

Polar and brown bear genomes reveal ancient admixture and demographic footprints of past climate change

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AUTHOR SUMMARY

There is overwhelming evidence that Earth's climate is changing. These changes are most noticeable in the Arctic, which has recently experienced a dramatic loss of sea-ice cover (1). Altered environments may lead to shifts in the distributions of closely related, formerly isolated species, permitting hybridization (i.e., admixture). Such phenomena likely shaped the evolution of many organisms but may become especially pronounced in the rapidly changing Arctic, where it could have tremendous impacts on polar biodiversity (2). The polar bear, a signature Arctic species, symbolizes the threat to biodiversity from climate change. We reasoned that knowledge of patterns of species divergence, migration, and extinction across past periods of severe climatic change during the Pleistocene might inform predictions about future organismal responses. Here, we analyzed the genomes of polar, brown, and American black bears. Although our results clearly demonstrate that these bear species have had largely independent evolutionary histories over millions of years, leaving imprints in the polar bear nuclear genome likely associated with ecological adaptation to the Arctic environment, our data also indicate ancient admixture between the species. Further, our results suggest that bear evolution has tracked key climate events, including a dramatic decline in their population for the past 500,000 y.

Based on fossil and mitochondrial DNA evidence, the polar bear has long been thought to have evolved recently from its lower-latitude sister species, the brown bear. For example, sequences of complete mitochondrial genomes have suggested a split of the maternal lineages of these species ~150 kya

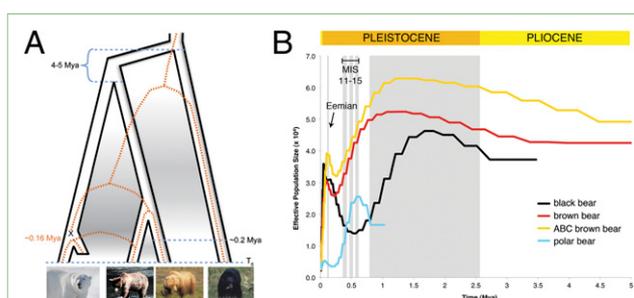


Fig. P1. Bear evolutionary history inferred from genome-scale data. (A) Evolutionary relationships of three bear species highlights discordance between maternally inherited mitochondrial (orange dashed line) and biparentally inherited nuclear (black outline) genomes likely as a result of ancient admixture events. The figure illustrates extinction ("X") and replacement of polar bear mitochondrial DNA with an ABC brown bear mitochondrial genome around 160,000 years ago, although the opposite scenario (i.e., capture of polar bear mitochondrial genome in the modern ABC brown bear) cannot be excluded. Also shown are proposed divergence times among American black, brown, and polar bear lineages (indicated in Mya), as well as the cessation of intermittent periods of gene flow (gray shading) between brown and black bears and between brown and polar bears. The lineage of the ancient polar bear, sequenced in this study, is indicated as extinct. (B) Estimates of changes in effective population size during the past 5 million years, as inferred from four bear genomes, American black bear, non-ABC brown bear, ABC brown bear, and polar bear. The larger gray-shaded area refers to the Early Pleistocene, at the end of which Earth's glaciations became more severe, and the smaller gray areas (from right to left) refer to interglacial Marine Isotope Stages (MIS) 15, 13, and 11, and the Eemian, respectively. (Photos: S. Farley, J. Schoen, Ø. Wiig.)

and confirmed a particularly close relationship between the polar bear and a genetically isolated population of brown bears from the Admiralty, Baranof, and Chichagof islands in Alaska's Alexander Archipelago (henceforth called ABC brown bears) (3). However, limited nuclear DNA sequence data acquired more recently suggests that polar bears may have diverged from brown bears approximately 450,000 y earlier (4). Our strategy was to examine such questions with far greater power, from a genome-wide perspective. Modern genomic tools have already provided tremendous analytical potential for increasing our understanding of complex speciation and genetic interchange among humans and domesticated

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species, whereas studies with whole-genome sequencing to investigate admixture in wildlife populations are only now beginning to emerge.

We performed deep, high-throughput sequencing of the genomes of a polar bear, two ABC brown bears, a non-ABC brown bear, and an American black bear. We also performed less-extensive sequencing of 23 other polar bear genomes, including a ~120,000-y-old specimen. We were able to identify more than 13 million single nucleotide polymorphisms (SNPs) that vary among individuals, including 26,000 SNPs within coding regions of corresponding dog genes that result in amino acid replacements. By computational predictions of the functional effect of each amino acid substitution (i.e., single amino acid polymorphisms), 7,000 were suggested to be damaging.

Our comparative analyses demonstrate that these bear species evolved largely independently over a period of millions of years, which is in sharp contrast to the more recent estimates of polar bear origin (Fig. P1). Moreover, 5% to 10% of the nuclear genome of the ABC brown bears is most closely related to polar bears, indicating ancient admixture between the species. Previously used gene-by-gene sequencing of single nuclear loci lacked sufficient power to detect such ancient admixture (4).

Our results are consistent with an ancient split between brown and polar bears approximately 4 to 5 Mya (Fig. P1A), coinciding with the Miocene–Pliocene boundary, a period of environmental change that may have launched a radiation of bear species (5). This initial split was followed by occasional admixture until recently, leaving a clear polar-bear imprint on the nuclear genomes of ABC brown bears. Genome-based analysis of historical fluctuations in effective population size (i.e., number of interbreeding bear individuals) strongly indicates that polar bear evolution has tracked key climatic events since the Middle Pleistocene; moreover, it suggests that, although the number of interbreeding individuals polar bears may have been

considerably larger in the past, there have been significant population decreases during periods of long-term climate changes (Fig. P1B). A prolonged and considerable bottleneck in the polar bear population in addition to recent expansions from small founder populations could explain the observed lower genetic diversity in contemporary polar bears compared with brown bears. Since their divergence from brown bears, polar bears have embarked on their own evolutionary pathway and developed their own unique genomic signatures. Thus, we identified potential regions of genes that may relate to adaptation to the Arctic environment, including those controlling fatty-acid metabolism, hibernation, and pigmentation.

Because of the uncertain long-term status of the polar bear and its charismatic nature, this species has become a focal point for discussions concerning the impact of global climate change on biodiversity. Previously, only limited genetic resources have been available to investigate questions of divergence and adaptation of bears. Here, we describe extensive genome sequence data from three different species that contribute to a better understanding of past responses during polar bear evolution and provide investigators with extensive resources that allow the formulation of testable hypotheses to establish key morphological and physiological features associated with adaptation to polar climates.

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