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Deciphering the Genomic History of the Rendena Valley People

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Genomic studies have greatly increased our knowledge of the genetic history of human populations. Present-day Italians (i) harbour a higher genetic diversity compared to other European populations, an effect of various genetic contributions since the Upper Paleolithic, and (ii) cluster into four major groups: Sardinia, Northern, Central and Southern Italy. This diversity is being further explored at the microgeographic level. In this study, we analyzed the population genomics of a community from the Rendena Valley, in the Italian Region of Trentino-Alto Adige/Südtirol. A total of 125 individuals having ancestors living in this valley ('Rendeneri') were sampled. First, we assessed their mitochondrial DNA variation by sequencing complete mitogenomes from 95 Rendeneri maternally unrelated and with the grandmother from Val Rendena. Haplotype and haplogroup distributions show high internal variability and an unusually high frequency of haplogroup U5a (18%), the highest detected so far in Italy. Age estimates of the main subclades U5a1a1 and U5a2c suggest Bronze Age connections with central-eastern Europeans and populations from central Asia, including some nomadic groups (Yamnaya) from the Pontic-Caspian steppes. To further investigate this hypothesis, 48 individuals were selected for genome-wide genotyping with the Human Origin SNP chip. Exploratory analyses of allele frequencies provided new insights into the genomic makeup of the valley. They not only confirm the high level of variation in the Rendereni, but also highlight their peculiar placement relative to the genomic cline previously reported for the Italian Peninsula, thus raising additional questions about past demographic events.