inhibitors could ameliorate a broad range of diseases caused by accumulation of misfolded proteins. Because many signaling pathways operate on the same dynamic-phosphorylation in the activation phase terminated by dephosphorylationdelaying the termination phase of signaling pathways through the selective inhibition of phosphatases may be of broad relevance to safely and selectively manipulate cellular functions for therapeutic benefit.

REFERENCES AND NOTES

- 1. H. P. Harding, Y. Zhang, A. Bertolotti, H. Zeng, D. Ron, Mol. Cell 5. 897-904 (2000).
- 2. D. Scheuner et al., Mol. Cell 7, 1165–1176 (2001).
- H. P. Harding et al., Mol. Cell 11, 619-633 (2003).
- P. Tsaytler, H. P. Harding, D. Ron, A. Bertolotti, Science 332, 91-94 (2011).
- D. Scheuner et al., J. Biol. Chem. 281, 21458-21468
- W. E. Balch, R. I. Morimoto, A. Dillin, J. W. Kelly, Science 319, 916-919 (2008).
- B. Holmes, R. N. Brogden, R. C. Heel, T. M. Speight, G. S. Avery, Drugs 26, 212–229 (1983).
- M. A. Brostrom, X. J. Lin, C. Cade, D. Gmitter, C. O. Brostrom, J. Biol. Chem. 264, 1638-1643 (1989).
- I. Novoa et al., EMBO J. 22, 1180-1187 (2003).
- 10. H. P. Harding et al., Mol. Cell 6, 1099-1108 (2000).
- 11. R. H. Meacham et al., J. Pharmacol. Exp. Ther. 214, 594-598
- 12. A. H. Hall, S. C. Smolinske, K. W. Kulig, B. H. Rumack, Ann. Intern. Med. 102, 787-788 (1985).
- 13. D. Genoux et al., Nature 418, 970-975 (2002).
- 14. M. Costa-Mattioli et al., Cell 129, 195-206 (2007).
- 15. R. G. M. Morris, P. Garrud, J. N. P. Rawlins, J. O'Keefe, Nature 297, 681-683 (1982)
- 16. M. Pennuto et al., Neuron 57, 393-405 (2008).
- 17. M. D'Antonio et al., J. Exp. Med. 210, 821-838 (2013).
- 18. L. Wrabetz et al., J. Neurosci. 26, 2358-2368 (2006).
- 19. A. Nordlund, M. Oliveberg, HFSP J. 2, 354-364 (2008).
- 20. H. Nishitoh et al., Genes Dev. 22, 1451-1464 (2008).
- 21. L. Wang, B. Popko, R. P. Roos, Hum. Mol. Genet. 23, 2629-2638 (2014).
- 22. M. E. Gurney et al., Science 264, 1772-1775 (1994).
- 23. S. J. Marciniak et al., Genes Dev. 18, 3066-3077
- 24. J. Wang et al., Proc. Natl. Acad. Sci. U.S.A. 106, 1392-1397 (2009).
- 25. H. P. Harding et al., Proc. Natl. Acad. Sci. U.S.A. 106, 1832-1837 (2009).

ACKNOWLEDGMENTS

We thank P. Tsaytler for some initial experiments on Sephin1; H. Meziane for the studies on memory; E. Fisher for $SOD1^{G93A}$ mice; E. Pettinato and C. Ferri for technical assistance; R. Roberts for discussions on CMT: A. Segonds-Pichon for statistical analysis: and members of the Bertolotti laboratory, M. Goedert, M. Hastings, and S. Munro for discussions. A.B. is an honorary fellow of the Clinical Neurosciences Department of Cambridge University. This work was supported by the Medical Research Council (UK) and the European Research Council (ERC) under the European Union's Seventh Framework Programme (FP7/2007-2013)/ERC grant 309516. A.K. was supported by the European Molecular Biology Organization and Human Frontier Science Program, K.S. by the Swiss National Science Foundation, M.D. by the Italian Ministry of Health (GR-2011-02642791), and L.W. by NIH R01-NS55256. A.B. is a co-inventor on Great Britain patent WO 2014108520, covering benzylideneguanidine derivatives inhibitors of PPP1R15A. The data presented in this paper are tabulated in the main paper and the supplementary materials.

SUPPLEMENTARY MATERIALS

www.sciencemag.org/content/348/6231/239/suppl/DC1 Materials and Methods Figs. S1 to S6 References (26-29)

8 December 2014; accepted 6 March 2015 10.1126/science.aaa4484

PRIMATE GENOMICS

Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding

Yali Xue, 1x Javier Prado-Martinez, 2x Peter H. Sudmant, 3x Vagheesh Narasimhan, 1,4x Qasim Ayub, 1 Michal Szpak, 1 Peter Frandsen, 5 Yuan Chen, 1 Bryndis Yngvadottir, 1 David N. Cooper, 6 Marc de Manuel, 2 Jessica Hernandez-Rodriguez, 2 Irene Lobon, 2 Hans R. Siegismund,⁵ Luca Pagani,^{1,7} Michael A. Quail,¹ Christina Hvilsom,⁸ Antoine Mudakikwa, Evan E. Eichler, Michael R. Cranfield, Tomas Marques-Bonet, 2,12 Chris Tyler-Smith, 1+ Aylwyn Scally 13+

Mountain gorillas are an endangered great ape subspecies and a prominent focus for conservation, yet we know little about their genomic diversity and evolutionary past. We sequenced whole genomes from multiple wild individuals and compared the genomes of all four Gorilla subspecies. We found that the two eastern subspecies have experienced a prolonged population decline over the past 100,000 years, resulting in very low genetic diversity and an increased overall burden of deleterious variation. A further recent decline in the mountain gorilla population has led to extensive inbreeding, such that individuals are typically homozygous at 34% of their sequence, leading to the purging of severely deleterious recessive mutations from the population. We discuss the causes of their decline and the consequences for their future survival.

ountain gorillas came to the attention of the western world as recently as 1902 (1) and survive today only in two small and critically endangered populations in central Africa (2). Current estimates place their total population at around 800, of which just over half are in the Virunga volcanic mountain range on the borders of Rwanda, Uganda, and the Democratic Republic of Congo (Fig. 1A) (3, 4). Mountain gorillas face a number of threats to their continued existence (5), and their survival became an urgent concern during the 1960s, when a precipitous decline in their numbers appeared to put them at grave risk of extinction (6). Active conservation efforts have since reversed the decline (3), but a loss of genetic diversity associated with small population size may still threaten their long-term viability. Infectious diseases

¹Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton CB10 1SA, UK. 2ICREA at Institut de Biologia Evolutiva (CSIC/UPF), Parque de Investigación Biomédica de Barcelona (PRBB), Barcelona, Catalonia 08003, Spain. ³Department of Genome Sciences, University of Washington, Seattle, WA 98195, USA. ⁴Department of Applied Mathematics and Theoretical Physics, University of Cambridge, Cambridge CB3 OWA, UK. ⁵Department of Biology, University of Copenhagen, DK-2200 Copenhagen N, Denmark. ⁶Institute of Medical Genetics, Cardiff University, Cardiff CF14 4XN, UK. ⁷Department of Biological, Geological and Environmental Sciences, University of Bologna, 40134 Bologna, Italy. ⁸Research and Conservation, Copenhagen Zoo, DK-2000 Frederiksberg, Denmark. ⁹Rwanda Development Board, KG 9 Avenue, Kigali, Rwanda. ¹⁰Howard Hughes Medical Institute, Seattle, WA 91895, USA. 11Gorilla Doctors, Karen C. Draver Wildlife Health Center, University of California, Davis, CA 95616. USA. 12 Centro Nacional de Análisis Genómico (Parc Científic de Barcelona), Baldiri Reixac 4, 08028 Barcelona, Spain. ¹³Department of Genetics, University of Cambridge, Cambridge CB2 3EH, UK.

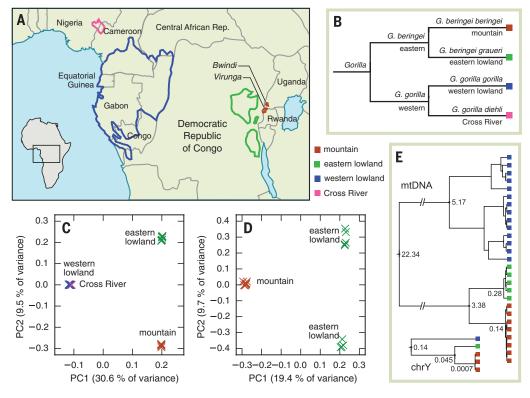
*These authors contributed equally to this work. †Corresponding author. E-mail: cts@sanger.ac.uk (C.T.-S.); aos21@cam.ac.uk (A.S.) such as Ebola, the cause of substantial recent mortality in the western lowland population (7), represent a particularly serious threat (8).

Despite extensive study of mountain gorillas in the field, few genetic analyses have been carried out, and these have been confined to mitochondrial sequences and a limited number of autosomal loci (9-11). Unlike the other great apes (12), mountain gorillas have not been studied on a genome-wide scale, which is key to understanding their biology, evolution, and relationship to sister taxa (Fig. 1B). It is also important in assessing their current status and in forming strategies for future conservation efforts. Phenotypic indicators of inbreeding such as syndactyly have been reported (13, 14), but the full genetic impact of their decline is unknown. Additionally, the severe population bottleneck experienced by mountain gorillas provides an opportunity to study processes that may have played a recurring role in hominin evolution and extinction.

We performed whole-genome sequencing and analysis for 13 eastern gorillas, comprising seven mountain gorillas from the Virunga volcanoes region and six eastern lowland gorillas. The mountain gorillas originate from both sides of the Virunga massif, including from three groups spanning a densely populated sector on the Rwandan side, and plausibly represent genetic variation and ancestry in the Virunga population at large (15). Sequencing was performed to an average $26 \times$ depth (table S1), and combining these data with published gorilla genome sequences (12) yielded a total data set of 44 samples spanning all four gorilla subspecies (table S2).

Sequences were aligned to the gorilla reference genome (16), and several tools were used to call variants across samples (15). Comparing

Fig. 1. Geography, taxonomy and genetic structure of gorilla species. (A) Distribution of gorilla subspecies (2). (B) Gorilla taxonomy. (C) PCA plot of SNP data for all four gorilla subspecies. (D) PCA plot of SNP data from mountain and eastern lowland gorilla samples only. (E) mtDNA and Y-chromosomal phylogenies. Node heights are in units of substitutions per base pair; each tree is drawn to a separate scale.



populations, we found that genetic diversity was lower in the eastern species than in the western by a factor of 2 to 3 (table S3), with mean autosomal heterozygosity (frequency of betweenchromosomal differences) of 6.5×10^{-4} per base pair in mountain and eastern lowland gorillas, versus 1.9×10^{-3} per base pair in the western lowland population. These values are consistent with the small reported census population sizes of eastern gorillas relative to those in the west (2). A detailed analysis of variation in mitochondrial DNA (mtDNA) showed little diversity in mountain gorillas, with only three haplotypes in total differing by just one to three mutations (15). A similar situation was observed on the Y chromosome, with only two sites differentiating the three male mountain gorillas (15).

A principal components analysis (PCA) of 11,743,407 single-nucleotide polymorphisms (SNPs) across all samples (Fig. 1C) showed a hierarchical structure consistent with the accepted Gorilla taxonomy and the geographical distribution of these populations. In particular, the separation between eastern lowland and mountain gorilla samples confirms them as genetically distinct populations. Another PCA focusing on the eastern species only (Fig. 1D) revealed no substructure within the mountain gorillas sampled here, but did show a separation of eastern lowland gorillas into two subgroups, which may reflect structure within the subspecies as a whole or may only be a feature of the individuals we have sampled. Similar patterns are also found in a genetic structure analysis using the program ADMIXTURE (fig. S3).

Copy number variation (CNV) among gorillas was assessed with a read depth-based approach

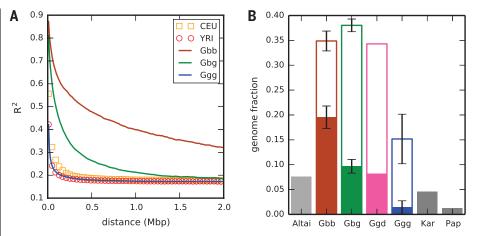


Fig. 2. Linkage disequilibrium and homozygosity in gorillas. (A) LD decay (*15*) in gorilla and human populations. Human samples are Utah residents with European ancestry (CEU) or Yoruba in Ibadan, Nigeria (YRI). (B) Mean per-sample genome fractions found in homozygous tracts. Open bars show total fractions for mountain (Gbb), eastern lowland (Gbg), Cross River (Ggd), and western lowland (Ggg) gorillas; solid bars show fractions in tracts of length 2.5 to 10 Mb (gorillas) or 2.5 to 10 cM in an Altai Neandertal and two human individuals [Karitiana (Kar) and Papuan (Pap)] (*19*). Error bars are ±1 SD.

(17), and the number of fixed loss and gain events between populations reflects the accepted taxonomy (table S5), although eastern lowland and mountain gorillas showed an excess of shared fixed deletions relative to western lowland gorillas, potentially reflecting a small ancestral population prior to their divergence. Diversity patterns at polymorphic CNVs also resembled those seen in SNPs (fig. S8) (15).

Accurate population monitoring and management are of central importance for conservation, and we cataloged 25,628 autosomal and 89 mtDNA $\,$

ancestry-informative SNP markers (AIMs) uniquely identifying eastern lowland and mountain gorillas (as sampled here), as well as 1127 lineage-specific CNV events (data files S1 and S2) (15).

We found that genome-wide linkage disequilibrium (LD) varies markedly between gorilla populations (Fig. 2A). Both eastern subspecies—particularly mountain gorillas—show much more extensive LD than western lowland gorillas, in whom the decay of LD with genomic distance is similar to that in African humans (15). These patterns reflect differing demographic histories and suggest

a recent population bottleneck in the two eastern subspecies.

An analysis of chromosomal sequence sharing within individuals (genomic tracts of homozygosity) provided insight into recent ancestry and a measure of parental relatedness. Within mountain and eastern lowland gorilla individuals, chromosomes are typically homozygous over one-third of their length (on average 34.5% and 38.4%, respectively) (Fig. 2B), much higher than in western lowland individuals (13.8%) and exceeding even the most inbred human populations (18). We observed longer tracts in the eastern species, particularly in mountain gorillas, and a clear distinction in tract length distribution between eastern lowland and mountain gorillas (fig. S15) (15). Very long tracts (2.5 to 10 Mb) are particularly indicative of recent inbreeding, and homozygosity on this scale in mountain gorillas exceeds not only that in other gorilla populations but also that observed in the Altai Neandertal (19), consistent with parental relatedness equivalent to that between two half-siblings (19). These data suggest that mountain gorillas may have experienced several recent generations of close inbreeding. We also examined haplotype matching between individuals, finding subclustering among eastern lowland gorillas consistent with that seen in Fig. 1D; individuals in one cluster displayed a 10% higher mean level of sequence sharing than those in the other (fig. S16).

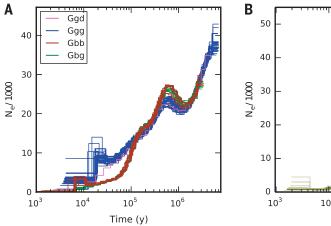
Whole-genome data also enabled us to explore the longer-term demographic history of these populations. Effective population size $(N_{\rm e})$ in each of the four subspecies, as inferred by the PSMC algorithm (20), has varied over time (Fig. 3A),

and we note the decline of $N_{\rm e}$ particularly in the eastern species within the past 100,000 years. The most recent values of inferred $N_{\rm e}$ are just 273 ± 54 in mountain gorillas and 290 ± 18 in eastern low-land gorillas. $N_{\rm e}$, which reflects genetic diversity, is usually lower than census population size; however, in 1981 the estimated census size in the Virunga region was as low as 254 (3).

As judged from the date when inferred N_e began to differ, the divergence of eastern and western gorillas began at least 150,000 years ago, but a more direct analysis using male Xchromosomal sequences suggests that they exchanged genetic material until around 20,000 years ago (Fig. 3B) (15). Given that this also coincides with a notable decline in western lowland gorilla $N_{\rm e}$ (Fig. 3A), it may be that environmental changes during the Last Glacial Maximum (26,000 to 19,000 years ago), when dry savannah replaced tropical forest over much of the Congo basin (21), triggered a collapse in the western population and complete separation of the two species. Indeed, changes in forestation across the center of the continent over the past 150,000 years may have had a substantial influence on speciation and diversity in African apes more generally, including ancient humans (12).

We found no evidence for more recent eastwest genetic contact, and *D*-statistic analysis provided no strong support for differences in east-west gene flow between subspecies pairs in either species (table S17).

These evolutionary differences between subspecies allowed us to investigate the relationship between demography and selection at protein-coding loci. To facilitate this, we classified coding sequence variants into three groups: synonymous, missense, and loss-of-function (LoF) (15). Accounting for potential reference bias, we found a relatively larger number of derived missense alleles in eastern gorillas, particularly in mountain



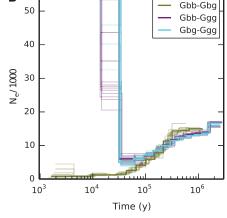
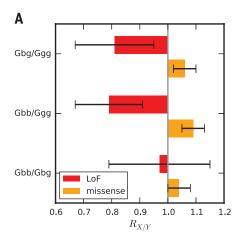
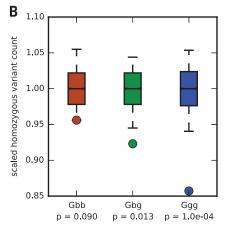


Fig. 3. Ancestral effective population size and gene flow between gorilla populations. (A) Inferred effective population size (N_e) history for each of the samples studied. (B) Cross-population N_e history, based on paired male X-chromosomal sequences. Both plots are scaled using a generation time of 19.3 years and an autosomal mutation rate of 1.25×10^{-8} per base pair per generation.





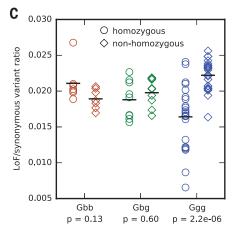


Fig. 4. The genetic burden of missense mutations and purging of LoF mutations in eastern gorillas. (A) Relative number of derived alleles at LoF (red) and missense (orange) sites that are frequent in one population and not another (15). Error bars represent ± 2 SD. (B) Circles indicate the scaled number of LoF variant sites in each population where at least one sample is homozygous for the derived allele. Boxplots show distributions of the same statistic for matched samples of synonymous sites (15); whiskers

show 5th and 95th percentiles; P values are the proportion of each sample distribution smaller than the corresponding LoF count. (\mathbf{C}) Circles show the rate of LoF variants relative to synonymous variants in homozygous tracts for each sample; diamonds show the same ratio in nonhomozygous regions. Horizontal bars indicate population means; P values for each subspecies correspond to a Kolmogorov-Smirnov test for difference in distribution between homozygous and nonhomozygous regions.

244 10 APRIL 2015 • VOL 348 ISSUE 6231 sciencemag.org SCIENCE

gorillas, relative to western lowland gorillas (Fig. 4A). This is consistent with long-term differences in N_e (Fig. 3A), because selection in a larger population is more effective at removing moderately deleterious variants. In contrast, we found that mountain gorillas have relatively fewer LoF variants than western lowland gorillas. This is again consistent with low N_e and inbreeding, wherein alleles are more frequently exposed in a homozygous state, and because LoF variants are more likely to be strongly deleterious, they are less likely to persist in the population even if recessive (22). Further evidence for the purging of severely deleterious mutations from mountain and eastern lowland gorillas comes from the lack of any deficit of homozygous LoF genotypes in mountain gorillas (Fig. 4B) and the observation that LoF variants in both subspecies occur at the same rate in homozygous and heterozygous tracts (Fig. 4C) (15).

Finally, we looked at variation in genes using function inferred from homology, focusing on possible selection and adaptation in mountain gorillas. Such adaptation might be expected from the fact that mountain gorillas range over high altitudes (1500 to 4000 m), with consequences for diet, morphology, and physiology (23). However, we found no significant enrichment in any functional category of genes, although there are interesting examples related to nervous system morphology, immunoglobulin quantity, and red blood cell morphology (15). Mountain gorillas carry a significant excess of variants in genes associated with blood coagulation in humans (fig. S21), perhaps linked to high-altitude living (15). We also identified variants associated with cardiomyopathy, including in one deceased individual (Kaboko) in whom post mortem analysis revealed evidence of muscular hypertrophy (15). Cardiovascular disease has been identified as a notable cause of death in captive western lowland gorillas (24).

Concern about the survival of mountain gorillas is focused largely on the threat from human encroachment on their habitat (2, 5), and their

severe recent population decline may be responsible for the high level of inbreeding that we observe. An increased burden of deleterious mutation and low genetic diversity—including at the major histocompatibility locus, of central importance to the immune system (fig. S22)—have likely reduced their resilience to environmental change and pathogen evolution. However, the origins of this condition extend far into their history, because both eastern subspecies have experienced a long decline over tens of millennia. Indeed, the demographic histories of mountain and eastern lowland gorillas (Fig. 3A) bear unhappy resemblance to similar histories inferred from Neandertals before their disappearance (19).

Nonetheless, the same evidence suggests that such a fate is not inevitable: These subspecies have survived for thousands of generations at low population levels and may have developed physiological and behavioral strategies to mitigate inbreeding, such as natal dispersal and gene flow between isolated populations (25). The purging of strongly deleterious mutations may also be an important factor. Our findings reinforce the imperative to sustain conservation efforts that have helped to keep mountain gorillas from the brink of extinction, and provide a genomic resource for future conservation and research.

REFERENCES AND NOTES

- G. B. Schaller, The Year of the Gorilla (Univ. of Chicago Press, Chicago, 2010), p. 7.
- M. Robbins et al., Gorilla beringei ssp. beringei (listing in IUCN Red List of Threatened Species, 2008); www.iucnredlist.org/ details/39999/0.
- 3. M. Gray et al., Biol. Conserv. 158, 230-238 (2013).
- 4. K. Guschanski et al., Biol. Conserv. 142, 290-300 (2009).
- 5. A. H. Harcourt, Biol. Conserv. 75, 165-176 (1996).
- A. W. Weber, A. Vedder, *Biol. Conserv.* 26, 341–366 (1983).
- 7. P. J. Le Gouar et al., PLOS ONE **4**, e8375 (2009).
- IUCN, "Ebola outbreak highlights critical links between biodiversity loss and human health, says IUCN's Wildlife Health Specialist Group" (2014); www.iucn.org/?18439.
- K. J. Garner, O. A. Ryder, Mol. Phylogenet. Evol. 6, 39–48 (1996).
- M. I. Jensen-Seaman, K. K. Kidd, Mol. Ecol. 10, 2241–2247 (2001).

- 11. J. Roy et al., Biol. Lett. 10, 2014.0811 (2014).
- 12. J. Prado-Martinez et al., Nature 499, 471-475 (2013).
- D. Fossey, Gorillas in the Mist (Harcourt Brace, New York, 1983), p. 72.
- A. Routh, J. Sleeman, in Proceedings of the British Veterinary Zoological Society (Howletts and Port Lympne Wild Animal Parks, Kent, 14–15 June 1997), pp. 22–25.
- 15. See supplementary materials on Science Online.
- 16. A. Scally et al., Nature 483, 169-175 (2012).
- 17. C. Alkan et al., Nat. Genet. 41, 1061-1067 (2009).
- 18. T. J. Pemberton et al., Am. J. Hum. Genet. **91**, 275–292 (2012).
- 19. K. Prüfer et al., Nature 505, 43-49 (2014).
- 20. H. Li, R. Durbin, Nature 475, 493-496 (2011).
- D. Anhuf et al., Palaeogeogr. Palaeoclimatol. Palaeoecol. 239, 510–527 (2006).
- 22. S. Glémin, Evolution 57, 2678-2687 (2003).
- 23. D. P. Watts, Am. J. Primatol. 7, 323-356 (1984).
- R. McManamon, L. Lowenstine, in Fowler's Zoo and Wild Animal Medicine (Elsevier, St. Louis, vol. 7, 2012), pp. 408–415.
- A. Pusey, M. Wolf, Trends Ecol. Evol. 11, 201–206 (1996).

ACKNOWLEDGMENTS

We thank the Institut Congolais pour la Conservation de la Nature and the Rwandan Development Board for the use of samples: J. Ramer, D. Zimmerman, J. F. Kinani, J. Bosco Noheri, M. Bahizi, J. P. Lukasa, J. Iyanya, M. Kabuyaya, E. Kambale, and J. Sohl of Gorilla Doctors for sample collection and processing; and M. Pollard, P. Hallast, M. Jobling, and L. Lowenstine for assistance. Supported by Royal Society grant RG130105 (A.S.), Wellcome Trust grants 098051 (Q.A., Y.C., V.N., L.P., M.A.Q., M.S., C.T.-S., Y.X., B.Y.) and 099769/Z/12/Z (V.N.), NIH grant HG002385 (E.E.E.), a European Research Council Starting Grant (260372), and Ministerio de Ciencia e Innovacion grant BFU2011-28549 (T.M.-B.). E.E.E. is an investigator of the Howard Hughes Medical Institute. Accession numbers for sequence data are given in table S1. Gorilla blood samples sequenced in this study were collected and transferred in compliance with CITES legislation and subject to a material transfer agreement between the Mountain Gorilla Veterinary Project and the Office Rwandais du Tourisme et des Parcs Nationaux. D.N.C. acknowledges support from BIOBASE GmbH through a license agreement with Cardiff University; E.E.E. is a member of the external scientific advisory board for DNAnexus Inc.

SUPPLEMENTARY MATERIALS

www.sciencemag.org/content/348/6231/242/suppl/DC1 Materials and Methods

Figs. S1 to S22 Tables S1 to S18 Data files S1 and S2

References (26–62)

30 November 2014; accepted 3 March 2015 10.1126/science.aaa3952



Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding

Yali Xue, Javier Prado-Martinez, Peter H. Sudmant, Vagheesh Narasimhan, Qasim Ayub, Michal Szpak, Peter Frandsen, Yuan Chen, Bryndis Yngvadottir, David N. Cooper, Marc de Manuel, Jessica Hernandez-Rodriguez, Irene Lobon, Hans R. Siegismund, Luca Pagani, Michael A. Quail, Christina Hvilsom, Antoine Mudakikwa, Evan E. Eichler, Michael R. Cranfield, Tomas Marques-Bonet, Chris Tyler-Smith and Aylwyn Scally

Science **348** (6231), 242-245. DOI: 10.1126/science.aaa3952

Genomes in the mist

The mountain gorilla is an iconic species that is at high risk of extinction. Xue *et al.* have sequenced 13 gorillas from two different populations to probe their genetic diversity. The genomes show large tracts of homozygosity and the loss of highly deleterious genetic variants, indicating population bottlenecks and inbreeding. This loss of genetic diversity appears to have started over 20,000 years ago and may have been caused by changes in climate and human-associated effects.

Science, this issue p. 242

ARTICLE TOOLS http://science.sciencemag.org/content/348/6231/242

SUPPLEMENTARY http://science.sciencemag.org/content/suppl/2015/04/08/348.6231.242.DC1

RELATED http://science.sciencemag.org/content/sci/348/6231/246.2.full

REFERENCES This article cites 52 articles, 11 of which you can access for free

http://science.sciencemag.org/content/348/6231/242#BIBL

PERMISSIONS http://www.sciencemag.org/help/reprints-and-permissions

Use of this article is subject to the Terms of Service