruence with the linguistic tree. This may reflect the information gained by using many independent loci. Furthermore, the mitochondrial tree shows more congruence with the linguistic tree than the Y-chromosome tree, an interesting result in these populations known to be patrilineal. Finally, we find several populations from one linguistic group to genetically cluster with the other linguistic group, which might reflect specific linguistic replacements.

POSTER

THE COEVOLUTION OF LANGUAGES AND GENES: TRACKING DOWN MATCHES AND MISMATCHES

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The coevolution of languages and genes represents the ultimate Darwinian paradigm for the reconstruction of population dynamics in time and space, and is still one of the most evoked parallels between cultural and biological diversity. In recent years, scholars have focused on the congruence of linguistic and genetic histories to shed light on population origin, diversification and contact. Popular case studies include the diffusion of major language families, such as Indo-European and Austronesian, but smaller, regional cases of population contact have also been examined.
Mismatches between linguistic and genetic variation are usually disregarded as an exception to the general pattern. But how often do these mismatches actually occur? Can we estimate the incidence of language shift and reconstruct more realistic models of cultural evolution? And which circumstances drive such discontinuities in cultural transmission? To answer these questions we will assemble genetic databases for different regions and we will match the population data collected with relevant linguistic and cultural information. We will correlate these data while controlling for ecological factors and geographic proximity, which are natural constrains on cultural exchange and gene flow.
Preliminary analysis on Europe suggest that language shift could have occurred in 10 or 30% of the populations considered, depending on the genetic dataset used. Cases of genetic discontinuity over regions where the same language is spoken might be associated with the state policies of the past centuries.
Our final aim is to develop a more realistic understanding of the complex mechanisms behind cultural transmission. The change of cultural features through time not only impacts our ability of tracing back human prehistory, but also influences the definition of “population” as the unit of research.

POSTER

GENETIC HISTORY OF SOUTHERN AFRICAN KHOISAN POPULATIONS REVEALS TIME DEPENDENT INTENSITY OF SEX-BIASED GENE FLOW

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The genetic history of southern African populations is characterized by interactions between indigenous hunter-gatherers and a range of populations that moved into the region in the past 2000 years. The relationships among these populations show different patterns for the paternally inherited non-recombining region of the Y chromosome (NRY), maternally inherited mtDNA, and autosomes, suggesting a complex scenario of sex-biased admixture and population interactions. Here we synergistically use all three lines of evidence (mtDNA, NRY, and genome-wide genetic data) to elucidate the genetic history of Khoisan populations. Our analysis reveals a complex history of multiple admixture events with immigrant food-producing populations. We find high levels of discrepancy between estimates of Khoisan-related ancestry based on the NRY vs. mtDNA as a consequence of

heavily male-biased gene flow between the immigrant and autochthonous populations. Furthermore, we demonstrate that populations that experienced more recent admixture also show more heavily sex-biased gene flow. Finally, we propose a method for uniparental haplogroup ancestry assignment based on autosomal and mtDNA/NRY data. This method can potentially increase the precision of ancestry assignments for haplogroups that are currently uninformative regarding the place of their geographic origin; it can thus be of use in forensics as well as human population studies.

POSTER

GENETIC DIVERSITY AND DEMOGRAPHIC HISTORY OF SUB-SAHARAN HUMAN POPULATIONS BASED ON GENOME-WIDE MARKERS

BRETON Gwenna, Carina Schlebusch, Lluis Barreiro, Barry S Hewlett, Evelyne Heyer, Alain Froment, George Perry, Himla Soodyall, Paul Verdu, Mattias Jakobsson

Various affiliations

Human evolutionary history is studied in many fields: archeology, linguistics, anthropology, ethnology. Recent developments in molecular biology allowed this field to become essential for the study of the human past. Our understanding of human evolutionary history has greatly improved; for example, it is now accepted that modern humans evolved in Africa before spreading to the rest of the world. One evidence for this is the decrease of genetic diversity along the hypothesized “out of Africa” routes. In this project, we focus on genome-wide variation in Sub-Saharan African populations. We expect to observe high genetic diversity in these populations (compared to the rest of the world). We want to address open questions about human evolution in Africa, like 1-deep population structure within Africa (before the “out of Africa” event – that is, in the order of 50,000 to 200,000 years ago), or 2-more recent events (1,000 to 10,000 years ago), for example the spreading of lifestyles (pastoralism and agriculture) to southern Africa, or the interactions between neighboring populations.
To that end, we are using modern DNA from several Central and Southern Africa populations: rainforest hunter-gatherers and neighboring farming populations, as well as Khoe-San populations, who are either hunter-gatherers or pastoralists. The ancestral populations of the rain forest hunter-gatherers and of the Khoe-San were the first to diverge from the rest of the tree of modern humans (Schlebusch et al 2012, Patin et al 2009. We obtained a dataset of genome-wide markers for these populations. We will apply a range of tools to this dataset, for example to describe diversity and infer potential ancestral populations. Approximate Bayesian Computation approaches will allow us to compare different potential demographic models. For some analyses, we combine our dataset to data from other human populations across the world.
First we will describe genetic diversity in our dataset. This will give us indications about how the populations relate to each other and allow us to make hypotheses about their past that we will then be able to test using model-based approaches. We are also interested in how well different categories – for example based on population identification, on geography or on subsistence pattern – correlate with the genetic diversity.
We will later focus on several precise demographic events: timing of divergence events between these populations and other modern humans, as well as old admixture events. We will also study more recent events that can be linked to a change of lifestyle and correlated with evidence in other fields; for example, we found evidence for admixture from an East African group into Khoe-San populations, that might be associated with the spread of pastoralism (Breton et al 2014. Concerning the Central African samples, one question of particular interest is the nature of the relationships between the hunter-gatherers and their neighbors; indeed, there is evidence of recent sex-biased admixture (Verdu et al 2013, that can be explained by the current organization of these societies.
This project will allow us to deepen our understanding of human evolutionary history in Africa, the cradle of modern humans. It combines events at different time scales, from the oldest divergence
events to historical and contemporary times. Finally, we can combine evidence based on molecular data with cultural evidence, which makes it particularly exciting.

POSTER

GENETIC LEGACY OF THE INDIAN OCEAN TRADING NETWORK

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For over 4000 years, the maritime routes of the Indian Ocean trading network favored the interactions between far-distanced human populations. As one of the earliest proto-globalization processes, it put into contact populations with highly differentiated genetic backgrounds such as Southeast Asians (various Austronesian-speaking groups), East Africans (ex: Swahili), and South Asians (ex: Indians). Eventually it led to the emergence of particular admixed groups. The best example of extensively admixed population, resulting from overseas historical trading activity, is the Malagasy, the descendants of a unique admixture event between South African Bantu and Southeast Borneo individuals. Although Malagasy is a clear result of gene flow between distant geographical areas of the Indian Ocean rim, the extent of the diverse gene flows along the dense trading network of maritime routes, remains to be examined. We gathered an exhaustive genome-wide SNP dataset of 3057 individuals from 189 populations along the Indian Ocean rim - including data generated by our group for 302 Island Southeast Asians and 100 East Africans. Using both SNP-based and haplotype-based analyses, we quantified and dated several gene flows that can be linked to anthropologically and/or archaeologically documented maritime trading routes. In addition to the Malagasy case study, we will discuss the important Austronesian input detected in Northeast India and the South African legacy in the Middle East and South Asia. We will also enlighten the absence of detectable gene flow in areas with documented interactions between groups, such as the absence of Austronesian genetic trace in East Africa, or Middle East influence in Southeast Asia. Considering historical and archaeological data, our study sheds a new light on the complexity of interactions that took place in the Indian Ocean.

POSTER

GENETIC DIVERSITY DURING THE NEOLITHIC: WHAT ABOUT FRANCE?

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Methodological and technical advances in ancient DNA studies make it possible to have an ever deeper look into the genomes of individuals who lived several thousands of years ago.
Benefits from this are twofold : on one hand, we can directly address questions about the diversity of genomes and their evolution through time, and on the other hand relate this information to modern genetic variability.
Despite a rich archaeological record, little is known about how the multiple migrations that punctuate the history of presentday France have shaped its modern population. To tackle this question, our current project uses targeted enrichment to study informative SNPs and alleles in a wide panel of individuals from periods ranging from the Neolithic to the Middle Ages, sampled from various sites across France. In this talk, we will discuss the preliminary results obtained for samples dating to the Neolithic.

POSTER

PREVALENCE OF HAEMOGLOBINOPATHIES IN UPPER ASSAM, INDIA

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Haemoglobinopathies are the most common inherited disorders of globin, the protein component of haemoglobin and poses a major public health problem in many countries including India. The hereditary disorders of haemoglobin may be classified into two broad groups – the haemoglobinopathies and the thalassaemias. Haemoglobinopathies are generally common in malarial regions of the world and North-East India in general and the state of Assam in particular are no exception to this.

The present paper focusses on the prevalence of haemoglobinopathies in three population groups of upper Assam namely the Mishing (N = 318, Ahom (N = 238 and the tea garden communities (N = 397. It is found that the prevalence of haemoglobin E (HbE) is very high among the Mishing and Ahom. The migrant tea garden workers of Assam show moderate presence of sickle cell haemoglobin (HbS) in them besides beta thalassaemia trait (BTT), S-thal and HbE.

POSTER

OVERCOMING THE DICHOTOMY: NEW INSIGHTS INTO THE GENOMIC DIVERSITY OF OPEN AND ISOLATED EUROPEAN POPULATIONS

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Human populations are often dichotomized into "isolated" and "open" using cultural and/or geographical barriers to gene flow as differential criteria. Although widespread, the use of these alternative categories could obscure further heterogeneity due to inter-population differences in effective size, growth rate, and timing or amount of gene flow. We compared intra and inter-population variation measures combining novel and literature data relative to 87,818 autosomal SNPs in 14 open populations and 10 geographic and/or linguistic European isolates. Patterns of intra-population diversity were found to vary significantly more among
isolates, likely due to differential levels of drift and inbreeding. The relatively large effective size estimated for some populations isolates challenges the generalized view that they originate from small founding groups. Principal component scores based on measures of intra-population variation of isolated and open populations turned out to be distributed along a sort of continuum, with an area of intersection between the two groups. Patterns of inter-population diversity were even closer, as we were able to detect some differences between population groups only for a few multidimensional scaling dimensions. Therefore, different lines of evidence suggest that dichotomizing human populations into open and isolated fails to capture the actual relations among their genomic features.

POSTER

GENETIC ANALYSIS AND EVOLUTION OF RH BLOOD GROUP SYSTEM IN BASQUES

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Basque people have been in the limelight of many studies in the last decades due to their specific characteristics that have placed them as an isolated and unique population within Europe, highlighting their own non-Indo-European language, Euskara. They have been historically distributed along the West Pyrenees, between the Spanish and the French territories, which acted as one of the most important European glacial refugia during the Last Glacial Maximum [1]. One of the most striking characteristic is related to the Rhesus (Rh) blood group system, presenting very high frequencies of Rh-negative [2]. A variant that is expected to be selected against as consequence of the Hemolytic Disease of the Newborn (HDN) [3]. Nevertheless, the causes of this high Rh-negative frequency in Basques still remain unclear since it could be explained due to demographic or adaptive processes round the flanking regions of the Rh genes [4]. Most of the previous studies have been focused in serological and genotyping analyses.

In the present project, we carried out a genotyping analysis of the most important variants of the system: Rh+/Rh-, E/e and C/c; in our samples from Basque Country and other populations (Catalans, Moroccans, CEU, YRI and CHB) with a standard Rh-negative frequency. Then, haplotype-based analyses were performed for both the upstream and downstream flanking regions of the RHD gene and the upstream region of the RHCE gene by direct sequencing and phasing.

Our phylogeographic results do not show any special differentiation or evidence of selective pressure on flanking regions between Basques and the other populations. Instead, haplotypes grouped according to geography, with more diversity in Africans. Furthermore, two differentiated Rh+ and Rh- groups were identified among populations, being the Rh- mainly represented by Basques and less diverse than the Rh+. This observation is the expected as the Rh- variant derived from the ancestral Rh+ form [5]. We show that frequencies of Rh+/Rh-, E/e and C/c genotypes in populations are in agreement with previous data [6] as well as evidence of a correlation between C allele and Rh+ in non-African populations [4].

Thus, our results suggest that the origin of such high frequency of Rh-negative in Basques is the result of a demographic process, probably a bottleneck during the settlement of the glacial refugia in the Last Glacial Maximum, and then a subsequent drift by isolation.

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POSTER

A SIMPLE OVERVIEW OF MUSICS THROUGHOUT GABON

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Musics in Gabon share a number of common features, but reveals a great intrinsic diversity. Most of previous studies on these musics were focused on monographical description or small-scale comparisons. However, increasing knowledge about these musics, as well as intensive fieldwork throughout Gabon now allow a broader comparison of these musics.

We collected first-hand musical and contextual data in 20 ethnonymo-geographical units. These represent 10 ethnonymical groups, distributed in 7 different provinces of Gabon. About 250 pieces were recorded, from which

191 were transcribed and analysed thanks to musical systematics tools. The pieces recorded were all rhythmical-instrumental, typically with two or three vocal parts and two or three instrument parts played by drums or idiophones (stroken beam or wood board). In this region, pieces can be grouped according to their context of performance, the 'repertoires', which have been shown of cultural and musical relevance. Most of time, in each population, two different pieces of each repertoire has been recorded.
We compared three categories of data, concerning the internal organization of the sound produced. Metrical and rhythmical organization were identified from instrumental parts. We studied the diversity of metric organization through the number of beats per period and the type of subdivision of the pulsation. Rhythmical organization were studied through it segmentation in elementary rhythmic cells. Given the known relation between rhythmic figures and repertoires, rhythmic cells diversity were studied both globally and in each repertoire. Scales were identified from transcription of vocal parts. Scales diversity was studied through their interval sequence, the number of degrees and the number of half-tones they contain.
These data were compared by simple descriptive tools such as graphical representations and map plotting. This gives a first global overview and quantitative assessment of the diversity of Gabonese musics. Concerning metrics, we observed a neat geographical organization, whith ternary subdivision being more frequent on the east of the country, and binary subdivision more frequent on the west. Rhythmic cells repartition were found to be non homogeneous, neither inside a population nor at the country level. A wide variety of scales has been found, most of them being found in only one or two populations, whereas three of them are widespread.

POSTER

HUMAN POPULATION ISOLATES: CHALLENGES IN PHASING AND IMPUTATION

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In the search for genetic associations with complex traits, population isolates offer the advantage of reduced genetic and environmental heterogeneity. Proposed cost-efficient next-generation association approaches in such populations - where only a subset of study individuals is sequenced - require high quality genetic imputation and preliminary phasing. To identify an effective study-design for such genetic association studies in population isolates we compare a range of phasing and imputation methods by simulations, either recently developed for population isolates or previously established for general outbred populations. Genotyping errors and missing genotypes are simulated to observe their effects on the performance of each algorithm. We also compare different sequencing strategies by assessing the benefits of obtaining either a genome or exome sequenced local reference panel specific for the population isolate.
We simulated full sequence data on chromosome 10 over the large complex pedigree recorded in Campora, a village within the established population isolate of Cilento in southern Italy. Genotypes for Campora individuals with available DNA are simulated via gene-dropping along the pedigree with founder haplotypes sourced from the UK10K reference panel. We first assess the phasing performance of SHAPEIT2, EAGLE and SLRP software by comparing switch-error-rates. For imputation we compare IMPUTE2, BEAGLE and PRIMAL by computing correlations between imputed genotypes and true simulated genotypes. Furthermore, we examine the impact of genotyping errors and missing genotypes on each algorithm by considering the quality of phasing and imputation in the neighbourhoods of variants with simulated errors or missingness.

POSTER

ANCIENT DNA AND THE SPREAD OF THE NEOLITHIC IN EUROPE

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We have gathered a database with the mtDNA of 506 Neolithic individuals to quantify the spatial and temporal variation of the frequency of mtDNA haplogroups. For a relevant Neolithic marker, we compare the data to the results of numerical simulations, based on a mathematical model in which a Neolithic population spread from the Near East to Europe, interbreeding with Mesolithic individuals. Both the data and the simulations show that the percentage of the Neolithic marker considered decreases with increasing distance from the Near East and that, in each region, its percentage tends to decrease with increasing time after the arrival of farming. Comparing the data to the simulation results makes it possible to estimate quantitatively the relative importance of demic diffusion (i.e., the reproduction and dispersal of farmers) and cultural diffusion (incorporation of hunter-gatherers).

POSTER

REASSESSING THE INFLUENCE OF SOCIAL ORGANIZATION ON GENOMIC DIVERSITY: THE CASE OF AUSTROASIATIC POPULATIONS OF SOUTH-EAST ASIA

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At the worldwide scale, Y chromosome genetic differences between populations are larger than mitochondrial differences. This pattern has been attributed to the high prevalence of patrilocality among human populations. However at the local scale, the impact of marital residence rules on genetic patterns remains controversial. Sino-Tibetan matrilocal and patrilocal populations from northern Thailand exhibit mtDNA and Y chromosome diversities correlated with their residence rules. However, Hmong-Mien patrilocal populations from Thailand have uniparental genetic patterns very similar to matrilocal populations. Similarly, uniparental genetic diversity was not correlated with marital residence rules in Austro-Asiatic and Dravidian populations from India. More recently, a study of two populations from Sumatra found a signature of matrilocality, but not of patrilocality, on genetic diversity.

This lack of consensus may result from an approach of social organization focused on rules, that may or may not be followed, rather than on the actual patterns of residence which are more likely to influence genetic diversity. We tackled this issue by using a quantitative approach which allows us to delve into the patterns of residence and investigated their influence on uniparental and autosomal diversity. We sampled 12 ethnic groups from South-East Asia which exhibit different social organizations (matrilocal and patrilocal residence with matrilineal, cognatic or patrilineal descent) while sharing the same environment, the same lifestyle and speaking related languages. We estimated quantitative ethno-demographic variables for 535 households and produce uniparental and autosomal genetic data for over 800 individuals.

As expected, mitochondrial diversity was lower in matrilocal than in patrilocal populations and was correlated with female migration rates among villages estimated from ethno-demographic data. Unexpectedly, Y chromosome diversity was not reduced in patrilocal populations in comparison to matrilocal populations. Our ethno-demographic data confirmed that males do not migrate more among villages in matrilocal populations than in patrilocal populations, thus explaining these genetic patterns. This tendency for men to stay in their natal village in matrilocal populations may be related to the transmission of political power in these populations. Furthermore, total male and female migrations among villages are reduced in matrilocal populations in comparison to patrilocal

populations. This leads to a higher level of village endogamy, increasing consanguinity levels in matrilocal populations.
Our study highlights the importance of quantifying social organization patterns to better understand their influence on genetic diversity.

POSTER

DEMOGRAPHIC HISTORY AND DELETERIOUS VARIATION IN HUNTER-GATHERER AND FARMER AFRICAN POPULATIONS

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Population demographic history can impact the efficiency of purifying selection in removing deleterious mutations. In particular, extended periods of low effective population size (Ne) can lead to an acceleration of the accumulation of slightly deleterious mutations. Rainforest hunter-gatherer pygmies (RHG) in Africa are known to have experienced recent population bottlenecks. To both revisit the demographic history and evaluate its impact on the mutational burden of these populations we sequenced a total of 300 exomes from both pygmy and Bantu-speaking farmer (AGR) populations sampled in western and eastern sub-saharan Africa and 100 exomes of European descent to be used as a well-studied outgroup population.
We used the composite likelihood method implemented in fastSIMCOAL to fit demographic models to the synonymous site frequency spectrum (SFS) of the sampled populations. Our inclusion of a European sample allowed us to explicitly model the African demography along with the Out Of Africa event (OOA) . Strikingly, we found that a model where the RHG split from an ancestral African population before the OOA event had a much better fit to the data than a model where the RHG split at the same time or after the OOA event. Moreover, our best-fitted model revealed an expansion in the ancestral population of RHG and AGR followed by a severe bottleneck in RHG and a second expansion in AGR. We also found substantial recent admixture between AGR and RHG similarly to recent studies.
We then inferred the distribution of fitness effects of new nonsynonymous mutations (DFE) by using the maximum likelihood method DFE-alpha. This analysis revealed almost identical fractions in the proportions of new mutations in different selection intensities (neutral, mild, strongly deleterious and lethal) across all African populations. The number of deleterious alleles per individual were also not significantly different between populations even when looking at low recombination regions or within long runs of homozygosity.
Our study suggests that the split of RHG was older than the OOA event and demonstrates how inference of demographic history can shed light on differences in deleterious variation between populations. For our setting we discuss how the relatively short-lasting bottleneck for the pygmies and recent migration from larger-Ne farmers kept their deleterious load at similar levels.

POSTER

EXPLORING THE POPULATION HISTORY OF VIETNAM

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The country of Vietnam, situated in Mainland Southeast Asia, is characterized by great ethnolinguistic diversity. In particular, there are 54 official ethnic groups speaking languages that belong to five major language families: Austronesian, Austro-Asiatic, Hmong-Mien, Sino-Tibetan, and Tai-Kadai. In combination with the distribution of

these groups within the heterogeneous geographical profile of Vietnam - lowland river deltas in the east and south, and high altitude mountain areas in the northern and central regions -" the ethnolinguistic diversity indicates a potential importance in human migrations through the region. However, no comprehensive study of genetic diversity in Vietnam was conducted to date.
In this study we are focusing on Vietnam as one of the enigmatic parts of Mainland Southeast Asia. Current knowledge about the genetic history and composition of Vietnam is based on very limited data; in order to fully understand Vietnam's population history we investigate Vietnam's genetic diversity and structure on a broad scale. Based on a DNA sample set of 600 male individuals from various regions in Vietnam and representing all five language families, we are analyzing multiple lines of evidence: uniparental markers, namely full mitochondrial genomes and about 2.3 mb sequence data from the non-recombining portion of the Y chromosome, and single nucleotide polymorphism data typed on the Axiom Genome-Wide Human Origins 1 Arrays.
Preliminary analyses on the uniparental markers indicate high heterogeneity in the distribution of both mitochondrial and Y chromosome haplogroups among populations that does not correlate with geography or language, suggesting isolation and drift effects. The structure within the shared haplotypes and genetic distances among populations resemble a potential signals of post-marital residence patterns and sex-biased genetic contact.
Our aim is to investigate the demographic and historical processes that have shaped the history of Vietnam and Mainland Southeast Asia.

POSTER

PALAEOGENETIC ANALYSIS OF BRONZE AGE / IRON AGE TRANSITION IN SOUTHERN CENTRAL ASIA

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At the end of the Bronze Age, the proto-urban Oxus Civilisation in Southern Central Asia (Uzbekistan, Turkmenistan) disappeared and was replaced by Iron Age Yaz Cultures.Environmental changes such as aridification and geopolitical reasons are called for to explain this cultural transition. However, evidences of settlements from Andronovo populations during the late Bronze Age suggest that this transition was associated with migrations from northern steppe populations. Indeed, palaeogenetic studies (Allentoft et al., 2015; Haak et al., 2015 have already shown that gene flow from Yamnaya steppe populations occurred in Europe and Altai at the end of the Neolithic, suggesting that the steppe inhabitants spoke indo-european langages.
To investigate the role of migrations in the Bronze Age/Iron Age transition in Southern Central Asia, we turned to palaeogenetic studies. DNA was extracted from 17 skeletons excavated in Ulug Depe (Turkmenistan) archaeological site. The hypervariable region I of the mitochondrial (mt) genome was sequenced for 6 individuals from the Bronze Age and 4 from the Iron Age.
Criteria of authentication for ancient DNA were met: experiments were done in a clean room dedicated to ancient DNA analysis, and blank DNA extraction and PCR controls were performed. Indeed, we observed DNA damages specific for ancient DNA and an inverse correlation between the efficiency of the PCR and the length of the amplified DNA fragment. Thus, we first evidenced the preservation of ancient DNA in Southern Central Asia. After sequencing and assignment of individuals to human mitochondrial haplotypes, a high diversity of haplotypes at Ulug Depe was observed. All the haplogroups found in Ulug Depe belong to modern western Eurasian populations.
Haplogroups shared between steppe populations and Ulug Depe were evidenced, suggesting gene flow between Southern Central Asia and the Steppe. Genetic data suggest a close relationship between Yamnaya related populations and Iron Age Ulug Depe population. However, no significant genetic discontinuity between Bronze and Iron Age was shown, that may be due to a limited sample dataset and calls for nuclear DNA analysis.

Allentoft, M. E., Sikora, M., Sjögren, K.-G., Rasmussen, S., Rasmussen, M., Stenderup, J., … Willerslev, E. (2015. Population genomics of Bronze Age Eurasia. *Nature*, *522*(7555, 167–172. doi:10.1038/nature14507
Haak, W., Lazaridis, I., Patterson, N., Rohland, N., Mallick, S., Llamas, B., … Reich, D. (2015. Massive migration from the steppe was a source for Indo-European languages in Europe. *Nature*. doi:10.1038/nature14317

POSTER

COMPLEX ANCIENT GENETIC STRUCTURE AND CULTURAL TRANSITIONS IN SOUTHERN AFRICA POPULATIONS

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The characterization of the structure of southern Africa populations has been the subject of numerous genetic, medical, linguistic, archaeological and anthropological investigations.
Current diversity in the subcontinent is the result of complex episodes of genetic admixture and cultural contact between the early inhabitants and the migrants that have arrived in the region over the last 2,000 years, with some of the variation present in the past being now lost as the result of cultural and demographic assimilation by surrounding populations. Here we analyze 1,856 individuals from 91 populations, comprising novel and available genotype data to characterize the genetic ancestry profiles of 631 individuals from 51 southern African populations. Combining local ancestry and allele frequency analyses we identify a tripartite, ancient, Khoesan-related genetic structure, which correlates with geography, but not with linguistic affiliation or subsistence strategy, and probably originated in pre-historical times (~30 Kya).
The fine mapping of these components in southern African populations reveals admixture dynamics and episodes of cultural reversion involving several Khoesan groups and highlights different mixtures of ancestral components in Bantu speakers and Coloured individuals, probably reflecting different historical scenarios.

POSTER

ANALYSIS OF EXOME-SEQUENCING VARIANTS IN CILENTO ISOLATES

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Recent advances in sequencing technologies have led to a new era of population studies, enabling a more precise comprehension of the past of any population and supporting existing evidences from archaeology and historical data with unprecedented validation and definition. This type of analysis is interesting not only on the global scale, but also on a local scale, particularly in the case of special and isolated populations. Founder populations play significant roles in population genetics and trait mapping due to the effects of bottlenecks and drift on their genetic variation. Their specificity is expected to be particularly important for rare variants making these populations very useful resources in genetic association studies.
Cilento villages are located in an inland area of the "National Park of Cilento and Vallo di Diano". These villages underwent a bottleneck due to the plague of the XVII century. Campora population experienced isolation from its foundation until very recently. Gioi and Cardile are two isolated villages located 7 km away from each other. According to historical sources, the village of Gioi was settled at the beginning of the 11th century and by mid-18th century members of Gioi founded Cardile in a nearby location. A high level of reproductive isolation characterized the two villages until mid-20th century. Indeed, the majority (95%) of both Cardile and Gioi current populations are connected in a unique huge pedigree. Demographic characteristics of Cilento populations are

intermediate between the ones found in the Icelandic population (lower level of inbreeding and relatedness) and the Hutterite religious isolate in the USA (high level of inbreeding and relatedness). In Cilento, the mean kinship is 0.007, compared to a mean kinship of 0.00025 in Iceland and 0.035 in the Hutterites. We present an exploratory analysis of the exomic structure of the Cilento populations. By cataloguing exomic variants, investigating their frequencies and functional effects, we describe the allelic architecture of these isolated populations, providing the basis for a valuable resource for genetic studies focusing on disease susceptibility and population genetics.

POSTER

SUPPORT FOR A LINGUISTIC SERIAL FOUNDER EFFECT ORIGINATING IN AFRICA

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It has been proposed that a serial founder effect could have caused the present observed pattern of global phonemic diversity. Here we present a model that simulates the human range expansion out of Africa about 70 ky ago, and the subsequent spatial linguistic dynamics until today. It does not assume copying errors, Darwinian competition, reduced contrastive possibilities, or any other specific linguistic mechanism. We show that the decrease of linguistic diversity with distance (from the presumed origin of the expansion) arises simply from a repeated bottleneck (drift) effect, i.e. due to random sampling of cultural variants (phonemes in this case) in low-density pioneering populations during the range expansion. The same effect has previously explained the observed decrease in the diversity of cultural variants (crops in that case) of a European Neolithic culture. Numerical simulations show that the predictions of the model agree with the observed decrease of linguistic diversity with increasing distance from the most likely origin of the out-of-Africa dispersal. Our results therefore support the proposal that a serial founder effect could have caused the present observed pattern of global phonemic diversity.

POSTER

CONFRONTATION OF RED CELL BLOOD GROUPS DISTRIBUTION TO ENVIRONMENTAL DATA

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3 Etablissement Français du Sang Alpes Méditerranée

In Humans, thirty-five Red Cell Blood Group (RBG) systems have been defined and some susceptibilities to infectious diseases have been highlighted. Examples include interactions between the Knops system and tuberculosis, glycophorins and babesiosis, P antigen and parvovirus B19, the ABO system and cholera, the Duffy protein and malaria, or the RBG ABO-A and Rhesus-D negativity and West Nile virus. Some erythrocyte surface antigens would act as “anchor points” or “gateways” for viruses, parasites or bacteria. Moreover, for some RBG systems usually less known, geographical distribution similarities exist between their genetic diversity and the repartition of some pathogens. This may suggest potential interactions. Broadly, environmental constraints can influence the distribution of antigens, and can drive their retention or removal over several generations within human populations that express them. This natural selection process may let genomic signatures that we aim to highlight.
This project aims to confront the distribution of systems of high transfusional (ABO, Rhesus...) or anthropological (Indian, Diego...) interest, with potentially aggressive environments for red blood cell: climate, altitude, pathogen agents. The study is carried out by analyzing (with population genetics tools and statistic methods) genome-wide

genetic data obtained in populations spread over the five continents and evolving in various environmental conditions associated with specific habitats. An original reading strategy, with selection detection related to the environment by genome scans, of the information contained in the loci encoding, or in connection with the RBG systems, can thus be validated.
The objective is to reveal an environment footprint on the distribution of genetic polymorphisms of RBG between human populations evolving in different ecosystems. Our study will be able to contribute to the improvement of blood safety of populations and takes as such a strong interest for the *Établissement Français du Sang*, which supports the project.
Keywords: Red Cell Blood Group Systems, Human Populations, Selection, Environment, Pathogens, Population Genetics, Single Nucleotide Polymorphims, Genome-wide Data.

POSTER

GENOMIC ANCESTRY OF SOUTHERN ITALY: INSIGHTS INTO A COMPLEX HISTORY OF ADMIXTURE

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Assessing the genetic impact of different migration processes on present-day populations is challenged by the extent of admixture layers involving ancestral groups. The cross-cultural gateway linking Southern Italy with the Balkans and Aegean islands has represented the theatre of multi-layered migrations of peoples and cultures both in pre-historical and historical times. However, their relationship patterns are largely unexplored from a high-resolution genomic perspective.
In this study, we genotyped 511 samples from 23 populations of Sicily, Southern Italy, Greece and Albania with the Illumina GenoChip Array (~150,000 SNPs). New remarkable samples from Albanian- and Greek-speaking ethno-linguistic minorities of Southern Italy were also included. In order to disentangle admixture layers and assess recent cultural heritages, we compared our data with a large collection of modern and ancient Euro-Mediterranean individuals.
Our results reveal that Sicily and Southern Italy belong to a vast Mediterranean genetic continuum, extending from Sicily to Cyprus, through Crete and Anatolian Greek Islands. Besides a predominant Neolithic heritage, these populations show significant impacts of Post-Neolithic Caucasus- and Levant-related ancestries. In addition, further historical genetic contribution from North-Central Balkans and Eastern Europe characterize the continental Southern Balkan groups of Greece and Albania.
The detected genetic texture helps clarifying some cultural changes associated to the spread of Italian Arbereshe and Grecani ethno-linguistic minorities during classical and medieval times. Albanian-speaking Arbereshe trace their recent heritage to Southern Balkans. On the other hand, the substantial genetic resemblance of Greek-speaking Grecani with the populations of the continuum and particularly their Italian neighbors suggest a longer history of presence in Southern Italy.
While contributing new details from both genetic and cultural viewpoints, our results emphasize the importance of considering complementary scales of investigations and detailed population panels to assess processes involving tightly related ancestries that repeatedly interacted at different times in the past.

POSTER

IRON AGE ITALIC POPULATION GENETICS: THE PICENI FROM NOVILARA (8TH-7TH CENTURY BC)

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During the Iron Age, in Italy, the archaeological data provide documentary evidence of the appearance of the first communities with a strong and well defined cultural identities.
At the moment, only few studies report genetic data about the Italian populations of this period and, in particular, the Piceni have never been studied. The traditional ethnogenesis describes the “Picenum Culture” as a pre-Roman population who lived in the northern Adriatic coastal plain of Italy.
A pilot research project, based on both genetic and archaeological approaches, has been started in the Novilara necropolis (dated at 8th-7th century BC) localized in the Marche region (central Italy). This archaeological site represents an exceptional evidence due to the presence of more than 300 graves excavated so far, characterized by abundance of grave goods and also a good conservation status of the skeletal remains. To shed light on the ancient genetic diversity of Italy of this period, the HVS-1 region of mitochondrial DNA (mtDNA) in a first set of 27 individuals from Novilara necropolis was examined by high-coverage next-generation sequencing (NGS). Typical ancient DNA damage pattern in the analysed sequences and their comparison with those of the researchers involved in this project confirm the authenticity of the obtained data. Moreover, we performed a forensic analysis on these same individuals obtaining partial profiles for both autosomal STRs (Globalfiler PCR Amplification kit and Powerplex ESX 17 System) and Indels (Investigator DIPplex Kit).
With the combination of these data, we used genetic tests in order to establish the possible kinship relationships among the individuals here analysed. By these analyses, we will try to be validate the archaeological technique of seriation based on the analysis of the material culture, and to understand the contribution of the Piceni population in shaping the modern Italian gene pool.

POSTER

COMPARATIVE GENOMICS OF INNATE IMMUNITY IN HUMAN AND NON-HUMAN PRIMATES

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Innate immunity constitutes the front line of host defence and provides a valuable model for the study of the selective pressures imposed by microorganisms on host genomes. Population genetic studies in humans have shown that the impact of selection on some families of innate immune receptors and downstream signaling molecules varies considerably. Despite humans and closest relatives share most of their genome, increasing evidence suggests that humans and other apes exhibit important differences in susceptibility to, and severity of, infectious diseases (e.g. HIV, malaria). In fact, it is possible that many of such differences emerged as a result of the action of natural selection leading to species-specific adaptations in response to environmental changes. Nevertheless, the effects of natural selection in shaping innate immunity in our closest relatives remain largely unknown.

In this study, we aim at uncovering shared and unique signatures of natural selection acting on innate immunity genes in great apes, and identify species-specific adaptations that might be essential for individual and population survival. We first implemented coalescent simulations on great ape demographic history and demonstrate that the different statistics used present high power to uncover targets of purifying, positive, and balancing selection in the genomes of great apes. We then analyzed whole-genome sequence data for different populations of great apes, covering a total of 11 great ape subspecies. To do so, we focused on comparing selection signatures on a set of more than 1,500 loci involved in innate immunity functions with the remainder of the genome. Our analyses show that, taken as a whole, innate immunity genes are privileged targets of natural selection in the genomes of great apes. Moreover, we provide evidence that some loci (and their related biological functions) exhibit extensive differences in the form and intensity of selection across species. These findings suggest important differences across primates in the mechanisms involved in host adaptation to pathogen pressures, informing about the pressures imposed by their respective ecological habitat. Together, our results provide a thorough understanding of selective forces shaping the evolution of innate immunity in great apes. To our knowledge, this study represents the first attempt of bringing to light the evolutionary mechanisms that operated for millions of years as a response to pathogen infection.

POSTER

GENETIC MAPPING OF HISTORICAL ARMENIA

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The origins of Armenians are difficult to reconstruct due to several reasons. The vast majority of historical evidence has been destroyed or lost during numerous invasions and earthquakes. Further, the modern political division of historical Armenia among neighbouring states has made archaeological, anthropological and archival research a sensitive and difficult task. Though most of historical Armenian provinces were forcibly depopulated during the last few centuries, fortunately, the descendants of these refugees retain information providing the possibility of reconstructing the genetic legacy of their ancestors. Here, we present the first ever attempt to create a genetic map of historical Armenia that reproduces the rich spatial mosaic of the Armenian gene pool. Our primary findings show that the Armenian paternal genetic legacy displays immense diversity of lineages, indicating a large number of "founding fathers". The vast majority of Y chromosomes belongs to the haplogroups originated and expanded during or following the Neolithic. The modern Armenian gene pool, studied by genome-wide SNP data, consists of traces of an origin from a mixture of various populations taking place from 3000 to 2000 BCE, with dramatic decrease of admixture signals after 1200 BCE. Armenians had no significant mixture with other populations also in their recent history. Of the primary reasons that have impeded genetic contact of Armenians with foreigners, the highland geography, early adoption of Christianity, strong ethnic and cultural identity can be considered the most likely. Recently, the origins of Armenians have been examined based on ancient DNA. A strong genetic continuity between the Bronze Age population and modern Armenians has been revealed. Further aDNA studies will cover time span from the Neolithic to the Middle Ages while addressing other principal questions on the Armenian genetic history.

WORKSHOP 1 *Measuring Culture*

Measuring cultural differences and evolution

Since about twenty years, computational methods and approaches have been applied to cultural differences. The purpose was to measure and quantify traits that, previously, were seen as computationally intractable by the specialists of the disciplines involved (cultural anthropology, linguistics, musicology, etc.).
Artefacts, languages and music are now coded as vectors, multistate matrices, distance matrices that allow an unprecedented degree of historical and geographical inference. But… What are we inferring? *From black boxes to new hypotheses, is the signal robust enough? The purpose of this workshop is to attract a panel of specialists, as diverse as possible, in order to address the weaknesses and strengths of computational methods that go beyond the understanding of cultural anthropologists, linguists, musicologists, etc.*

December, 9

WORKSHOP *Measuring Culture*
Measuring cultural differences and evolution.
Organized by Pierre Darlu and Franz Manni

10:00 am – 1:30 pm *Talks of the morning in the following order:*

Pierre DARLU and Franz MANNI
General overview of the aims and themes of the workshop
Franz MANNI
The descendants of the Middle-Ages Spanish population do not speak Spanish and live by the coast
Leonardo ARIAS
Flowing genes and languages, the role of rivers in structuring genetic variation in native American populations from Nort-West Amazonia
Kristina TAMBETS
The genetic structure of the Uralic-speaking populations - Do genetic and linguistic data tell the same story?
Daniel CORACH
Genetic Relationships of Southern South-American Tribal Groups Inhabiting Argentinean Territory Assessed by Uni and Biparentally Transmitted Polymorphic Markers Correlates with Linguistic Affinities.
Eugenio BORTOLINI
Isolation by distance, demic diffusion, and cultural transmission in the genomic era: A case study based on folktale distribution over Eurasia and Africa
Valentin THOUZEAU
Reconstructing demographic and cultural history of human populations from genetic and linguistic polymorphism data
Franz MANNI
The cultura package? Comparing the diversity of music and languages in Gabon (Africa)
Pierre DARLU
Cladistics, Networks or Distance methods? Controversial representations of the geographical distribution of linguistic traits

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS
Jeremy GARDENT *Page 30*
A simple overview of music throughout Gabon
(10' talk) Goki LY *Page 32*
Reassessing the influence of social organization on genomic diversity: the case of Austroasiatic populations of South-East Asia
(10' talk) Joaquim PEREZ-LOSADA *Page 36*
Support for a linguistic serial founder effect originating in Africa

END

WORKSHOP 1 *Measuring Culture*
Abstracts in alphabetical order

TALK WORKSHOP MEASURING CULTURE

FLOWING GENES AND LANGUAGES: THE ROLE OF RIVERS IN STRUCTURING GENETIC VARIATION IN NATIVE AMERICAN POPULATIONS FROM NORTHWESTERN AMAZONIA

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Geographic distances and linguistic differences are often important factors that influence genetic variation among human populations. Here, we address the impact of these two factors, as well as cultural practices, on the genetic variation observed among Native American populations from Northwestern Amazonia, a huge area extending from the Andean foothills to both the Orinoco and the Amazon basins and crossed by a vast network of rivers. This area harbors tremendous human diversity in terms of languages, various cultural practices, and subsistence strategies.

We have generated sequences of the complete mitochondrial genome sequence and 2 Mb of the non-recombining region of the Y-chromosome in a sample of 460 individuals belonging to 40 different ethnolinguistic groups from Northwestern Amazonia, in Colombia. The analysis of these data shows that the distribution of groups along rivers is a more important factor explaining the structure of genetic variation than geography or language affiliation. Furthermore, linguistic exogamy –a marriage system in which the spouse has to belong to a different language group- among groups from the Tukanoan, Arawakan, and Maku-Puinave language families boosts the movement of women among different linguistic groups while men often stay in their homeland, thus leaving a clear signal in the patterns of genetic variation observed among and within groups. Finally, we show how differences in subsistence strategies are related to levels of genetic diversity. While the majority of the groups included are riverine agriculturalists and harbor higher levels of genetic diversity, those with incipient agriculture as well as hunter-gatherer populations show lower levels of diversity, with one of the two hunter-gatherer groups included showing an extreme reduction in diversity.

To conclude, the evidence provided by these populations constitutes an important source of information to understand the peopling of Amazonia and contributes to our understanding of the initial peopling of South America.

TALK WORKSHOP MEASURING CULTURE

ISOLATION BY DISTANCE, DEMIC DIFFUSION, AND CULTURAL TRANSMISSION IN THE GENOMIC ERA: A CASE STUDY BASED ON FOLKTALE DISTRIBUTION OVER EURASIA AND AFRICA

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Over the past thirty years, many quantitative studies on the mechanisms of gene-culture coevolution focused on the spread of cultural information through demic and/or cultural diffusion (Ammerman and Cavalli-Sforza, 1984; Collard et al. 2006; Ackland et al. 2007;
Bell et al. 2009; Itan et al. 2009; Fort 2012; Creanza et al. 2015). The most accepted null model against which instances of demic processes have been assessed draws on the expectation that non-selective markers (both cultural and genetic) would form geographic clines produced over time by Isolation-by-Distance processes (IBD; Pinhasi and von Cramon-Taubadel 2009). At the same time, advances in DNA sequencing have opened new avenues for exploring the demographic histories and geographic trajectories of human populations, shedding new light on the origins of genetic and cultural diversity around the world.
The present study explores the impact of demic diffusion on the distribution of 596 folktales (Uther 2004; Graça da Silva and Tehrani 2016) at a cross-continental scale by bringing newly available whole-genome sequences into the picture. To investigate this process in 33 populations of Africa and Eurasia, the relationship between cultural, genetic, and geographic variability is formally assessed by using both a frequentist hypothesis-testing approach, and a model-selection approach based on Akaike's Information Criterion.
This research also addresses methodological questions concerning inferential problems emerging when cultural and genetic evidence are directly compared, and the applicability of standard null models based on isolation by geographic distance considering the increasing availability of genetic evidence.

TALK WORKSHOP MEASURING CULTURE

GENETIC RELATIONSHIPS OF SOUTHERN SOUH AMERICAN TRIBAL GROUPS INHABITING ARGENTINEAN TERRITORY ASSESSED BY UNI AND BIPARENTALLY TRANSMITTED POLYMORPHIC MARKERS CORRELATES WITH LINGUISTIC AFFINITIES

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Besides available information offered by the Archaeology, little is known about the aboriginal American populations at pre-European contact times. Currently, an increasing wealth of molecular knowledge is being produced that might shed light on the pre-hispanic history of the Americas.
Accordingly, with the aim of investigating the phylogeographic histories of the descendants of Southern Native Americans inhabiting nowadays the territory of Argentina eight tribal groups were selected, including: Tehuelche and Mapuche from South West area: Guarani and Chiriguanos from North, Wichi from North West and Pilaga, Toba and Mocoví from North Central area. A collection of over 300 individuals were analyzed by means of autosomal STRs, ancestry informative autosomal markers, Y-STRs, rapid mutating Y-STR and Y-SNPs, coding region mtDNA SNPS and complete mtDNA control region sequences.
All samples included in this study belong to the Y chromosome Haplogroup (HG) that characterize Native Americans: Qa1a3, and to one of the four Native American mtDNA HG: A,B,C,D. Our results showed a clear correlation between genetic information and the linguistic families of the groups they belong to. In addition, Toba group might represent the ancestral ethnicity of Guaycurúan speakers tribes. Even though, we employed different genetic systems, most of them provide concordant results. The most remarkable result was the lack of non-admixed individuals among the Native American descendants included in the study. The less admixed group was Toba displaying little over 95% Native American contribution. The last continental territory to be occupied by the modern humans showed to be the one with highest genetic admixture. Descendants of Native American lineages might represent a case-study for genetic globalization at world-wide extent.

TALK WORKSHOP MEASURING CULTURE

CLADISTICS, NETWORKS OR DISTANCE METHODS? CONTROVERSIAL REPRESENTATIONS OF THE GEOGRAPHICAL DISTRIBUTION OF LINGUISTIC TRAITS

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We will examine various methods for representing the geographical structure of linguistic traits at different scales, using different sets of data. We will compare the main and common methods that all take as a starting point the modelling the changes of the linguistic characters, while diverging later on *i)* either by estimating pairwise distances based on the degree of dissimilarity between traits and between linguistic units, or *ii)* by applying various cladistic or probabilistic algorithms to handle the changing of the linguistic features at stake, and to draw clusters or trees representing the linguistic units. In both cases, the results have then to be plotted into space. Networks can usually be derived from the two previous methods in a way that will be discussed. We will also examine how to take into account the informativeness and the robustness of the results, depending on the method being used. These comparisons will be applied on various linguistic data sets about which we have already some experiences: i) the Euskararen Atlas Sociolinguistikoa (EAS, UPV University) survey, which includes dialect variations of Basque speakers (elders), on the basis of verbal morphology (aditza); ii) Mazatec and other Mesoamerican languages (from ALMaz/Kirk database 1966, 2010-13), such as Tseltal (the ALTO project (Ciesas Sureste & University Paris-Sorbonne); iii) Dyen's Indo-European database. Extension to other kind of data will also be proposed. The discussions will focus on the advantages and disadvantages of these different approaches, and their degree of congruence when interpreting the results.

TALK WORKSHOP MEASURING CULTURE

THE CULTURAL PACKAGE? COMPARING THE DIVERSITY OF MUSIC AND LANGUAGES IN GABON (AFRICA)

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With reference to the talk given in the plenary session (8th of December – This conference) and titled '*The Bantu expansion and the peopling of Gabon, a genetic and linguistic overview',* I have tried to compare the computational-linguistics classification of the Bantu languages spoken in Gabon with a classification of the musical practices of the same country. Le Bomin *et al.* (2016) coded musical differences as multistate characters and obtained a phylogenetic tree by cladistics methods, suggesting that the transmission of musical cultural traits can be vertical also according to the patrilinarity and matrilinearity of the ethnic groups.
This article is interesting because it addresses inherent aspects of the Culture that might be part of a package transmitted as a whole (for example gastronomic traditions and language).
In reality, the classification of musical diversity, as published, does not match the classification of the languages spoken by the same populations. Nevertheless, when independently re-analyzing the data of Le Bomin *et al.* (2016) by using resampling techniques, new results are obtained and they are compatible with linguistic data. This example raises theoretical and methodological questions that are relevant to the workshop.

Le Bomin et al. (2016). The evolution of musical diversity: the key role of vertical transmission. PLoS One http://dx.doi.org/10.1371/journal.pone.0151570

TALK WORKSHOP MEASURING CULTURE

THE DESCENDANTS OF THE MIDDLE-AGES SPANISH POPULATION DO NOT SPEAK SPANISH AND LIVE BY THE COAST

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A previous study concerning 33,753 single Spanish surnames (considered as tokens) occurring 51,419,788 times has shown that the geography of contemporary surname variability in Spain corresponds to the political geography of the country at the end of the Middle-Ages. The synchronicity between surname adoption and the political and cultural effects of the *Reconquista* have permanently forged a Spanish identity that subsequent migrations, internal or external, did not deface (Rodriguez-Diaz *et al.* 2015).
The same database has been analyzed differently, that is by clustering together surnames having a similar geographic distribution and, among them, considering only the clusters exhibiting a clear peak of frequency in one of the 47 continental provinces of Spain, meaning that such provinces can correspond to the areas where these surnames were first adopted (about five centuries ago, when family names started to be transmitted in a rigid way from generation to generation). From these results, we computed the frequency of the surnames that are 'autochthonous' of each province (endogenous origin) with respect to the others (of exogenous origin). It appears that the provinces having the higher frequency of endogenous surnames are distributed along the coast, especially in Catalonia and in the area of Valencia, together with the northern shore of Spain. Interestingly, these are also the regions where languages other than Castilian are spoken.
This preliminary result can be interpreted in two ways. Existing linguistic differences might have been reinforced by a lower level of migrations from other areas (a characteristic of provinces having a higher frequency of autochthonous' surnames) and may have discouraged migrations to culturally different areas.

Rodriguez-Diaz et al. (2015). Footprints of Middle Ages Kingdoms are still visible in the contemporary surname structure of Spain. *PLoS One* http://dx.doi.org/10.1371/journal.pone.0121472

TALK WORKSHOP MEASURING CULTURE

THE GENETIC STRUCTURE OF THE URALIC-SPEAKING POPULATIONS: DO GENETIC AND LINGUISTIC DATA TELL THE SAME STORY?

TAMBETS Kristiina [1] , Anne-Mai Ilumäe[1], Bayazit Yunusbayev[1], Georgi Hudjashov[1], Siiri Rootsi[1], Terhi Honkola[2], Outi Vesakoski[2], Quentin Atkinson[3], Alena Kushniarevich[1], Sergey Litvinov[1,4], Pontus Skoglund[5], Monika Karmin[1], Ene Metspalu[1], Maere Reidla[1], Marina Gubina[6], Sergey Zhadanov[1,6], Larisa Damba[1,6], Khadizhat Dibirova[1,7], Toomas Kivisild[1,8], Oleg Balanovsky[5], Elena Balanovska[5], Irina Evseeva[5], Mikhail Voevoda[6], Ludmila Osipova[6], Elza Khusnutdinova[4], Mait Metspalu[1], Richard Villems[1]

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[8] University of Cambridge, United Kingdom

The Uralic languages have a patchy spread pattern across a vast territory in North Eurasia, from the Scandinavian Peninsula in Europe to the West Siberian Plain in Asia. The number of the speakers of Uralic languages, however, is only 20.5 million people. The language family splits into two major branches, Finno-Ugric and Samoyedic, that separated ca 4000 ya and are mostly spread in Europe and in Siberia, respectively. The

mitochondrial DNA (mtDNA) studies of the Uralic-speakers have revealed that the proportions of their western and eastern Eurasian maternal ancestry components are determined by geography – they belong to the common western Eurasian mtDNA package in Europe and are part of eastern Eurasian package in Siberia. However, there is a considerable common legacy in the paternal lineages of North Eurasian populations: both European and Siberian Uralic-speaking populations share a noticable proportion of Y chromosomal haplogroup N, which originates from southeast Asia. Thus, there is a clear paternal gene flow from Asia to Europe not seen in mtDNA variation, part of what might be connected with the spread of Finno-Ugric languages. Here we present the results of autosomal genetic variation of 16 Uralic-speaking populations genotyped for over 600 000 genome-wide SNPs. We analyze the genetic structure of the studied populations and compare that with the results obtained from uniparental markers. We study whether the patterns in genetic differences follow the linguistic landscape or are governed by geography and test for the correlation between genetic and linguistic variation of the Uralic-speakers. We conclude that the history of the present spread of Uralic languages and peoples is a result of complex processes including gene flow over many generations and possible language shifts associated with some but not substantial change of genes.

TALK WORKSHOP MEASURING CULTURE

RECONSTRUCTING DEMOGRAPHIC AND CULTURAL HISTORY OF HUMAN POPULATIONS FROM GENETIC AND LINGUISTIC POLYMORPHISM DATA

THOUZEAU Valentin, Philippe Mennecier, Paul Verdu, Frédéric Austerlitz

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The main aim of our research is to develop methods for analyzing language diversity and genetic polymorphism data in a unique methodological framework, in order to infer the past history of separation, exchanges and admixture among human populations. For this purpose, we have developed a new computer program that simulates, simultaneously, the evolution of genetic and linguistic diversity in a set of populations for which both kinds of data are available.
Simulations are then compared to real genetic and cognate polymorphism data, using Approximate Bayesian Computation (ABC) methods to identify the most realistic historical scenarios underlying each type of data, and to infer the parameters of the corresponding model.
So far, we have applied this approach to Central Asia, an area where Turkic-Mongol and Indo-Iranian speaking populations historically met, and where our laboratory has gathered both genetic data (sequences, microsatellites, genome-wide genotypes) and linguistic data (lists of basic vocabulary later coded into cognates).

WORKSHOP 2 *Genomic Demography*

Inferring demography, gene flow and admixture from genome-wide data

Population genetics has enabled a reconstruction of the past demographic history of human populations. The increasing availability of genome-wide datasets at regional or global scales allows, in principle, to investigate in a much greater detail the demographic history of populations. Classical population genetics has to be adapted to genome-wide data and the development of new analytical tools has become crucial.
In this workshop, we aim at discussing the strengths and limitations of existing and forthcoming approaches to real genomic data, with a special focus on demographic changes over the time, complex gene-flow and admixture processes.

.December, 9

WORKSHOP *Genomic Demography*
Inferring demography, gene flow and admixture
from genome-wide data
Organized by Frederic Austerlitz and Paul Verdu

10:00 am – 1:30 pm *Talks of the morning in the following order:*

Frederic AUSTERLITZ and Paul VERDU
General overview of the aims and themes of the workshop
Simone Andrea BIAGINI
Human population genomics of the western Mediterranean
Ashot MARGARYAN
7800 years of mitochondrial genetic continuity in Armenia
Gerard SERRA-VIDAL
Study of the human north African genome landscape through the analysis of complete genomes
Lara R. ARAUNA
Recent historical migrations have shaped the gene pool of Arabs and Berbers in North Africa
Sandra OLIVEIRA
The maternal history of south-west Angola
Alex MAS-SANDOVAL
Native American pre-colonization genetic history through admixed Brazilians

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS

(10' talk) Nicolas BRUCATO *Page 28*
Genetic legacy of the Indian Ocean trading network
(10' talk) Enrico MACHOLDT *Page 33*
Exploring the population history of Vietnam
(10' talk) Stefania SARNO *Page 37*
Genomic ancestry of southern Italy, insights into a complex history of admixture

END

WORKSHOP 2 *Genomic Demography*
Abstracts in alphabetical order

TALK WORKSHOP MEASURING CULTURE

RECENT HISTORICAL MIGRATIONS HAVE SHAPED THE GENE POOL OF ARABS AND BERBERS IN NORTH AFRICA

Lara R. ARAUNA1, Javier Mendoza-Revilla1,2, Hassan Izaabel3, Asmahan Bekada4, Soraya Benhamamouch4, Karima Fadhlaoui-Zid5, Pierre Zalloua6, Garrett Hellenthal2, David Comas1

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5 University El Manar, Tunis, Tunisia
6 The Lebanese American University, Chouran, Beirut, Lebanon

North Africa is characterized by its diverse cultural and linguistic groups, which mainly include Berbers and Arabs. The genome-wide structure of North African populations has been described as an amalgam of autochthonous, Middle Eastern, European and Sub-Saharan ancestries. The indigenous North African genetic ancestry has been estimated to be derived from a “back to Africa” migration dated in pre-Holocean times, and suggested to be in highest frequency in Berber populations (Henn et al. 2012), which are considered the autochthonous populations of North Africa. No differences have been found in uniparental and classical markers between Berbers and Arabs, although the scanty genomic data available have highlighted the singularity of Berbers. We analyze genome-wide autosomal data (~900,000 SNPs) in five Berber and six Arab groups. Haplotype-based methods show a high degree of genetic heterogeneity within the geographical sampled populations and find genetic clusters spread among them without a clear geographical pattern and without strong differences between Berbers and Arabs. No differential Middle Eastern, European and sub-Saharan Africa ancestries are found between Berbers and Arabs. However, differential ancestry is found within the geographical locations, dividing the sampled population in different genetic clusters found across North Africa. These differences might obey to other social categories rather than the ethnical dicotomy Berber/Arab. However, some Berber groups are found to be isolated and endogamous, with high autochthonous component frequencies, large homozigosity runs and low effective population sizes, while none of the Arab populations show this pattern. We find a continuous gene flow from the Middle East with a peak around the 7th century AD coincident with the Arabization of the region; and an incoming demographical wave from sub-Saharan Africa in the 1st century AD, in agreement with the Roman slave trade, and a strong migration in the 16th century AD, coincident with a huge impact of the global trans-Atlantic and trans-Saharan trade of sub-Saharan slaves in the Modern Era. Therefore, a complex pattern of recent historical migrations has shaped the genetic and social structure in North Africa.

Henn BM, Botigué LR, Gravel S, Wang W, Brisbin A, Byrnes JK, Fadhlaoui-Zid K, et al. Genomic ancestry of North Africans supports back-to-Africa migrations. PLoS Genet. 2012; 8(1):e1002397.

TALK WORKSHOP GENOMIC DEMOGRAPHY

HUMAN POPULATION GENOMICS OF THE WESTERN MEDITERRANEAN

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The Western Mediterranean basin represents one of the most lively core of cultural crossroads in the past, and its history is still alive in the present-day populations. Both pre-historical and historical migrations affected the genetic structure of the whole Mediterranean human groups(1): the existence of a genetic gradient across Europe has often been highlighted, and a Near East origin for most of the current Europeans has been suggested(2). So far, a large number of genetic studies have been carried out in the Mediterranean area using different markers, but no consensus has been reached on the genetic landscape of the Mediterranean populations, and often-contradictory results have been achieved(1). Because of the intricacy of its past, today identify a genetic profile of the Western Mediterranean populations is if nothing else a complex challenge.
Using the array technology, in this work we enhance the resolution on a local scale, and try to detect finer substructures related to the internal and external contributions, thus highlighting the dynamics of the peopling of this area. For this purpose, more than 1200 samples coming from all the regions surrounding the Western Mediterranean basin have been included in the study. In particular, data comes from samples we have gathered from Catalonia, the Balearic Islands, Valencia, seven regions covering most of France, and Naples, and that have been or are being typed with the Axiom®Genome-Wide Human Origins (~629K SNPs) array. The dataset is also completed with published data for Spain(3) (also with the Axiom®Genome-Wide Human Origins), Italy (Illumina HumanOmni2.5 BeadChip Array, ~2.5M SNPs), and North Africa (Genome-Wide Human SNP Array 6.0, ~900K SNPs).
With this approach we aim to shed a light on the evolutionary history of the peopling of the Western Mediterranean, even considering the role of the Mediterranean Sea as a putative barrier to the gene flow, and the impact of the historical events in shaping the different human Western Mediterranean groups.

1. Sazzini M, Sarno S, Luiselli D (2014) The Mediterranean Human Population: An Anthropological Genetics Perspective in: Goffredo S and Dubinsky Z, The Mediterranean Sea: Its History and Present Challenges, Berlin, Springer, 2014, pp. 529 - 551 [book chapter]
2. Falchi A, Giovannoni L, Calo CM et al. (2006) Genetic history of some western Mediterranean human isolates through mtDNA HVR1 polymorphisms. J Hum Genet. 2006;51(1):9-14
3. Lazaridis I et al. (2014) Ancient human genomes suggest three ancestral populations for present-day Europeans Nature. 2014 Sep 18; 513(7518): 409–413

TALK WORKSHOP GENOMIC DEMOGRAPHY

7800 YEARS OF MITOCHONDRIAL GENETIC CONTINUITY IN ARMENIA

MARGARYAN Ashot[1,2], Miroslava Derenko[3], Hrant Hovhannisyan[2], Boris A. Malyarchuk[3], Rasmus Heller[4], Levon Yepiskoposyan[2], Eske Willerslev[1], Morten Erik Allentoft[1]

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The origin of the Armenian people is heavily debated among historians and archaeologists. Despite a long history and vast archaeological records in Armenia, it has proven very challenging to infer the demographic events that led to the formation of Armenians as a distinct ethno-cultural group. To obtain a detailed understanding of the demographic events in Armenia across millennia, we study complete mitochondrial genomes from 49 ancient individuals covering 7800 years and compare them with with that of modern Armenians (n=206) and seven neighboring populations (n=482). In this context, the lowest genetic distance was observed between the modern and ancient Armenians and this was also reflected in network analyses and discriminant analysis of principal components (dapc) showing genetic proximity between the ancient individuals and modern Armenians. We used Approximate Bayesian Computation (ABC) to test five different demographic scenarios of the Armenian population, and the simulations favored a model where both ancient and modern Armenians derive from the same source population. We conclude that there is a strong signal of continuity in the maternal Armenian gene pool during the last 7800 years.

TALK WORKSHOP GENOMIC DEMOGRAPHY

NATIVE AMERICAN PRE-COLONIZATION GENETIC HISTORY THROUGH ADMIXED BRAZILIANS

MAS-SANDOVAL Alex(1)(2), ARAUNA Lara R(2), TARAZONA-SANTOS Eduardo(3), BORTOLINI Maria-Cátira(1), COMAS David(2)

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(2)Institut de Biologia Evolutiva (CSIC-UPF), Departament de Ciències de la Salut i de la Vida, Universitat Pompeu Fabra, Barcelona, Spain.
(3)Departamento de Biologia Geral, Instituto de Ciências Biológicas, Universidade Federal de Minas Gerais, Belo Horizonte, Brazil.

Brazilian urban admixed populations are composed of three main genetic components: Native American, European and African. Although their proportions between and within populations are different, Native American component is usually the one found in lowest proportions (6-8%), and was mostly admixed in east Brazil soon after the European Conquest. We take advantage of 6487 urban admixed Brazilians genotyped at 2,5M SNPs (Kehdy et al 2015) in three locations of eastern Brazil (North-East, South-East and South regions) to extract the Native American haplotypes and rearrange them in Assembled Individuals with full Native American ancestry, emulating a historical landscape where Native Americans had not admixed with Europeans and Africans. This method allows us to analyze the Native Americans population structure in a region where most of Native American populations disappeared after European colonization. While the structure of Native American component in admixed individuals from Caribean, Mesoamerican, and Andean regions has been described in a fine scale (Moreno-Estrada et al 2013, Moreno-Estrada et al 2014, Homburger et al 2015) few approaches have analyzed it in the admixed individuals from the South American east coast. We find that population structure of Native American Assembled Individuals is driven by the geography of the sampling location while the populations structure of urban admixed Brazilians is modulated by the European or the African ancestry. In order to explore in depth the origin and the demographic history of the Brazilian Native American component we analyze the Native American Assembled Individuals together with a dataset of non-admixed Native Americans of Brazil to see differential ancestry of Native American main genetic groups in Brazilian urban admixed populations.

Kehdy, F. S. G. et al. Origin and dynamics of admixture in Brazilians and its effect on the pattern of deleterious mutations. Proc. Natl. Acad. Sci. 112, 8696–8701 (2015).
Moreno-Estrada, A. et al. Reconstructing the Population Genetic History of the Caribbean. PLoS Genet. 9, (2013).
Moreno-Estrada, A. et al. The genetics of Mexico recapitulates Native American substructure and affects biomedical traits. Science (80-.). 344, 1280–1285 (2014).
Homburger, J. R. et al. Genomic Insights into the Ancestry and Demographic History of South America. PLoS Genet. 11, 1–26 (2015).

TALK WORKSHOP GENOMIC DEMOGRAPHY

THE MATERNAL HISTORY OF SOUTHWEST ANGOLA

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(2) Departamento de Biologia, Faculdade de Ciências, Universidade do Porto, Porto, Portugal
(3) Max Planck Institute for the Science of Human History, Jena, Germany
(4) Dynamique du Langage, UMR5596, CNRS & Université Lyon 2, Lyon Cedex 07, France
(5) Department of Evolutionary Genetics, Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany

Southern Angola is an important, yet genetically understudied, region of sub-Saharan Africa, located at the southwestern edge of the Bantu expansion and peopled by different foraging and pastoral groups that speak languages from the Bantu (Herero), Kx'a (Ju) and Khoe-Kwadi (Kwadi) families. Since this linguistic and bio-cultural diversity is likely to have been generated by the confluence of different migratory movements, the study of contemporary populations from southern Angola not only provides an ideal framework to investigate the history and consequences of population contact in the area, but will also give insights for understanding human settlement in the wider region of southern Africa.

Here, we present an analysis of complete mtDNA sequences from seven southern Angolan populations (Kuvale, Himba, Tchimba, Kwisi, Twa, Kwepe and Sekele) potentially associated with the 3 major layers of peopling of southern Africa: indigenous foragers, a pre-Bantu pastoralist intrusion from eastern Africa, and ultimately the Bantu migrations. We assessed the relationships between the mtDNA lineages of these populations, as well as their correlation with a matriclanic system in which clans are inherited through the maternal line. Additionally, we use an ABC framework to specifically investigate the demographic history of the two dominant Herero-speaking groups and the closely related Nyaneka from Angola.
We found: i) the probability of sampling closely related pairs of sequences from the same clan is much higher (0.51) than the one obtatined from pairs of sequences randomly chosen (0.10), indicating a remarkable correlation between matriclans and mtDNA lineages; ii) despite the linguistic and cultural proximity of the two dominant Herero groups - Kuvale and Himba, a considerable level of differentiation is observed between them, with the former presenting a very high frequency (~50%) of L0d mtDNA lineages , also found in populations from Botswana, Zambia and Namibia; iii) nonetheless, by testing different demographic histories we found that the model of an early divergence of Nyaneka and subsequent split of Herero+Himba+Damara from Kuvale had the highest support (posterior probability = 0.74); iv) the Kwisi/Twa are genetically distant from all the other groups, presenting particularly high frequencies of L1c1b and L0a lineages, a pattern that might have emerged by lineage fission and genetic drift or might indicate a more distant relation to the Bantu groups from the Namib; v) absence of characteristic Khoisan mtDNA haplogroups (L0d or L0k) in the Kwepe, who are the present-day descendants of the speakers of the extinct Kwadi language; vi) absence of sequence sharing between the Ju-speaking Sekele and the other groups from the Namib, showing that despite their geographic proximity, there is no evidence of admixture.
Overall, our study confirms the presence of characteristic mtDNA lineages from indigenous foragers and Bantu speaking peoples, but shows no evidence of a pre-Bantu intrusion from the East. Morover, the finding that the Kwisi/Twa are genetically distinct from the presently sampled groups of southern Africa, suggests the presence of previously unknow structure in the studied region.

TALK WORKSHOP GENOMIC DEMOGRAPHY

STUDY OF THE HUMAN NORTH AFRICAN GENOME LANDSCAPE THROUGH THE ANALYSIS OF COMPLETE GENOMES

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The African continent is known be the place of origin for modern humans, which makes the understanding of its internal genetic variation specially relevant to for reconstructing human complex demographic history. Nevertheless, the North of the continent has a somewhat independent history from the rest and has a particularly highly complex demographic history due to its singular geographical location – between the Sahara desert, the Mediterranean Sea and the Middle East – which has historically favoured a complex demographic history (including back migrations, gene flow, bottlenecks or genetic isolation) involving both autochthonous and bordering (European, Asian and Sub-Saharan) populations [Henn et al., 2012].
Here, we study the recent history and relationships among ten different North African populations, by analyzing twenty-five individual whole-genome sequence data at deep coverage covering all major geographic and ethnic groups within North Africa (Western Sahara, Morocco, Algeria, Tunisia, Libya and Egypt, including Arab and Berber groups). We analysed these data and compared them to eight Sub-Saharan, four Middle Eastern and four European individual genomes to have them in a broader genomic context.
More than 10 million single nucleotide substitutions were found, providing an unprecedented rich picture of the genome diversity and population history in North Africa, which allows a deeper insight into complex demographic features, such as the evolution of effective population sizes, split times, migration rates within North African groups and between surrounding populations, and other population dynamics patterns. North African samples were found to be genetically heterogeneous and show different patterns of admixture and demographic histories, such as relevant endogamy levels or high sub-Saharan component within certain Berber and non-Berber groups, though genetic diversity and geographic origins or ethnicity were not found to follow a clear correlation pattern.

Henn B.M. et al. 2012. Genomic Ancestry of North Africans Supports Back-to-Africa Migrations. PLOS Genetics., 8:1

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WORKSHOP 3 *Ancient DNA*

Ancient DNA, from archaeological sites to genome analysis.

Ancient genomes provide a direct insight into the evolution of populations and species. In this workshop, we will review the different sources of genetic material that are available in archaeological sites, concerning vegetal and animal genomes, humans being included under the latter rubric.
During the workshop, classical aspects of ancient-DNA studies will be discussed, from the development of on-site genetic analyses to specific laboratory protocols and the estimation of population genetics statistics using ancient, low-coverage genomes. A variety of data sources will be reviewed, including ancient microbiome data. Thus, this workshop will cover up-to-date protocols in palaeogenetic analyses and their contributions to archaeological or historical investigations.

December, 9

WORKSHOP *Ancient DNA*
Ancient DNA: from archaeological sites to genome analyses
Organized by Celine Bon and Jean-Marc Elalouf

10:00 am – 1:30 pm *Talks of the morning in the following order:*

Celine BON
General overview of the aims and themes of the workshop
Jean-Marc ELALOUF
Validation of a portable system for ancient DNA analysis on archeological sites
Lasse VINNER
Comparison of target enrichment methods for ancient human samples
Andrew E. BENNET
Ancient DNA bares its teeth: What ancient dental calculus can tell us about past populations
E.A.MATISOO-SMITH
Mitochondrial Genomes of Ancient Phoenicians
PAGANI Luca
Genome-wide allele frequency estimates from population level ultra-low coverage aDNA samples.
Etienne GUICHARD
Impact of non-LTR retrotransposons in the differentiation and evolution of the Homo genus

1:30 pm – 2:30 pm *Lunch at the Trocadéro Business Centre*
(5 minutes walk -- included in the registration fee)

2:45 pm – 3:30 pm Flash presentations concerning relevant POSTERS
(5' talk) Antony HERZIG *Page 31*
Human Population Isolates: Challenges in Phasing and Imputation
(5' talk) Samantha BRUNEL *Page 28*
Genetic diversity during the Neolithic: what about France?
(5' talk) Neus ISERN *Page 32*
Ancient DNA and the spread of the Neolithic in Europe
(5' talk) Aurore MONNEREAU *Page 34*
Palaeogenetic analysis of Bronze Age/Iron Age transition in Southern Central Asia
(5' talk) Patrizia SERVENTI *Page 38*
Iron Age Italic population genetics: the Piceni from Novilara (8th-7th century BC)

END

WORKSHOP 3 *Ancient DNA*
Abstracts in alphabetical order

TALK WORKSHOP ANCIENT DNA

ANCIENT DNA BARES ITS TEETH: WHAT ANCIENT DENTAL CALCULUS CAN TELL US ABOUT PAST POPULATIONS?

BENNET Andrew E., Olivier Gorge, Samantha Brunel, Thierry Grange, Eva-Maria Geigl, Melanie Pruvost

Jacques Monod Institute, Paris, France

The discovery of well-preserved DNA in dental calculus of ancient humans has opened a rich frontier in the reconstruction of past oral microbiomes. This information has been used to trace the evolution of oral pathogens, origins of antibiotic resistance, and to begin to understand the impact regional variation and dietary practices have had on our oral microbiota over the millennia.
We present a comparative study of metagenomic data from dental calculus recovered from multiple archeological sites in France at various time points from the Neolithic to Middle Ages. We will also discuss the special insights and limitations that accompany this new perspective in the study of past populations.

TALK WORKSHOP ANCIENT DNA

VALIDATION OF A PORTABLE SYSTEM FOR ANCIENT DNA ANALYSIS ON ARCHEOLOGICAL SITES

ELALOUF Jean-Marc[1, 2], Mélanie Flaender[3], Delphine Plaire[1,2], Josie Lambourdiere[5], Jérôme Ventosa[3], Remco Den Dulk[3], Anne-Gaëlle Bourdat[3], Jean-Baptiste Mallye[4], Myriam Boudadi-Maligne[4]
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Mobile devices for DNA analysis have been used for medical diagnosis, detection of plant or animal pathogens and biodefense research, but no attempt has been made to take benefits of such systems for ancient DNA analysis. In this study, we evaluated the potential of microfluidic technology to analyze the DNA content of samples recovered during an archeological excavation campaign. The mobile microfluidic devices were set up 20-m away from the cave site and consisted in an automated system for sample gridding, and in a platform for real-time PCR analysis. DNA detection was carried out using species-specific TaqMan probes targeting mitochondrial DNA fragments. The whole process, from sample collection in the cave to DNA analysis, was completed in three hours. This new approach is referred to as real-time molecular archeology and was successfully used to analyze carnivore remains from Aurignacian (> 30,000-year-old) layers of the Maldidier cave (Dordogne, France). Results of these studies will be presented, and the application of the method to a variety of species from different sources (freshly excavated material, museum collections) will be discussed.

TALK WORKSHOP ANCIENT DNA

IMPACT OF NON-LTR RETROTRANSPOSONS IN THE DIFFERENTIATION AND EVOLUTION OF THE *HOMO* GENUS

GUICHARD Etienne*1, Lucia Abitante1, Margherita Musella1, Jimmy Caroli2, Valentina Peona1, Marco Ricci1, Davide Pettener1, Cristian Taccioli3, Alessio Boattini1

1 Department of Biological, Geological and Environmental Sciences – University of Bologna, Bologna (Italy)
2 Department of Life Sciences, University of Modena and Reggio Emilia, Modena (Italy)
3 MA.P.S Department, University of Padova, Padova (Italy)
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Transposable Elements (TEs) have generated at least 46 % of the human genomic material. Although these elements have often been dismissed as "selfish", "parasites" or simply "junk", the advent of whole genome DNA sequencing, in conjunction with molecular genetic, biochemical, genomic and functional studies, is revealing that TEs are biologically important components of eukaryote genomes. In modern humans, only some TE subfamilies of the non LTR-retrotransposon sub-class have recently been active. These elements often contain internal promoters and Transcription Factor Binding Sites, are able to drive adjacent gene expression, can produce alternative transcripts for existing genes and have generated both new genes and pseudogenes. These characteristics make them one of the primary sources for genomic mutations and variability.
Despite all the evidence, the effects and implications of retrotransposon activity throughout the evolution of the human lineage are still understudied.
This study aims at the identification of the role of such retrotransposons in the differentiation and evolution of the genus *Homo*, by comparing insertions in the genome of modern humans with those of our closest extinct relatives, Neanderthals and Denisovans.
Because of the fragmentation of Ancient DNA (as well as the reads length limits implicit in NGS sequencing), plus the inability to work with assembled genomes due to the difficulties that occur while aligning repetitive genomic regions, a new *in silico* methodology for identifying species-specific insertions was developed.
The procedure hereby presented only relies on the BLAST+ package and custom R scripts; it operates on a modern reference genome and the raw reads of archaic DNA sequencing. Briefly, the retrotransposons' 3' ends and flanking sequences are identified in both the compared genomes, then the putative species-specific insertions 3' portions are isolated by comparing the flanking sites in the two species. All the putative species-specific insertions are confirmed via the identification of the empty site in the other species' genome, which in turn allows finding the 5' portion of the insertion. Only insertions of which both 3' and 5' portions are present and have confirmed empty (pre-insertion) sites in the other species' genome are kept.
The precise annotation of the species-specific retrotransposon insertions, the characterization of their genomic surroundings and the comparison of the site's activity and functionality both with and without the inserted elements further clarifies the impact and role of retrotransposons in recent *Homo* evolution.

TALK WORKSHOP ANCIENT DNA

MITOCHONDRIAL GENOMES OF ANCIENT PHOENICIANS

MATISOO-SMITH E.A., A.L. Gosling, J. Boocock, Y. Kurumilian, M. Guirguis, B. Costa, W. Khalil and P.A. Zalloua.

Various affiliations

Previous research, based on DNA variation in modern populations associated with Phoenician influence, identified a number of Y chromosome STR markers that were likely to have been spread across the Mediterranean as a result of Phoenician trade networks. We now have the unique opportunity to study the genetic makeup of the original Phoenicians themselves through aDNA analyses of ancient Phoenician burials. Here we present ancient mitochondrial genome data from Phoenician and Punic tooth and bone samples collected from key archaeological sites from across the Mediterranean region including Lebanon, Sardinia, Spain and Tunisia.

We investigate the patterns of genetic variation of the Phoenicians to identify their origins and track their settlement patterns in the Mediterranean and possibly beyond.

TALK WORKSHOP ANCIENT DNA

GENOME-WIDE ALLELE FREQUENCY ESTIMATES FROM POPULATION LEVEL ULTRA-LOW COVERAGE ADNA SAMPLES

PAGANI Luca(1), SCHEIB Christiana L (2), CESSFORD Craig(2), ROBB John E(2), KIVISILD Toomas(2)

(1) *Estonian Biocentre, Tartu, Estonia*
(2) *Department of Archaeology and Anthropology, University of Cambridge, Cambridge, United Kingdom*

INTRODUCTION: Recent improvements in DNA extraction techniques from ancient human remains have dramatically expanded the availability of starting material for population level genetic studies. However sequencing costs remain the main limiting factor shaping the balance between number of samples and sequencing depth in any such study design. Consequently, population level studies are often characterized by low or ultra-low coverage sequences, invariably affecting the quality of the obtained genotype calls.
METHOD: Here we introduce an approach to obtain population level allele frequency estimates a pool of 29 ultra-low (0.1-1x) coverage samples. Particularly we focus on two British populations from the same geographic site, before and after the 14th century Plague epidemic. Additionally we down-sample modern sequence data to assess how reads coming from multiple individuals from the same population can be combined to form a "chimeric" genome, representative of an average good-quality individual from that population.
RESULTS: The population level allele frequencies hence estimated, can be used to detect selective sweeps occurred during the Plague epidemic and, compared with the modern GBR samples (The 1000 Genomes Project Consortium 2015) inform us on putative adaptive responses to pathogens. We find putative signs of differentiation on the TLR genes, involved in the immune response and reported to have played a role in the genetic adaptation to Plague (Laayouni et al. 2014). Furthermore we show that mosaic individuals made of random alleles from a given population can serve as PCA proxies for the population itself when data is too scarce to plot individual samples.

The 1000 Genomes Project Consortium 2015. A global reference for human genetic variation. Nature, 526, 68-74.
Laayouni, H. et al. 2014. Convergent evolution in European and Rroma populations reveals pressure exerted by plague on Toll-like receptors. PNAS 111, 2668-2673.

TALK WORKSHOP ANCIENT DNA

COMPARISON OF TARGET ENRICHMENT METHODS FOR ANCIENT HUMAN SAMPLES

VINNER Lasse, Victor Moreno-Mayar, Morten Rasmussen, Ludovic Orlando, Eske Willerslev

Centre for GeoGenetics, Natural History Museum, Copenhagen, Denmark.

High sequencing depth is required to call genotypes that are a prerequisite for various sequence analysis methods that compare allele frequencies. Low endogenous DNA content in most ancient human samples has so far cost prohibited generation of high sequencing depth of coverage by shotgun sequencing. Only relatively few human genomes have been sequenced to high depth. Target enrichment by hybridisation capture is one strategy to improve the efficiency of sequencing while reducing the sequencing costs. For modern samples the method allows for sequencing of particular genomic regions. However, capture reaction conditions are affected by the short average length of DNA fragments in most ancient samples. In this study we compared commercially available capture methods using ancient human samples through space and time.

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