The Use of Forensic DNA Phenotyping in Anthropology

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Objective

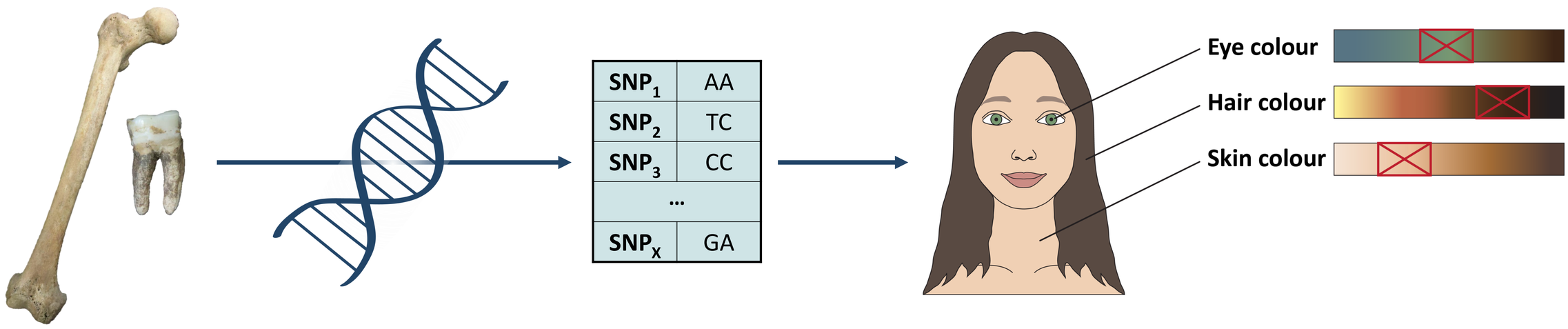
My thesis aims to show that forensic DNA phenotyping using the Dynamic Array™ IFC by Fluidigm® is a useful tool in answering various anthropological research questions.

The method will be applied to DNA of different conservation and degradation stages with samples from the Meso- and Neolithic over the Middle Ages to modern history to conclude the general suitability of forensic DNA phenotyping for anthropological questions. Exemplarily four research subjects are addressed.

Background

In forensic casework, the analysis of STRs and hence the use of genetic fingerprints to identify a person are routinely in use. Recently, due to new scientific findings as well as legislative changes the method of forensic DNA phenotyping has become increasingly important. Compared to DNA typing, it does not rely on comparison with other samples or a database. Rather, its outcome can serve as a biological witness and therefore provide new investigative leads.

Retrieving information about eye, hair and skin colour from DNA is, however, equally interesting in an anthropological context. The challenge here is that ancient DNA is often highly degraded and therefore of low quantity and quality. Although forensic DNA phenotyping was successfully applied to DNA from bones and teeth, practical case applications are still scarce. However, forensic DNA phenotyping has great potential for future anthropological investigations as it can shed new light on old debates and uncover phenotypic traits that are inaccessible to traditional methods.



Research Subjects

European Phenotype

When and how did the light skin colour and the variety of eye and hair colour shades develop? Were various selection pressures or migratory movements responsible? Which SNPs and their alleles were predominant at which time? For this project, 27 samples from the Mesolithic and Neolithic up to the Iron Age will be examined. The aim is to contribute to current research regarding the development of the European phenotype. Here, each phenotyped individual can bring new insights or challenge previous hypotheses.

Identification

This project will deal with identification in an anthropological and forensic context, to show, specifically, how phenotyping can contribute to the success of an investigation. Exemplary for the identification of historical individuals serves skeletal material from a crypt in Oldenburg. The main objective here is to unambiguously identify Christoph von Oldenburg who is described as red-haired and red-bearded in historical reports.

Museal Portrayal

Exhibitions in museums often deal with the culture and living conditions of certain populations or groups by focusing on individuals and their life stories. Genetic information on a phenotype can support visual representations. This project, therefore, includes reconstructing the external appearance for the presentation in a museum exhibition. One individual for which a facial reconstruction is planned and nine individuals from the Napoleonic Army are analysed.

Provenance Research

In anthropological research, the question of provenance concerns human remains that were acquired (illegally) during the period of colonial rule overseas. There are currently great efforts to clarify the provenance of questionable objects in anthropological collections and possibly facilitate restitutions. Here, phenotyping can provide additional information that is inaccessible with the standard methods and therefore contribute to the current discussions. For this project, eight samples with an unclear provenance will be analysed.

Methods

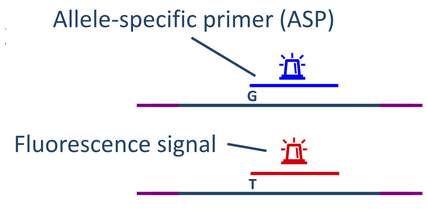
Genotyping with the Dynamic Array™ IFC

A total of 48 SNPs concerning eye, hair and skin colour published in Chaitanya et al. (2018), Maronas et al. (2014) and Söchtig et al. (2015) were chosen for the genotyping. The Dynamic Array™ IFC (Fig.1) allows for easy handling and combines rapid throughput with excellent call rates, high concordance and low cost. The genotyping is divided into three parts:

1. ***Specific target amplification (STA)***

2. ***Allele-specific amplification***

Allele-specific amplification is performed on the Dynamic Array™ IFC by Fluidigm (Fig.1). If the allele-specific primer bind to their respective allele, they emit a fluorescence signal. These signals will be detected by a high-resolution camera.



3. ***Scatter-plot based determination of the genotype.***

The detected fluorescence for each sample is plotted into a cluster plot with three clusters for homozygote Allele 1, homozygote Allele 2 and heterozygote.

Phenotyping

The genotype of the samples is translated into an eye, hair and skin colour phenotype using two freely accessible online tools: Snipper Classifier (http://mathgene.usc.es/snipper/) and HIrisPlex-S Webtool (https://hirisplex.erasmusmc.nl/).

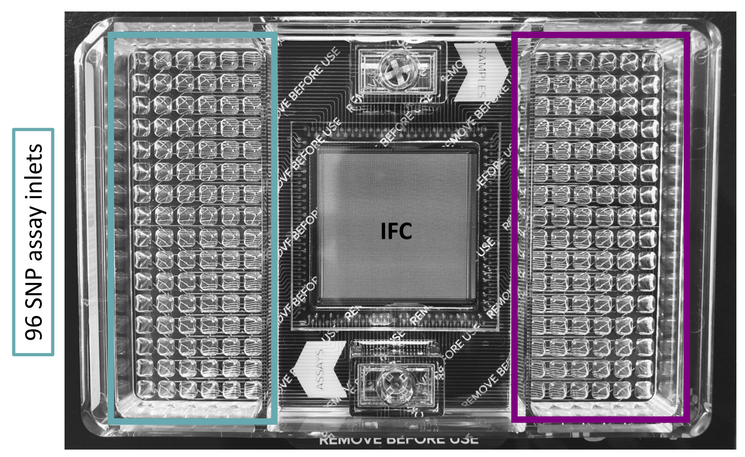


Figure 1: 96x96 Dynamic Array™ IFC by Fluidigm. The Dynamic Array™ is loaded with the PCR products of the STA and custom-designed SNP assays with the ASPs. The IFC then allows for 9216 separate reactions for each sample with each SNP.

Outlook

The first results are very promising. Full genotypes could be generated for a majority of the samples after the first analysis run. The second run will be used to adjust the parameters and replicate the results. For samples that showed only some genotyping success the DNA input as well as the PCR cycles in the specific target amplification will be increased.

Schmidt et al. (2019) have already demonstrated that the Dynamic Array is suitable for well-preserved aDNA samples. My first results now show that it can also be used for challenging aDNA samples, and that phenotyping will enrich anthropological research in many ways in the future.

References