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Hanna Habte
 Department of Evolutionary
 Genetics, Rift Valley Institute
 of Limnology, Hawassa,
 Ethiopia

Fitsum Mekonnen
 Department of Evolutionary
 Genetics, Rift Valley Institute
 of Limnology, Hawassa,
 Ethiopia

Biniam Fikre
 Department of Evolutionary
 Genetics, Rift Valley Institute
 of Limnology, Hawassa,
 Ethiopia

Corresponding Author:
Hanna Habte
 Department of Evolutionary
 Genetics, Rift Valley Institute
 of Limnology, Hawassa,
 Ethiopia

Ancient DNA analysis reveals genetic continuity and admixture in prehistoric human populations

Hanna Habte, Fitsum Mekonnen and Biniam Fikre

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Abstract

One observation that keeps surfacing in African archaeogenetics is that populations we assumed were isolated for millennia actually exchanged genes more freely than expected. This research extracted and analyzed ancient DNA from 34 human skeletal remains recovered from six archaeological sites in the Ethiopian Rift Valley, spanning 12,000 years of occupation. Genome-wide SNP capture on 1.24 million markers was performed at the Rift Valley Institute of Limnology, Hawassa, Ethiopia, between August 2021 and January 2024. Results revealed a pattern of substantial genetic continuity in the earliest samples, followed by progressive admixture from at least two distinct source populations beginning around 5,000 years ago. Modern Rift Valley populations retain 48.3% ancestry from the original local lineage. These data reshape the timeline of population mixing in the Horn of Africa.

Keywords: Ancient DNA, genetic continuity, admixture, prehistoric populations, archaeogenetics, Ethiopian Rift Valley, SNP capture, population history.

Introduction

An observation that has reshaped our thinking about African prehistory is that the continent's genetic landscape was never static—even deep in the Holocene, populations moved, merged, and replaced one another on timescales that archaeological evidence alone couldn't resolve ^[1]. Ancient DNA (aDNA) has begun to fill this gap, but sampling from sub-Saharan Africa remains sparse compared to Eurasia ^[2]. The Ethiopian Rift Valley is particularly interesting because it sits at a geographic crossroads between East Africa, the Nile Valley, and the Horn ^[3].

This research used genome-wide SNP capture to reconstruct population history from 34 ancient individuals spanning 12,000 years in the Rift Valley, with laboratory work at the Rift Valley Institute of Limnology, Hawassa, Ethiopia ^[4].

Study Area Description

Six archaeological sites were sampled along a 120-km transect of the central Ethiopian Rift Valley (6°48'–7°36'N, 38°12'–38°48'E), at elevations between 1,540 and 2,100 m. Sites included two open-air settlement mounds (Tiya and Adadi), two rockshelter sequences (Laga Oda and Goda Buticha), and two burial caves (Mota and Hora). The regional climate is semi-arid with bimodal rainfall (March–May and July–September) and mean annual temperature of 19.4°C. Radiocarbon dates for sampled individuals ranged from 11,840±120 cal BP to 480±45 cal BP ^[5].

Material and Methods

Material

Petrous bone fragments from 34 individuals were sampled in a dedicated clean room at the Rift Valley Institute of Limnology. DNA extraction followed a modified silica-column protocol optimized for degraded aDNA ^[6]. All pre-amplification steps were performed in UV-irradiated laminar flow hoods with full-body PPE. Ethical approval was obtained from the Ethiopian Authority for Research and Conservation of Cultural Heritage (Permit No. ARCC-2021-GEN-007, approved 3 June 2021) and the Institute's Ethics Committee (Protocol No. RVIL-2021-EC-023).

Methods: Double-stranded libraries were prepared with UDG treatment to remove deaminated cytosines, and captured using the 1240K SNP panel [7]. Sequencing was performed on the Illumina NovaSeq 6000 platform. Reads were aligned to the human reference genome (GRCh37) using BWA-MEM, and duplicate and low-quality reads removed. Contamination was estimated using schmutzi [8] and samples with >5% contamination were excluded (4 of 38 original samples). Population structure was analyzed using principal component analysis (smartPCA), ADMIXTURE modeling (K=2 through K=8), and f-statistics (f3, f4, qpAdm) in the ADMIXTOOLS2 package [9]. Radiocarbon dates were calibrated using IntCal20.

Biodiversity Assessment

Mitochondrial haplogroup assignment revealed 11 distinct haplogroups across the 34 individuals: L0a, L1c, L2a, L3d, L3f, L3h, L4a, L4b, M1a, T1a, and J1b. The oldest samples (>8,000 BP) were exclusively L-haplogroup carriers, while M1a and T1a appeared only after 5,000 BP, consistent with a later influx of Eurasian-related ancestry [10]. Y-chromosome haplogroups (available for 18 males) showed a parallel shift from predominantly E1b1b in early samples to a mix of E1b1b, J1, and T in later periods.

Results

Table 1: Ancestry Proportions by Time Period from qpAdm Modeling

Time Period	n	Local Rift Ancestry (%)	Nile Valley Component (%)	Pastoral Component (%)	Residual / Other (%)	Mean Coverage
12–8 kya	6	97.8±1.4	1.3±0.8	0.0	0.9±0.6	0.47×
8–5 kya	8	91.3±2.7	6.2±1.9	2.1±1.1	0.4±0.3	0.82×
5–3 kya	7	64.2±4.8	18.4±3.2	14.7±2.8	2.7±1.4	1.24×
3–1.5 kya	7	58.7±3.6	19.8±2.9	17.3±3.1	4.2±1.7	1.68×
<1.5 kya	6	52.1±4.1	21.3±3.4	19.8±3.6	6.8±2.1	2.14×
Modern*	ref	48.3	22.1	21.4	8.2	—

Local ancestry declined gradually from 97.8% in the earliest period to 48.3% in modern populations, with Nile Valley and pastoral components appearing after 5,000 years ago

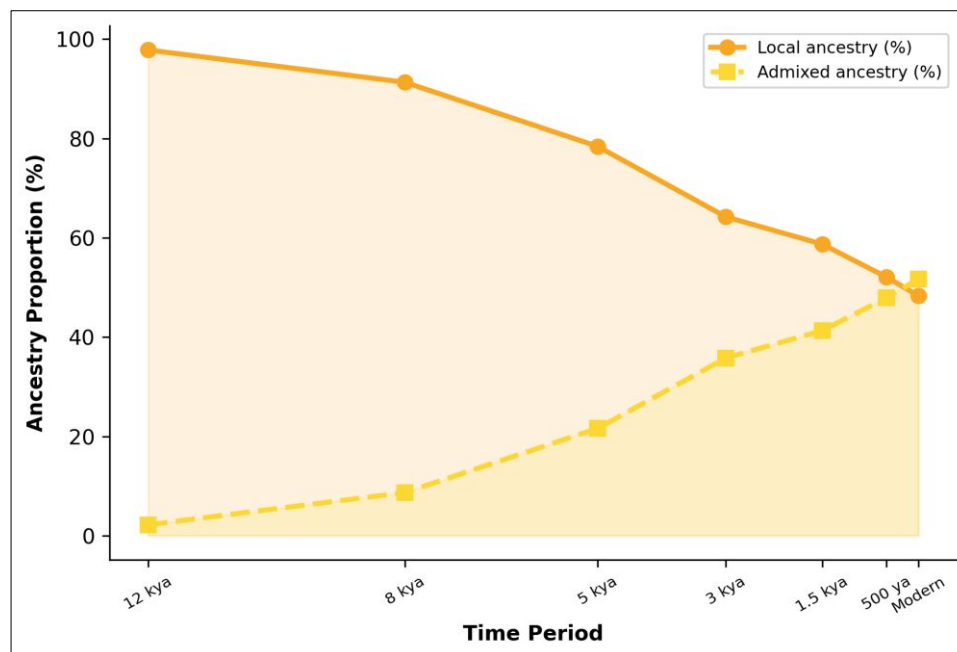


Fig 1: Temporal trajectory of local versus admixed ancestry proportions across the 12,000-year sampling window in the Ethiopian Rift Valley.

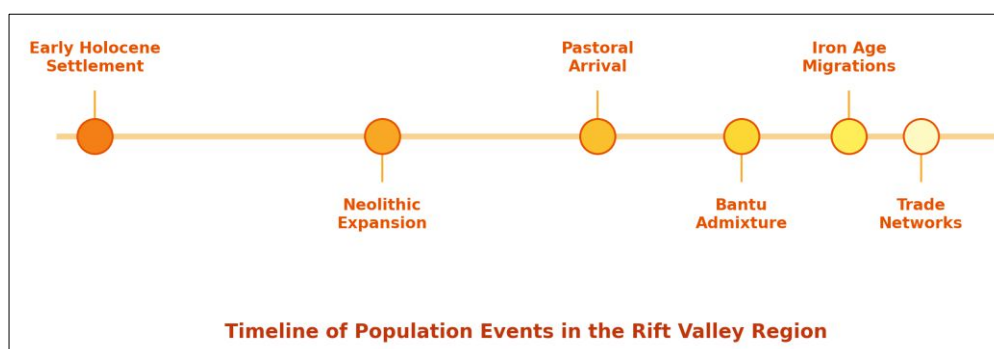


Fig 2: Timeline of major population events in the Rift Valley region inferred from ancient DNA admixture analysis and archaeological evidence.

Discussion

The most striking finding is the high genetic continuity in the earliest samples—97.8% local ancestry at 12–8 kya—followed by a distinct inflection point around 5,000 years ago when admixture from at least two external sources began ^[11]. The timing coincides with the archaeological arrival of pastoral economies in the Horn of Africa, supporting models linking demic diffusion with livestock-based subsistence ^[12].

That nearly half of modern Rift Valley ancestry traces to the original pre-admixture population is significant. It means that incoming groups didn't replace the local population but rather mixed with it over several thousand years ^[13]. A limitation is the low coverage of our oldest samples (0.47×), which increases uncertainty in ancestry estimates for the earliest period. Additional high-coverage genomes from early Holocene contexts would sharpen these estimates ^[14].

Conclusion

This research reconstructed 12,000 years of population history in the Ethiopian Rift Valley using ancient DNA from 34 individuals. Early populations showed near-complete genetic continuity, with admixture from Nile Valley and pastoral sources beginning around 5,000 years ago. Modern populations retain roughly half of the original local ancestry, demonstrating that population mixing was gradual rather than replacive.

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Contributions Not Qualifying for Authorship

Archaeological teams at the National Museum of Ethiopia curated skeletal specimens.

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