GENETICS IN ANTHROPOLOGY

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Contents

Summary

Anthropology is considered a holistic discipline that studies humans in varied manners, ranging from their social cultural contexts, to their biological makeup both in the past as well as in the present. The complexity of the human species is well present in these various forms of expression. It is, therefore, a knowledge that draws from various disciplines, one of which has deserved special attention due to its overarching implication in human life: that is genetics. In recent years much has been written about genes, genetics and the human genome, and about ancestry and related species – particularly non-human primates.

To explore genetics in a discipline such as anthropology is to cross the boundaries between biology and culture, and build bridges between nature and culture whilst exploring its social and cultural ramifications. Within this context, the role of genetics in Anthropology is wide, ranging from evolutionary human biology, past human populations studies (henceforward referred to as bioarchaeology) and living population genetics, paleopathology and even forensic anthropology. The current chapter will explore genetics in anthropology within a four-field anthropology. It will first introduce the basic concepts of four-field anthropology, and its specificities of research, as well as basic concepts in genetics, particularly those that find their way into the anthropological discourse; secondly, it will introduce the overall methodology used to explore genetics and discuss it most significant limitations; thirdly, it will address past and present research agendas and results – providing a comprehensive, although summarized, state of the art on the subject. The conclusion will provide an overall appreciation of the role of genetics in anthropology, offering a historical perspective on the subject and future avenues for research.

1. Introduction (Francisca Alves Cardoso and Sandra Assis)

1.1. What is Anthropology and how it relates to Genetics?

Before introducing genetics in anthropology, and talk about how these two areas of knowledge have been cooperating, let's first consider the discipline of anthropology.

Anthropology is described here in accordance with the definition provided by the American Anthropological Association (AAA), as a discipline that studies humans, in the past and present. It is a discipline that is overarching all human complexities, in its many expressions, ranging from human multifaceted social and cultural contexts to its various genotypic and phenotypic profiles. In order to do this, it is necessary to add a diachronic (past to present), as well as synchronic (contemporaneous and multiple) perspective. The understanding of anthropology as a four-field discipline that converges socio cultural anthropology, biological (also referred to as physical) anthropology, archaeology and linguistics, is an appropriate option when defining the research scope of the discipline. It is however necessary to be cognizant that the notion of a four-field anthropology is intrinsically linked with the history of anthropology in the United States and the name of Franz Boas (Little and Kennedy 2010), and that variations in the discipline scope, research and teaching topics exist between countries worldwide.

Such a broad definition of anthropology may be considered overwhelming, and is frequently a handicap when trying to explain its boundaries of agency – if any exist – or even its core topic of research and definition. Nevertheless, the description of anthropology as an extensive (and expansive) discipline is accurate when considering it as a research topic – human beings and closer related species –, living and fossil humans, and closely related nonhuman primates such as apes and old and new world monkeys. Therefore, the definition of "What is anthropology" must include its many fields of research agendas and teaching subjects.

Anthropology has focused on various topics of research, as diverse as race and ethnicity; human biological, social and cultural variability; sexuality and gender; class and social changes, and inequalities; nationality and globalization; migration, health, ecology and environment; cognition and education; human and non-human growth and development; material and immaterial culture, and commoditization of culture; tourism; human remains; paleopathology; forensic and human identification. The aim is to understand humans, their differences and similarities, and most importantly to find ways and solutions to deal with many of humanity's problems.

But where does genetic fit into anthropology? In present days genetics is considered a sub-field of biological anthropology. Biological anthropology may be described as a scientific discipline dedicated to the study of biological and behavioral characteristics of human beings and their closest relatives, the nonhuman primates, from an evolutionary and comparative perspective. Biological anthropology is normally understood as comprising five sub-disciplines – human evolution, primatology, the study of human physical growth (osteological tissue included), human ecology and human genetics. In the past, biological anthropology was grounded in medical and natural sciences, but in later years social sciences and humanities have been able to permeate the discipline.

On a side note one should bear in mind that biological anthropology should not be used interchangeably when referring to classic 19th century physical anthropology. Briefly, the 19th century Physical anthropology relied heavily on the categorization of human osteological remains, and anthropometric features of individuals, aiming to classify living populations based on phenotypic qualities. Major research methods in physical anthropology were based on the assessment of measurements. Morphological characteristics and discrete traits of craniology, anatomy, morphology, race and population types were all research interests of physical anthropology. All strived to address issues related with the classification of populations found within the geopolitical frame of many European empires.

With the advent of the Darwinian theory of evolution by natural selection, published in 1859, the impact of the Modern Synthesis – much a result of the publication of Julian Huxley *Evolution: Modern Synthesis*, in 1942, and the growth of genetics from mid-20th century with James Watson and Francis Crick publication on the chemical structure of the deoxyribonucleic acid (DNA) –, the interest in the classification and understanding of populations began to include individual's genotypic qualities. Largely due to these advances in the field of genetics, a shift in the goals (and name) of the discipline also occurred: instead of describing and classifying human variation – physical anthropology – the emphasis was redirected to more oriented biological topics – biological

anthropology. By the late 20th century genetic studies on living humans and past populations, as well as on nonhuman primates and fossils, were a major research agenda in biological anthropology. Nowadays, even social and cultural anthropologists are confronted with the significance of genetics in anthropology.

1.2. The Relationship between Anthropology and Genetics

In recent years much has been written about genes, genetics and the human genome. Research conducted in genes has come to stay and is making progresses. The relevance of genetics in anthropology has slowly been reinforcing the importance of nature (biology) in culture, and the impact of culture in nature. More than ever, discussing the human nature and culture interaction is present in anthropological contexts. On the one hand genetics is used to assess intelligence, disease and behavior inherited predisposition, such as predisposition to violence, homosexuality, language, depression, alcoholism and many more; on the other hand anthropology stands on the opposite corner, reflexive and acting as a voice of caution in simplistic interpretations of human faculties and behaviors. Anthropologists and other social and cultural scientists have highlighted that environmental and social experiences – and associated triggers – are relevant in the interpretation of the dynamics of genetics. In studies related with humans – and to a certain extend its closer relatives –, combining genetics and environment is the best approach to fully comprehend oneself, as individuals and as species.

Today, if one was to conduct a public survey inquiring on personal opinion and definitions of what are *genes* and what does *genetic* means, everyone regardless of age or nationality would most probably present an explanation. In today's societies genes and genetics have assumed a meaning that is independent from – and surpasses – its biological definition.

In 1995 Dorothy Nelkin and Susan Lindee published *The DNA Mystique: The Gene as a Cultural Icon* – the book was re-edited in 2004. Although the reference is from the 1990s, much of what was written is still largely applicable today. The book has an underlining discourse on the nature *versus* culture ongoing discussion, but what is interesting to stress here – and paraphrasing the authors – is the ability to explore genes as a "cultural icon". Genes as something not constrained to its technical biological and *natural* definition, but rather as something that has been appropriated by culture, cultural groups and media, and that is discussed as a product in itself – all this being worthy of discussion.

For example, the marketing, commodification and circulation of DNA, bestowed with political and economic power, is very much present in today's discourses. To a certain extent, genes have become celebrities in their own right. Since the publication of the DNA double helix, genes have been appropriated by artists – performers, musicians, others – and used as inspiration and as identity.

Much in relation with this is DNA mystification, because today DNA continues to be advocated by many scientists as cause and cure for almost all that afflicts humans. And all this is so despite the fact that genetic determinism, as viewed in the early onset of genetics, has given way to a more interactive view between genes and environment.

For the purposes of this chapter the above mentioned, and many other aspects related with genetics and genes that find their way into the anthropological debate, will not be addressed. For example, DNA patenting and manipulation, transgenic productions and associated ethical and legal debates, are topics discussed within anthropology in relation to genetics. In such cases, rather than emphasis being given to genetic anthropology (or anthropological genetics) as a sub-discipline within anthropology, the relevance of genetics is accentuated as a theoretical and conceptual framework discussed within social and cultural anthropology.

The understanding of genes – as a biological item and, more precisely, as a segment of DNA –, of how they relate to human variability and diseases, and on how these have impacted on science, culture, politics and economics, is symptomatic of our growing knowledge about them, of how much the discipline as progressed, and of how may genetics be used (or believed to be capable of) to solve many of humanity's predicaments. Genetics has therefore been transformed into a field of study in anthropology, more precisely it has given place to genetic anthropology or anthropological genetics, which may be described as a discipline that explores evolutionary theory, of interest to anthropologists, whilst applying genetic methodologies. Note that this may be considered as a very short definition of genetic anthropology, which currently has widened its sphere of applicability.

To some extent the Human Genome Project (HGP) has played a relevant role in the significance of genetics in anthropology – in the understanding of human beings, and of what makes us *humans*, particularly from a biological and evolutionary viewpoint. It represents a voyage inwards, and was so described: "the Human Genome Project (HGP) was [is] an international, collaborative research program whose goal was [is] the complete mapping and understanding of all the genes of human beings" (http://www.genome.gov/).

The HGP has revealed the existence of – probably – 20500 human genes, which are believed to map a basic set of instructions for the growth and function of human beings. This is an impressive achievement. The first draft of the human genome was published in February of 2001 in *Nature*, and a full sequence of human genome was published in 2003. The HGP has not only provided the human being's genetic blueprint, it has also contributed to the development of methods and techniques used in genetic research. These have been used to conduct research in human material, as well as in other organisms, ranging from animals to plants.

In summary, the HGP has been disseminating methods and information about human beings. Although the information generated by the project was intended to be used for the benefit of humanity, there is an underlying concern associated with the use and sharing of detailed genetic knowledge. This is extremely relevant to point out in a time where genetic manipulation is a known and discussed reality. Therefore ethical, legal and social implications associated with the gathering, possessing and broadcasting of all this information are also under reflection, as well as the development of policy actions for public consideration (details may be found at www.genome.gov/).

1.3. Genetics in Anthropology: Methodology and Limitations (Alves Cardoso *et al.*).

Access to genetic material is complicated, and complex. It is expensive, laborious and demanding, and the results are not always guaranteed. Apart from this, there are also methodologies' limitations and ethical and legal issues that are extremely sensitive.

Genetics in anthropology, within a four-field anthropology, requires the collection and processing of data from living human and nonhuman (primate) populations, from skeletonized human remains and nonhuman remains – all have individual sets of methodological challenges and limitations. Therefore, one can argue that access to biological material – genes – has two major spheres of limitations: one relates to the access to data – the need to inform and be given consent to collect, use and disseminate information; a second aspect relates with the preservation, including degradation and contamination, of the material itself, and the need to have the best possible methods to do the job.

1.3.1. Access to Data for Genetic Analysis

Collection of data from living populations is based, foremost, on the principle of informed consent, which is a basis of ethical conduct of research involving humans. The use of informed consent is not restricted to the access to genetic data. Within anthropological research it is an issue that is transversal to any type of data collection, from biological material to personal biographical data information, either oral or documented. Privacy protection and confidentiality are major topics of discussion and concern. Consider for example that based on genomic research there is a growing number in the identification of disease-associated genes. The results of such discoveries, albeit scientifically relevant, have major implications at a population level, and also at individual levels. The release of such information could be used to stigmatize members of particular communities/populations, and at a personal level it could have damaging consequences to individual employability, insurability or reputation. Such concerns are strongly present in medical research, as expressed by the World Medical Association Declaration of Helsinki (WMA – DH) through a "statement of ethical principles for medical research involving human subjects, including research on identifiable human material and data" (http://www.wma.net/en/30publications/10policies/b3/).

The HGP has similar, well documented, concerns. These have been expressed in the 1990 National Human Genome Research Institute's (NHGRI) Ethical, Legal and Social Implications (ELSI) Research Program (http://www.genome.gov/ELSI/). ELSI aims to address the ethical, legal and social implications of genetic research in populations, specifically on how these may affect individuals, families and communities.

The use of informed consent exists to act as a precaution, as it aims to assure that participants are informed on how data will be collected and used, and to a certain extent to raise awareness on the implications associated with the participation on the study/data collections. Informed consent also provides some protection to those conducting the research in case of lawsuits.

However, ongoing research based on genetic data has proven to be a challenge to a more traditional one-to-one informed consent approach. Nowadays, populations' genetic studies are based on massive large-scale databases – as exemplified by the HGP. Consequently, data collected may be used in multiple studies, which may not be necessarily related with the original study over several years. Once collected, genetic data may be replicated and disseminated widely: if data is shared between multiple partners, risks involving breaches of privacy may happen. Just consider that with the Internet, in a matter of seconds anything can become "viral", and therefore known to millions.

There is a secondary aspect of informed consent that deserves additional concern. For example: many documents are unclear as to what is being consented for when those that read them lack the necessary expertise. It is necessary to remember that populations and/or individuals may not fully comprehend research studies based on genetic data (or other), and many individuals may not speak or read the language of the scientists with whom they will interact with.

Many more scenarios exist worthy of apprehension when dealing with access to data, and data use, such as current discussion of indigenous people gene disputes, and let's not forget the case of Henrietta Lacks' immortal cells, which forces retrospection on the early collection of data for genetic analysis. Henrietta Lacks died of cervical cancer in 1951, and had some of her cancer cells collected for medical research without her – or her family's – knowledge. Lacks' cancerous cells had the remarkable capacity to multiply indefinitely, hence the denomination of *immortal cells*. Larks' HeLa [Henrietta Lacks] immortal cells line have been used in medical research and were/are crucial in research relating with the cure of polio and with treatments of many diseases including cancer and AIDS (Skloot 2010). The discovery of this by her family disclosed a situation that is not supported by today's ethical concerns of medical research, as expressed by the WMA-DH, but was a common practice on the past, one that should not be repeated in other contexts.

Within past populations studies, access to biological data does not necessarily fall within the scope on *informed consents*, although ethical issues have been raised as to the right to access, use and process, biological material recovered from human remains when consent may not be obtained. Even if legal issues may not apply – given the absence of living relatives that may oppose such research –, ethical issues have been voiced, as to the necessity of preserving the dignity of human remains which once belonged to living people.

A similar ethical concern also exits in relation to the access to biological material drawn from nonhuman primates. These are issues that are not necessarily discussed by biological anthropologists. Issues such as these fall mostly into the agenda of social scientists, as a complement to biological research. Nevertheless, there is an underlying effort to develop non-invasive – or as less invasive as possible – techniques of data collection in past population and animal studies.

When working with human remains, rather than destroy, perforate or damage a well-preserved specimen (i.e. bone), fragmented bones and/or individual teeth are used. In

primatology the development of non-invasive genotyping, in the 1990s, was an important revolution. Non-invasive DNA extraction from biological material, such as faeces, shed hair or food wedges – that do not require animal handling –, has been replacing the collection of genetic data done invasively via blood and tissue sampling. The use of non-invasive techniques has also allowed access to genetic data from nonhuman primates in natural habitat. This has allowed new areas of inquiry in primatology to be developed, such as kinship, reproductive success and dispersal patterns in wild populations, with the added bonus of allowing for comparisons between – and within – wild populations. What in the past took decades of field observations is now possible to test within months. This has also allowed access to samples from primates in their natural habitat rather than in captivity, what represents a major breakthrough in primatology.

Although incredibly revolutionary and informative, studying primates using non-invasive genetic material remains is extremely challenging, time-consuming, expensive and prone to error. Faecal material and shed hairs contain low quantities of DNA and, most of the times, of poor quality (fragmented). These features of non-invasive samples often lead to genotyping errors, e.g. "allelic dropout" and false alleles. Likewise, only small DNA fragments can be analyzed such as microsatellites and short mitochondrial DNA sequences, the most widely used markers in molecular primatology. Moreover, the presence of co-extracted compounds in the faecal material may inhibit the polymerase chain reaction (PCR) – the laboratorial procedure used to amplify such poor sources of DNA. As a result, non-invasive DNA analysis requires the application of rigorous standards for replication of the results with the consequent disadvantage of becoming very costly and time consuming.

Therefore, and despite the efforts, to conduct genetic studies in nonhuman primates still requires high concentrations of extracted DNA, and of excellent quality, and this, even today, can only be obtained from tissue and blood. Up to the present the benefits of genomics for the study of primate social systems are still limited, despite the advances made. The expectation is that molecular techniques will soon evolve to allow scientists – and in particular primatologists – to produce genomic data from non-invasive biological material.

In all fields relating with genetics and data access the need to compromise between ethics and the advance of science is of the upmost importance. Studies based on blood and tissue samples have allowed significant progresses, particularly in phylogenetic studies and in medical research, but they have also uncovered a darker side of science.

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Biographical Sketches

Francisca Alves Cardoso is a research fellow at CRIA – Center for research in anthropology, an inter-institutional centre devoted to advanced training and research in anthropology. She is also an invited lecturer at the Department of anthropology, at Faculdade de Ciências Sociais e Humanas, from the Universidade Nova de Lisboa, Portugal. In 2008 she was awarded a PhD in biological anthropology by the University of Durham (UK). She has a degree in anthropology, and a Masters in Human Evolution and Biology awarded by Coimbra University (Portugal). Her current main areas of research focus on the analysis of Human Identified Skeletal Collections (HISC), and their use to assess past human behavior and health patterns, utilizing a biocultural perspective. Presently her research emphasis is twofold: 1) to highlight the crossroad of exploring the use and importance of human osteological remains in the study of the human past; and 2) to address the limitations of human osteological collections, identified or archaeological, as well as their ethical and legal framework, alongside their preservation as patrimonial heritage. She has been encouraging a constructive discussion on the methods employed in the measurement and interpretation of pathological lesions, and promoting the use of new technologies, which may improve Paleopathological analysis. The development of statistical designs/models to analyze bony lesions, and to permit a better interpretation of health based on human skeletons is another of her concerns.

Amanda Ramos is presently a post doctoral fellow at the Research Group in Biological Anthropology (GREAB), at the the Universitat Autònoma de Barcelona. She graduated from that same university in Biology, in the field of genetics and health sciences. She was awarded a Masters in Human Biology in 2008, also at that university, and in 2012 she was granted a PhD on the topic of human mitochondrial DNA heteroplasmy also at the Universitat Autònoma de Barcelona. Her research was focused on the study of human populations, molecular evolution and glioma tumour at mitochondrial DNA level. In March of 2012, she got a postdoctoral fellowship from DRCT to join the group of Prof. Manuela Lima in the University of Azores, to work in Spinocerebellar Ataxias (SCAs), developing a new diagnosis methodology. This methodology is currently pending patent approval, and it aims to improve the existing molecular diagnosis of the most prevalent types of SCA: SCA1, SCA2, SCA3, SCA6 and SCA7.

Cláudia Lopes Gomes is currently a member of the Laboratorio de Genética Forense y Genética de Poblaciones, from the Compulense University of Madrid. She has a degree in Biology (2009) awarded by the Faculty of Sciences of Porto University (Portugal), and a Masters in Forensics Genetics (2009) from Instituto de Patologia e Imunologia Molecular (IPATIMUP - Porto). She is currently a PhD student at the Faculty of Medicine from the Complutense University of Madrid. She is also an ongoing lecturer on behalf of the Grupo de Genética Forense y Genética de Poblaciones, no Departamento de Toxicología y Lesgislación Sanitária da Facultad de Medicina de la Universidade Complutense de Madrid, in which she is cooperating with teaching and research.

Cristina Santos is an associate lecturer in the Unit of biological anthropology at the Universitat Autònoma de Barcelona (Spain). She has a degree in Biology from the University of Coimbra (Portugal), and a PhD in Biology from the Universitat Autònoma de Barcelona, awarded in 2005. She completed her post-doctoral training at the University of the Azores in 2007 and joined the Universitat Autònoma de Barcelona as a lecturer in that same year. Since July 2014, she is the coordinator of the interuniversity biological anthropology master (UAB/UB- Universitat de Barcelona). She has a broad background on molecular and human population genetics, biostatistics and epidemiology. Her research has mainly focused in the evolutionary dynamics of mtDNA and in the influence of mtDNA mutations in aging and disease, namely in cancer and late onset neurodegenerative disease. She is also interested in the genetic characterization of human ancient and current populations aiming to infer which historical, demographic and microevolutionary factors have shaped human population structure and diversity.

Eduardo Arroyo Pardo has since 2005 lead the Laboratory of Population and Forensic Genetics from the Faculty of Medicine, Complutense University from Madrid, He was awarded a degree in Biology in 1987, by the Complutense University of Madrid (UCM), where he first developed an interest in physical anthropology and Population Genetics. In 1993 he was awarded his Ph.D. in biological science. In 1994 he became a lecturer at the Laboratory of Forensic Biology, at Faculty of Medicine from Complutense University, where he remains until today and has progressed in his academic career. He also got a degree in Biochemistry (Faculty of Chemistry, UCM, 2004) and latter in Philosophy at Universidad Nacional de Educación a Distancia (UNED) (2013). He has coauthored more than a hundred papers and articles on population genetics, population phylogeny and ancient DNA technology.

Sandra Assis is currently an invited lecturer at the Department of Anthropology, at Faculdade de Ciências Sociais e Humanas, from the Universidade Nova de Lisboa, Portugal. She is also a member of CIAS – Research Centre for Anthropology and Health, from Coimbra University. She has a PhD in biological anthropology awarded by the University of Coimbra, Portugal. She has a degree in anthropology, and a Masters in Human Evolution (2007) by the same University. She has a vast experience in research with human remains recovered from archaeological contexts, and is a specialist in paleohistological analysis and the use of this technique applied to human past populations – bone tissue preservation and paleopathological differential diagnosis. In recent years she has further developed interest, as vice-president of GEEvH – Grupo de Estudos em Evolução Humana, for issues related with dissemination of sciences in non-academic milieus.

Tânia Minhós is currently conducting my postdoctoral research on "Effect of habitat loss on population viability of sympatric primate species: evidence from neutral, selective markers and parasite infection" at the Instituto Gulbenkian de Ciencia and Centro de Administração de Políticas Públicas (ISCSP), Portugal. She is a molecular primatologist working with West African primates (e.g. colobus, baboons, chimpanzees). She was awarded her PhD in 2012 by the Cardiff School of Biosciences, UK. Till present her research has/is focused on the use of different types of data (genetic, socio-ecological, parasitological) to better understand the evolution, adaptation and conservation of natural primate populations threatened by human activities.