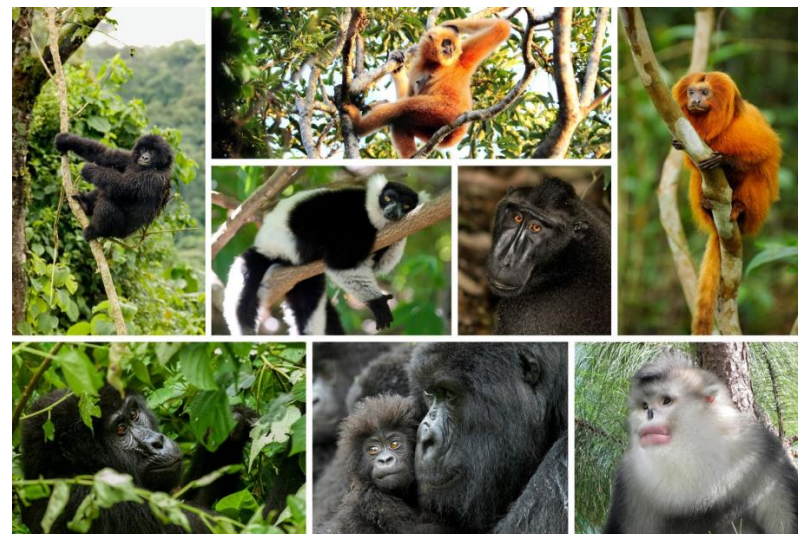
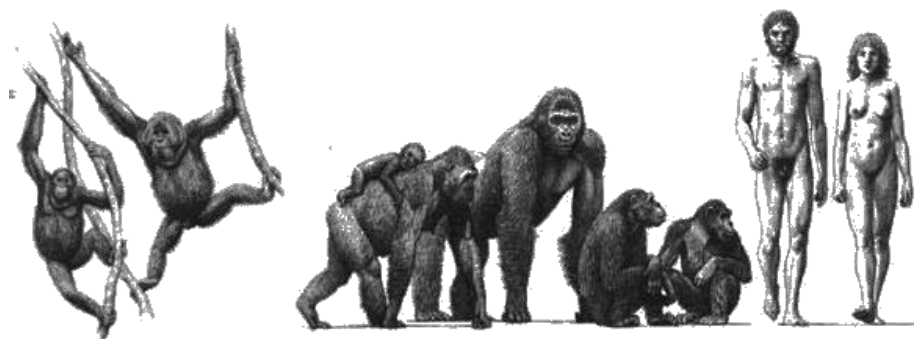


ELS PRIMATS COM A MODEL D'EVOLUCIÓ, BIODIVERSITAT I BIOMEDICINA

Tomas Marques-Bonet
Universitat Pompeu Fabra



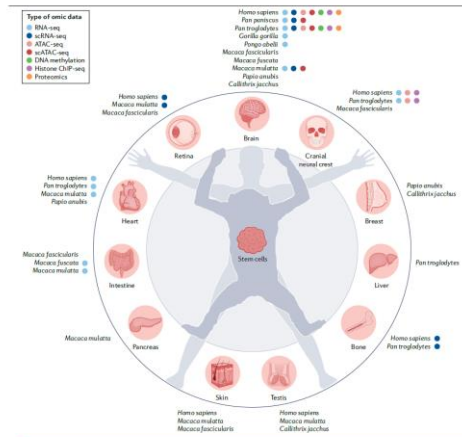
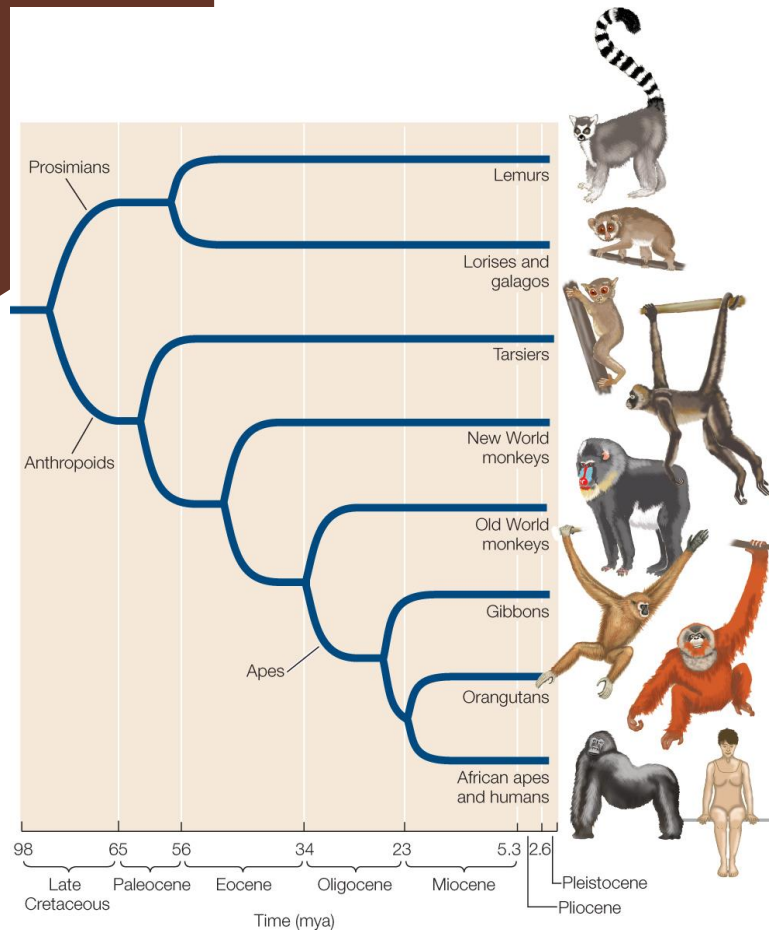
Primats i simis



One Health



Genòmica Comparativa



nature reviews genetics

<https://doi.org/10.1038/s41576-022-00554-w>

Review article

 Check for updates

Current advances in primate genomics: novel approaches for understanding evolution and disease

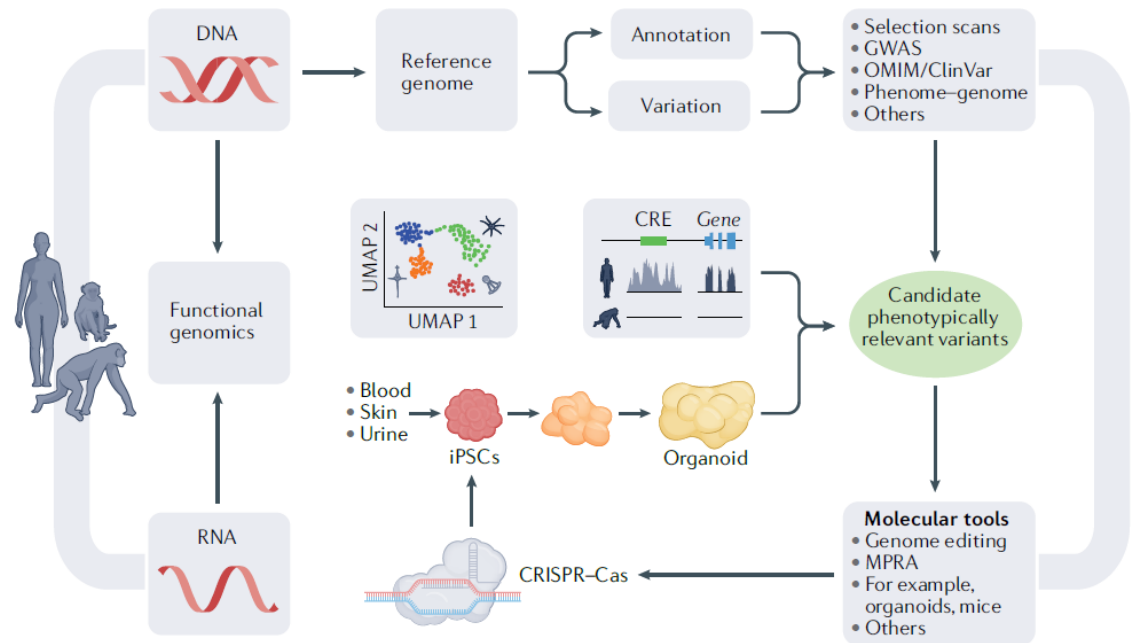
David Juan^{1,9}, Gabriel Santpere^{2,3,9}, Joanna L. Kelley^{4,8}, Omar E. Cornejo^{4,8} & Tomas Marques-Bonet^{1,5,6,7}✉

Abstract

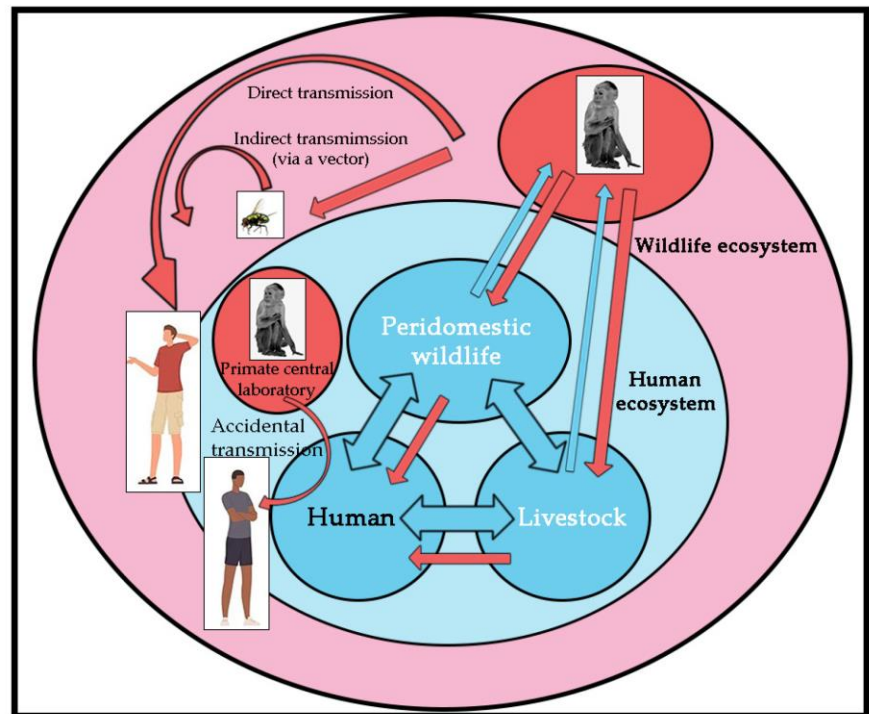
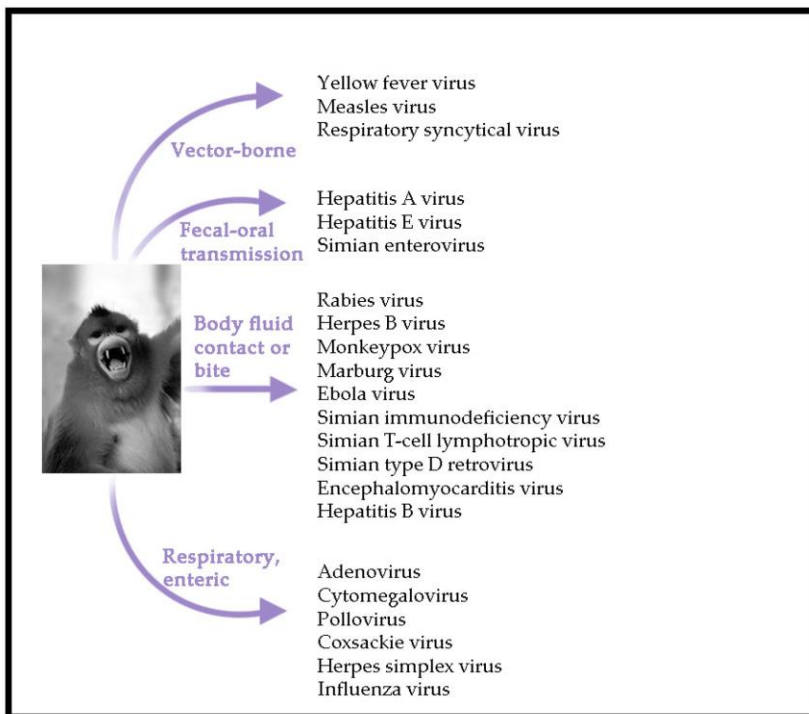
Sections

Juan et al. 2023

Perquè
hauríem
d'estudiar
primats?



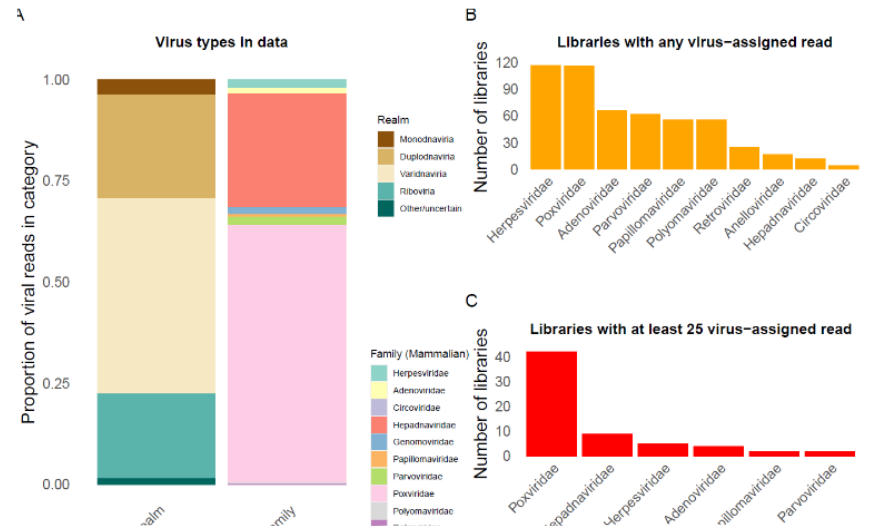
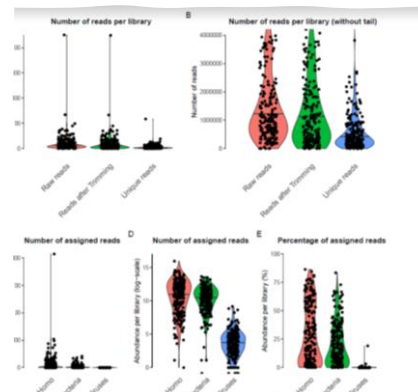
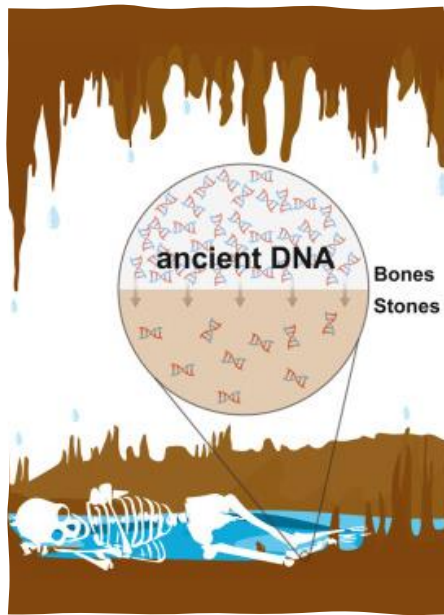
Pathogens



Una visió al passat



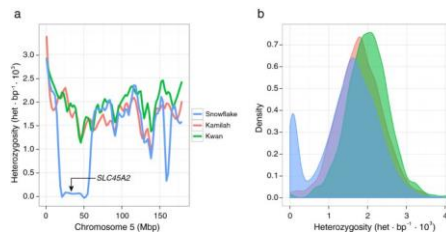
Una visió al passat



I sobre malalties?

The genome sequencing of an albino Western lowland gorilla reveals inbreeding in the wild

Prado-Martinez *et al.*



Prado-Martinez *et al.* 2013

Potential damaging mutation in *LRP5* from genome sequencing of the first reported chimpanzee with the Chiari malformation

Manuel Solís-Moruno¹, Marc de Manuel¹, Jessica Hernandez-Rodriguez², Claudia Fontseré¹, Alba Gomara-Castaño², Cristina Valseira-Naranjo¹, Dietmar Crailsheim³, Arcadi Navarro^{1,3,4}, Miquel Llorente¹, Laura Riera¹, Olga Feliu-Olleta² & Tomas Marques-Bonet^{1,3,4}



Figure 1. Noto.

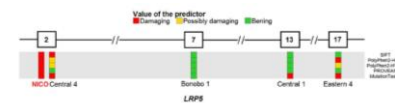


Figure 4. Schematic and zoomed representation of exons 2-17 of the *LRP5* gene along with all the mutations having a score for the used predictors. Each box is a predictor, from top to bottom: SIFT, PolyPhen2, HVAR, PolyPhen2-HVAR, PROVEAN and MutationTaster. The five mutations are missense variants in heterozygosity. The rest of the mutations that do not appear here are all of them considered as low by SIFT, being most of them synonymous variants. For a full view of the mutations in *LRP5* in all the individuals, see Supplementary Fig. S17. Note: the mutation of the Central 4 have been moved to the right to make the visualization more comprehensive.

Phenotype	Phenotype MIM number	Substance
Hypoplasia, unilateral	144750	AD
Osteopetrosis, autosomal dominant	607631	AD
Osteopetrosis	144750	AD
van Buchem disease, type	607631	AD
Bone mineral density variability	601884	AD
Osteopetrosis	144750	AD

Table 2. Phenotypes associated to mutations in *LRP5* gene. According to OMIM's code, brackets indicate "autosomal", while braces indicate "susceptibility to multifactorial disorder".

Solis-Moruno *et al.* et al. 2017

Un catàleg global de la diversitat genòmica a través de la radiació dels primats



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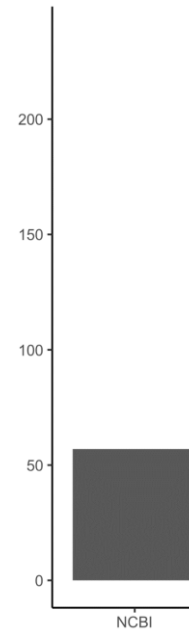
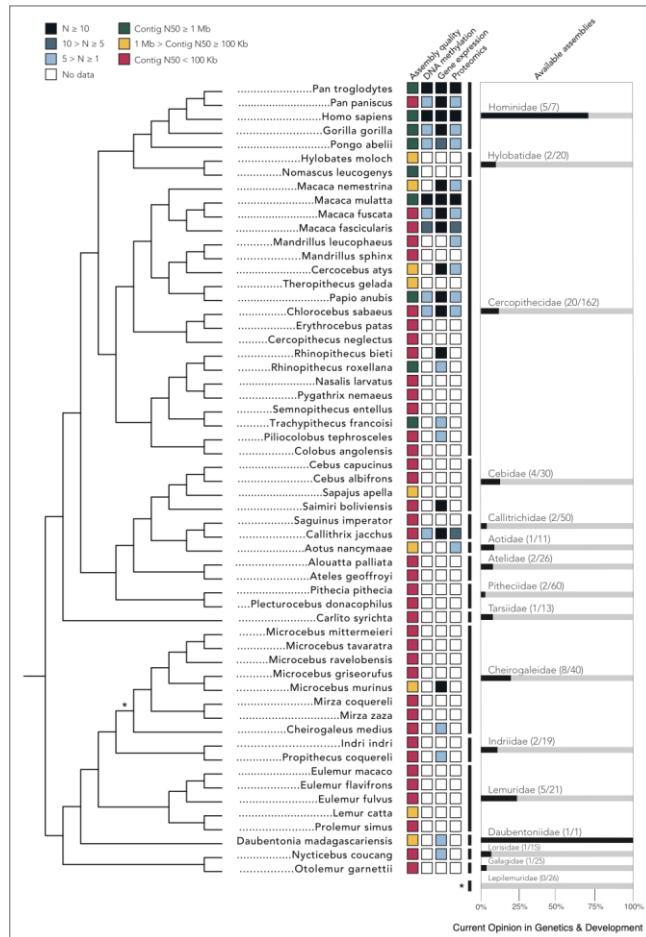
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Medicine



upf. Universitat
Pompeu Fabra
Barcelona



Pocs recursos genòmics en primats



Xarxa de primatòlegs

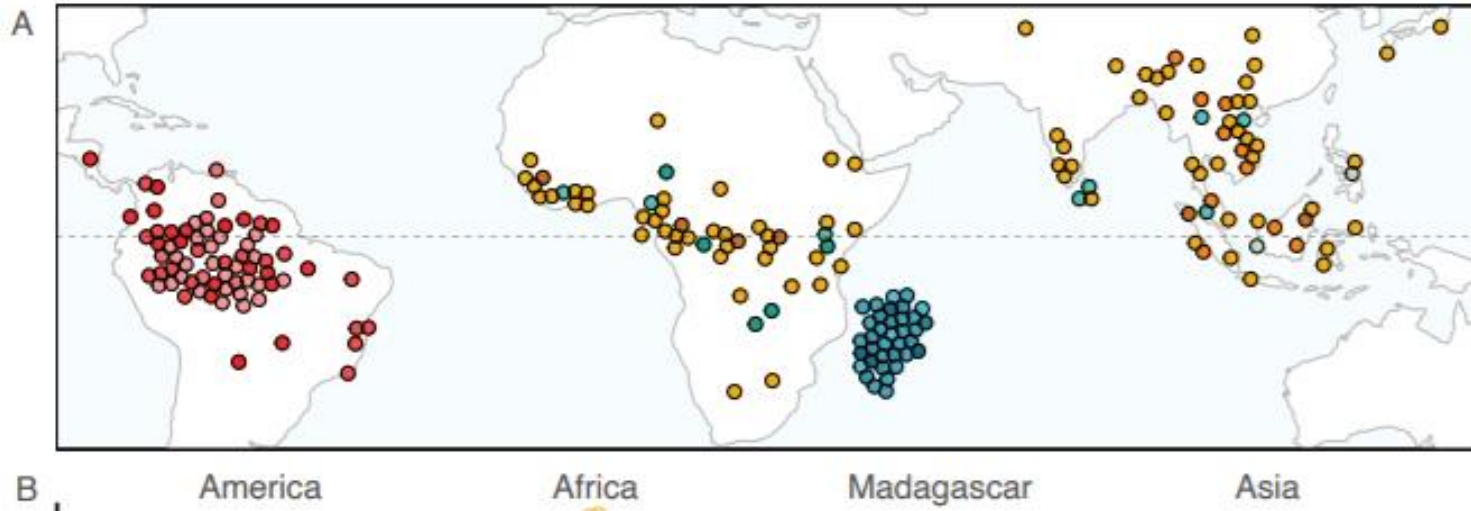


Jean Boubli
Christian Roos
Katherina Gushanski
Amanda Melin
Govindhaswamy Umapathy
Jessica Lee
Lim Weng Khon
Julie Horvath

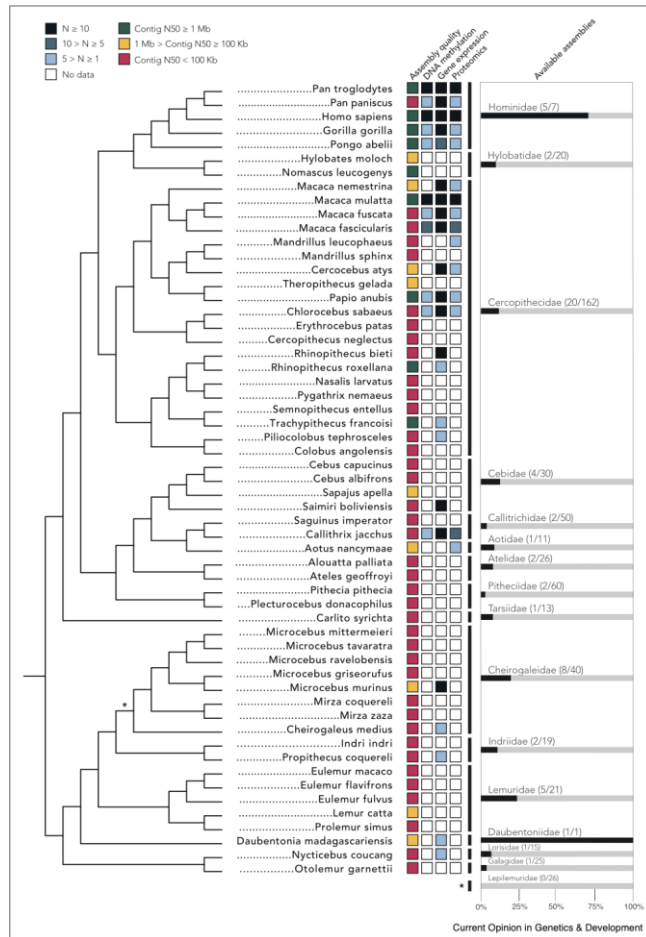
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Duke-NUS Singapore
Duke Lemur Center



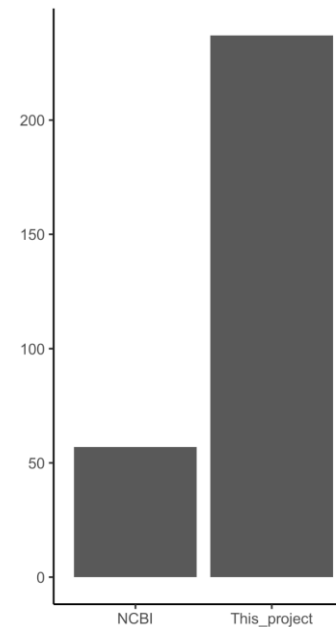
Xarxa de primatòlegs



Pocs recursos genòmics en primats

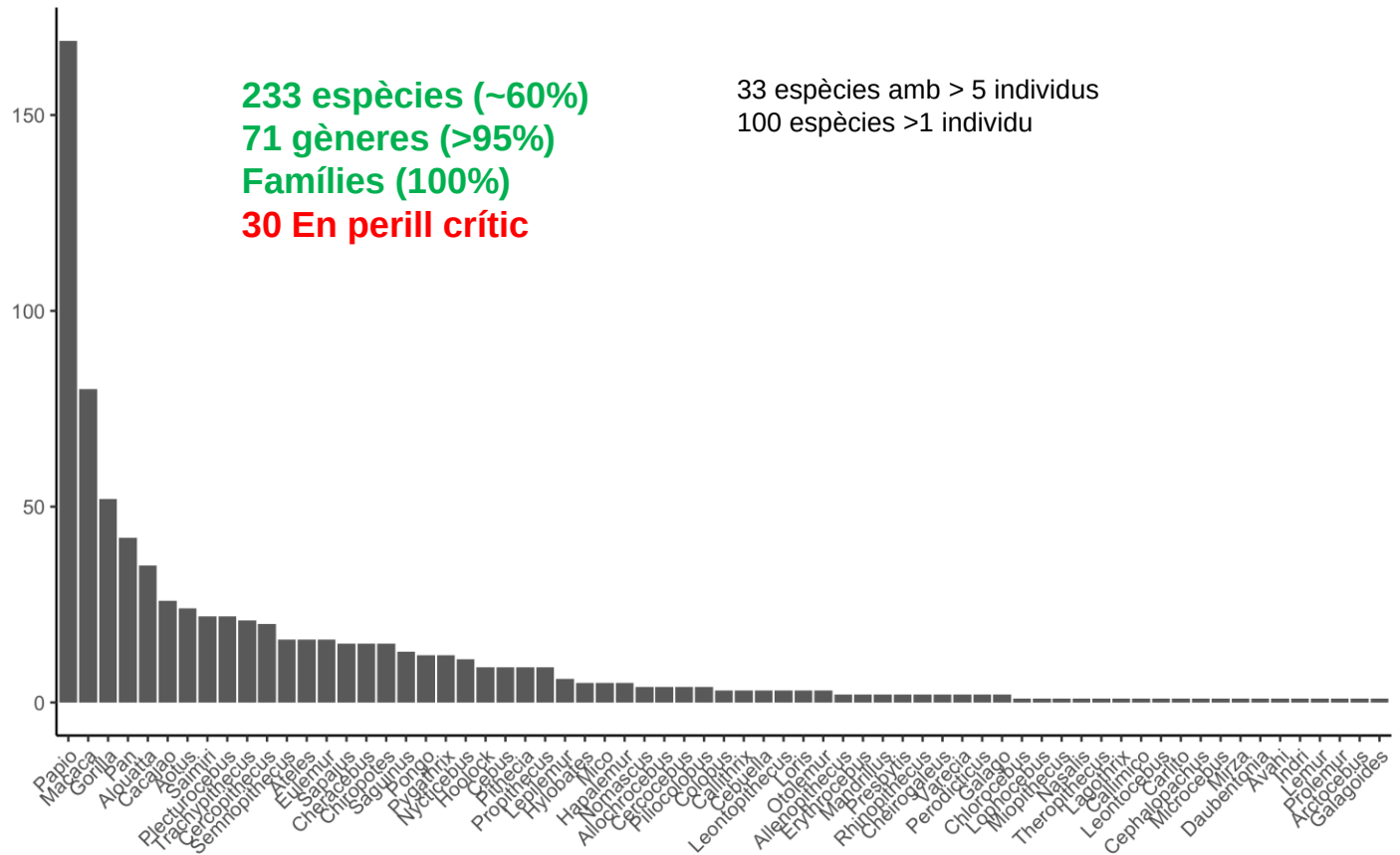


Kuderna, Esteller-Cucala, Marques-Bonet, 2020, Curr. Op. Gen. Dev.



El nombre d'espècies de primats amb seqüències de genomes disponibles va augmentar un > un 400%

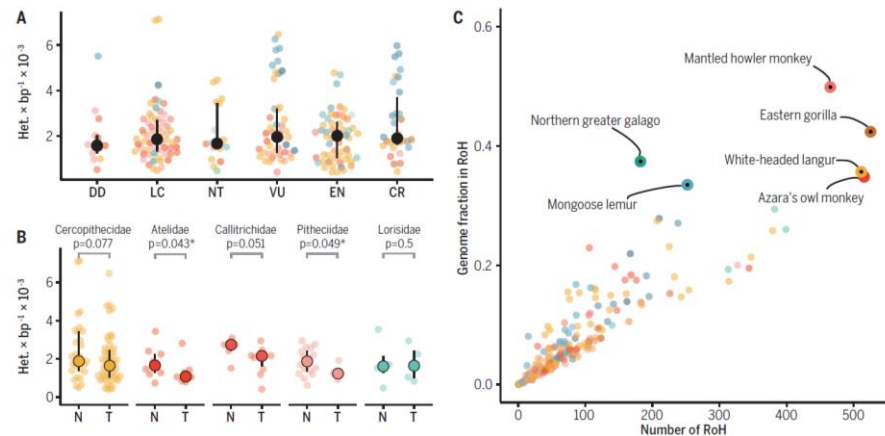
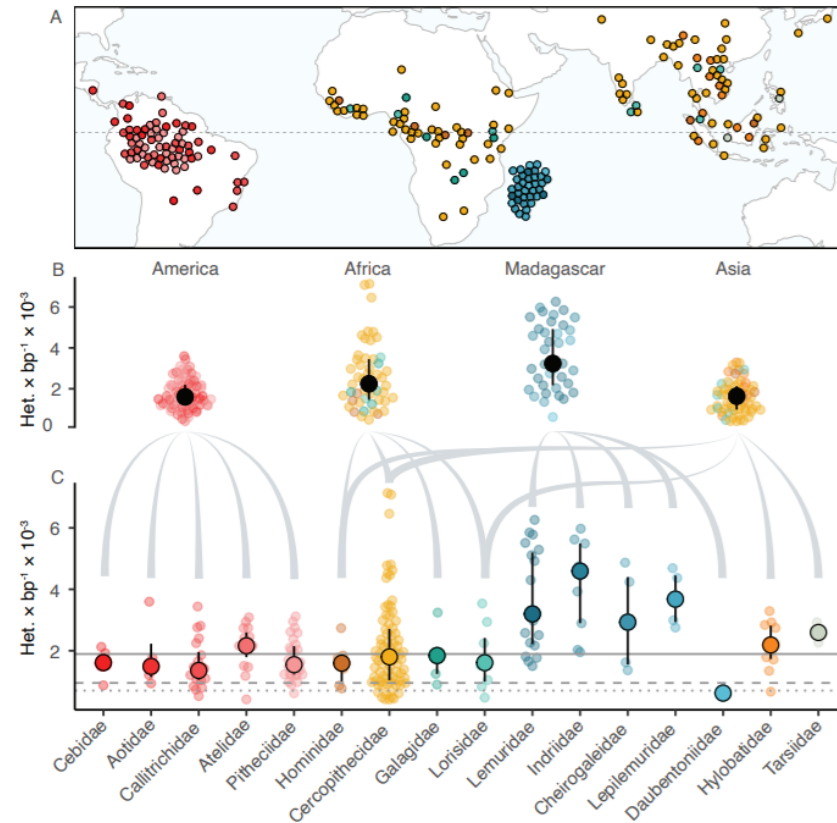
(N=233 vs 57)



Però... i què podem fer amb aquestes dades?

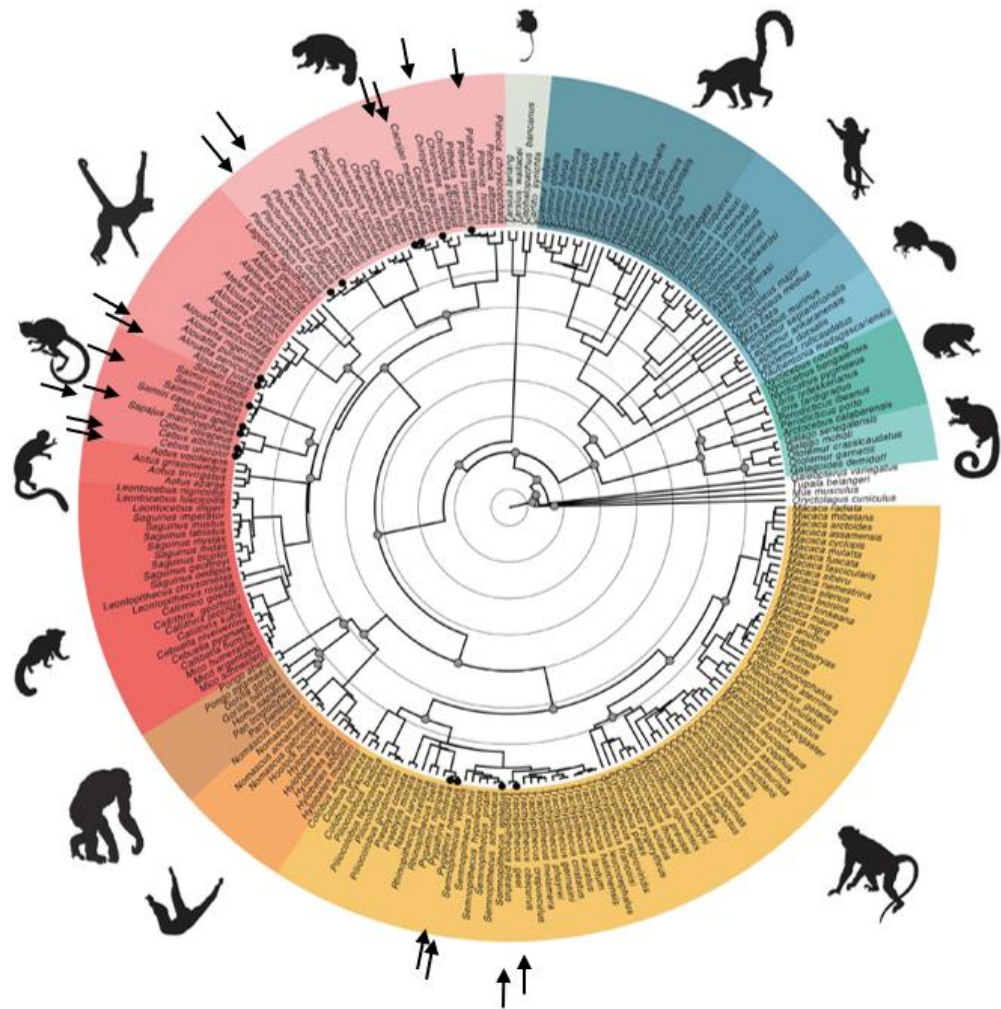


Biodiversitat i variabilitat genòmica



Les espècies més amenaçades tenen senyals genòmiques?

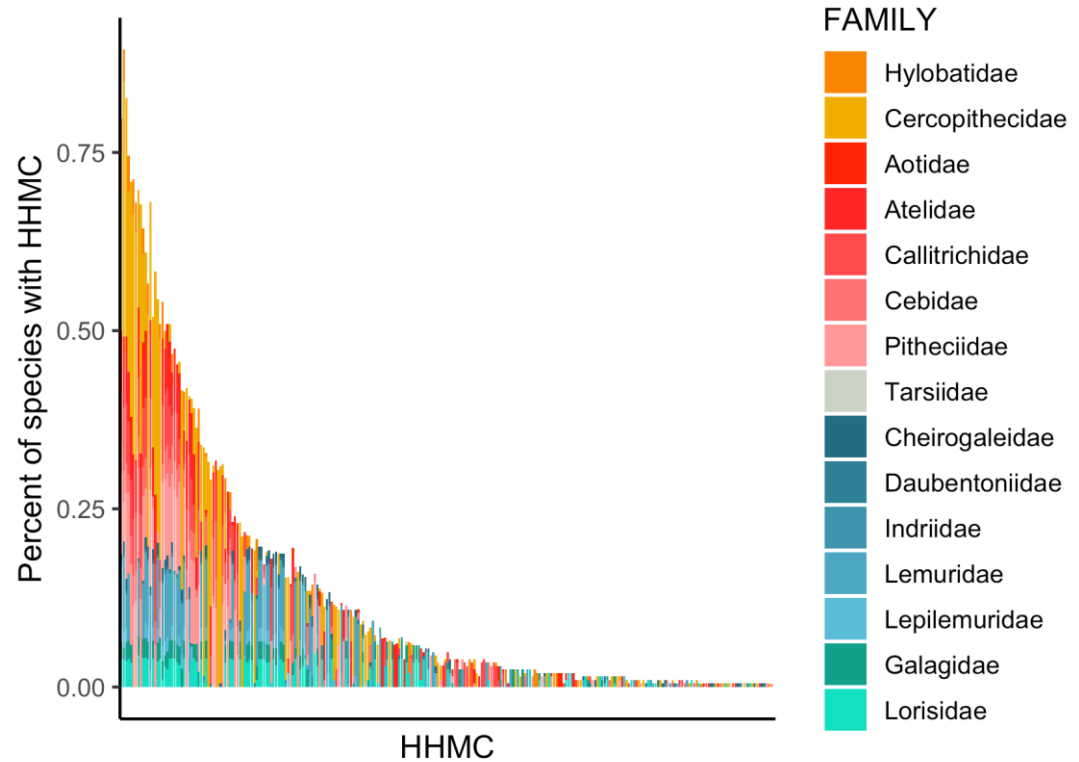
Filogènies i taxonomies



Què ens fa humans?

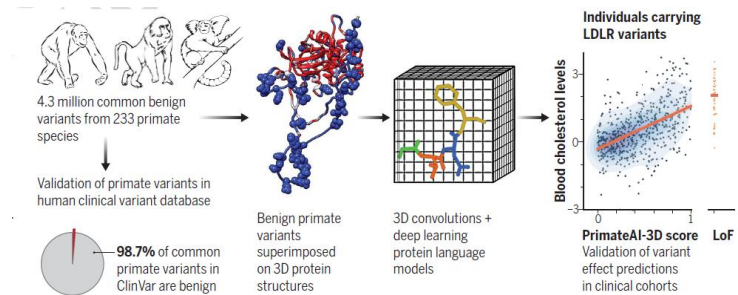
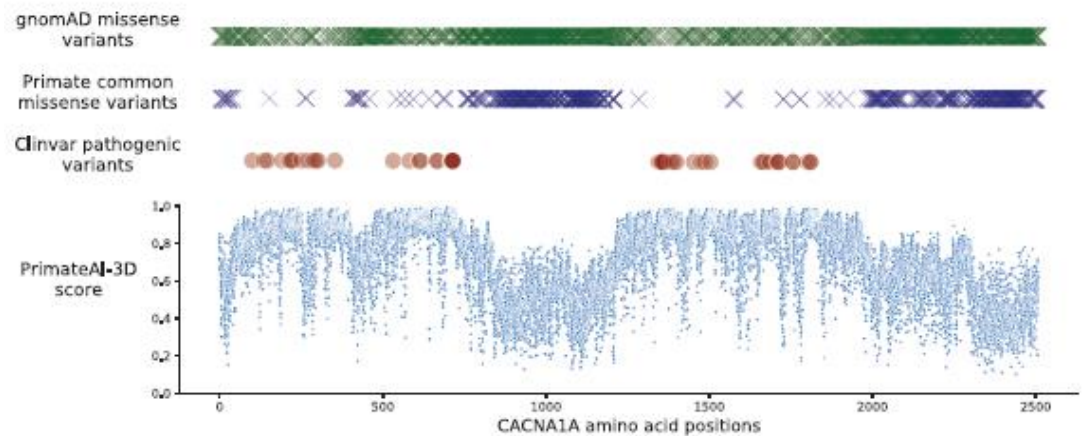


80 gens



HHMC: human lineage -specific
high-frequency
missense changes

Una nova visió del genoma humà i de com buscar mutacions de malalties



Resultat

RESEARCH ARTICLE SUMMARY

PRIMATE GENOMES

Rare penetrant mutations confer severe risk of common diseases

Petko P. Fizev¹, Jeremy McEl
Pierick Wainschein, Zijian Ni
François Aguet, Marc Fasnach
Heidi L. Rehm, Anne O'Donnell

RESEARCH ARTICLE SUMMARY

PRIMATE GENOMES

The landscape of tolerated genetic variation in humans and primates

Hong Gao¹, Tobias Hamp¹, Jeffrey Ede, Joshua G. Schraiber, Jeremy McRae, Mori

PRIMATE GENOMES

Genome-wide coancestry reveals details of ancient and recent male-driven reticulation in baboons

Erik F. Sørensen et al.

RESEARCH ARTICLE

PRIMATE GENOMES

A global catalog of whole-genome diversity from 233 primate species

Lukas F. K. Kuderna^{1,2,*}, Hong Gao², Mareike C. Janiak³, Martin Kuhlwiilm^{1,4,5}, Joseph D. Orkin^{1,6}, Thomas Bataillon⁷, Shivakumara Manu^{8,9}, Alejandro Valenzuela¹, Juraj Bergman^{7,10}, Marjolaine Rousselle⁷, Felipe Ennes Silva^{11,12}, Lidia Agueda¹³, Julie Blanc¹³, Marta Gut¹³, Dorien de Vries¹, Ian Goodhead¹, R. Alan Harris¹⁴, Muthuswamy Raveendran¹⁴, Axel Jensen¹⁵, Idrissa S. Chuma¹⁶, Julie E. Horvath^{17,18,19,20,21}, Christina Hvilsom²², David Juan¹, Peter Frandsen²², Joshua G. Schraiber², Fabiano R. de Melo²³, Fabricio Bertuol²⁴, Hazel Byrne²⁵, Iracilda Sampaio²⁶, Izeni Farias²⁴, João Valsecchi^{27,28,29}, Malu Messias¹⁰, Maria N. F. da Silva³¹, Mihir Trivedi⁹, Rogerio Rossi³², Tomas Hrbek^{24,33}, Nicole Andriaholinirina³⁴, Clément J. Rabarivola³⁴, Alphonse Zaramody³⁴, Clifford J. Jolly³⁵, Jane Phillips-Conroy³⁶, Gregory Wilkerson³⁷, Christian Abec³⁷, Joe H. Simmons³⁷, Eduardo Fernandez-Duque³⁸, Sree Kanthaswamy³⁹, Fekadu Shiferaw⁴⁰, Dongdong Wu⁴¹, Long Zhou⁴², Yong Shao⁴¹, Guojie Zhang^{43,42,44,45,46}, Julius D. Keyyu⁴⁷, Sascha Knaut⁴⁸, Minh D. Le⁴⁹, Esther Lizano^{1,50}, Stefan Merker⁵¹, Arcadi Navarro^{1,52,53,54}, Tilo Nadler⁵⁵, Chiea Chuen Khor⁵⁶, Jessica Lee⁵⁷, Patrick Tan^{58,59,56}, Weng Khong Lim^{59,58,60}, Andrew C. Kitchener⁶¹, Dietmar Zinner^{62,63,64}, Ivo Gut¹³, Amanda D. Melin^{65,66,67}, Katerina Guschanski^{68,15}, Mikkel Heide Schierup⁷, Robin M. D. Beck³, Govindhaswamy Umapathy^{8,9}, Christian Roos⁶⁹, Jean P. Boubli³, Jeffrey Rogers¹⁴, Kyle Kai-How Farh², Tomas Marques Bonet^{1,50,13,52}



Article

Identification of constrained sequence elements across 239 primate genomes

<https://doi.org/10.1038/s41586-023-06798-8>

Received: 9 February 2023

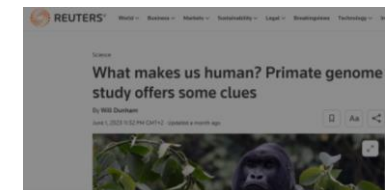
Accepted: 30 October 2023

Published online: 29 November 2023

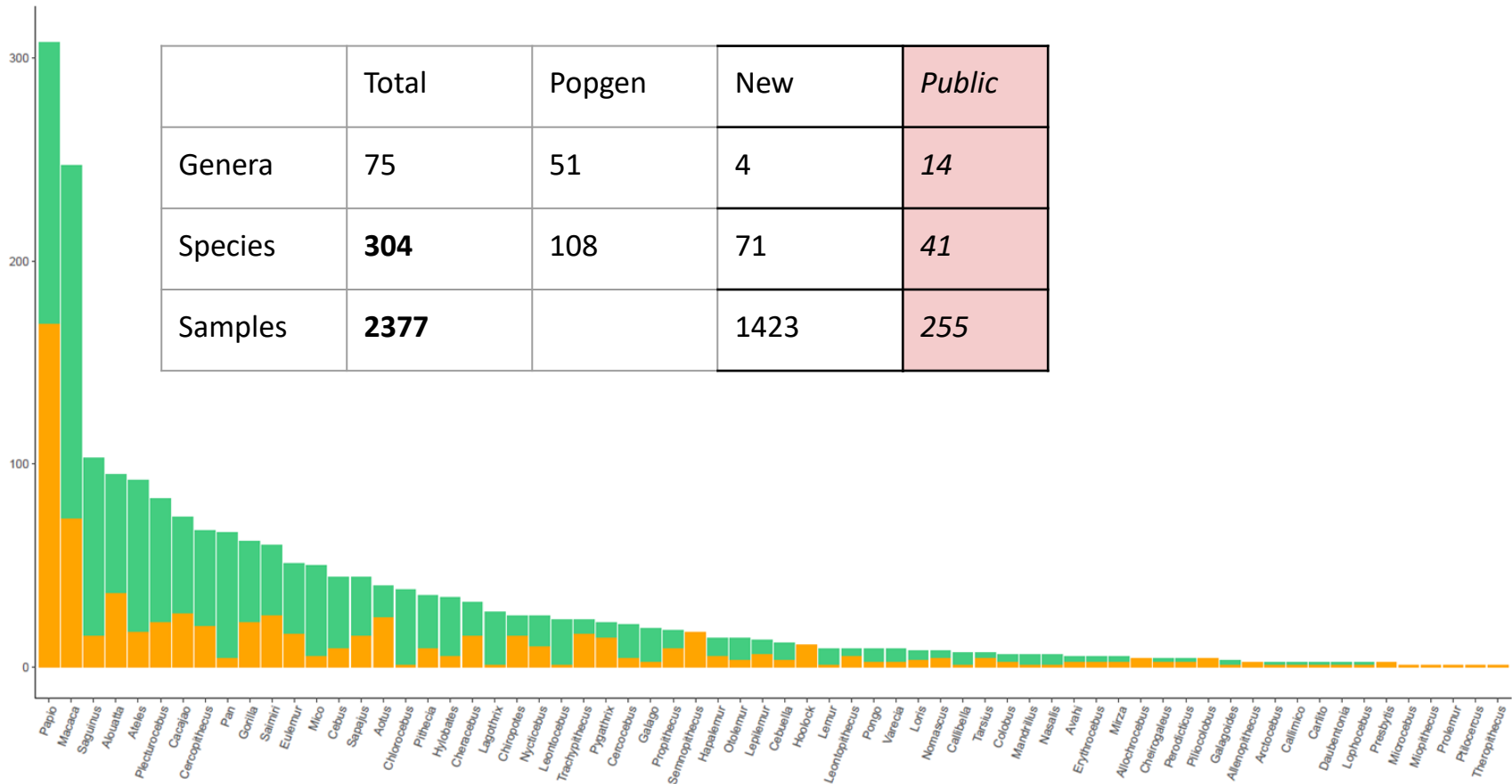
Open access

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Noncoding DNA is central to our understanding of human gene regulation and complex diseases^{1,2}, and measuring the evolutionary sequence constraint can establish the functional relevance of putative regulatory elements in the human genome^{3–5}. Identifying the genomic elements that have become constrained specifically in primates has been hampered by the faster evolution of noncoding DNA compared to protein-coding DNA⁶, the relatively short timescales separating primate species⁷, and the previously limited availability of whole-genome sequences⁸. Here we reconstruct a whole-genome alignment of 730 species, representing nearly half of all



Això no ha acabat.... (II)



One Health



Agraïments



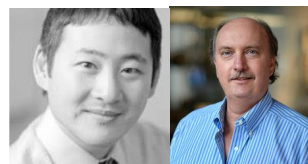
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Joshua G. Schraiber	Illumina
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Mareike Janiak	University of Salford
Dorien de Vries	University of Salford
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