



Investigating the Italian genetic variation through time with ancient complete genomes

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Background

Italy has some peculiar characteristics:

- Located in the heart of the Mediterranean Sea;
- Presents diverse environments and climates;
- Is a peninsula accessible from both the European mainland and the sea;
- Was a crossroads for migration, trade and cultural exchange for thousands of years;
- Presents genetic contributions from across Eurasia, with various populations and cultures.

Ancient DNA provides valuable insights into these complex dynamics thanks to

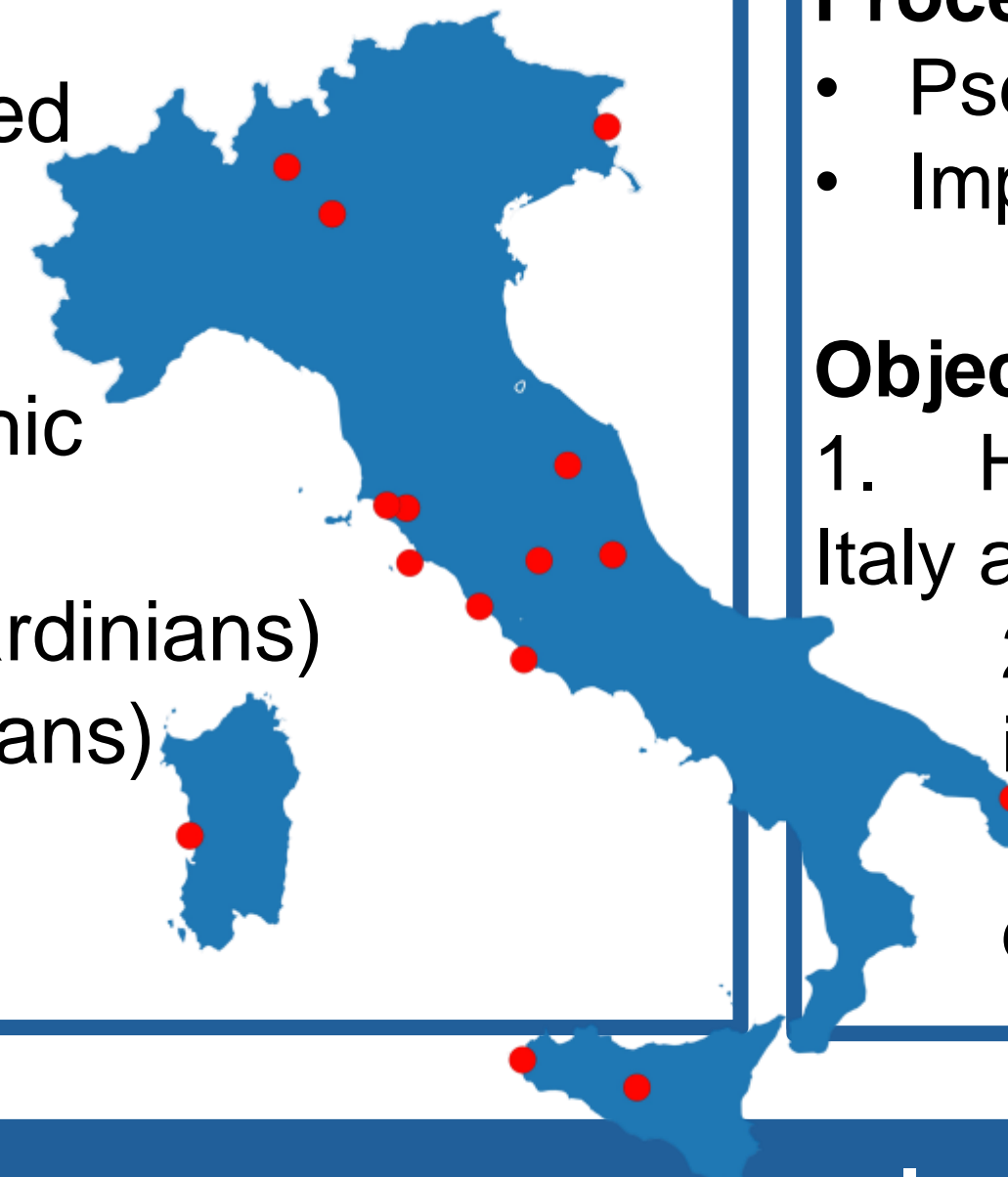
- growing availability of complete ancient genomes;
- enhancement of statistical tools for analysis.

New Ancient Samples

We analyzed 43 new ancient whole genome sequences from 16 Italian necropoleis from the Neolithic to the Middle Ages. 2 individuals have been discarded.

The remaining **41** are subdivided temporally as follows:

- 2 individuals from the Neolithic
- 14 from the Chalcolithic
- 5 from the Bronze Age (3 Sardinians)
- 11 from the Iron Age (4 Sicilians)
- 9 from the Middle Ages



Methods and Objectives

Data used as reference:

- 292 published ancient samples;
- 792 modern samples.

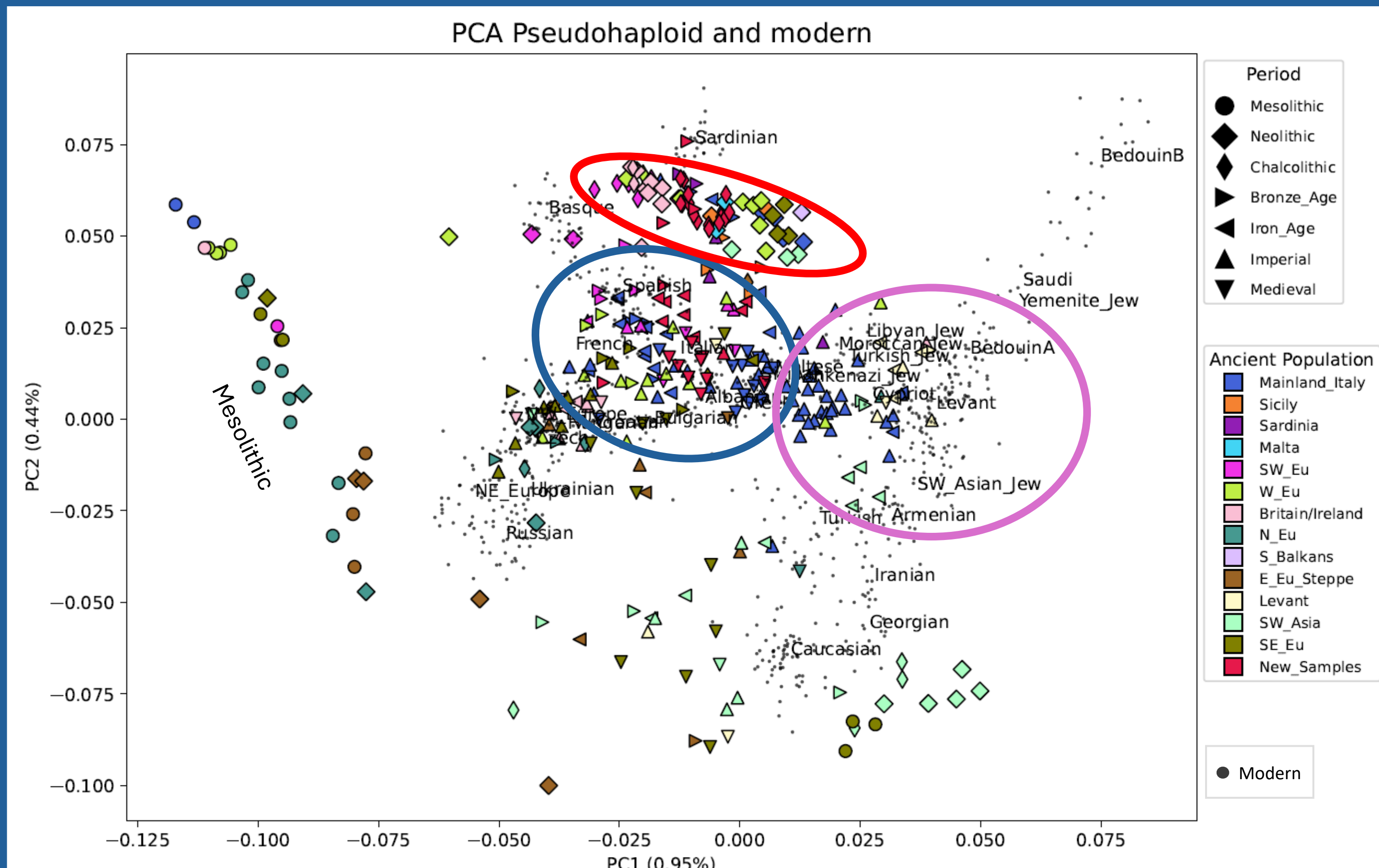
Processed in two different ways:

- Pseudo-haploidization;
- Imputation.

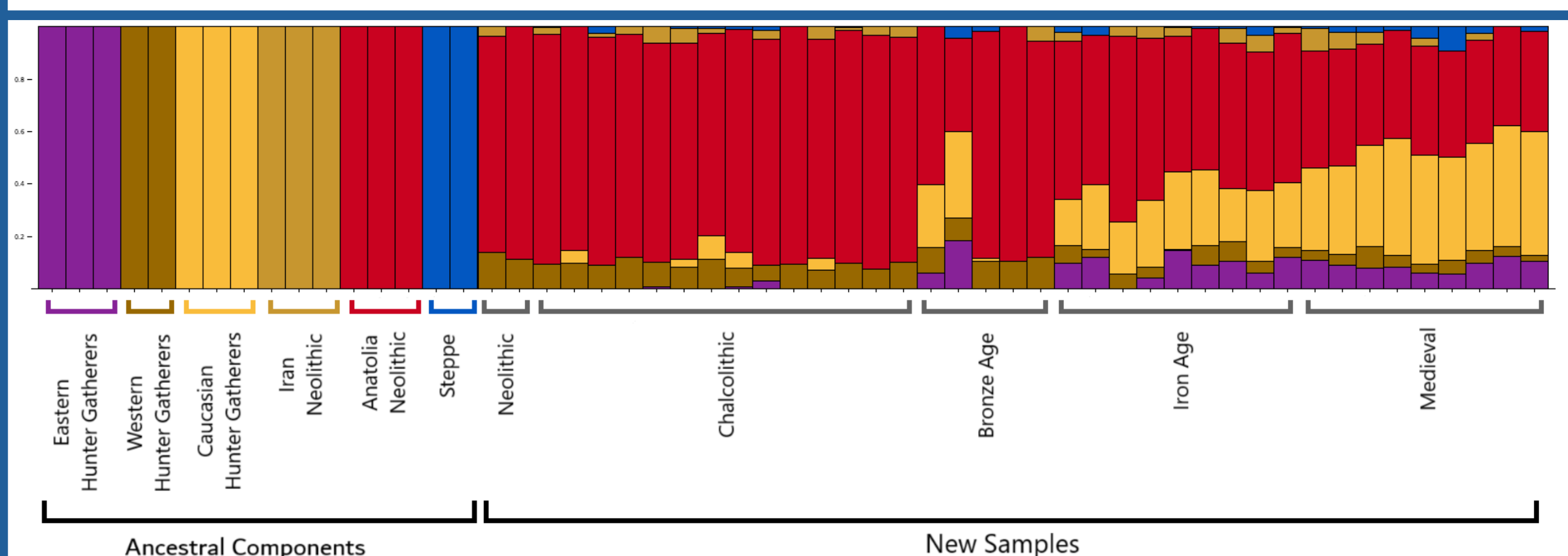
Objectives:

1. How migrations affect the genetic composition of Italy and how it relates to the European context;
2. How Ancient genetic components have influenced the current Italian genetic variation;
3. What Demographic scenario best describes the migrations that occurred in Italy.

Pseudo-haploid Data

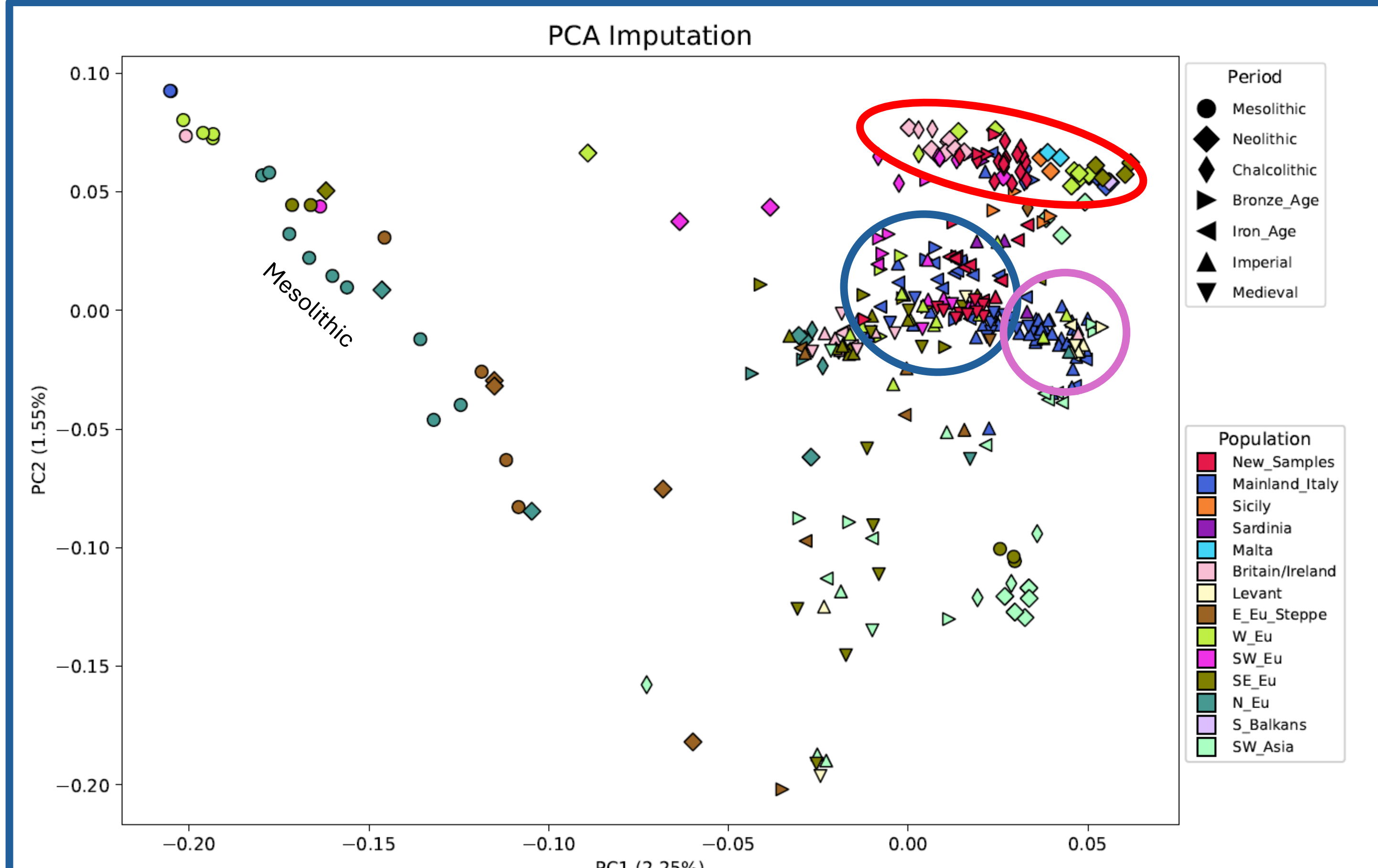


- Mesolithic individuals are distant from every Modern Individual;
- Neolithic and Chalcolithic individuals are close to Modern Sardinians (Red);
- Iron Age and Medieval individuals are close to Modern Europeans (Blue);
- Imperial individuals are closer to modern Middle Easterners and Levantine (Pink).

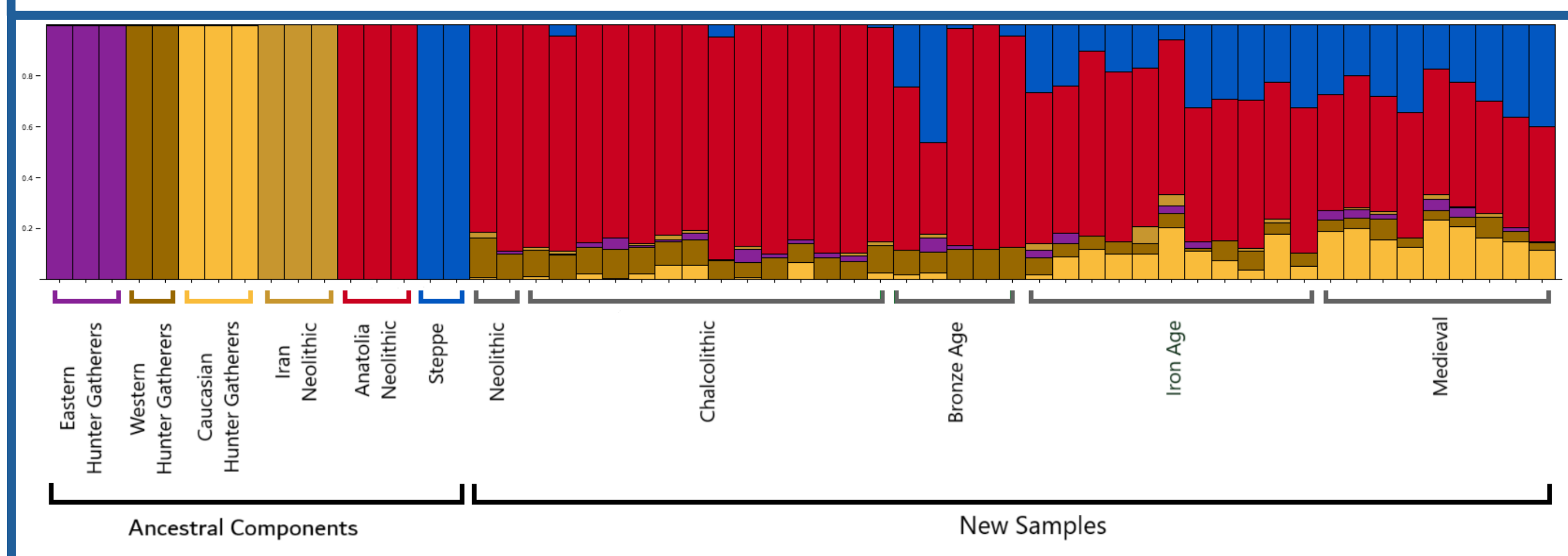


- Great similarity among Neolithic and Chalcolithic individuals with a main Anatolia Neolithic component and a minor Western Hunter gatherers one.
- With the Bronze age samples new eastern components appear (CHG, EHG). These components are absent in the three Sardinians.
- The CHG component is slightly higher in Medieval individuals compared to Iron Age individuals.

Imputed Data



- PC1 separates Mesolithic individuals from all the subsequent periods.
- Neolithic and Chalcolithic Italian early farmers are separated from the rest (red)
- Iron Age and Medieval Italians cluster with other Europeans (Blue)
- Imperial Italians are close to ancient South-western Asians and Levantine people (Pink)
- Bronze age Sardinians cluster with early farmers



- Similarity among Neolithic and Chalcolithic individuals with a main Anatolia Neolithic component and a minor Western Hunter gatherers one.
- With the Bronze Age new eastern components appears, mainly a STEPPE component, as expected from literature. These components are absent in Sardinians
- The CHG component is present only from the Iron Age onwards.

Conclusions

We recognize three major events during Italian Prehistory and early history:

- **Neolithic transition** that separates Mesolithic individuals
- The arrival of eastern **populations from the steppe** during the Bronze age that separates clearly Europeans before and after this period.
- The expansion of the **Roman Empire** towards east that brings a new Middle-Eastern/Levantine component. During the Middle ages Italians probably continued admixing with other Europeans but not with the Middle-Easterners, diluting the Middle-Eastern component.

