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Letter to the editor

### Whole-genome sequences shed light on the demographic history and contemporary genetic erosion of free-ranging jaguar (*Panthera onca*) populations



Whole-genome sequences provide massive amounts of data that can illuminate many aspects of a species' biology and evolutionary history, as well as valuable information for conservation planning. This is particularly important for large carnivores, whose large area requirements and typical low densities make them prone to undergo rapid genetic erosion (e.g. accumulation of runs of homozygosity, Díez del Molino et al., 2018; Leroy et al., 2018) in the face of habitat loss and human persecution. One such species is the jaguar (*Panthera onca*), the largest extant felid in the Americas, whose range has already declined by >50% and some of whose remaining populations have been severely impacted by human-induced habitat fragmentation (De La Torre et al., 2018).

To characterize jaguar genomic diversity and demographic history, we analyzed 13 complete jaguar genomes from individuals sampled in 7 ecoregions across the species' distribution, from its main core in South America (Amazon and Pantanal biomes) towards both the southern and northern ends of the present-day range, in the Atlantic Forest of southern Brazil, and the Mexico/USA border region, respectively (Fig. 1A). The methods of genome resequencing, variation detection, and downstream analyses are shown in the online Supplementary Data. The genomes were sequenced at 21× to 37× depth of coverage (Table S1), allowing the identification of 8,991,736 SNPs, of which 6,429,763 were shared among individuals and 2,561,973 were private. The latter ranged from 83,298 in Atlantic Forest individual AF052 to 334,655 in Amazonian individual AM404 (Table S2).

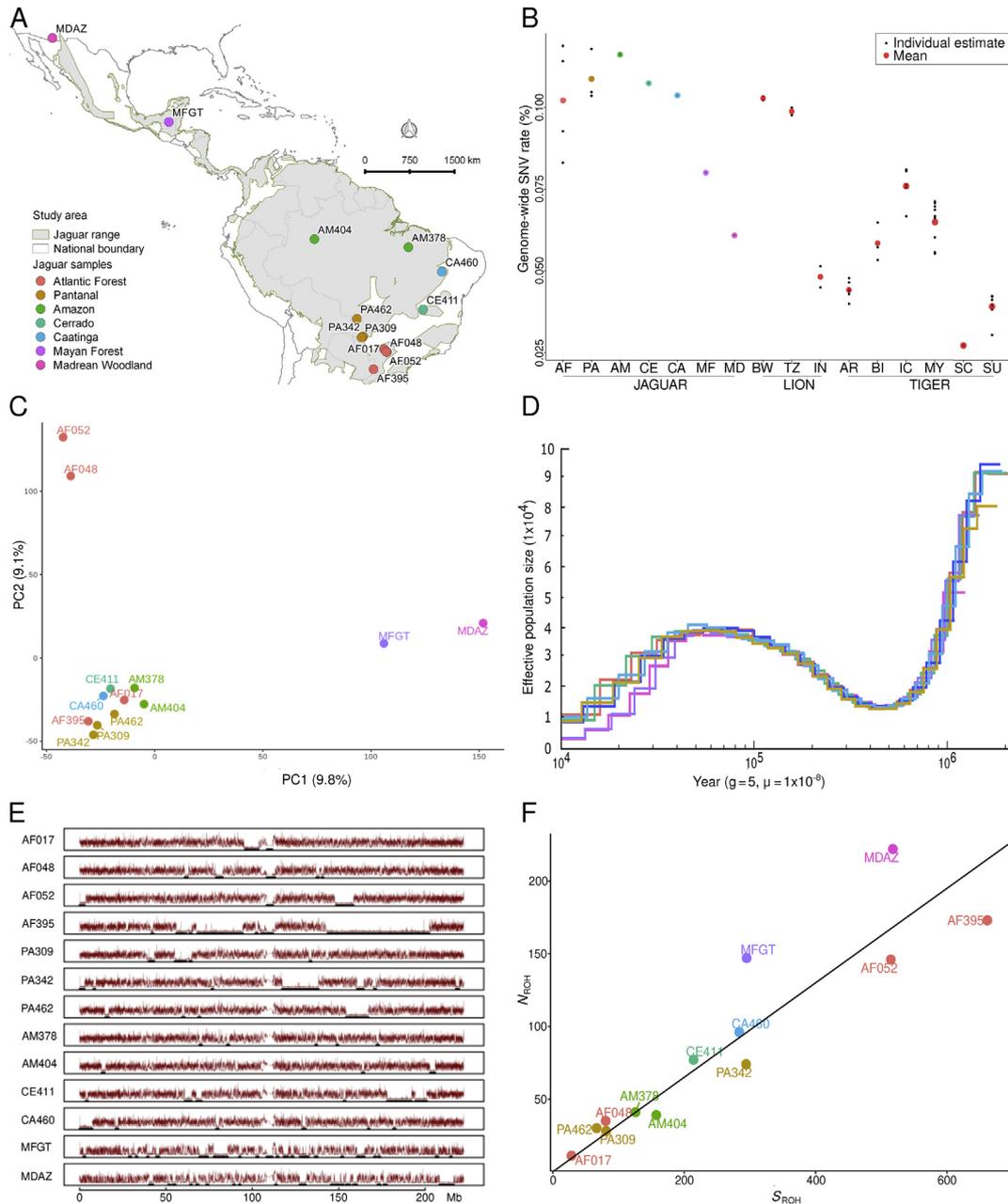
South American jaguars (except for the Atlantic Forest AF052 and AF395 individuals) exhibited consistently higher levels of genome-wide diversity than those sampled in Central and North America (Fig. 1B). For example, the heterozygous SNV rate (in %) of the Madrean and Mayan individuals was half (0.06) and two-thirds (0.08), the amount observed in the most diverse individuals from the Amazon, Pantanal, and Atlantic Forest biomes (ca. 0.12). A consistent pattern was observed regarding mean heterozygosity (Fig. S1). These results could be attributed to the joint effects of historical and contemporaneous processes, mainly range expansions and contractions across the Americas, and the increased isolation (both natural and human-induced) of some of the remaining breeding populations, as discussed below.

We also compared jaguar genomic diversity (SNV rate) with that observed in lions (*Panthera leo*) and tigers (*P. tigris*), which led to the following observations: South American jaguars exhibited at least as much diversity as African lions and a consistently higher diversity than any sampled tiger population. Central/North American jaguars

showed less diversity than African lions and similar SNV rates to the most diverse tiger populations. The lowest levels of diversity among these pantherine species were observed in the Asiatic lions and Amur, South China, and Sumatran tiger populations, all of them severely endangered (Liu et al., 2018; De Manuel et al., 2020).

To investigate population differentiation in jaguars, we performed Principal Component (PCA) and admixture analyses. They revealed a discernible structure between North and South American jaguars, with a less marked structure within the latter group, following a pattern that suggests isolation by distance (Figs. 1C and S2). The only exceptions were three individuals from the Atlantic Forest (AF048, AF052, and AF395): the former two (which were sampled at the same site) were very distinct from all others on PC2 (Fig. 1C) and in the Admixture analysis (with the best-fit  $k = 3$ ), while the latter was very distinct on PC3 (Fig. S3). The strong differentiation signal observed on those three individuals can be attributed to genetic drift due to habitat loss and fragmentation that occurred in the last few decades in the Atlantic Forest (Haag et al., 2010; Srbek-Araujo et al., 2018).

We then estimated past fluctuations in jaguar effective population size ( $N_e$ ) with the pairwise sequentially Markovian coalescent (PSMC) method (Li and Durbin, 2011). The demographic trajectories were very congruent among all the individuals throughout most of the inferred time. All of them begin 1–2 Mya, with initial  $N_e$  values ranging from  $4 \times 10^4$  to  $8.5 \times 10^4$ . Then all plots show a marked decline in  $N_e$ , decreasing to ca.  $1.5 \times 10^4$  ca. 0.5 Mya. Subsequently, a gradual population expansion ensued, reaching ca.  $2 \times 10^4$  around 30,000 years ago, followed by a new round of reduction that affected all individuals (Fig. 1D). Interestingly, the trajectories of all South American individuals remain similar after this point, reaching a  $N_e$  of ca.  $1 \times 10^4$  at the end of the plot, 10,000 years ago, while both the Mayan forest and Madrean individuals show a distinct path, with a more pronounced decline in this final phase, ending with a  $N_e$  of ca.  $3 \times 10^3$ . The congruent trajectories corroborate inferences from previous studies (e.g. Eizirik et al., 2001; Lorenzana et al., 2020) that jaguar populations have remained highly connected across the species' range until recent times. We hypothesize that the departure from this shared pattern observed in the most recent phase of the assessed history (<30,000 years ago) for the Mayan and Madrean individuals (Fig. 1D) could have resulted from a more recent process of colonization of this region, located at the northern end of the species' current range, most likely driven by an expansion of South American lineages, as suggested previously (Eizirik et al., 2001). Such



**Fig. 1.** Jaguar (*Panthera onca*) genome-wide diversity and population history. **A:** Free-ranging individuals ( $n = 13$ ) sampled in seven different biomes/ecoregions (see internal legend) across the species' range in the Americas. **B:** Genome-wide heterozygous SNV rate (in %) in three pantherine cats: jaguars (data from this study; AF, Atlantic Forest; PA, Pantanal; AM, Amazonia; CE, Cerrado; CA, Caatinga; MF, Mayan Forest; MD, Madrean Woodland), lions (*P. leo*, data from De Manuel et al. [2020]; BW, Botswana; TZ, Tanzania; IN, India) and tigers (*P. tigris*, data from Liu et al. [2018]; AR, Amur; BI, Bengal; IC, Indo-Chinese; MY, Malayan; SC, South China; SU, Sumatra). **C:** Jaguar population structure based on a Principal Component Analysis (PCA) plot showing PC1 x PC2. **D:** Reconstruction of historical fluctuations in effective population size ( $N_e$ ) based on the genome-wide distribution of heterozygous sites, using the pairwise sequential Markovian coalescent (PSMC) applied to the 13 jaguar whole-genome sequences. For clarity, only one individual per biome is shown, as their trajectories are very similar, and there is virtually no within-biome variation in the reconstructed pattern. **E:** Runs of homozygosity (ROH) > 1 Mb (thick dark lines) detected on chromosome C1 in jaguars. The full set of ROH plots for every chromosome is shown in Fig. S8. **F:** Jaguar relative ROH burden based on the total number of ROH ( $N_{ROH}$ ) and the sum total length of ROH ( $S_{ROH}$ ). An approximation of the recent demographic history of each population can be obtained by the relative position of each individual along the plot (cf. Fig. 1 in Ceballos et al., [2018]).

a scenario implies the extinction of pre-existing North American jaguar lineages and recolonization from South America, in a process that may have included a succession of founder events, analogous to the pattern exhibited by the puma (*Puma concolor*) in the Americas (Culver et al., 2000; Saremi et al., 2019). For the jaguar, the lower productivity of habitats in North America north of Mexico, relative to moister environments farther south, may have contributed to keeping jaguar densities and effective sizes at lower levels in this area (Jędrzejewski et al., 2018).

Finally, we investigated genomic patterns indicative of recent demographic history based on the detection of runs of homozygosity (ROH). ROHs were detected in each individual after parsing  $8.98 \times 10^5$  biallelic SNPs shared by the 13 individuals (i.e. no missing data), which were identified with a sliding window analysis (Kardos et al., 2018) of the jaguar's 18 autosomes across the full set of whole-genome sequences (Table S3). After discarding short ROH, which are indicative of ancient inbreeding episodes, we identified 1119 runs of homozygosity longer than 1 Mb, adding up to 3.32

Gb of IBD segments along the 13 genomes altogether (Figs. S4 and S5; Table S4). The Atlantic Forest AF395 jaguar showed the heaviest ROH burden, with 661 Mb distributed in 164 long (1–10 Mb) and 9 very long ROHs (>10 Mb), implying that 27.5% of its genome is homozygous as a consequence of recent inbreeding. On the other hand, individuals AF017 and PA462 exhibited the lowest cumulative sums of ROH ( $S_{ROH}$ ), with 28.3 and 66.7 Mb distributed in 11 and 30 segments, respectively, representing ca. 1%–3% of their genomes in IBD tracts of recent origin (Fig. S5; Table S4). Considering the 13 jaguar genomes altogether, the mean proportion ( $F_{ROH}$ ) comprised by long ROHs was ca. 10%, while very long ROHs were detected in half of the individual genomes (7 out of 13), totaling 451 Mb in 33 different segments (Figs. S6–S8; Table S4). Again, AF395 showed the heaviest load of very long ROH (126 Mb in 9 segments), including the single longest homozygous tract detected in our sample, spanning nearly 25 Mb on chromosome C1 (Fig. 1E). Next, another Atlantic Forest individual, AF052, exhibited 115 Mb in nine very long ROH. For those seven individuals, the total proportion of the genome comprised by very long IBD segments ranged from 0.94% in CA460 to 5.27% in AF395. The mean pairwise genome-wide ROH overlap was 34 Mb. Chromosomes A2 and A3 showed the most pairwise- and multiple-individual overlapping tracts, respectively (Tables S5–S7).

Overall, our results indicate that most South American jaguar populations have been relatively large until very recently but are clearly very sensitive to fast genomic erosion triggered by anthropogenic habitat fragmentation leading to rapid drift and inbreeding. For instance, although the rate of heterozygous SNV documented for most jaguars in this study is considerably higher than those found in free-ranging tigers (Liu et al., 2018), the Atlantic Forest AF395 individual showed a 40% reduction in genome-wide diversity compared to its most diverse conspecifics in our sample. With respect to homozygosity tracts, the longest single ROH we identified across the sampled jaguars reached 25 Mb in length in this same individual (AF395; Table S4). ROH >15 Mb could be indicative of close inbreeding, with a common ancestor < 3 generations ago (Saremi et al., 2019), and we detected 5 of those instances in 3 of the sampled biomes (Atlantic Forest, Amazon, and Pantanal; Table S4). The comparison of relative ROH burden allowed us to approximate the recent jaguar population history, following the approach described by Ceballos et al. (2018). Amazon jaguars (AM378, AM404) and two Pantanal individuals (PA309, PA462) exhibited signatures of relatively larger populations, as their overall ROH burden was very slight (ca. 6%), while Cerrado and Caatinga samples indicate smaller populations (Fig. 1F). One Pantanal animal (PA342) showed an intriguing pattern suggestive of local inbreeding, which should be investigated with additional samples from that site. The Madrean (MDAZ), and to a lesser degree the Mayan (MFGT) jaguars, exhibited potential signals of both historical and recent bottlenecks, likely influenced by their location at the northern edge of the species' range. In the case of MDAZ, that pattern is probably exacerbated by suboptimal conditions of habitat productivity and prey biomass, perhaps also affected by recent pressure from human disturbances (Fig. 1F). A noteworthy case was that of individual AF017, sampled in a transitional area between the Atlantic Forest and the Cerrado and Pantanal biomes (and whose jaguar population has subsequently been extirpated; Haag et al., [2010]), which seems to be admixed, harboring signatures of regional gene flow, before the onset of anthropic-driven population size reductions that seem to characterize remaining Atlantic Forest fragments (Fig. 1F). The major source of concern arising from our ROH results pertains to jaguars AF395 and AF052 (both from the Inner Atlantic Forest), which bear signs of a bottlenecked and potentially inbred population, supported by the fact that at least 20% of their genomes are homozygous due to recent IBD tracts (Table S4). The former individual, sampled in

the Atlantic Forest Green Corridor, showed worrisome signals of inbreeding, even more severe, than those observed for other Inner Atlantic Forest individuals (AF048 and AF052) coming from populations that had been previously identified as having comparatively stronger signals of genetic drift due to habitat loss and fragmentation (Haag et al., 2010). This can be attributed to the fact that this individual was sampled more recently (in 2010, as a juvenile) than the other analyzed animals from this biome, especially AF017 (which was first captured in 1994 as a ~8-year-old adult). It is remarkable that the Inner Atlantic Forest harbors two individuals that are on the opposite sides of the spectrum in Fig. 1F, i.e. the ones with the highest (AF395) and lowest (AF017) ROH burden. Based on its estimated time of birth (the late 1980s), which was prior to more intense anthropogenic disturbance, and the transitional nature of the area in which it was sampled, we infer that AF017 represents an individual bearing signature of inter-biome connectivity that occurred until very recently. This outcome suggests that increased inbreeding has occurred very quickly after recent massive deforestation in that biome (Ribeiro et al., 2009), leaving forest fragments with only a few related individuals (Canale et al., 2012; Paviolo et al., 2016).

The genome-wide results presented here add to the sense of urgency of restoring connectivity among remaining jaguar populations in Atlantic Forest fragments (as highlighted by Srbek-Araújo et al., [2018]) so as to rescue their genetic diversity and retain their evolutionary potential, likely already impacted by the loss of adaptive alleles and inbreeding. Overall, our study illustrates the power of genome-wide analyses to investigate the demographic history of large carnivores across a broad geographic range, illuminating not only evolutionary trajectories leading to present-day populations but also providing critical information that can be incorporated into conservation strategies on their behalf.

#### Data availability

Variant sites (*vcf*) and sequence (*psmcf*) files are available at Mendeley Data server (DOI: [10.17632/bgmg4rd2nw.1](https://doi.org/10.17632/bgmg4rd2nw.1)).

#### Conflict of interest

All authors declare no conflict of interest.

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#### Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.jgg.2021.10.006>.

#### References

- Canale, G.R., Peres, C.A., Guidorizzi, C.E., Gatto, C.A.F., Kierulff, M.C.M., 2012. Pervasive defaunation of forest remnants in a tropical biodiversity hotspot. *PLoS One* 7, e41671.
- Ceballos, F.C., Joshi, P.K., Clark, D.W., Ramsay, M., Wilson, J.F., 2018. Runs of homozygosity: windows into population history and trait architecture. *Nat. Rev. Genet.* 19, 220–234.
- Culver, M., Johnson, W.E., Pecon-Slatery, J., O'Brien, S.J., 2000. Genomic ancestry of the American puma (*Puma concolor*). *J. Hered.* 91, 186–197.
- De La Torre, J.A., González-Maya, J.F., Zarza, H., Ceballos, G., Medellín, R.A., 2018. The jaguar's spots are darker than they appear: assessing the global conservation status of the jaguar *Panthera onca*. *Oryx* 52, 300–315.
- De Manuel, M., Barnett, R., Sandoval-Velasco, M., Yamaguchi, N., Vieira, F.G., Mendoza, M.L.Z., Liu, S., Martin, M.D., Sinding, M.K.S., Mak, S.S.T., et al.,

2020. The evolutionary history of extinct and living lions. *Proc. Natl. Acad. Sci. U. S. A.* 117, 10927–10934.
- Díez del Molino, D., Sánchez-Barreiro, F., Barnes, I., Gilbert, M.T.P., Dalén, L., 2018. Quantifying temporal genomic erosion in endangered species. *Trends Ecol. Evol.* 33, 176–185.
- Eizirik, E., Kim, J.H., Menotti-Raymond, M., Crawshaw Jr., P.G., O'Brien, S.J., Johnson, W.E., 2001. Phylogeography, population history and conservation genetics of jaguars (*Panthera onca*, Mammalia, Felidae). *Mol. Ecol.* 10, 65–79.
- Haag, T., Santos, A.S., Sana, D.A., Morato, R.G., Cullen Jr., L., Crawshaw Jr., P.G., De Angelo, C., Di Bitetti, M.S., Salzano, F.M., Eizirik, E., 2018. The effect of habitat fragmentation on the genetic structure of a top predator: loss of diversity and high differentiation among remnant populations of Atlantic Forest jaguars (*Panthera onca*). *Mol. Ecol.* 19, 4906–4921.
- Jędrzejewski, W., Robinson, H.S., Abarca, M., Zeller, K.A., Velasquez, G., Paemelaere, E.A., Goldberg, J.F., Payan, E., Hoogesteijn, R., Boede, E.O., et al., 2018. Estimating large carnivore populations at global scale based on spatial predictions of density and distribution—Application to the jaguar (*Panthera onca*). *PLoS One* 13, e0194719.
- Kardos, M., Åkesson, M., Fountain, T., Flagstad, Ø., Liberg, O., Olason, P., Sand, H., Wabakken, P., Wikenros, C., Ellegren, H., 2018. Genomic consequences of intensive inbreeding in an isolated wolf population. *Nat. Ecol. Evol.* 2, 124–131.
- Leroy, G., Carroll, E.L., Bruford, M.W., DeWoody, J.A., Strand, A., Waits, L., Wang, J., 2018. Next-generation metrics for monitoring genetic erosion within populations of conservation concern. *Evol. Appl.* 11, 1066–1083.
- Lí, H., Durbin, R., 2011. Inference of human population history from individual whole-genome sequences. *Nature* 475, 493–496.
- Liu, Y.C., Sun, X., Driscoll, C., Miquelle, D.G., Xu, X., Martelli, P., Uphyrkina, O., Smith, J.L.D., O'Brien, S.J., Luo, S.J., 2018. Genome-wide evolutionary analysis of natural history and adaptation in the world's tigers. *Curr. Biol.* 28, 3840–3849.
- Lorenzana, G., Heidtmann, L., Haag, T., Ramalho, E., Dias, G., Hrbek, T., Farias, I., Eizirik, E., 2020. Large-scale assessment of genetic diversity and population connectivity of Amazonian jaguars (*Panthera onca*) provides a baseline for their conservation and monitoring in fragmented landscapes. *Biol. Conserv.* 242, 108417.
- Paviolo, A., De Angelo, C., Ferraz, K.M., Morato, R.G., Pardo, J.M., Srbeek-Araujo, A.C., Beisiegel, B.M., Lima, F., Sana, D., da Silva, M.X., et al., 2016. A biodiversity hotspot losing its top predator: the challenge of jaguar conservation in the Atlantic Forest of South America. *Sci. Rep.* 6, 1–16.
- Ribeiro, M.C., Metzger, J.P., Martensen, A.C., Ponzoni, F.J., Hirota, M.M., 2009. The Brazilian Atlantic Forest: how much is left, and how is the remaining forest distributed? Implications for conservation. *Biol. Conserv.* 142, 1141–1153.
- Saremi, N.F., Supple, M.A., Byrne, A., Cahill, J.A., Coutinho, L.L., Dalén, L., Figueiró, H.V., Johnson, W.E., Milne, H.J., O'Brien, S.J., et al., 2019. Puma genomes from North and South America provide insights into the genomic consequences of inbreeding. *Nat. Commun.* 10, 1–10.
- Srbeek-Araujo, A.C., Haag, T., Chiarello, A.G., Salzano, F.M., Eizirik, E., 2018. Worrisome isolation: noninvasive genetic analyses shed light on the critical status of a remnant jaguar population. *J. Mammal.* 99, 397–407.

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