Ancient genomes show social and reproductive behavior of early Upper Paleolithic foragers

Sikora, Martin; Seguin-Orlando, Andaine; Sousa, Vitor C.; Albrechtsen, Anders; Korneliussen, Thorfinn Sand; Ko, Amy; Rasmussen, Simon; Dupanloup, Isabelle; Nigst, Philip R.; Bosch, Marijolein D.; Renaud, Gabriel; Allentoft, Morten Erik; Margaryan, Ashot; Vasilyev, Sergey V.; Veselovskaya, Elizaveta V.; Borutskaya, Svetlana B.; Deviese, Thibaut; Comeskey, Dan; Higham, Tom; Manica, Andrea; Foley, Robert; Meltzer, David J.; Nielsen, Rasmus; Excoffler, Laurent; Lahr, Marta Mirazon; Orlando, Ludovic Antoine Alexandre; Willerslev, Eske

Published in:
Science

DOI:
10.1126/science.aao1807

Publication date:
2017

Document version
Publisher's PDF, also known as Version of record

Citation for published version (APA):
Ancient genomes show social and reproductive behavior of early Upper Paleolithic foragers

Martin Sikora,1 Andaine Seguin-Orlando,1,6 Vitor C. Sousa,2,5,6 Anders Albrechtsen,5 Thorfinn Korneliusson,3 Amy Ko,7 Simon Rasmussen,7 Isabelle Dupanloup,2,3 Philip R. Nigst,8 Marjolein D. Bosch,9,10 Gabriel Renaud,1 Morten E. Allentoft,1 Ashot Margaryan,1,11 Sergey V. Vasilyev,12 Elizaveta V. Veselovskaya,12 Svetlana B. Borutskaya,13 Thibaut Deviese,14 Dan Comeskey,14 Tom Higham,14 Andrea Manica,15 Robert Foley,1,16 David J. Meltzer,1,17 Rasmus Nielsen,1,5 Laurent Excoffier,2,3 Marta Mirazon Lahr,1,16 Ludovic Orlando,1,18 Eske Willerslev1,19,20

Present-day hunter-gatherers (HGs) live in multilevel social groups essential to sustain a population structure characterized by limited levels of within-band relatedness and inbreeding. When these wider social networks evolved among HGs is unknown. To investigate the distribution of relatedness among Upper Paleolithic individuals buried at the Sunghir site, we sequenced complete genome sequences (from Sunghir 1, SI) and compared them with panels of modern and ancient humans. We found that SI is from ~34,000 years before the present, and that its maternal and paternal haplotypes are distantly related to two other Upper Paleolithic individuals, SIIB and SIIC. We screened six complete genome sequences (from SI to SIIV) for evidence of inbreeding, and found that SI and SIIC are closely related, possibly as extensive as the ~36,000-year-old Kostenki 12; SIIB and SIIC are more distant but still related, possibly as extensive as the ~100,000-year-old Vindija 20. We found that SI and SIIC are more closely related to each other than to SIIB and SIIC, suggesting that SI and SIIC were the offspring of a single couple. We also found evidence of inbreeding in SI and SIIC, which may represent a single social group. One such example of multiple burials is Sunghir, a site harboring two of the most extraordinary Upper Paleolithic burials (3, 4) (fig. S1 and S2 and tables S1 to S6): one of an adult male (Sunghir 1 (SI)) and another of two subadults (Sunghir 2 and 3 (SII and SIII)), originally thought to be a boy and girl, interred head-to-head. All remains were covered in ochre and were accompanied by rich grave goods including ivory beads and spears, armbrads, and carvings, as well as arctic fox canines. Adjacent to SII was the femoral diaphysis of an adult [Sunghir 4 (SIV)] that had been polished, hollowed out, and filled with red ochre. The site also yielded other less complete human remains, some of which were stratigraphic provenance [Sunghir 5 to 9 (SV to SIX)]. Radiocarbon analyses place the age of SII to SIV between 33,600 and 34,600 years (5, 6). The homogeneity in morphological traits (e.g., metopism) among the remains, as well as signs of possible congenital pathologies in SIII, have been interpreted as evidence of inbreeding (3). Other Upper Paleolithic individuals with reported congenital or degenerative pathologies (e.g., at Barma Grande and Doini Vêstonico) (3) reinforce the view that Upper Paleolithic groups were small and susceptible to inbreeding, possibly as extensive as what has been reported for the Altai Neandertal (7). However, genomic data available for some of those individuals (8) were of insufficient coverage to infer population sizes or inbreeding levels. We screened six of the Sunghir individuals (SI to SIV) to assess DNA preservation: five of them (all but SV) yielded sufficient endogenous DNA for genome sequencing. We sequenced these genomes to an average depth of coverage ranging from 11X to 107X (figs. S3 and S4 and tables S5 and S6) and compared them to panels of modern and ancient human genomes (4). All individuals were genetically male on the basis of the fraction of Y chromosome reads (table S7), including SIIB, who was previously identified as female (3). Contamination levels from X chromosome heterozygosity were low (0.33 to 0.90%; table S5), except for SVI (13.1%). Radiocarbon dates indicate that whereas SV is only slightly more recent than the other individuals, SI is from ~900 years before the present (figs. S5 and S6 and tables S9 to S10). Together with mitochondrial DNA (mtDNA) and Y chromosome haplogroups (W3a1 and I2a1b2, respectively), these data indicate that SI is not associated with the Upper Paleolithic burials at the site; SVI was therefore excluded from further analyses.

Analyses of mtDNA genomes place SI to SIV in haplogroup U, consistent with West Eurasian and Siberian Paleolithic and Mesolithic genomes (9) (fig. S7 and table S6). SI belongs to haplogroup U8c; the sequences for the three individuals from the double burial (SIIV to SIV) are identical and belong to haplogroup U2, which is closely related to the Upper Paleolithic Kostenki 12 (8) and Kostenki 14 (10) individuals. Phylogenetic analyses of the Y chromosome sequences place all Sunghir individuals in an early divergent lineage of haplogroup C1a2 (fig. S8 and tables S12 to S23). Y chromosome haplogroup C1, which is rare among contemporary Estonians, has been found in other early European individuals, including the ~36,000-year-old Kostenki 14 (11).

We investigated the degree of relatedness among the Sunghir individuals with a method that allows relationships inferences up to the third degree but does not rely on allele frequencies (4). Surprisingly, none of them were found to be closely related (that is, third degree or closer), even though the SI to SIV individuals buried together share both mitochondrial and Y chromosome lineages (Fig. 1 and tables S16 to S23). We then inferred genomic segments that were identical by descent (IBD) and homzygous by descent (HBD) from three-higher-content Sunghir genomes (SI to SIV) and a panel of ancient and contemporary humans (4). We compared their distributions to those inferred from whole genomes obtained using coalescent simulations (12) of randomly mating populations with varying effective population sizes (Ne) (fig. S9). The distributions of HBD between different AMHSs and archaic humans, indicating small effective population sizes and/or recent inbreeding in archaic

1Centre for GeoGenetics, Natural History Museum of Denmark, University of Copenhagen, 1350 Copenhagen, Denmark. 2Institute of Ecology and Evolution, University of Bern, CH-3012 Bern, Switzerland. 3Swiss Institute of Bioinformatics, 2015 Lausanne, Switzerland. 4Centre for Ecology, Evolution and Environmental Changes, Faculdade de Ciências, Universidade de Lisboa, 1749-016 Lisboa, Portugal. 5Department of Biology, University of Copenhagen, 2200 Copenhagen N, Denmark. 6Department of Integrative Biology, University of California, Berkeley, CA 94720, USA. 7Centre for Biological Sequence Analysis, Department of Systems Biology, Technical University of Denmark, Kemitorvet, 2800 Kongens Lyngby, Denmark. 8Division of Archaeology, Department of Archaeology and Anthropology, University of Cambridge, Cambridge CB2 3EJ, UK. 9McDonald Institute for Archaeological Research, University of Cambridge, Cambridge CB2 3ER, UK. 10Department of Human Evolution, Max Planck Institute for Evolutionary Anthropology, 04103 Leipzig, Germany. 11Institute of Molecular Biology, National Academy of Sciences, 01044 Yerevan, Armenia. 12Centre of Physical Anthropology, Institute Ethnology and Anthropology, Russian Academy of Science, 191534 Moscow, Russia. 13Department of Anthropology, Biological Faculty, Lomonosov’s Moscow State University, 119991 Moscow, Russia. 14Oxford Radiocarbon Accelerator Unit, University of Oxford, Oxford OX1 3QY, UK. 15Department of Zoology, University of Cambridge, Cambridge CB2 3JE, UK. 16Leverhulme Centre for Human Evolutionary Studies, Department of Archaeology and Anthropology, University of Cambridge, Cambridge CB2 1QH, UK. 17Department of Anthropology, Southern Methodist University, Dallas, TX 75275, USA. 18Laboratoire d’Anthropologie Moléculaire et d’Imagerie de Synthèse, CNRS UMR 5288, Université de Toulouse, Université Paul Sabatier, 31000 Toulouse, France. 19GeoGenetics Groups, Department of Zoology, University of Cambridge, Cambridge CB2 3JE, UK. 20Wellcome Trust Sanger Institute, Hinxton, Cambridge CB10 1SA, UK.

*These authors contributed equally to this work. †Corresponding author. Email: ewillerslev@snm.ku.dk
Fig. 1. Relatedness among ancient Eurasians. Kinship coefficients and R1 ratios (4) were inferred from IBS counts. (A) Pairs of Upper Paleolithic individuals, using 1000 Genomes Phase 3 single-nucleotide polymorphism (SNP) sites. (B) Pairs of ancient Eurasians, using 1240K capture SNP sites. Within-group pairs of Sunghir are highlighted.

Fig. 2. Identity by descent and recent effective population sizes. (A and B) Distributions of the number and total length of HBD and IBD segments in modern, ancient, and archaic humans (Altai Neandertal and Denisovan). Ellipses indicate 95th percentile (dark gray) and 99th percentile (light gray) of the distributions inferred from simulated data of various $N_E$ values. Individuals with previously described close relatedness and their degree are indicated in (B). (C) Distributions of inferred recent effective population sizes for modern and ancient HGs with a minimum of three individuals, as well as simulated data sets of randomly mating populations with a range of $N_E$ values. For each population, a pair of symbols and box plots indicates the distributions of population sizes inferred from individual HBD (left) and pairwise IBD tracts (right).
individuals, particularly the Altai Neandertal (7, 13) (Fig. 2A and figs. S10 to S14). Patterns of pairwise IBD sharing successfully detect close genetic relatives in modern individuals (Fig. 2B). However, the Sunghir pairs do not share sufficiently long IBD tracts to suggest relatedness at the first or second degree, consistent with the results from genome-wide identity-by-state (IBS) counts (Fig. 2B and fig. S15). Interestingly, the effective population sizes tended to be higher ($N_e \approx 500$) for two of three Sunghir pairs than those estimated from HBD segments ($N_e \approx 200$). $N_e$ from both HBD and IBD tracts (6) was within the range of, or slightly higher than, that of contemporary non-African HG populations (Fig. 2C), particularly from genetically isolated groups (14).

Genetic clustering of ancient individuals using outgroup-$f_2$ statistics ($f_2$(Mbuti; Ancient1, Ancient2)) indicates shared genetic drift and tight clustering of the Sunghir individuals, which form a cline to the exclusion of all other individuals (Fig. 3, figs. S16 to S20, and tables S28 and S29). Furthermore, we find genetic affinities between the Sunghir individuals and those from Kostenki (12 and 14), as well as with the “Vestonice cluster” (6) associated with the Upper Paleolithic Gravettian culture.

Individuals mapped onto a previously inferred admixture graph of early Eurasians (4, 8) placed the Sunghir cluster as a descendent of a lineage related to the Kostenki 14 individual, contributing the major fraction of the ancestry of the Vêstonice cluster (Fig. 3C and figs. S21 to S24). Adding the shared genetic drift [outgroup-$f_2$ statistics $f_2$(Mbuti; Ancient1, Ancient2)].

Our results suggest a social and population network of HG demes that preferentially mated within subgroups, with exogamy and regular exchanges between demes. Among contemporary HGs, primary kin constitute <10% of residential
groups, leading to low genetic relatedness (20, 21). Some modern human groups exhibit increased levels of inbreeding, including populations where consanguineous marriage practices are encouraged, or geographically isolated HG groups such as those from the Amazon rainforest region (Fig. 2, A and C). In contrast, patterns of HBD among the Upper Paleolithic individuals are consistent with randomly mating populations of moderate effective size ($N_e \approx 200$), which suggests that close consanguineous matings were avoided (Fig. 2, A and C). Although our findings are currently limited to a single Upper Paleolithic site, if they are representative of early Upper Paleolithic HGs more generally, they reveal a social structure and cultural practices that emphasized exogamy. This is consistent with archaeological evidence of high mobility in the Upper Paleolithic (22), perhaps comparable to the scale of mobility seen ethnographically among small foraging bands at high latitudes (23). We note that this interpretation relies on the evidence that all individuals at Sunghir were contemporaneous and members of the same social group. This is clearly the case for the two children in the double burial (SII and SIII). It is possible that both SIV and SI were members of different social groups, potentially separated in time from SII and SIII. However, the shared material culture, overlapping radiocarbon date intervals, and close genetic relationship among all individuals support this inference.

Although the number of ancient genomes available remains small, the differences in inbreeding levels, and thus group organization, between AMH groups in the Upper Paleolithic and Neandertals are intriguing. The small reproductive groups of Upper Paleolithic AMHs at Sunghir apparently avoided inbreeding and its deleterious consequences, in contrast to what has been observed for the Altai Neandertals. We caution that more genomic data on Neandertals from other regions is necessary to conclude whether the patterns observed in the Altai are representative of their genetic diversity more generally, or whether that individual was an outlier. Assuming the former, whether this would reflect ongoing extinction of Neandertals or a more general difference in social behavior and cultural practices also remains unknown. Our results nonetheless suggest that the human HG social structure of low levels of within-band relatedness, complex family residence patterns, relatively high individual mobility, and multilevel social networks were already in place among Upper Paleolithic societies 34,000 years ago. This social structure may have affected the development of cooperation and information transfer that underlie the evolution of culture in humans (20, 21, 24, 25) and may be crucial to understanding our species’ unique evolutionary resilience and trajectory.

REFERENCES AND NOTES


3. See supplementary materials.


ACKNOWLEDGMENTS

Supported by the FPT Marie Curie program (CIG Nr 322661), Isaac Newton Trust, Leakey Foundation, and McDonald Grants and Awards Fund (P.R.N.), the H2020 Marie Skłodowska Curie program (EF Nr 656325) (M.D.B.); Swiss NSF grant 310033A-143393 (L.E., I.D., and V.C.S.); ERC Consolidator Grant 647787/LocalAdaptation (A. Manica); and the ERC’s Seventh Framework Programme ("PalaearChnot" grant no. 324139) (T.H., T.D., and D.C.). GeoGenetics is supported by the Lundbeck Foundation, Danish National Research Foundation grant DNRF94, and the University of Copenhagen (KU2016 programme). We thank L. Moreau, M. Gerassimova, and A. Sinistyn for helpful discussions, and the staff of the Danish National High-Throughput DNA Sequencing Centre and the laboratory technicians of the Centre for GeoGenetics for technical assistance. Genomic data are available for download at the European Nucleotide Archive, accession number PRJEB22592.

SUPPLEMENTARY MATERIALS

www.sciencemag.org/content/358/6363/659/suppl/DC1

Supplementary Text

Figs. S1 to S37

Tables S1 to S29

References (20-159)

22 June 2017, accepted 25 September 2017

Published online 5 October 2017

10.1126/science.aao3807
Ancient genomes show social and reproductive behavior of early Upper Paleolithic foragers


Science 358 (6363), 659-662. DOI: 10.1126/science.aao1807 originally published online October 5, 2017

How early human groups were organized

Sequencing ancient hominin remains has provided insights into the relatedness between individuals. However, it is not clear whether ancient humans bred among close relatives, as is common in some modern human cultures. Sikora et al. report genome sequences from four early humans buried close together in western Russia about 34,000 years ago (see the Perspective by Bergström and Tyler-Smith). The individuals clustered together genetically and came from a population with a small effective size, but they were not very closely related. Thus, these people may represent a single social group that was part of a larger mating network, similar to contemporary hunter-gatherers. The lack of close inbreeding might help to explain the survival advantage of anatomically modern humans.

Science, this issue p. 659; see also p. 586