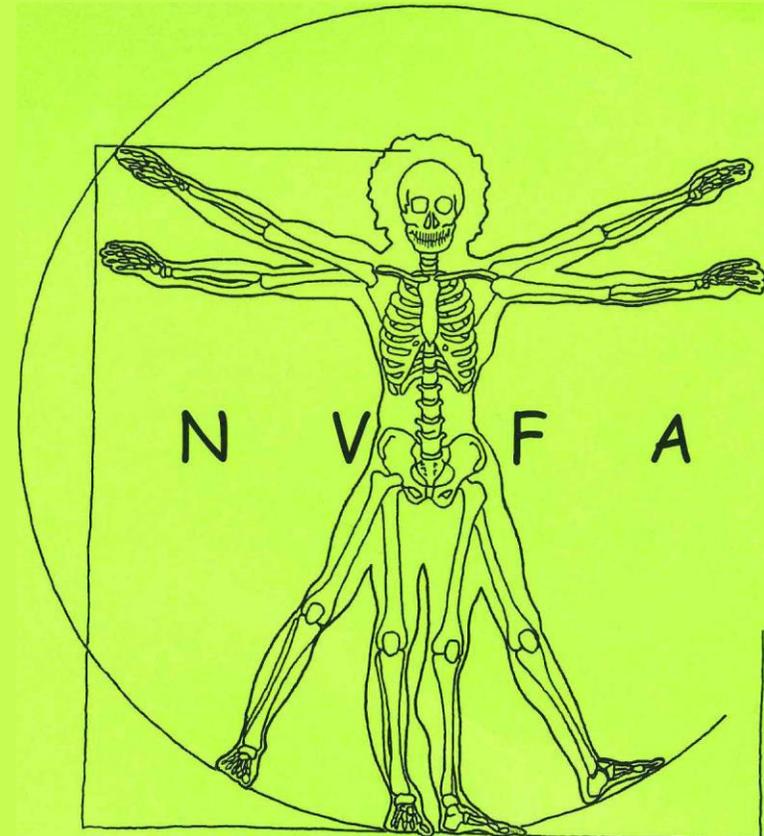


Nederlandse Vereniging voor Fysische Antropologie

DNA TODAY



Symposium in celebration of the 30th anniversary of the NVFA
30 November 2013
Programme and Abstracts

We gratefully acknowledge the support of:

Nederlandse Stichting voor Anthropobiologie



Stichting Nederlands Museum voor Anthropologie en
Praehistorie



Leids Universitair Medisch Center

Programme

- 10.30 a.m. Registration and Coffee/tea
- 11.00 a.m. Opening by Piero Giordano, chairman of the NVFA
- 11.10 a.m. Introduction to DNA
Peter de Knijff
- 11.30 a.m. Session 1: Population genetics (chair Toineke Westen)
Mark Thomas
The Co-Evolution of Lactase Persistence and Dairying
Eveline Altena
- 12.30 p.m. The Dutch genetic landscape from past to present
- 1.00-2.00 p.m. Lunch
- 2.00 p.m. Session 2: Forensic research (chair Eveline Altena)
Peter de Knijff
STR genotyping 3.0: a new human identification method
Toineke Westen
- 3.00 p.m. Who is in my sample? Identifying multiple contributors
and phenotypic traits with SNPs
- 3.30-4.00 p.m. Coffee/tea
- 4.00 p.m. Session 3: Palaeoepidemiology (chair Risha Smeding)
Kirsten Bos
Ancient Pathogen Genomics: What we can learn from
historic Pathogens
- 5.00 p.m. Christos Economou
Health in ancient Scandinavia: infections and genetic
diseases
- 5.30 p.m. Close (Piero Giordano)
- 5.45 p.m. Drinks

Abstracts

Mark Thomas

*Research Dept. of Genetics, Evolution and Environment
University College London*

The Co-Evolution of Lactase Persistence and Dairying

Most Europeans take drinking milk for granted; it's the everyday consumption of an everyday drink. But for most adult humans, indeed, for most adult mammals, milk is very far from an everyday drink. Milk is something that we have specifically evolved to be able to consume in the relatively recent past. The ability to digest the sugar in milk is called Lactase Persistence and Darwin's engine of evolutionary change, natural selection, has probably worked harder on this trait than on any other biological characteristic of Europeans in the last 10,000 years. In this presentation we will see how Genetics, Archaeology, Anthropology, Physiology, ancient DNA and computer simulations can be combined to understand where, when and how Lactase persistence co-evolved with the culture of dairying in Europeans.

Eveline Altena

Forensic Laboratory for DNA Research, LUMC

The Dutch genetic landscape from past to present

For several decades population genetic research on modern human populations has been used to infer large scale (pre)historic human demographic events all over the world. Mainly due to recent technical advances in the field of DNA sequencing it is now also possible to analyse large sample collections of ancient individuals. By incorporating data from archaeological populations in population genetics we will be able to test many more hypotheses and get a detailed insight in past demographic events. Since 2006 archaeological DNA research is carried out on several large and small scale human skeletal collections across the Netherlands. The majority of these samples date from the medieval and post-medieval periods. They enable us to reconstruct (part of) the genetic history of the Netherlands.

This presentation mainly focuses on the results of the male specific Y-chromosome. By comparing Y-chromosomal information of Dutch archaeological sites and an extensive survey study on modern Dutch males, we are now able for the first time to get a glimpse of not only the regional, but also the temporal genetic variation of the Y-chromosome in the Netherlands. This will be a useful tool in understanding the extent and complexity of historical demographic processes that took place and shaped the present Dutch genetic landscape.

Peter de Knijff

Forensic Laboratory for DNA research, LUMC

STR genotyping 3.0: a new human identification method

Forensic DNA research is, strictly speaking, a relatively simple kind of genetic research made complex because results are used to, sometimes, convict suspects of serious crimes (meaning that simple science has to be explained to notoriously difficult students such as judges and lawyers or members of a jury, one of the most difficult teaching tasks one can imagine). The basic scientific principle is easy enough. You make DNA profiles from biological samples found at crime scenes and compare these with similar profiles from suspects and victims of crime. If, subsequently, a profile from a crime scene sample matches with a suspect, it seems likely (note: not proven!) that this suspect is the donor of that sample and can at least be linked to the crime. Ideally, DNA profiles should be easy to detect in very small DNA quantities. They should be so unique that each profile is only found in a single human individual and the method to detect these profiles should be fast and robust.

It was therefore extremely fortunate that, in 1991, Al Edwards and Tom Caskey from Baylor College of Medicine, Houston, Texas, USA, were the first to describe the use of short tandem repeats (STRs, also called microsatellites), for a number of different genetic diagnostic purposes (1). STRs represent small pieces of DNA that can be found in different lengths in different human individuals, and since we, humans, have always been very good in measuring lengths, with or without the help of complex technology,

these STRs caused the most important revolution in forensic DNA research of the past 50 years. To illustrate its importance, there are now worldwide at least 50,000,000 STR-DNA profiles in criminal DNA databases.

Since 1991, in just four years, the initial genotyping method (using different gel-type separation techniques and various band staining protocols) was replaced by the multiplex, fluorescent based capillary separation method using the ABI 310 in 1995. STR genotyping 2.0, in vogue for close to 20 years, did not improve substantially, and it is about time to move on. With the introduction of a new generation of DNA sequence technologies, starting with the Roche-454 massive parallel sequence platform in 2005 (2), it seems that we are very close to the introduction of STR genotyping 3.0, that will revolutionize routine STR genotyping. Exactly how will be explained and shown in detail in this presentation.

(1) Edwards A, Civitello A, Hammond HA and Caskey CT (1991) DNA typing and genetic mapping with trimeric and tetrameric tandem repeats. *Am J Hum Genet* 49: 746-756.

(2) Margulies M, et al. (2005) Genome sequencing in microfabricated high-density picolitre reactors. *Nature* 437: 326-327.

Toineke Westen

Netherlands Forensic Institute

Who is in my sample? Identifying multiple contributors and phenotypic traits with SNPs

Apart from medicine, human DNA typing is performed in forensic cases to determine the cell donor of a stain, in kinship analysis and in human identification cases. DNA typing is usually based on the analysis of short tandem repeats (STRs). STRs are characterised by differences in length, due to a difference in the number of repeated DNA elements. STRs are the "golden standard" in forensic DNA typing, and national DNA databases are based on STR data. However, other types of markers, such as single nucleotide polymorphisms (SNPs), are also available for forensic use. SNPs are characterised by a change in the DNA sequence of only one nucleotide (i.e. DNA building block) in length. Even though SNP information is not available in the national DNA database, SNPs possess several characteristics that make them interesting for one-to-one comparisons between a reference sample and a stain. As these markers are very small, they are suited for the analysis of (highly) degraded DNA. Additionally, SNPs may be used to predict someone's bio-geographical ancestry or externally visible characteristics. The potential of the use of SNPs to obtain information on the donor of a sample will be explained in this presentation.

Kirsten Bos

Dept. Paleogenetics, Prehistory and Archaeology Science, Eberhard-Karls-Universität Tübingen

Ancient Pathogen Genomics: What we can learn from historic Pathogens

Genome-wide data from ancient microbes may help to uncover mechanisms of pathogen evolution and adaptation for many diseases. Using high throughput DNA sequencing in combination with targeted DNA enrichment we have reconstructed medieval bacterial genomes of *Yersinia pestis* and *Mycobacterium leprae* from skeletal remains. Phylogenetic analyses indicate that the ancient *Y.pestis* strain from the Black Death pandemic is ancestral to most extant strains, and falls very close to the ancestral node of many *Y. pestis* bacteria. Temporal estimates suggest that the Black Death of 1346—1351 was the main historical event responsible for the worldwide dissemination of most currently circulating *Y. pestis* strains. In contrast, the medieval *M. leprae* strains fall within current genetic diversity and are found on at least two main branches in the phylogenetic tree of leprosy bacteria. Dating analyses reveal a most recent common ancestor of both *Y.pestis* and *M.leprae* within the last 4000 years, suggesting that both diseases may have a Neolithic origin. The extraordinary preservation of the *M.leprae* DNA' allowed for the first *de novo* genome assembly of an ancient organism and indicates that some bacterial DNA may survive longer than vertebrate DNA in ancient remains. This may permit tracing the history of many infectious organisms back to their prehistoric origins.

Christos Economou

Dept. of Archaeology and Classical Studies, Stockholm University

Health in ancient Scandinavia: infections and genetic diseases

Infectious diseases have always played a major role in shaping the history of the societies of the past and Palaeopathology is the scientific field that approaches their occurrence and effects on human populations through the use of archaeological remains. Ancient-DNA has proven to be of invaluable assistance to such studies as it can provide answers not only for the mere presence of the pathogens in the remains but it can shed light on their geographical distribution, evolution and host's immunity as well. Leprosy and Plague had been two of the most talked-about diseases in Middle-Ages Europe, either due to the social stigma that they carried or the high mortality rate. The main purpose of our current studies is to molecularly analyse human remains from Medieval Sweden that had been infected by those two diseases (using osteological or historical evidence) and make a map of the health conditions in Scandinavia of that period. Apart from the molecular identification of the pathogens in the remains, the infected individuals were also tested for the presence or absence of alleles that are thought to provide immunity to those particular diseases. A connection between the bacteria and the host's immune system during that particular period of time is therefore tackled and comparisons with published modern data can be made.

CVs

Mark Thomas

Mark has worked extensively on understanding how humans have evolved and migrated around the World. He has used genetic data — including ancient DNA — computer simulations and archaeological information to examine the origins and past migrations of a number of specific human populations including Jewish and Judaic groups, British populations and a number of enigmatic European and African peoples. In recent years he has worked on using 14C data as a proxy for past demography, on modelling cultural evolution to better understand the origins of modern human behaviour, and to examine ethnic structuring in past populations, on recent natural selection using genetic data — particularly in relation to diet and infectious disease — and on gene-culture co-evolution, particularly the origins of lactase persistence and dairying in Europe and Africa.

Eveline Altena

Eveline did an MA in the archaeology of Meso-America at the Faculty of Archaeology, University of Leiden with a strong emphasis on physical anthropology and funerary archaeology. After several years of fieldwork in Dutch archaeology and some campaigns in the Caribbean to escape the Dutch clay, she started a PhD in human archaeological DNA research in 2006 at the Forensic Laboratory for DNA Research, Leiden University Medical Center. For the PhD she works on several Dutch (post-)medieval populations from, amongst others, Eindhoven, Vlissingen and Vlaardingen.

Peter de Knijff

Peter is full professor in population genetics and evolutionary genetics and head of the Forensic Laboratory for DNA Research, at the department of human genetics of Leiden University Medical Center. He enjoys many different research interests, varying from developing, using and understanding human Y-chromosome polymorphisms to disentangling the evolutionary relationships of large gulls.

Toineke Westen

Toineke (Antoinette) Westen has a background in Biomedical Sciences. Since 2006, she works at the Netherlands Forensic Institute at the department of Human biological traces within the research and development group. In spring of this year she finished her PhD project on "Human identification & forensic analyses of degraded or low level DNA".

Kirsten Bos

Kirsten holds a Master's degree in skeletal biology, and in 2011 she received her PhD from McMaster University in Canada with a specialization in ancient DNA. Since January of 2012, she has been a postdoctoral researcher at the University of Tuebingen working in the Paleogenetics group run by Johannes Krause, where she specializes in ancient infectious diseases and host-host-pathogen interactions.

Christos Economou

Christos is a Biologist with postgraduate studies in Biomolecular archaeology. He is currently working on ancient-DNA as a PhD candidate in Stockholm. His main research is on palaeo-diseases, both infectious and genetic. Current projects include studies on Leprosy, Plague, Cholera and Osteoporosis in Swedish populations of the past (particularly Medieval times).

Organizing committee:

Eveline Altena

Paul Storm

Rachel Schats

Matty Spinder

Kelly Fennema

George Maat