Abstract Book

SMBE Satellite Meeting

29-31 July 2024
Trinity College Dublin

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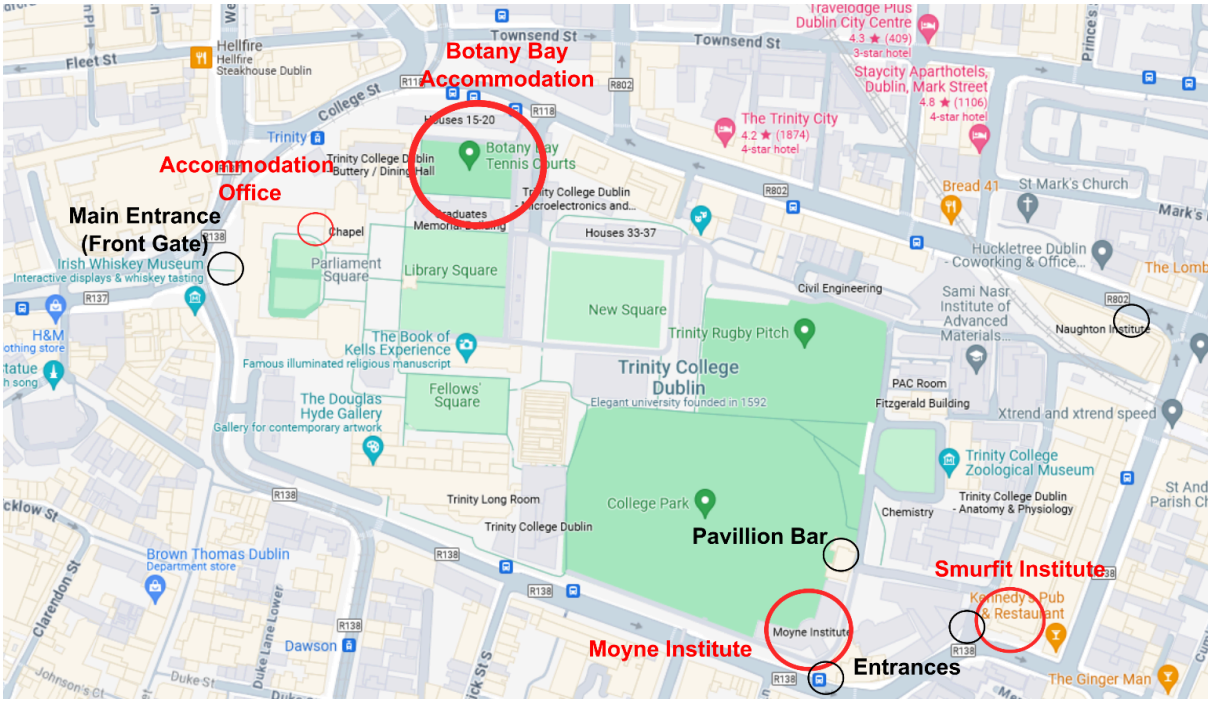
Venue Information



More details can be found in the Info Package

Programme

Day	Time	Session	Location	Abstract	Presenter/Notes	Type of Talk
29/07/2024	11:00-15:15	Registration Desk	Smurfit Atrium	-	Refreshments provided	-
	15:15-15:30	Organizers Welcome	Moyne Theater	-	-	-
	15:30-16:15	Plenary Speaker	Moyne Theater	PL1	Lara Cassidy	35+10
	16:15-16:30	S1) Humans 1	Moyne Theater	H1-01	Théo Cavinato	12+3
	16:30-16:45	S1) Humans 1	Moyne Theater	H1-02	Rui Martiniano	12+3
	16:45-17:00	S1) Humans 1	Moyne Theater	H1-03	Iseult Jackson	12+3
	17:00-17:15	S1) Humans 1	Moyne Theater	H1-04	Davide Marnetto	12+3
	17:15-17:22	S1) Humans 1	Moyne Theater	H1-05	Nicola Rambaldi Migliore	5+2
	17:23-17:30	S1) Humans 1	Moyne Theater	H1-06	Viridiana Villa Islas	5+2
	17:30-18:30	Check-in Break	-	-	Free time	-
	18:30-20:30	Round Table - Networking	Smurfit Atrium	-	Refreshments and pizza provided	-
30/07/2024	9:00-9:30	Coffee and pastries	Smurfit Atrium	-	-	-
	9:30-10:15	Plenary Speaker	Moyne Theater	PL2	Leo Speidel	35+10
	10:15-10:30	S2) Methods	Moyne Theater	M1-01	Aina Colomer	12+3
	10:30-10:45	S2) Methods	Moyne Theater	M1-02	Bárbara Sousa da Mota	12+3
	10:45-11:00	S2) Methods	Moyne Theater	M1-03	Jazeps Medina Tretmanis	12+3
	11:00-11:15	S2) Methods	Moyne Theater	M1-04	Yilei Huang	12+3
	11:15-11:30	Flash Talks	Moyne Theater	-	Alacamli, Bird, Mercuri, Özkan, Rix, Silva	2
	11:30-13:00	Poster session 1 (P1)	Smurfit Atrium	-	Coffee and snacks provided	-
	13:00-14:00	Lunch Break	-	-	-	-
	14:00-14:15	S3) Animals	Moyne Theater	A1-01	Jolijn Erven	12+3
	14:15-14:30	S3) Animals	Moyne Theater	A1-02	Andy Foote	12+3
	14:30-14:45	S3) Animals	Moyne Theater	A1-03	Katia Bougiouri	12+3
	14:45-15:00	S3) Animals	Moyne Theater	A1-04	Stephen Gaughran	12+3
	15:00-15:07	S3) Animals	Moyne Theater	A1-05	Marco De Martino	5+2
	15:08-15:15	S3) Animals	Moyne Theater	A1-06	Federico Sanchez-Quinto	5+2
	15:15-16:15	Coffee Break	Smurfit Atrium	-	Coffee and snacks. Free time	-
	16:15-18:00	Round Table - Group Discussions	Smurfit Atrium	-	-	-
	18:30-late	PAV event	PAV	-	-	-
31/07/2024	9:30-10:00	Coffee and pastries	Smurfit Atrium	-	-	-
	10:00-10:45	Plenary Speaker	Moyne Theater	PL3	Ben Peter	35+10
	10:45-11:00	S4) Open Session	Moyne Theater	OS-01	Torsten Günther	12+3
	11:00-11:15	S4) Open Session	Moyne Theater	OS-02	Léo Planche	12+3
	11:15-11:30	S4) Open Session	Moyne Theater	OS-03	Natassja Brien	12+3

	11:30-11:45	S4) Open Session	Moyne Theater	OS-04	Pnina Cohen	12+3
	11:45-13:00	Poster session 2 (P2)	Smurfit Atrium	-	Coffee and snacks provided	-
	13:00-14:00	Lunch Break	Smurfit Atrium	-	-	-
	14:00-14:15	S5) Humans 2	Moyne Theater	H2-01	Caitlin Martin	12+3
	14:15-14:30	S5) Humans 2	Moyne Theater	H2-02	Alena Kushniarevich	12+3
	14:30-14:45	S5) Humans 2	Moyne Theater	H2-03	Shai Carmi	12+3
	14:45-15:00	S5) Humans 2	Moyne Theater	H2-04	David Peede	12+3
	15:00-15:07	S5) Humans 2	Moyne Theater	H2-05	Mayra Bañuelos	5+2
	15:08-15:15	S5) Humans 2	Moyne Theater	H2-06	Ruairidh Macleod	5+2
	15:15-15:22	S5) Humans 2	Moyne Theater	H2-07	Lehti Saag	5+2
	15:23-15:30	S5) Humans 2	Moyne Theater	H2-08	Sandra Penske	5+2
	15:30-16:00	Coffee Break	Moyne Atrium	-	Coffee and snacks provided	-
	16:00-17:30	Panel Discussion	Moyne Theater	-	-	-
	17:30-18:00	Closing Cerimony	Moyne Theater	-	-	-

Abstracts

Plenary speakers

PLENARY 1

Dr. Lara Cassidy

Trinity College Dublin, Ireland

Tracing population history with haplotypes on the Atlantic Edge

This talk focuses on the ancient genomics of northwest Europe. I examine two key features of human societies - migration and kinship - through the lens of haplotypic analyses. Through case studies, I show how fine-grained dissection of genetic ancestry can allow us to better appreciate the complexity of demographic transitions on the islands. Patterns of recent genetic relatedness and inbreeding are also explored, both within and between communities, which inform on marriage practices and the shaping of group identities.

PLENARY 2

Dr. Leo Speidel

UCL Genetics Institute and the Francis Crick Institute, London, UK

High-resolution ancestry reconstruction using joint genealogies of ancient and modern humans

Over the past decade, ancient DNA has revealed a rich and dynamic genetic past across many species and humans in particular. Today, large cohorts of high-quality ancient genomes are becoming available. We have a unique opportunity to develop the tools that can leverage these genomes to paint increasingly high-resolution images of our shared genetic past. A powerful technique for this goal is the reconstruction of joint genealogical trees that relate the genomes of modern and ancient individuals through their shared ancestors back in time. In this talk, I will introduce how we can now infer such trees and how demographic change, migration and mixtures, selection, mutation rate changes and other evolutionary forces are implicated in these. These may range from key events in the deeper human past to fine-scale ancestry transformations in a nation's recent past. In particular, I will introduce Twigstats, a new approach to leverage this information for fine-scale time-stratified ancestry inference. Twigstats boosts f-statistics and can improve statistical power by an order of magnitude by focusing on coalescences in recent times, while remaining unbiased by population-specific drift. Applied to >1000 available ancient genomes focussed on Northern Europe, we reveal waves of mobility during the early historical period in Europe.

PLENARY 3

Dr. Benjamin Peter

Max Planck Institute for Evolutionary Anthropology, Germany

Neandertal ancestry through time: Insights from genomes of ancient and present-day humans

Gene flow from Neandertals has shaped the landscape of genetic and phenotypic variation in modern humans. We present a new method to identify the location and size of introgressed Neandertal ancestry segments from low-coverage data, and apply it to more than 300 genomes spanning the last 50,000 years. We study how Neandertal ancestry is shared among individuals to infer the time and duration of the Neandertal gene flow. We find the correlation of Neandertal segment locations across individuals and their divergence to sequenced Neandertals, both support a model of single major Neandertal gene flow. Our catalog of introgressed segments through time confirms that most natural selection–positive and negative–on Neandertal ancestry variants occurred immediately after the gene flow, and provides new insights into how the contact with Neandertals shaped human origins and adaptation.

Abstracts

Oral Presentations

Abstract: H1-01
Solving ambiguous genealogical structures in ancient pedigrees using IBD detection

Speaker: Théo Cavinato
Graduate Student (PhD, Masters)
University of Lausanne, Switzerland

Kinship inference between individuals from an archeological site allows pedigree reconstruction and improves our knowledge on past social structures. However, reconstructing a pedigree can be challenging: kinship inference methods often only return the degree of relatedness between individuals, which could be explained by multiple underlying genealogical structures. For instance, an avuncular pair (uncle/aunt - niece/nephew), a half-sibling pair, or a grand-parent grand-child pair would have the same degree of relatedness, but distinct pedigrees. Recent improvements in the detection of Identical-By-Descent (IBD) segments between ancient genomes now allow identifying which part of the genome is shared between two ancient relatives. In this work, we developed a method that leverages the analysis of IBD segments to solve complex genealogical structures. Briefly, in cases where siblings show a 2nd-degree relatedness with a third individual, (i) we first infer shared IBD between the siblings and the third individual and (ii) then verify which of the third individual's haplotypes contain the detected IBD segments. Regions where the third individual shares IBD segments with the siblings on both haplotypes reveals that they are avuncular, while their absence reveals another relatedness. We benchmarked our approach on simulated low coverage genomes and we applied it to the case study of the Koszyce archeological site to correctly place a 7-year-old child in a reconstructed genealogy.

Abstract: H1-02
Ancient genomes illuminate Eastern Arabian population history and adaptation against malaria

Speaker: Rui Martiniano
Group Leader
Liverpool John Moores University, UK

The harsh climate of Arabia has posed challenges in generating ancient DNA from the region, hindering the direct examination of ancient genomes for understanding the demographic processes that shaped Arabian populations. In this study, we report whole genome sequence data obtained from four Tylos-period individuals from Bahrain. Their genetic ancestry can be modelled as a mixture of sources from ancient Anatolia, Levant and Iran/Caucasus, with variation between individuals suggesting population heterogeneity in Bahrain before the onset of Islam. We identify the G6PD Mediterranean mutation associated with malaria-resistance in three out of four ancient Bahraini samples and estimate that it rose in frequency in Eastern Arabia from 5-6 kya onwards, around the time agriculture appeared in the region. Our study characterises the genetic composition of ancient Arabians, shedding light on the population history of Bahrain and demonstrating the feasibility of aDNA studies in the region.

Abstract: H1-03
Disease and Demography in Medieval Croatia and Slovenia

Speaker: Iseult Jackson
Graduate Student (PhD, Masters)
Trinity College Dublin, Ireland

Excavations of the graveyard in Kranj (Slovenia) yielded a number of early medieval human remains with an unusual joint phenotype. The observed pathology included irregular, lytic lesions, no periosteal reaction, no new bone formation and the formation of subchondral cysts, some of which were calcified. Sex bias towards females and a wide age range of affected individuals was also observed. These features were inconsistent with any known joint condition, so a subset of the 50 individuals identified with these lesions were sampled for ancient genomic sequencing. In parallel, contemporaneous individuals from Croatian excavations were sequenced in order to act as controls for genomic analyses: this also allowed for population genomic analysis of this dataset. This dataset provides a snapshot in time during the early medieval period in the Balkans, and allows us to see the genetic diversity of this region reaching back a thousand years. Analysis of autosomal and X chromosome IBD segments allows us to reconstruct pedigrees and assess the evidence for a genetic basis of disease. Here we present genomic and metagenomic analyses of Kranj, giving us new hypotheses for the aetiology of this joint phenotype.

Abstract: H1-04
Revenant gene regulatory profiles from high quality ancient genomes

Speaker: Davide Marnetto
Postdoc
University of Turin, Italy

Learning methodologies that leverage large-scale training data potentially allow to predict molecular data-points that would otherwise be lost to time. However, the reliability of these predictions depends on how well the training and discovery datasets match. To date, to infer gene expression in ancient genomes, researchers had to tackle a low genotyping quality and density in the discovery dataset, with 700k to 1024k haploid calls available in ancient datasets under best scenarios. Relying on the recent publication of 1664 phased and imputed ancient genomes we now have the opportunity to predict gene expression and splicing patterns in samples spanning 10,000 years of history, largely matching the quality and completeness of the contemporary training data. We inferred gene expression and splicing profiles for these samples and a contemporary reference. We then fitted these profiles onto multivariate models including time, space, archaeologically-derived lifestyle and genomic ancestry, in order to reveal gene regulatory patterns matching chronological, environmental and genetic axes of variation. Preliminary results suggest that the high quality of the discovery dataset can yield better gene expression inferences, thus revealing yet undiscovered patterns through time and space, complemented with alternative splicing profiles. We will showcase the results and discuss the discernible transcriptomic patterns that could be explained by divergence, migration and adaptation.

Abstract: H1-05
Fine-scale genomic analyses of peculiar pre-contact burials in Panama

Speaker: Nicola Rambaldi Migliore
Postdoc
University of Pavia, Italy

We investigate ancient DNA data from two peculiar pre-contact burials in Panama. In both cases, a female skeleton, accompanied by ornamental artifacts, was found surrounded by male crania. Their arrangement stands out among the diversity of pre-Hispanic burials, both locally and regionally. Previous genetic analyses indicated no biological relationships and the radiocarbon dating spanned hundreds of years for both tombs, leading to the hypothesis of female seers buried together with skulls of prestigious enemies, used as tools in their practices. Here, we increased the genomic coverage of the primary individuals from both sites, along with six male skulls, three from each tomb, and applied haplotype-based methods. We imputed the shotgun sequence data and evaluated imputation accuracy in the context of pre-contact Indigenous American genomes. We investigated the demography and genetic histories of these individuals leveraging the higher resolution of haplotypes, to explore their connections, possibly suggesting origins from various pre-Hispanic groups of the Isthmus.

Abstract: H1-06
Benchmarking of imputation in Ancient DNA datasets of pre-Columbian individuals from Mexico

Speaker: Viridiana Villa Islas

Postdoc

International Laboratory for Human Genome Research, Universidad Nacional Autónoma de México (UNAM), Querétaro, México

Imputation for ancient individuals in the Americas has been assessed using genomes with relatively high coverage (>10x) and downsampling to lower depths, with results suggesting that imputation is reliable for aDNA datasets with > 0.5x genome coverage, for variants with MAF >5%. Here, we tested the reliability of imputation on lower depth genomes (<4.7X) from pre-Columbian individuals from Mexico. We evaluated the influence of biases introduced during imputation of these low coverage pre-Columbian genomes for which there is not a good representation of Native American individuals in the reference panel. First, we used one genomic dataset of a pre-Columbian (671-867 CE) individual from Mexico (2417Q) with a coverage of 4.7x , and down-sampled it to 2x, 1x, 0.5x, 0.2x, and 0.1x. We then, imputed each down-sampled genome at 77.8 million biallelic SNPs using the 1000 Genomes Project haplotype dataset as the reference panel. The evaluation of concordance between the variants observed in the original dataset and the imputed down-sampled genomes were low (0.67-0.72). Next steps of the project involve testing the effect of different parameters that could be influencing the imputation quality (e.g. trimming or soft clipping of reads, genotype likelihoods) and evaluation of concordance (e.g. in function of the minor allele frequency). Also, the imputed down-sampled genomes will be assessed to observe whether they conserve the same population structure as the original data set.

Abstract: M1-01
Evaluating allele frequency trajectory and selection coefficient estimates from genealogies including ancient DNA

Speaker: Aina Colomer
Graduate Student (PhD, Masters)
Universitat Autònoma de Barcelona, Spain

During the dispersal across continents, humans have faced vastly different environments, pathogenic exposure, and technological innovations. Yet, a question still unsolved is the extent to which selection has played a role in shaping our genomes across different time periods of human evolution. The sequencing of increasingly large ancient DNA cohorts from single populations has enabled the inference of allele frequency trajectories and the associated selection coefficients and has provided new opportunities to study our genetic past. Recently, new methods for inferring genealogies jointly for modern and ancient genomes - such as Relate, tsinfer, and ARGneedle -, have made it possible to fully leverage haplotype information for the inference of selection, including to infer allele frequency trajectories and selection coefficients. Here, we present a framework to simulate genomes undergoing positive selection that allows the sampling of ancient DNA. We benchmark several new techniques for inferring selection on this data, with and without genealogies and with varying density of ancient genomes, under different selection regimes, showing that aDNA substantially improves selection estimates derived from genealogies.

Abstract: M1-02
Imputing ancient human genomes

Speaker: Bárbara Sousa da Mota
Graduate Student (PhD, Masters)
University of Lausanne, Switzerland

Genotype imputation is used to infer genotypes when there is missing data. It therefore holds great potential for ancient DNA (aDNA), given the prevalence of DNA fragmentation, postmortem damage (PMD) and microbial contamination that lead to low genome coverage. Here, we tested this potential by performing downsampling and imputation experiments of high-coverage ancient human genomes to assess imputation accuracy using different reference panels and benchmark against different degrees of PMD and contamination. Firstly, we found that common variants can be accurately imputed (minor allele frequency, MAF>5%) for genomes >0.5x using the 1000 Genomes (1KG) reference panel. Secondly, rare variant imputation accuracy by GLIMPSE2 massively improved by using the larger 150K whole-genome sequencing (WGS) UK Biobank (UKB) as reference panel, with equivalent results for a 0.25x genome compared to a 1x genome when using the 1KG panel. Thirdly, we showed that imputation (1) is robust to modest amounts of PMD by comparing a PMD-agnostic genotype caller with one that takes it into account prior to imputing, but (2) breaks down for contamination rates >5%. In summary, our results reinforce imputation as a reliable approach that will allow us to unearth a wealth of information on human history and evolution that has hitherto remained obscure.

Abstract: M1-03
Comprehensive Simulation of Ancient Datasets

Speaker: Jazeps Medina Tretmanis
Graduate Student (PhD, Masters)
Brown University, USA

The availability of larger and higher quality datasets has helped establish paleogenomics as a crucial field for the continued exploration of human evolution. However, the comparatively small amount of data available still remains one of the main challenges for the field. The use of simulated data provides a partial, but essential, solution to the low availability of real ancient data. Simulated ancient DNA (aDNA) has been used to train and test new population genetics methods. As more methods to analyze aDNA are developed it is crucial to be able to simulate aDNA to benchmark these methods. Missing data, contamination, deamination, etc. all introduce additional complexities when simulating aDNA. Here, we present the most thorough method for aDNA simulation, integrating the process of simulating aDNA through all stages: coalescent simulation, read and sequence simulation, sample quality specification, and variant calling. Our method provides all of this functionality through a single python package installation. We show how our method achieves better runtime and memory footprint performance for both sequence and read simulation when compared to popular methods for these tasks, allowing for large-scale simulations of thousands of ancient individuals. We also show how these aDNA simulations can be leveraged to measure the error introduced by using ancient data for various analyses, namely imputation, haplotype estimation, and population structure reconstruction.

Abstract: M1-04
Detection of Identity-by-descent (IBD) segments in ancient DNA and its applications

Speaker: Yilei Huang
Graduate Student (PhD, Masters)
Max Planck Institute for Evolutionary Anthropology, Germany

We have recently developed ancIBD that can robustly identify identity-by-descent(IBD) segments shared between pairs of ancient humans. These segments provide an ideal genomic signal for studying recent genealogical connections, as they must be co-inherited from a recent common ancestor, with recombination breaking the segments and thus acting as a rapid clock. Population genetic studies based on IBD segments have inferred the recent demographic history of present-day populations, including population size dynamics and geographic mobility. With the growing availability of aDNA samples, the study of IBD segments in ancient humans can offer an equally powerful opportunity. To realize this potential, we have developed a new method to utilize IBD sharing in time-transect aDNA data to better infer population size dynamics. Applying our method to individuals associated with Corded Ware Culture, we inferred a rapid population growth from about 5000BP. We also observed population expansion in Medieval Britain up to the onset of black death in 1348AD. In addition, we demonstrate that with IBD sharing on X chromosome, one can distinguish maternal and paternal relatedness, providing insights into the social structure of ancient societies.

Abstract: A1-01
Imputation of a large dataset of low-coverage ancient cattle: Exploring herd structure

Speaker: Jolijn Erven
Graduate Student (PhD, Masters)
Smurfit Institute of Genetics, Trinity College Dublin, Ireland

As of recently, genotype imputation has been shown to be a feasible method for ancient cattle. This presents the opportunity to look at haplotype-focused and genealogical methods on low-coverage ancient cattle. Here, we employ these methods to investigate herd structure within settlements and potential movement of cattle between settlements in Northwest Europe. To do so, we sequenced 17 Neolithic cattle from 2 archaeological sites (Swifterbant and Schipluiden) from the Netherlands and imputed in total 165 ancient cattle. Our analyses suggest Identity-By-Descent (IBD) sharing patterns that are indicative of close relatives (1st to 3rd degree) within settlements but this is not universal for most settlements in Europe, where varying IBD sharing patterns are observed. Within Schipluiden a cluster of individuals have IBD sharing patterns that are indicative of 4th to 7th degree relatives, while another cluster shares little to no IBD with other Schipluiden individuals. The possibility to explore more distant relatives (>8th degree) needs to be further explored, these distant relatives might give more insight in movement between settlements over time. Altogether, this indicates that genotype imputation is a viable method on a large dataset of low-coverage ancient cattle and that haplotype-focused analyses offers the opportunity to explore new questions.

Abstract: A1-02

Late Pleistocene stickleback genomes from bones and sediments reveal the chronology of freshwater adaptation

Speaker: Andy Foote
Group Leader
University of Oslo, Norway

Directly observing the chronology and tempo of adaptation in response to ecological change is rarely possible in natural ecosystems. Ancient DNA has been shown to be a tractable source of genome-scale data of long-dead organisms and to thereby potentially provide an understanding of the evolutionary histories of past populations. We maximise sequence coverage by extracting DNA from ~50x more sediment per sample than the majority of previous studies to achieve genotype resolution. From a time series of Late Pleistocene sediments spanning from a marine to freshwater ecosystem, we compare adaptive genotypes within haploblocks re- constructed from the environmental genomes of three-spined stickleback at key time points of this transition. We find a staggered temporal dynamic in which freshwater alleles at known loci of large effect in marine-fresh- water divergence of three-spined stickleback were already established during the brackish phase of the formation of the isolation basin. However, marine alleles were still detected across the majority of marine- freshwater divergence-associated loci, even after the complete isolation of the lake from marine ingression. Our retrospective approach to studying adaptation from environmental genomes of three-spined sticklebacks at the end of the last glacial period complements contemporary experimental approaches and highlights the untapped potential for retrospective ‘‘evolve and resequence’’ natural experiments using ancient genomes.

Abstract: A1-03
Best practices for estimating local ancestry in imputed ancient dog genomes

Speaker: Katia Bougiouri
Graduate Student (PhD, Masters)
Globe Institute, University of Copenhagen, Denmark

An individual's genome can be thought of as a mosaic of segments inherited from different ancestral lineages. Researchers can infer local ancestry in admixed populations by sampling genomes from different candidate populations as sources and using them to "paint" a set of admixed genomes of interest. This "painting" process is particularly challenging when working with ancient genomes, due to the scarcity of samples from specific locations and time periods, either pre- or post-dating the admixture event. This is further compounded by cases where the continuity of populations through time has been interrupted by admixture and population turn-overs. Using genetic simulations within the slendr framework, we have tested how local ancestry painting performs under different demographic scenarios, using two common methods. Furthermore, we assessed whether the number of source individuals may impact our findings, as well as whether it is best to use present-day sources, ancient sources, or a mixture of sources from different time periods. Based on these results, we inferred local ancestry and dated the admixture time in a set of imputed ancient dog genomes from the Mesolithic and Neolithic periods. Our study suggests it is very important to carefully assess the sources chosen for local ancestry inference in ancient samples. This is particularly vital for ancient populations with limited genomic samples, as well as for those that have complex demographic histories.

Abstract: A1-04
Temporal genomics of an extreme bottleneck in the northern elephant seal

Speaker: Stephen Gaughran
Postdoc
Princeton University (Dept. of Ecology & Evolutionary Biology), USA

In the 19th Century, the northern elephant seal (Mirounga angustirostris) was hunted so intensely that it was thought to be extinct. Despite this extreme harvesting, a small population re-appeared in the early 1900s and was given legal protection. Since then, the species has experienced almost exponential growth and is now common along the Pacific coast of North America. I sequenced genomes of archeological, museum, and modern specimens of elephant seals to reconstruct their demographic history and track their genomic evolution before, during, and after the bottleneck. Genetic diversity was clearly lost during the bottleneck, although pre-bottleneck heterozygosity was still not as high as heterozygosity observed in other abundant seal species. To assess the accuracy of genotype imputation in pre-bottleneck samples using modern references, I progressively downsampled read depth of the ancient samples and attempted genotype imputation to see if the full-coverage genotype was recovered. Finally, I used machine learning take a temporal view of natural selection on the elephant seal genome while accounting for ancient DNA damage to identify regions that show signs of ancient or ongoing positive selection. This study sheds light on the impact of extreme bottlenecks on the evolution of species and provides new ways to study selection and demography through temporal genomic sampling.

Abstract: A1-05
Shall we impute low-coverage ancient cat genomes?

Speaker: Marco De Martino
Graduate Student (PhD, Masters)
University of Rome "Tor Vergata", Italy

Shall we impute low-coverage ancient cat genomes? From wild animals to pest controllers and finally to pets, the evolutionary success of domestic cats is indisputable. However, cats are largely understudied, especially when compared to other domesticates and their wild relatives. For this reason, current hypotheses about the original places of cat domestication (the Levant and/or Egypt) and the timing of cat dispersal remain still open. Modern and ancient genomes proved to be key tools to investigate animal domestication. Recently, the optimization of low-coverage imputation algorithms laid the grounds for the application of haplotype-based methods in ancient domesticates. Yet, in cats, a significant gap is represented by the poor knowledge of present-day wild genetic variation. In fact, to date, only three F. s. lybica wildcat genomes (from Israel) are publicly available, thus hampering accurate imputation in low-coverage genomes. Here, we illustrate the analysis of 60 ancient European cat genomes providing novel insights into the dispersal of domestic cats. In addition, we provide a broader view of wildcat present-day genetic variation by reporting 15 high-coverage modern wildcat genomes from two areas so far largely unexplored, North Africa and Italy. Finally, we discuss limitations and possibilities for the adoption of haplotype-based methods in cat paleogenomics in the future.

Abstract: A1-06
Mammoth paleogenomic data from Mexico reveals a complex arrival of the Columbian mammoth to America

Speaker: Federico Sanchez-Quinto
Group Leader
LiiGH-UNAM, Mexico

The evolutionary history of mammoths has been a subject of great interest across different disciplines. The woolly mammoth and the Columbian mammoth co-habitated America towards the end of the Pleistocene. While woolly mammoth inhabited the holarctic circle , the Columbian mammoth lived from southern Canada to Costa Rica. To date, there is no genetic data from Columbian mammoth samples southern than the US. The recent discovery of megafauna remains during the construction of Mexico's new International airport provides a unique opportunity to investigate the evolutionary trajectory of the Columbian mammoth across America and its relationship to the woolly mammoth. To address that objective we extracted DNA from 83 paleontological mammoth remains, from which we obtained 63 mitochondrial genomes and autosomal data from two samples. We contrasted this data to the genetic variation of the Columbian mammoths from North America, as well as that of the woolly mammoth across the world, in order to better understand the initial arrival and migration dynamics of the Columbian mammoth into the continent. Our findings suggest that the initial settlement of the Columbian into America is more complex than previously reported and requires further investigation.

Abstract: OS-01
Adjusting genotype likelihoods for mapping bias

Speaker: Torsten Günther
Group Leader
Uppsala University, Sweden

One of the first steps of each palaeogenomic analysis is to map short reads to a reference genome, among other things, to separate endogenous DNA from environmental contamination. In most species, this linear reference genome will represent a single allele from a specific population of the species. A consequence is that DNA fragments carrying the reference allele will be more likely to map successfully, or receive higher quality scores. This makes the sequenced individual seemingly more similar to the reference population and the extent of this effect correlates with fragment size and post-mortem damage. We have previously shown that mapping ancient DNA sequencing reads to a linear reference genome can bias population genetic inference, and different filtering strategies were proposed by us and others. Here, we introduce an approach to empirically adjust genotype likelihoods (GL) for mapping bias. GLs are a powerful approach to accommodate uncertainty about genotypes as they can be directly used in downstream analysis. We simulated ancient DNA sequencing data, calculated GLs and used them as inputs for different tools for estimating ancestry proportions. The corrected GLs largely mitigate the effect of mapping bias on ancestry estimates but there seem to be software-specific biases that it cannot account for. As GLs have many other applications, including as input for imputation, the corrected GL calculation could have many applications beyond allele frequency-based methods.

Abstract: OS-02
Detecting multiple Archaic introgression using multiple reference populations

Speaker: Léo Planche
Postdoc
Université Paris Saclay, France

Modern humans admixed with archaic populations that are now extinct, Neanderthal and Denisovian are two of such populations. The date of Neanderthal admixture is estimated to be around 50,000 years ago, but this number and the context in which the admixture occurred are still debated. We are building mathematical models which could provide new insights on these questions. The study of ancient admixture can be done in two main ways, global ancestry inference, which consists of determining for an individual the percentage of its genome which comes from some particular ancestry. The second, which is the subject touched upon here, local ancestry inference (LAI) gives the parts of the genome that come from said ancestry. In HMMMix (Skov 2018), a Hidden Markov Model (HMM) using a single outgroup is built to detect between Archaic and modern ancestry. We extend this method to allow the use of multiple reference populations and the detection of more than two ancestries in a single run. We show that on top of facilitating the detection of multiple archaic introgression, it also increases the accuracy of the detection, in both simulations and real-world data. As our HMM may use multiple reference populations, it also allows the use of both modern and Archaic reference genomes simultaneously to further increase the accuracy.

Abstract: OS-03
Methylation analysis of loci related to age and bone mineral density in ancient DNA

Speaker: Natassja Brien
Graduate Student (PhD, Masters)
McMaster University, Canada

Recent improvements to ancient DNA (aDNA) methodology, including high-throughput sequencing methods, have enabled the analysis of methylation patterns. In humans, methylation patterns are established at birth, but changes can occur over the life course due to disease, stress, and trauma. Areas of the genome that are sensitive to these experiences can become hypo- or hypermethylated, causing changes to gene expression. Therefore, examining patterns of methylation in well-characterized loci can inform our understanding of the lived experience of people in the past. This research focuses on methylation patterns at specific loci associated with chronological age and bone mineral density, that have already been well-characterized in modern clinical studies. The first stage of this research is to examine methylation patterns at these loci for a population where age-at-death is known (via medical records), to characterize the relationship between methylation and chronological age. This research also has the potential to improve age-at-death estimates in bioarchaeological research, particularly in older adults where skeletal age estimation is more difficult.

Abstract: OS-04
Reconstructing our evolutionary past using sediment DNA: Best practices and Applications

Speaker: Pnina Cohen
Postdoc
Tel Aviv University, Israel

Ancient DNA (aDNA) analyses— the study of genetic material from individuals that died hundreds or thousands of years ago— have revolutionized the research in human evolutionary genetics. At most archaeological sites dated to the Middle or Late Pleistocene (780,000-12,000 years ago), no human remains have been found. However, recent studies have shown that aDNA can be recovered from archaeological sediments, even in the absence of any skeletal remains, providing an exciting new avenue to learn about our evolutionary past. Despite that, so far only a limited number of studies have successfully recovered, identified and authenticated ancient human DNA from prehistoric sediments. We focus on advancing and developing analytical frameworks to support the study of ancient human DNA from sediments. Here, we evaluate current methods for imputation, phasing and kinship analyses of genetic variants testing them for accuracy on both empirical data and on simulated datasets mimicking the characteristics of ancient sedimentary DNA, including very low genetic coverage, restricted relevant reference panels, mixture of individuals within a single sample, and the presence of non-human ancient DNA. Based on this, we propose appropriate filters and parameters for the tested software, when applied to the study of ancient human DNA from sedimentary samples.

Abstract: H2-01

From bones to biographies: paleopathology and paleogenomics reveal the history and evolution of genetic disease in Celtic populations

Speaker: Caitlin Martin
Graduate Student (PhD, Masters)
Institut Jacques Monod, Université Paris Cité, France

Thanks to advances in genomic imputation combined with ever-expanding global genome datasets, we are able to ask increasingly complex questions, even from ancient low-coverage genomes. In this case-study of French medieval populations, we exploit these advances to link paleopathological phenotype with genetic disease genotype. First, we analysed ancient genomes from a population previously identified, through paleopathology, as showing high frequencies of a specific genetic disease expressed on the skeleton. We imputed the resulting whole-genomes to assess genotypes of ancient affected and unaffected individuals at specific 'risk' SNPs derived from modern populations. These imputation results were then validated through targeted hybridization capture, to ensure the observed risk variants were not products of imputational bias nor ancient deamination. Finally, through comparison with other ancient and modern genomes, we investigated the larger haplotype background upon which these risk variants arose, to trace their prevalence from the medieval to the modern era, and to better understand their lingering genetic impact in living populations. To our knowledge, this study represents the first time that a specific genetic disease, known to disproportionately affect a modern population, has been both paleopathologically identified and genomically characterized in the ancestral population.

Abstract: H2-02
Dynamics of the genetic structure of human populations during the last two Millennia in Eastern Europe

Speaker: Alena Kushniarevich
Postdoc
Institute of Genomics, University of Tartu, Estonia

Large-scale movements of people in the last two thousand years have reshaped the cultural landscape of Eastern Europe. To study the dynamics of the genetic structure in East Europe and to better understand the relationships of cultural and genetic changes, we assembled two datasets: a) ancient low coverage shotgun genome sequences (coverage >=0,08x; N=260) from the late Iron Age, Medieval and Early Modern time Eastern Europe covering the areas of distribution of Eastern Slavic, Baltic, and Finnic tribes; b) genome-wide genotype data from modern Eastern Europeans. We imputed with QUILT the genotypes of ancient genomes using 2000 high-coverage genomes from the Estonian Biobank as a reference panel. We inferred genomic segments that are shared identical by descent (ibd) between ancient and modern individuals. Further we aim at constructing ibd-sharing networks that are region-, time- and language-specific. Our preliminary analysis demonstrates differential relatedness between the Baltic and the Finnic groups from medieval Eastern Europe at the micro-regional scale.

Abstract: H2-03
The founder event in medieval Ashkenazi Jews based on haplotype sharing in modern and ancient genomes

Speaker: Shai Carmi
Group Leader
The Hebrew University of Jerusalem, Israel

The high prevalence of severe recessive diseases in Ashkenazi Jews (AJ), noted already decades ago, hinted of a founder event. Evidence of a severe medieval founder event has accumulated more recently with the discovery of AJ-private high frequency mtDNA haplogroups and high levels of identical-by-descent (IBD) genomic sharing. Estimates based on modern data suggested a medieval reduction in effective population size to merely a few hundreds. We hypothesized that ancient DNA could clarify the timing and extent of the AJ founder event. We generated genome-wide data for 33 individuals whose remains were rescued from the medieval Jewish cemetery in Erfurt, Germany and were dated to the 14th century. Multiple lines of evidence supported a medieval founder event: a single mtDNA haplogroup shared by one-third of the individuals; detection of multiple known AJ pathogenic founder variants; and high levels of runs of homozygosity (ROH). Joint modeling of ROH in ancient genomes and IBD in modern genomes using a novel framework suggested a bottleneck of effective size about 400-1000 throughout the entire second half of the Middle Ages and that a subset of the population underwent a milder bottleneck (Waldman et al, Cell 2022). Unpublished analyses of IBD sharing between the ancient AJ genomes demonstrated very high sharing levels. Further, the IBD analysis confirmed medieval substructure in AJ previously suggested based on allele-frequency methods.

Abstract: H2-04

The MUC19 gene in Denisovans, Neanderthals, and Modern Humans: An Evolutionary History of Recurrent Introgression and Natural Selection

Speaker: David Peede
Graduate Student (PhD, Masters)
Brown University, USA

Humans carry archaic ancestry from Neanderthals and Denisovans, while the impact of Neanderthal introgression has been well-studied, less is known about the genetic contributions from Denisovans. We highlight MUC19, a gene linked to gel-forming mucin proteins affecting lacrimal and salivary glands, as a candidate for Denisovan adaptive introgression. Notably, the Denisovan-like haplotype of MUC19 is prevalent in admixed American populations, carrying six derived missense mutations that are fixed in individuals with this haplotype. Additionally, using phasing approaches we observe that late-stage Neanderthals possessed the Denisovan-like haplotype, including the six fixed mutations, indicating a complex evolutionary history of recurrent introgression.

Abstract: H2-05

Leveraging archaic and modern human genomes with machine learning approaches to elucidate the evolutionary history of our species

Speaker: Mayra Bañuelos
Graduate Student (PhD, Masters)
Brown University, USA

Archaic introgression, the interbreeding between archaic hominins and modern humans (MH), is an evolutionary process capable of introducing novel genetic variation into humans. In MH populations, archaically introgressed variants have been found to facilitate expansions and adaptations to different environments. Thus, the identification and correct classification of archaic variants in MH are central steps in understanding human evolutionary history. Currently, we have excellent methods for identifying putatively archaically introgressed haplotypes in MH populations. However, classifying these archaic segments into Neanderthal, Denisovan or other archaic hominin is often an ad-hoc process that requires a series of filtering steps, and different criteria are often applied for each study. Here, we develop a more principled statistical approach to classify archaic segments into Denisovan, Neanderthal or an unknown archaic population. Our statistical method leverages genetic affinities of putatively archaic segments in MH samples from simulated data to train a Naïve Bayes classifier. In simulated data, classification between Neanderthal and Denisovan segments in a sampled MH population produces a 95% accuracy after cross-validation. We apply this method to classify putatively introgressed segments in empirical data and explore its performance under various demographic scenarios.

Abstract: H2-06
Human demography beyond ancient genomes alone: mid-Holocene Communities at Lake Baikal

Speaker: Ruairidh Macleod
Graduate Student (PhD, Masters)
University of Cambridge, UK

Human demographic history is substantially the outcome of ecology and environment. While population genomics studies are well-equipped to answer 'how?' questions in demographic history, 'why?' questions often require different data. To elucidate causative processes in human-ecosystem interactions affecting demography at a local scale, we combined a large dataset of ancient genomes from mid-Holocene hunter-gatherer communities at Lake Baikal with an ancient environmental DNA record of ecosystem evolution in the same region. By explicitly studying associations between demography and ecosystem succession, human prehistory can be situated in community ecology. Extensive archaeological investigation of hunter-gatherer cemeteries (8000-4000BP) undertaken by the Baikal Archaeology Project furnishes extensive bioarchaeological and cultural insights into spatiotemporal variation. We analyse 200 novel shotgun-sequenced ancient humans (and apply diploid imputation) to generate a high-resolution chronology of human communities (including many insights into familial and social organisation). Combining this with an analysis of high-resolution environmental DNA sampling from three sediment cores yields insights into causative processes affecting human demography, beyond simply its inference. This has implications in particular for major archaeological questions such as the apparent middle Neolithic 'hiatus' in the region.

Abstract: H2-07

A genetic bridge over the Gulf of Finland: tracing the origin of genetic connectedness between Finns and Estonians

Speaker: Lehti Saag
Postdoc
University of Tartu, Estonia

Previous studies have suggested that the migrations associated with Uralic languages reached the circum-Baltic region by the 1st millennium BC. However, the high linguistic similarity of Finnish and Estonian points to a more recent split of the languages. We set out to study genetic relatedness of Finns and Estonians in time. First, by searching for long shared allele intervals in unphased data for >143,000 present-day Estonians, 99 Finns, and 14 imputed ancient genomes from Estonia, we found unexpectedly high levels of individual connectedness between Estonians and Finns for the last 8 centuries in contrast to their clear differentiation by allele frequencies. Next, we produced ancient DNA data from 16 additional individuals from Iron Age Estonia. Using imputed genotypes, we show that already Roman/Middle Iron Age individuals had higher levels of individual connectedness with modern Estonians and Finns than individuals from earlier time periods. However, unlike medieval genomes, they do not show region-specific connectedness within Estonia, suggesting that the local regional genetic structure in Estonia had not formed by the Middle Iron Age.

Abstract: H2-08
Kinship practices at the Early Bronze Age burial site of Leubingen in Central Germany

Speaker: Sandra Penske
Graduate Student (PhD, Masters)
Max Planck Institute for Evolutionary Anthropology, Germany

The retrieval of ancient DNA in recent years has transformed our understanding of the human past. We have learned about large-scale genetic changes which were linked to migrations of past peoples who shaped the gene pool of modern-day populations. A drastic increase in the number of genomes per study and site, and new analytical approaches allow us to address questions regarding genetic relatedness, kin and social organization, and the distribution of wealth. The Early Bronze Age (EBA) in Central Europe ~2200 BC experienced a marked change in social organization. A small number of individuals in positions of power were set apart from other, non-elite individuals by their rich graves and extensive burial constructions. To address questions about social organization and wealth distribution within a local farmer community, we generated genome-wide data from 46 individuals from the EBA burial ground of Leubingen. We reconstruct five pedigrees, each made up of close genetic kinship groups (parents and their offspring), as well as finding individuals who are not genetically related to anyone. By integrating archaeological, genetic and strontium isotope data, we identified patrilineality as the predominant kinship structure involving female exogamy. In addition, we find that individuals at Leubingen were buried with a different amount of grave goods based on their genetic sex, age at death and locality but that these factors made no difference to the types of grave goods.

Abstracts

Poster Presentations

Abstract: P1-1
Region-specific reference panel for Genotype Imputation in ancient human genomes from Eastern Europe

Presenter: Erkin Alacamli
Graduate Student (PhD, Masters)
University of Tartu, Estonia

As sequencing technologies advance, retrieval of genome-wide sequences from ancient samples rapidly grows as well. However, degraded samples lead to highly fragmented and contaminated sequences, resulting in low endogenous DNA content with low genomic coverage. Therefore, most ancient samples do not yield sufficient coverage for diploid genotype calling. Genotype imputation facilitates a way to overcome this issue by inferring the missing sites often using large reference panels. Although this sounds straightforward to use, there are several aspects on which imputation accuracy depends. This includes coverage limitation, usage of a representative (population- or region-specific) and a big enough reference panel that also represents the rare variants sufficiently. This study aims to create an enriched region-specific reference panel by merging the available high-coverage genome sequences from the Estonian Biobank, Haplotype Reference Consortium, and The Thousand Polish Genome Project to be used for genotype imputation in ancient DNA samples from Eastern Baltic region. The performance of the enriched and “single-source” panels in the genotype imputation of low-coverage genomes from various archaeological contexts and locations will be assessed. The imputed genotypes will be used in various haplotype- or allele frequency-based analyses to study the fine-scale genetic structure and its dynamics in Eastern Europe during the last two Millennia.

Abstract: P1-2
The reliability of inferred archaic segments in human genomes

Presenter: Nancy Bird
Postdoc
UCL, UK

Many or all present-day human genomes include segments of DNA inherited from archaic humans due to admixture events occurring >30,000 years ago. Several methods have been published to detect such segments. These variously require phased haplotypes, an archaic reference sequence and/or an outgroup with little related archaic introgression. Inference from these approaches have been used to (e.g.) document purifying selection of archaic segments and understand their influence on traits. However, the comparative accuracy of different methods at detecting archaic segments remains underexplored. Little is known about how genomic features correlate with assignment accuracy and the consequences of such non-random bias. We examined accuracy in detecting archaic ancestry under four widely-used approaches: Sprime, diCal, IBDMix and HMMix, and two unpublished ones. In simulations we find substantial variation across methods in their false positive and false negative rates. We also find an inconsistency across approaches in identified archaic segments in real data. For example, out of all SNPs identified as Neanderthal in any of the published methods, only 29% of these are found in more than two methods. Our benchmarking of different methods highlights the difficulties in robustly identifying archaic segments in modern human genomes. We discuss the implications for downstream analyses that make use of inferred archaic segments.

Abstract: P1-3
Museomics of Eastern African baboons provide insights on their population structure

Presenter: Matteo Caldon
Graduate Student (PhD, Masters)
University of Parma, Italy

Museomics is an emerging field of research that involves the study of genomic data obtained from specimens in museum collections. The analysis of museum samples presents the same methodological challenges characterizing ancient DNA studies but enables the examination of historical populations for which direct sampling in the wild is currently challenging or impossible. This is particularly relevant for studying baboons (genus Papio) whose diversity is currently only partially represented in modern genomic data. This study focuses on two baboon species, Papio anubis and Papio hamadryas, occurring from western to eastern Africa and extending to the western coast of the Arabian Peninsula. We combined genomic data from extant populations with data gathered from specimens conserved in two Italian Natural History Museums (Milan and Genoa). Specimens were collected in areas from Eastern Africa in the late 19th and early 20th centuries, unrepresented in modern data. Our objective is to investigate the degree of population structure present in these two species, explore their interactions and characterize their demographic dynamics.

Abstract: P1-4
Museomics of Eastern African baboons provide insights on their population structure

Presenter: Giacomo Mercuri
Graduate Student (PhD, Masters)
University of Parma, Italy

Museomics is an emerging field of research that involves the study of genomic data obtained from specimens in museum collections. The analysis of museum samples presents the same methodological challenges characterizing ancient DNA studies but enables the examination of historical populations for which direct sampling in the wild is currently challenging or impossible. This is particularly relevant for studying baboons (genus Papio) whose diversity is currently only partially represented in modern genomic data. This study focuses on two baboon species, Papio anubis and Papio hamadryas, occurring from western to eastern Africa and extending to the western coast of the Arabian Peninsula. We combined genomic data from extant populations with data gathered from specimens conserved in two Italian Natural History Museums (Milan and Genoa). Specimens were collected in areas from Eastern Africa in the late 19th and early 20th centuries, unrepresented in modern data. Our objective is to investigate the degree of population structure present in these two species, explore their interactions and characterize their demographic dynamics.

Abstract: P1-5
Archaeogenomics of Donkey Remains of Anatolia

Presenter: Mustafa Özkan
Postdoc
Middle East Technical University, Turkey

Donkeys were domesticated from African wild asses in ancient Egypt. Recent studies based on ancient DNA recovered from donkey remains confirmed their origin of domestication and laid a foundation for research on how donkeys dispersed other continents. Due to its proximity to putative domestication centers, Anatolia can be regarded as an important station in donkey dispersal. Hybrids between donkeys and wild asses can be traced back to 3rd millennium BC in Northern Syria, namely kungas. This, however, provides a dispersal date to Levant but not to Anatolia. In order to elucidate the history of the earliest donkeys of Anatolia, we sequenced 39 equid samples from Arslantepe and Kuriki Höyük from Central-East Anatolia, and generated ancient genomes of 6 individuals. These individuals were found to be in genetic vicinity of modern day donkeys in demographical analyses. One of the Arslantepe donkeys, which was excavated from a Late Chalcolithic context, suggests the arrival of donkeys into Anatolia between 3900-3400 BC, contemporary with Early and Middle Uruk cultures in Mesopotamia, and represents the oldest known genetically confirmed donkey on Anatolian landscape so far.

Abstract: P1-6
Snow Petrel Evolution and Diet through Time

Presenter: Anna Rix
Postdoc
Durham University, UK

The cold, dry environment of Antarctic results in the preservation of novel biological archives that can be used as proxy for environmental conditions. One Antarctic bird with a well-preserved record is snow petrels. As snow petrels are provisioning their young and defending their nest sites they excrete stomach oil. Over time this accumulates at nesting sites in a layered manner. From these deposits we are able to extract DNA to determine what the snow petrels have been eating over various time scales. The fish and krill content of snow petrel diet is thought to be related to the amount of sea ice present in the environment, through temperature or polynyas. We have deposits spanning from modern to radiocarbon dead. Additionally, we are using the population history of snow petrels to track sea ice as snow petrels are reliant on sea ice. We can obtain some genetic information about snow petrels directly from the deposits, but also have samples from modern birds and six ancient snow petrel chicks ranging in age from 3,000 years old to radiocarbon dead. Through the whole genome resequencing of these samples we hope to be able to illuminate areas of the genome that have undergone strong selection and determine snow petrel demography. To enable these analyses, we have constructed a high-quality snow petrel genome from a modern female. Combined these data sets will provide a unique perspective on how these birds have persevered in the past and how they will fare in the future.

Abstract: P1-7

Using temporal genomics to investigate the genetic basis of speciation and hybridization

Presenter: Eugénio Silva

Graduate Student (PhD, Masters)
BIOPOLIS-CIBIO, Portugal

Understanding how a species is formed is a major question in evolutionary biology. However, divergence and reproductive isolation are slow processes, what prevents their direct observation in real-time. Thus, most of what we know about speciation and hybridization is derived from the genetic variation segregating in contemporary populations. We aim to fill this gap by integrating ancient and modern genomic data of an iconic example of speciation: the European rabbit (Oryctolagus cuniculus). This species is native to the Iberian Peninsula where two subspecies that diverged for over 1 Mya co-exist. These subspecies were separated, but with the end of the last glacial maximum, their range expanded and a hybrid zone was created. Nonetheless, reproductive barriers exist between subspecies and hybrids are strongly negatively selected. This species is also commonly found in Iberian archaeological sites, due its close relationship with humans. The persistent reproductive barriers together with their ubiquitous archaeological presence provide an opportunity to study incipient stages of population divergence using ancient DNA. We will generate and analyze exomes of archaeological rabbit samples across the species distribution range over the last 40,000 years. By investigating genetic changes that have accumulated between these two subspecies across time and space, we will shed light on the underlying mechanisms of reproductive isolation that lead to speciation.

Abstract: P1-8
Genetic Time Travel in Philippine Fishes

Presenter: John Whalen
Graduate Student (PhD, Masters)
Old Dominion University, USA

The Philippines is the global epicenter of marine fish biodiversity. The Philippines also ranks highest globally for threats to marine biodiversity and coral reefs. Therefore, identifying the impacts of human activity, such as overexploitation and habitat degradation, on the adaptive potential of fishes in the Philippines is of growing concern. The goal of this study is to identify what genetic level changes have occurred over the past century in the Philippines. The USS Albatross research vessel conducted sampling expeditions across the Philippines between 1907 and 1910. Contemporary duplicate collections were made to sample the same populations. Shotgun capture and low coverage whole genome sequencing was used to investigate centennial genetic changes. We observed a loss of genetic diversity (heterozygosity, theta, allelic richness) in several species. Site frequency spectrum models and positive temporal shifts in Tajima's D suggest recent population bottleneck events in several species. This study is unique because it successfully sequenced 100-year-old specimens using two different methods. Furthermore, the results indicate that genetic diversity is declining in marine fishes in the tropics at a rate similar to other temperate vertebrates. Differences in declines between species may be due to connectivity differences or localized fisheries and habitat impacts. Our findings emphasize the urgent need to protect the adaptive potential of marine fishes in the Philippines.

Abstract: P2-1
Museomics are enabling conservation of the Galapagos giant tortoises

Presenter: Evelyn Jensen
Group Leader
Newcastle University, UK

The Galapagos giant tortoises are charismatic emblems of evolutionary biology, and conservation icons. Despite decades of study, new discoveries about them are still being made, thanks in large part to specimens in museum collections. In this talk I will present recent developments on the conservation genetics of Galapagos tortoises, including how genomes from museum specimens are playing a critical role in managing a one-of-a-kind captive breeding program, and the exciting discovery of new, undescribed extinct lineages.

Abstract: P2-2

Reexamining Beachy Head Woman: using aDNA techniques to clarify the ancestry of a Roman female previously deemed Britain's oldest sub-Saharan migrant

Presenter: William Marsh

Postdoc
Natural History Museum, UK

The application of ancient DNA techniques offer opportunities to further our understanding of the demography of ancient populations. This study revisits a a female skeleton excavated from the south coast of the UK dating to the Roman occupation of Britain. Initial craniometric analysis suggested this individual was sub-Saharan in origin and thus represented the earliest sub-Saharan migrant into the UK, a designation that led to widespread public interest. However, aDNA analysis carried out in the late 2010s hinted at a continental European origin for the individual, although insights were limited given poor DNA preservation. Here, through the employment of a novel aDNA-specific hybridisation capture method, we overcame the challenges posed by poor DNA preservation. The individual exhibits no evidence of a sub-Saharan origin, and instead is most similar genetically to modern Europeans. Comparative analysis with ancient samples from the Roman period reveals a close genetic affinity to Iron Age Northern Europeans, with phenotypic analysis suggesting the individual was most likely light skinned, light haired, and blue eyed. These genetic results indicate a local British origin for this individual, with little support for the initial designation of this individual as sub-Saharan African. The study highlights the utility of ancient DNA in untangling historic population movement, and the importance of using a multi proxy approach when examining skeletal material of unknown origin.

Abstract: P2-3
Exploring Drought Effects on Elephant Gene Pool through Ancient DNA Analysis in Kenya

Presenter: Rodney Omukuti
Graduate Student (PhD, Masters)
Pwani University, Kilifi, Kenya

Climate change-induced droughts pose significant threats to wildlife populations, particularly in arid regions like Tsavo National Park, Kenya. The elephant population in Tsavo has been significantly impacted by drought-related deaths, raising concerns about the long-term genetic consequences for this iconic species. This study proposes to utilize ancient DNA analysis to investigate the effect of drought-related deaths on the overall gene pool of elephants in Tsavo National Park. Ancient DNA will extracted from elephant bones collected from different time periods and locations within the park. Through genetic analysis, we aim to assess changes in genetic diversity, inbreeding levels, and population structure over time. By elucidating the genetic impacts of drought-related deaths, our findings will inform evidence-based conservation strategies for mitigating the effects of climate change on elephant populations in Tsavo National Park and similar ecosystems.

Abstract: P2-4
Archeogenomic insights on the Late-Antique italian society

Presenter: Flavia Risi
Graduate Student (PhD, Masters)
Sapienza University of Rome, Italy

Late Antiquity (LA) is the historical period between the decline of the Western Roman Empire (WRE) and the onset of the Medieval era. It was characterized in the Western regions by great political instability, socio-economic changes, and massive migrations of mostly Germanic people. As the focal center of the Roman Empire, and given its strategic geographic position throughout European history, the Italian peninsula is an area of great importance to shed light on the dynamics and effects of such a major transitional period. My research is focusing on the genetic landscape of Italy, through the analysis of about 30 samples from two LA burial areas from central Italy (Fossombrone, 5th-6th cc. CE and Castel Trosino, 6th-8th cc. CE). Notably, preliminary analyses on the first one reveal a genetic shift towards Near Eastern populations, possibly suggesting the enduring of a great mobility from the Eastern provinces even after the decline of WRE. Nevertheless, it is interesting to understand how the society in Italy was re-shaped by the migrations of Germanic peoples; in particular if any demographic collapse occurred and new social structures based on kin groups or strongly subdivided by social classes were established. Innovative approaches are crucial in this regard, such as aDNA imputation and the analysis of identity-by-descent segments as well as the estimate of the inbreeding levels to uncover close and distant genetic relationships and make inferences about the social ties.

Abstract: P2-5

Estonian Medieval elite: who were they? A study to investigate the ancestry of the ruling elite in Medieval Estonia

Presenter: Stefania Sasso
Graduate Student (PhD, Masters)
Institute of Genomics, University of Tartu, Estonia

The 13th century in the Eastern Baltic was the period of crusades, aiming to spread Christianity in the region of historical Livonia, overlapping with the current territories of Southern Estonia and Northern Latvia (Livonian crusade). By 1227, the area of modern Estonia was conquered. This meant the beginning of a new era. The ruling elite of mostly German origin brought along new rules and traditions, and the social stratification of the population. These changes included new burial practices: elite people were buried inside the churches, while local people were buried in the churchyard or in rural village cemeteries. Was this difference only cultural or also genetic? To study if these changes had an effect mostly on culture or did they change also the population, we collected and sequenced 58 medieval individuals (1407-1640 CE) buried inside 8 different churches, six of them located in Tartu and two in Tallinn. We also included 230 individuals from rural sites for comparison. PCA reveals that studied individuals form two different groups: “local” group overlapping with modern Estonians, and “elite”group shifted towards modern Western European (WE) populations. The low-coverage data of the subsample of medieval individuals (n = 31) will be imputed using a local reference panel (N=2000). Imputed genotypes will allow us to perform haplotype-based analyses to investigate the genetic ancestries of medieval individuals.

Abstract: P2-6
Beyond SNPs: Investigating Deletions in Ancient Sheep Genomes

Presenter: Muhammed Sıddık Kılıç
Graduate Student (PhD, Masters)
Middle East Technical University, Turkey

Ancient genome analyses have predominantly focused on single nucleotide polymorphisms (SNPs). However, many copy number variants (CNVs) play a crucial role in both diseases and evolutionary adaptation. In this study, we will be studying >1kb deletions in ancient and modern-day sheep genomes, and how dispersal and bottlenecks may have shaped temporal and geographic variation in deletion frequencies. We start by de novo ascertaining deletions from 125 high coverage (>10x) wild and domestic modern sheep genomes. Using the CONGA algorithm, designed to call deletions from low-coverage genomes, we will then genotype these deletions across 9 (>0.5x) ancient sheep genomes sourced from various regions of Eurasia and dated between c.9000 BP to c.2000 BP, as well as modern-day mouflon genomes. In addition to examining the frequency of these deletions in time and across regions, we will also investigate the functions of the genes affected by these deletions. We expect our results to expand our insight into sheep evolution, including the effects of bottlenecks and intense breeding on large deletions.

Abstract: P2-7

Quantifying genomic variation before and after decades of decline in the highly gregarious Tricolored blackbirds of North America

Presenter: Megha Srigyan
Graduate Student (PhD, Masters)
University of California, Santa Cruz, USA

Temporal sampling of species that have undergone anthropogenic declines are useful to understand past genetic variation and inform conservation efforts. Tricolored blackbirds, a highly gregarious, near-endemic species to California, sustained massive population declines in the past century, declining from millions of individuals in the early 1900s to ~200,000 currently, representing a ~95% reduction in census size. This was primarily driven by conversion of wetlands, their preferred nesting habitats, to agricultural fields and plantations. Despite widespread habitat loss and fragmentation, modern tricolored populations have been shown to be panmictic across California. In this study, we analyze historical variation across 34 museum specimens collected in the early 20th century across breeding sites in California and compare it to that in modern populations. We show that modern tricolored blackbirds exhibit reduced genetic diversity and harbor lower effective population size than expected. However, there are no significant differences in population structure or levels of inbreeding across time. This indicates that their breeding colonies are sufficiently large to prevent strongly negative effects of bottlenecks which would be expected for declines of this magnitude. These results are important to understand determinants of long-term population persistence for highly social species of conservation concern that show potential of demographic recovery.

Abstract: P2-8
soibean: High-Resolution Taxonomic Identification of Ancient Environmental DNA Using Mitochondrial Pangenome Graphs

Presenter: Nicola Vogel
Graduate Student (PhD, Masters)
Denmarks Techical University (DTU), Denmark

Ancient environmental DNA (aeDNA) offers insights into past ecosystems, particularly when traditional fossil records are lacking. However, studying aeDNA has many challenges such as ancient damage, genomic divergence, the presence of multiple DNA sources, and sparse, fragmentary data. Additionally, a lack of reference genomes makes species identification and phylogenetic placement more difficult. Existing methods, are mostly tailored for modern DNA, anticipate high coverage and similarity to reference genomes to provide results. So far only one method adresses some idiosynchrasies of aeDNA but only supports the identification of a single-source. To overcome these limitations, we introduce soibean, a tool for species identification of aeDNA from FASTQ inputs using mitochondrial pangenomic graphs. soibean can identify multiple closely related contributing sources in an aeDNA sample and accurately place each source in their phylogenetic context. Using reconstructed ancestral states and a damage-aware maximum likelihood model, we are able to identify ancestral sources as well as estimate the contribution of each source.. Our Markov Chain Monte Carlo sampling algorithm on a phylogenetic tree makes accurate assignment possible at low coverage depths up to ~0.1X, as shown in tests on simulated data. We were able to show consistency with empirical data as well as discover previously overlooked species in published datasets.