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The peopling of the African continent and the diaspora into the new world

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Africa is the birthplace of anatomically modern humans, and is the geographic origin of human migration across the globe within the last 100,000 years. The history of African populations has consisted of a number of demographic events that have influenced patterns of genetic and phenotypic variation across the continent. With the increasing amount of genomic data and corresponding developments in computational methods, researchers are able to explore long-standing evolutionary questions, expanding our understanding of human history within and outside of Africa. This review will summarize some of the recent findings regarding African demographic history, including the African Diaspora, and will briefly explore their implications for disease susceptibility in populations of African descent.

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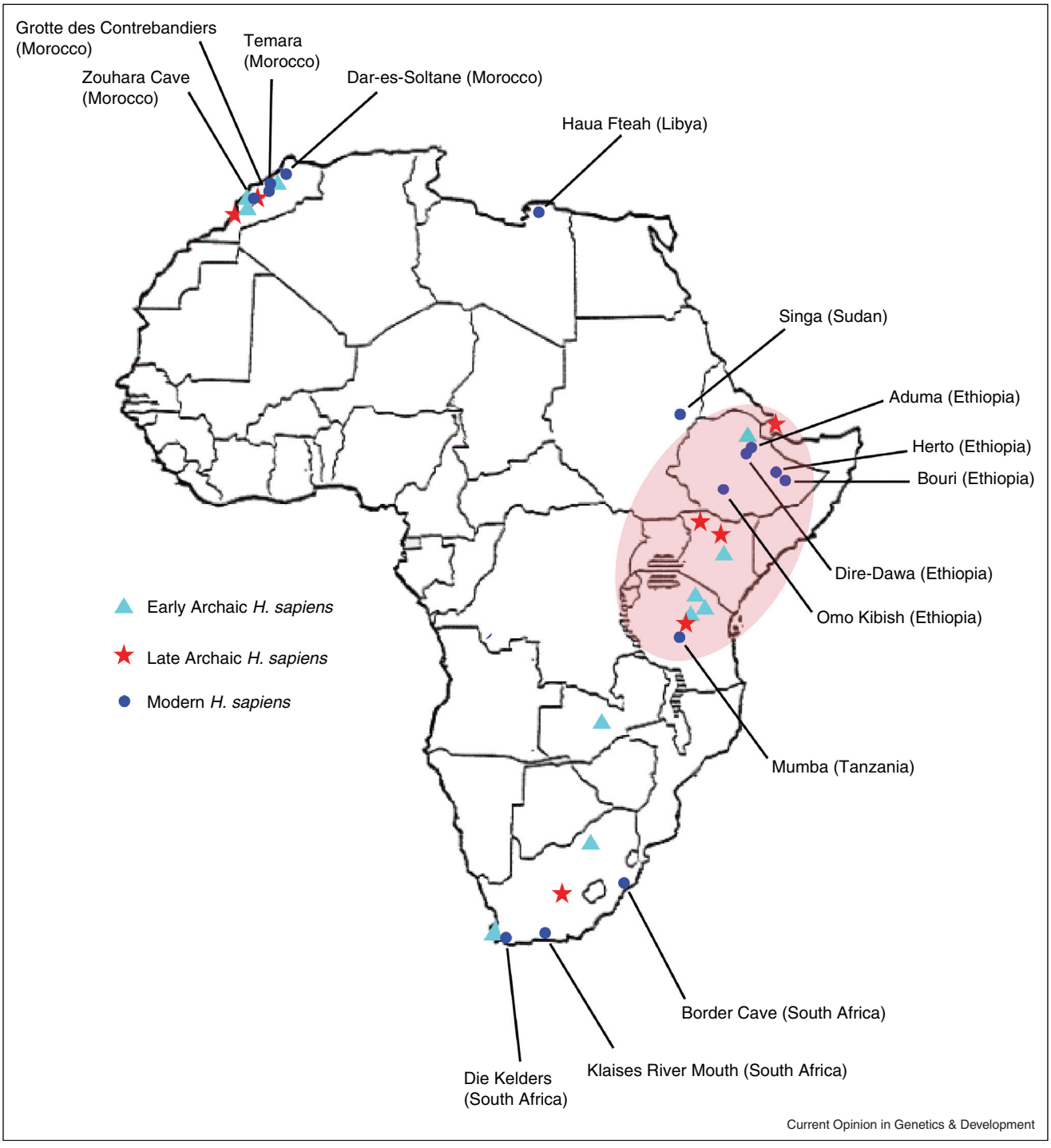
Introduction

Current paleontological and genetic evidence indicates that anatomically modern humans (AMHs) arose in Africa ~200 thousand years ago (kya) and have lived continuously on the African continent longer than in any other geographic region. African populations are characterized by higher levels of within-population and between-population genetic diversity relative to non-Africans consistent with a larger long-term effective population size of ancestral African populations [1–3]. The history of Africans has encompassed other demographic events such as population structure, admixture, long-range and short-range migration that have shaped patterns of genetic variation in modern populations [1,4]. In recent years, the resequencing of large portions of the genome and their analysis with new computational methods have increased power to infer past demographic events at an unprecedented resolution. Most notably, the recent finding that non-African populations share ancestry with Neanderthals, consistent with a model of archaic introgression, has provided additional insights into human evolutionary history. However, the extent of archaic admixture in diverse African populations still remains unclear. Given the central role of Africa in human evolution, characterizing extant genomic variation in diverse Africans will be important for reconstructing both ancient and recent demographic events, and for identifying variants that play a role in disease susceptibility in African populations. Here, we summarize our current knowledge of modern human origins and patterns of genetic diversity in populations of African descent, as well as explore their implications for the risk of complex disease.

The origin of anatomically modern humans in Africa

The earliest suite of derived morphological traits associated with AMHs was identified in fossils from Ethiopia dating to ~160–195 kya (Figure 1; Table 1) [5–7]. Other early AMHs displaying modern features were also found in Ethiopia, Sudan, Tanzania and South Africa dating to >100 kya and in the Middle East dating to ~100 kya (Figure 1; Table 1) [8–12]. Although eastern Africa has often been considered the geographic location of modern human origins ~200 kya, some have argued that South Africa is the site where AMHs originated. Indeed, a recent study suggested that the geographic distribution of genetic diversity in Africa, as measured by linkage disequilibrium (LD), is more consistent with a South African origin of modern humans [13]. However, this inference does not account for the possibility that the geographic location of populations in the present may have differed in the past. Furthermore, a large-scale analysis of southern African populations demonstrated the difficulty of localizing the origin of modern humans using summary statistics of diversity, such as LD [14••]. Nevertheless, regardless of the precise location of origin, paleontological and genetic evidence indicates that AMHs evolved on the African continent.

Figure 1



The geography of major *Homo sapiens* (*H. sapiens*) fossil sites. This map, adapted from [8], illustrates the geographic distribution of sites in Africa where early archaic, late archaic and modern *H. sapiens* have been found. The labeled sites are the locations of fossil remains that have been designated as modern *H. sapiens*. A more detailed description of the 'modern' features of these fossils is given in Table 1.

Recent archaeological data also showed that modern behavior (such as symbolic culture and complex tool production) arose at a relatively early stage of human evolution, contrary to prior studies that argued for the later development of complex cognition ~45 kya [15,16]. In particular, technological advances in the form of heat-treated microlith stone tools were found in southern Africa dating to ~71 kya [17•,18]. The use of pigment,