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# Epigenetics: Anthropological Implications

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## Abstract

This paper examines the rapidly emerging field of epigenetics and specifically focuses on its implications for anthropology both social and biological. A number of themes are reviewed: Social inequality, the evolution of human and non-human primates, the migration of ancient populations, the impact of early experience on development, new conceptualisations of race and ethnicity. Finally, I discuss issues in epigenetic research.

**Keywords:** Development; Epigenetics; Evolution; Mental illness; Race; Social inequality

## Introduction

Epigenetics is the study of chemical modifications to DNA that are associated with changes in the way genes are expressed, or 'turned on,' or 'turned off' and are essential for normal development within mammals. The term was introduced more than 70 years ago by C. H. Waddington, an embryologist, philosopher of science and geneticist. Epigenetics can be defined as the "stable and heritable changes in gene expression that are not caused by changes in underlying DNA sequence" [1]. It involves the study of mechanisms of gene regulation beyond the genome whereby social-environmental information is transferred into genetic information. Meloni [2] notes that epigenetic mutations are often seen a case of developmental plasticity, the way by which a 'fixed genome' can respond in a more plastic and flexible way to the solicitations from a changing and dynamic environment.

Epigenetic markers include many types of chemical modifications. One common modification is the addition of a methyl group to the DNA itself or the addition or removal of a methyl, acetyl, or other type of group to the proteins, called histones, around which the DNA is wrapped. DNA methylation is recognized as an epigenetic mechanism that typically silences genes and is defined by the addition of a methyl group to particular locations on DNA.

Epigenetic marks are of significant interest to anthropologists because, unlike DNA, they are very sensitive to environmental exposures [3]. Slavich and Cole [4] point out how human genetic expression may not be as fixed as was previously thought. Our genetic expression depends on the ever changing social and environ-mental factors that occur throughout an individual's lifecourse. Similarly, [4] notes how a growing literature in human social genomics examines how everyday life experiences are able to influence human gene expression. Furthermore, these marks can be acquired throughout the lifetime of the individual and, when carried in their gametes, these marks are inheritable.

Current research is showing how environmental factors can modify epigenetic processes, thereby affecting epi-genetic marks and downstream patterns of gene expression in specific cells and cell lineages. These effects account for how early life exposures, such as prenatal nutrition or stress, can result in a phenotypic 'memory that persists into later ages to influence adult physiological function, health and risk for disease. Strong biological effects result from physical and emotional abuse (and other similarly extreme childhood events) and have a significant impact on health and developmental consequences. More recent research indicates that less obvious but more regular adversities of early childhood also have a lasting influence on later health and development.

Jablonka [5] argues that Studying epigenetics has the potential to forge new experimental and conceptual bridges between biology, the social sciences and the humanities. In a similar vein [6] avers: The Epigenetics Revolution, 'we are finally starting to unravel the missing link between nature and nurture; how our environment talks to us and alters us, sometimes forever'.

As [2] notes, 'no longer are genes seen as absolutely sovereign in the process of development, but as contextually

dependent (and elusive) entities that cooperate extensively with a large variety of postgenomic factors'. Biology is becoming more social. The body is becoming linked to its spatiotemporal contexts in which it lives:

'What has become increasingly evident in the last years, especially after the completion of the Human Genome Project, is that only a very small fraction of the genome (slightly more than 1 per cent in fact) is 'devoted to protein-codingsequences' - the orthodox definition of gene - whereas the large part of the genome is employed in regulation, that is, in responding to environmental signals, from the cell, the organism and the environment around it. In sum, the more genetic research has gone forward, the more genomes are seen to 'respond in a flexible manner to signals from a massive regulatoryarchi-tecture that is, increasingly, the real focus of research in "genetics".

## Anthropology and Epigenetics

Epigenetics raises many issues pertinent to the nature nurture debate- one that has caused longstanding divisions between life and social scientists and, according to [7] in *The Mirage of a Space Between Nature and Nurture*, continues to capture public attention. In her view the terms nature and nurture are inseparable; to insert the 'and' is to make them into a false dichotomy. Fox- Keller asserts, it is erroneus to think that heredity and the environment can be separated when 'the entanglement' of these two factors in humans "is not only immensely intricate but is there from the start.

Jablonka [8] notes how human populations have very different local cultures: they differ in their food preferences, in the way men and women live and are treated, where they choose to live and their type of dwelling, in the way they care for their children, and so on. All of these persistent cultural traditions are likely to have epigenetic correlates, and diet-related cultural variations will have additional correlated changes on the constitution of the microbiome of group members [9,10]. Furthermore, changes in the social landscape is likely to involve a change in the epigenetic landscape. [11] examine how the field of epigenetics is relevant to anthropology. This review develops their ideas.

## Social Inequality

It is well recognized that poverty and socioeconomic deprivation can result in premature mortality and morbidity and despite universal access to free health care, inequitable healthcare outcomes continue to be found in socioeconomically deprived populations [12]. The embodiment of social and racial inequalities may be mediated by epigenetic mechanisms. Epigenetics may explain how social environments can become embodied and impact health and more specifically health inequalities [13-15]. There are recent epidemiological reports of associations between socioeconomic status and epigenetic markers that predict vulnerability to diseases. These under-score the significant biological effects of social inequalities. This striking epidemiological phenomenon is illustrated by the city of Glasgow, UK, where people in the most deprived neighbourhoods are likely to live 12 years less than their counterparts in the richest parts of the city. Poverty accounts for part of these disparities: a steady gradient of health outcomes across social classes has been observed for many conditions, even among groups that are fully above the threshold of poverty [16,17]. Mc Guiness et al similarly examined social inequality in Glasgow and found that the poorest people, as well as the manual workers, were born with low levels of an epigenetic marker for global methylation. The most socioeconomically deprived groups had hypomethylation. Lower methylation has been linked to the risk of developing diabetes and cardiovascular disease and related to the stressful conditions experienced by expectant mothers in poor areas.

Loi [18] note that epigenetic markers and health inequalities may be related in a number of ways:

- Sensitivity to social structures: Some epigenetic phenomena are highly responsive to environmental changes, which are affected by social institutions.
- Early programing: Several epigenetic traits are established early-on in development, and their effects on health unfold throughout the life course.
- Trans-generational transmission: There is evidence in both animal models and epidemiological studies that epigenetic traits can be trans-generationally inherited. In addition to genetic inheritance that provides adaptive flexibility in the long (evolutionary) time span, epigenetics constitutes other, semi-stable, biological mechanisms through which features are inherited through generations.
- Epigenetic findings raise important questions that have significant policy implications for improving public health and promoting social justice.

## Evolution of Human and Non-Human Primates

Epigenetics has implications for the study of evolution. Epigenetic modifications have the potential to influence evolutionary processes [19]. Studying the epigones of our related species facilitates understanding of the relatively large divergence that has occurred since our emergence from our distant cousins, a significantly large divergence that cannot be solely explained by nucleotide changes [20]. Recent studies point to the fact that dramatic differences occur between humans and chimps in regard to the methylation aspect of epigenetics. Evidence exists that dramatic differences exist between humans and chimps in regard to the methylation aspect of epigenetics. It has been argued that epigenetics can silence older genes that are no longer required, under the influence of selective pressure, thus introducing more plasticity into the expression of genes [21]. Similarly, [22]

proposes that epigenetic changes also function as a time-keeping mechanism, mitigating the negative effects of genetic relics acquired by ancestor populations under different evolutionary pressures. This silencing of older genes that once served a vital purpose epigenetics prevents the build-up of complexity in an organism, silencing older, less frequently transcribed genes

As [11] argue, given that epigenetic marks can be inherited across generations, it is possible that these marks facilitate intergenerational transmission of environmental information and therefore impact the direction of evolutionary change. Biological anthropologists who study nonhuman primates can learn about the evolution of hominids. Indeed, contemporary techniques enable biologists to study differences in DNA methylation in long-dead animals and even in long-extinct species. A recent study of the methylomes of Neanderthals, Denisovans and present-day *Homo sapiens* identified around 2,000 differentially methylated regions in archaic and present-day humans, some of which are related to genes associated with anatomical differences and diseases. These findings suggest that epigenetic variations may have been one of the many factors driving hominid evolution [23].

It is well recognised that modern day humans have survived and thrived in diverse environments for millennia, ranging from the Arctic tundra to Saharan deserts. This ability is directly dependent on the uniquely human ability to adapt quickly with epigenetics playing a central role here. The current main contemporary theory of evolution is Neo-Darwinism, which is based on population genetics and combine Mendelian genetics with Darwin's theory of natural selection. Epigenetic findings suggest that both neo-Darwinian and neo-Lamarckian mechanisms drive evolution, and they appear to be intertwined. Furthermore, new evidence suggests that epigenetic changes, culture and ecological inheritance could be strong drivers of human evolution.

## Migration of Ancient Populations

Epigenetic markers may enhance understanding of patterns of migration among ancient populations. Advances in genetic sequencing help us track prehistoric and historic population movements and allowed migration to be described both as a biological and socioeconomic process. To date, hundreds of DNA methylation differences have been discovered between samples from European, African, and Asian Americans [20]. If relatively stable epigenetic modifications are found among populations, these could provide mechanisms for tracing population histories for related populations that have diverged too recently for sufficient genetic variation to develop. Epigenetic marks may provide some information about the nutrition and stresses experienced by ancient populations. While genetic adaptation can take millions of years, epigenetic changes are much quicker, occurring in just one or two generations. Epigenetics has also helped us confirm other theories of evolution, like the theory that modern humans originated from the Horn of Africa.

## The Impact of Early Experience on Human Development

Epigenetic research may provide valuable information concerning the epigenetic impacts of primarily adverse environmental factors, such as exposure to war trauma and famine [24,25] and how and why some groups develop resistance to these stressors. By now there is voluminous confirmatory evidence that prenatal exposure to maternal stress, anxiety, and depression can result in lasting effects on infant development linked to the appearance of psychopathology later in life. Similarly, postnatal care can impact either positively or negatively on later mental health. Links have been drawn between external stress, epigenetic change and later depression and anxiety. Epigeneticists can now map segments of the pathways whereby environmentally induced epigenetic marks are apparently associated with behavioural outcomes pre- and postnatally [26].

In their seminal text *Revisioning Psychiatry*, [27] argue for a dynamic 'ecosystemic' model in psychiatry: interacting systems are 'ecological' in involving 'biological organisms embedded in and in constant transaction with the environment' at multiple levels (Preface xxi). Epigenetic research into health and illness focuses upon with the ways in which 'the environment' leaves its mark on the genome, and how this then produces particular phenotypic effects. This model argues that stress influences health via an epigenetic mechanism involving altered DNA methylation and gene expression

It is well recognised that childhood adversity is a significant risk factor for poor mental and physical health. Much of the work on epigenetics and mental health has concentrated on child abuse. Adverse Childhood Experiences, including physical, sexual, and emotional abuse, result in a broad range of negative health consequences including adult psychopathology, cardiovascular, and immune disease. A review of the area by [28] reveals increasing evidence deriving from animal, clinical, and epidemiological studies, highlighting the central role of epigenetic programming, such as DNA methylation and histone modification, in altering gene expression, brain structure and function, and ultimately impacting life-course trajectories.

While variation in epigenetic processes, such as DNA methylation emerges as a potential pathway mediating this association; importantly the extent to which different forms of maltreatment may be characterized by unique vs shared epigenetic signatures is currently unknown [29-30] provides evidence that childhood adversity is associated with epigenetic alterations in the promoters of several genes in hippocampal neurons. Finally, [31] observes that genome-wide methylation profiles in adult DNA associated with childhood abuse. For these authors the results justify the further exploration of epigenetic regulation as a mediating mechanism for long-term health outcomes.

The postulation that psychosocial factors act via epigenetic mechanisms to lead to psychiatric disorders has been increasingly supported by experimental data deriving from numerous epigenetic studies on humans [32]. Compared to epigenetic studies on psychiatric disorders like Schizophrenia, Bipolar disorder, and Major depressive disorder, genetic mapping studies have not definitively found any genetic mutation or polymorphism predisposing to these disorders. In addition to these, other psychiatric disorders like Alzheimer's disease, intellectual disability, and autism spectrum disorders are increasing recognised as having an epigenetic role in their pathogenesis

It is possible that the impact of traumatic experiences may be epigenetically inherited via molecular memory that is passed down through generations. There is evidence that traumatic memories may be intergenerationally transmitted through epigenetic mechanisms. Previous research assumed that such transmission was caused by environmental factors, such as the parents' childrearing behaviour. Recent research indicates that these transgenerational effects may have been also (epi) genetically transmitted to their children [33].

Genetic changes stemming from the trauma suffered by Holocaust survivors are capable of being passed on to their children, indicating that one person's life experience can affect subsequent generations. Yehuda et al. [34] studied the genetic makeup of 32 Jewish men and women who had either been interned in a Nazi concentration camp, witnessed or experienced torture or who had had to hide during the second world war. They also examined the genes of their children, who are known to have increased likelihood of stress disorders and compared the results with Jewish families who were living outside of Europe during the war. She observed that gene changes in the children could only be attributed to Holocaust exposure in the parents. Thus Holocaust trauma may transmitted inter -generationally via epigenetic mechanisms.

## New Conceptualisations of Race and Ethnicity

Epigenetic research has led to new conceptualisations of ethnicity, race and genetic ancestry. Galanter [35] notes how race and ethnicity are social constructs and poor markers for genetic diversity, failing to capture the heterogeneity present within racial/ethnic groups and in admixed populations. To explain these heterogenei-ties and in order to avoid social and political controversies, geneticists have instead grouped individuals by genetic ancestry instead of race and ethnicity [36]. Importantly however, racial and ethnic categories also reflect the shared experiences and exposures to known risk factors for disease, such as air pollution and tobacco smoke, poverty, and inadequate access to medical services, which have all resulted in worse disease outcomes in certain populations.

Galanter et al. [34] demonstrate how in a diverse population of Latinos, a substantial number of loci are differentially methylated between ethnic sub-groups. Approximately one-quarter of the epigenetic difference between the two ethnic sub-groups could not be accounted for by differences in the children's genetic ancestry. This difference, the authors suggested, could reflect a biological stamp made by the different experiences, practices, and environmental exposures distinctly related to the two ethnic subgroups.

## Issues in Epigenetic Research

It is important to note that epigenetic research is problematic on a number of grounds: over interpretation of epigenetic findings; data obtained from peripheral tissues rather than brain tissues and some have argued that it is a form of somatic reductionism. Lock [14] argues that with epigenetics, neobiological reductionism is currently occurring due to molecularization of the environment by epigeneticists. She states (p154)

'Epigenetic findings of this kind have certainly opened doors to what has been described as "an explosion of interest in so-called epigenetic mechanisms of gene regulation in the brain" [2], but cultural anthropologists will be wary that we may be entering an era bent on embracing a new round of somatic determinism, this time with attention directed at the cell as the site for primary attention.'

There are several epistemological and social problems pertaining to the rise of epigenetics. These range from the emergence of a new determinism not so dissimilar from an old-fashioned genetic [16], to an intense molecu-larization of sociological and psychological categories [37] from a new exaggerated rhetoric of plasticity and mastery of the genes via epigenetic factors, to the moralistic literature around the role of the maternal body. Some even argue that epigenetics is far from novel and that rather it is simply a variant of established research on gene expression. Such critiques aside, [2] notes that the cultural impact of epigenetics has been profound in showing how permeable the boundaries between nature and nurture, biological and social factors have become.

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