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# ARCHAEOGENETICS AND HEALTH GEOGRAPHY OF DISEASE IN ASSESSING THE EFFECTS OF PANDEMICS

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ABSTRACT

*Recent outbreaks of various deadliest diseases provide a histrionic example of the destruction and dread of epidemics, especially those brought on by newly discovered or remerging diseases. Most research on epidemic diseases is dominated by a focus on managing and preventing infections in living populations. Therefore, a systematic study is critically needed to mitigate the risk and impacts of pandemics before it hits globally in an unprecedented manner. In this regard, the historical study of epidemics, alongside the investigation of the health geography, adds temporal depth to our understanding of the causes and effects of diseases essential for making future predictions about how diseases may affect human biology and demography. This review summarizes some of the advancements in our knowledge of the genetic foundations of diseases, recent human changes, and long evolutionary history that can all contribute to understanding how and why people become vulnerable to epidemics. Analyzing the recent COVID-19 pandemic and Cancer data in this review, it has become evident that evolutionary genetics gradually increase our understanding of geographies of disease by combining the knowledge with the evolutionary history recorded in the human genomes. The increasing availability of diverse genetic information from different populations will help us define an individual's disease risk more precisely in the future.*



## INTRODUCTION

Modern humans have widely migrated over the past 100,000 years to live in every habitable region of the world (Pickrell & Reich, 2014). The history of this expansion draws attention among different disciplines, including genetics, archaeology, linguistics, and physical anthropology, to explore the same question on the evolution of the modern race of humans. Two parallel hypotheses have often been considered to answer the question. On the one side of the debate, demographic stasis arguments presume the idea that residents of a territory are the direct descendants of their ancestors (Wootton, Young, & Winkler, 1991). Other views support the rapid demographic transition model where the present-day inhabitants of a region may have evolved through migrations among several populations due to technological and cultural advancements and the displacement of earlier inhabitants (Bongaarts, 2009).

The “pot versus people” concept in archaeology considers either human migration or cultural transmission as the answer to this debate (Renfrew & Bahn, 2012). The debate has been played out in physical anthropology by bringing up the concepts of in-situ evolution of morphological character change over time or the arrival of a new population to induce craniofacial change and adaptation.

In genetics, alongside the population replacements, “demic diffusion” (Fort, 2015) and “wave of advance” (Ammerman & Cavalli-Sforza, 1979) models have played out around the issue. These theories contend that the farmers’ movement through Europe from Near East partially or entirely replaced the native hunter-gatherers during the Neolithic Period. The “Serial founder effect’ model (Deshpande, Batzoglou, Feldman, & Cavalli-Sforza, 2009) proposed that people stayed in the regions they originally colonized following the out-of-African expansion and exchanged immigrants with their close neighbours at a low rate until the long-distance migrations of the past 500 years.

Several other genetic models were proposed prior to the onset of large-scale genomic data. In 1994 ‘The History and Geography of Human Genes’ was published based on about 100 protein polymorphisms information, including genetic, archaeological, historical, and linguistic details (Cavalli-Sforza, Cavalli-Sforza, Menozzi, & Piazza, 1994). Recent technological advances dramatically increase the quantity of available genomic data for learning the survival and evolution

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of human history through the expedition of archaeological evidence.

Archaeogenetics, in this context, has now dragged two disparate fields of scientific disciplines into one another orbits and uses large-scale genomic datasets to resolve various issues, including the correlation of genetic variation in infectious disease based on the geographical distribution of population. In recent years, emerging infectious diseases, either newly discovered in human populations or previously known but have increased frequency or geographic distribution, have drawn the attention of Archaeogenetics for their recent devastating outbreak (Morse, 1995).

Archaeological evidence provides an opportunity for researchers to examine past diseases and some of their presence in the present day allows them to assess the future trends of epidemic and emerging disease dynamics. Several epidemics in recent decades, including Ebola, severe acute respiratory syndrome (SARS), West Nile Virus, acquired immune deficiency syndrome (AIDS), and Middle East respiratory syndrome coronavirus (MERS-CoV) has created the concern of emerging new diseases in the near future (Morens, Folkers, & Fauci, 2004; Cleaveland, Haydon, & Taylor, 2007). However, the more devastating effects of recent epidemics than previous outbreaks put forward the question of which populations are at the highest risk of death and disease during the emergence of the new epidemic. With the aid of health geography, Archaeogenetics could reduce the lack of temporal depth required to analyse behavioural changes of newly emerging diseases and the relationships between human hosts and pathogens. This review summarizes some of the studies that connect the hypotheses of evolution with the onset of current diseases in a way that benefits people now and in the future.

## LITERATURE REVIEW

### COVID-19

Over six million deaths have already been linked to the COVID-19 pandemic, along with considerable morbidity and mortality (WHO, 2022). The severity of this disease caused by the SARS-CoV-2 virus can range from having few or no symptoms to progressing to respiratory failure (Vetter et al., 2020). It became evident in the early stage of the pandemic that advanced age, several co-morbidities and being a male are the significant risk factors (Zhou et al., 2020). However, it cannot fully explain why some persons experience no or minor symptoms while others do. Thus, the hypotheses emerged that genetic risk factors might influence the progression of the disease.

The severity of COVID-19 was first associated with two genomic areas, one comprised of six genes on chromosome 3, and the other specifies the ABO blood group on chromosome 9 (Ellinghaus et al., 2020). After being made available to the public, a dataset from the COVID-19 Host Genetics Initiative revealed that a small region of chromosome 3 (45,859,651-45,909,024 (hg19)) was linked with the COVID-19 severity on a genome-wide scale (Zeberg & Paabo, 2020). This core Neanderthal-inherited haplotype is present in 16% of Europeans and about 50% of South Asians. (Zeberg, 2022; Zeberg & Paabo, 2020). Compared to east Asia, where it is essentially non-existent, South Asia has a 30% prevalence of the Neanderthal risk haplotype, demonstrating the effect of selection in the past (Zeberg & Paabo, 2020). Thus, the Neanderthal haplotype might significantly increase the risk of COVID-19 in some populations when combined with advanced age-like risk factors.

Currently, it is unclear which features of the Neanderthal-derived region are responsible for the severity of COVID-19, as well as if any effects of such characteristics are specific to SARS-CoV-2, other coronaviruses, or other disorders. The current outbreak, however, makes it clear that Neanderthal gene flow has fatal effects (Luo, 2020). Further elucidation of functional features may speculate the Neanderthal's susceptibility to other pathogens.

### CANCER

Cancer, one of the leading causes of death, is responsible for one in six deaths worldwide (WHO, 2022). Chronic non-infectious disorders like cancer emerged in the early modern period of the second epidemiological transition where primary infectious disease mortality decreased along with the improved living conditions through advanced industrialization (McKeown, 2009). Initially, the first epidemiological transition led to the development of an agriculturally based civilization from hunting and gathering societies, while the world is currently going through the third epidemiological transition due to the resurgence of infectious diseases and novel infections combined with antibiotic resistance (McKeown, 2009). Economic, political, and environmental elements are usually considered to have an impact on both well-being and health at each transition.

However, the antiquity, evolution, and epidemiology of cancer in earlier human populations are very little known. Nevertheless, historical medical records suggest that both Egyptians and the Ancient Greeks were aware of pathological diseases that have been tentatively diagnosed as cancer. Recently the increased incidence of cancer (figure 1) in the world has mostly been blamed on environmental and lifestyle-related causes, including pollution, dietary constituents, smoking, and higher life expectancies (David & Zimmerman, 2010). In addition to lifestyle and environmental exposure, macro or micro-geographic origin can affect an individual's incidence and clinical outcome in cancer-promoting genomic variation (Danaei, Vander Hoorn, Lopez, Murray, & Ezzati, 2005; Siegel, Miller, & Jemal, 2017; Tan, Mok, & Rebbeck, 2016). Several well-known examples include the BRCA1 gene mutations in the ancestry of Ashkenazi Jewish against the mixed-referenced populations (Szabo & King, 1997), notable molecular differences between Caucasians and African Americans' prostate tumours (Khani et al., 2014) and a higher frequency of somatic mutations for TP53, NFE2L2 and EP300 in Chinese vs Caucasian patients diagnosed with ESCC (esophageal squamous cell carcinoma) (Deng et al., 2017).

Geographic disparities and ethnicity can be attributed to cancer cases, prognosis, and clinical outcomes. Recent large-scale sequencing initiatives worldwide started unravelling the genomic basis of cancer traits, although there is still much to learn about the underlying mechanism and causes of genomic diversity. Information based on geographic location can be used better to understand the health behaviours and screening of cancer patients while extending their chances of

survival (Goodwin et al., 2020; Møller et al., 2018). Our understanding of cancer genetics is anticipated to advance with the growing knowledge of hereditary and somatic genomes, resulting in better cancer detection, prevention, and treatment options. For instance, even while existing screening programmes continue to focus on high-risk cohorts with an emphasis on age, lifestyle, or family history, it is anticipated that demographically susceptible variances will create chances for risk categorization or risk mitigation interventions.

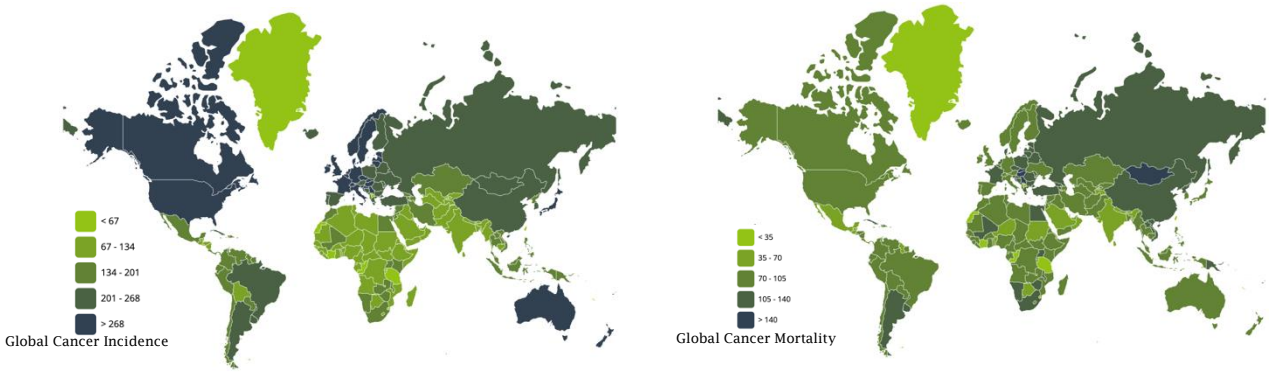


Figure 1. Age-standardized incidence and mortality rates (per 100000) of worldwide all cancers in both sexes. Data were obtained from the World Cancer Research Fund International (https://www.wcrf.org/cancer-trends/global-cancer-data-by-country/)

## CONCLUSIONS

Combining knowledge of the geographic distribution of disease and the evolutionary consequences of the human lineage from recent genomic studies is essential for assuming the disease risk and association framework. As recent epidemics have exposed the hypothesis that signatures of evolutionary histories are recorded in human genomes, insights from evolutionary genetics are now gradually enabling our understanding of these histories with high accuracy, depth, and resolution. The increasing availability of diverse genetic information from different populations nowadays also helps us map genetic traits precisely, which will define an individual's disease risk more accurately.

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