REZGEN-IBA
Red IberoAmericana sobre los recursos zoogenómicos y su resiliencia

GENOMIC TOOLS FOR THE CONSERVATION OF IBERO-AMERICAN DOMESTIC ANIMALS

Catarina Ginja

BIOPOLIS cibio U.PORTO

FRAMEWORK

- The **Creole populations** of various species of domestic animals in Latin America are the result of animals that were introduced over the years, especially in the early stages of transatlantic trade.
- Many Creole breeds have disappeared in the last century, although some **still survive in marginal regions**.
- The **REZGEN-IBA network** is funded by the Ibero-American Program of Science and Technology for Development – CYTED (Ref. 123RT0139; 2023-2027).
- It is based on **existing knowledge and networks**, especially CONBIAND, derived from our previous and ongoing national and international projects

THE REZGEN-IBA NETWORK

Major aim

The use of **genomic tools** will make it possible to understand, rescue, conserve, and value Creole livestock populations, which are the result of more than 500 years of **adaptation to the diverse environmental conditions** they encountered in America, and which represent a source of wealth and food for millions of Ibero-American citizens, especially in the most rural environments.

THE REZGEN-IBA NETWORK

Specific aims

- Consolidate an Ibero-American cooperative framework for the **characterization, conservation, recognition, and valorization** of the region's zoogenomic heritage through the application of genomic tools
- Define **standardized protocols** across participating groups, including sample collection, phenotyping, genotyping, and bioinformatics data processing.
- Addressing conservation and **genomic characterization** challenges while engaging a broad spectrum of groups, from those with substantial funding to those with limited financial resources.
- Promote scientific events, **training** sessions, and **knowledge transfer initiatives**.

TEAM

- A multidisciplinary team of 178 researchers, breeders, and companies from 15 Ibero-American countries, Italy and the U.S collaborating on training, research, and knowledge transfer activities.

BIOBOVIS
CONSORTIUM

scientific reports

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nature > scientific reports > articles > article

Article | Open access | Published: 07 August 2019

The genetic ancestry of American Creole cattle inferred from uniparental and autosomal genetic markers

Catarina Ginja, Luis Telo Gama, Oscar Cortés ✉, Inmaculada Martin Burriel, Jose Luis Vega-Pla, Cecilia Penedo, Phil Sponenberg, Javier Cañón, Arianne Sanz, Andrea Alves do Egito, Luz Angela Alvarez, Guillermo Giovambattista, Saif Agha, Andrés Rogberg-Muñoz, Maria Aparecida Cassiano Lara, BioBovis Consortium, Juan Vicente Delgado & Amparo Martinez

Scientific Reports **9**, Article number: 11486 (2019) | Cite this article

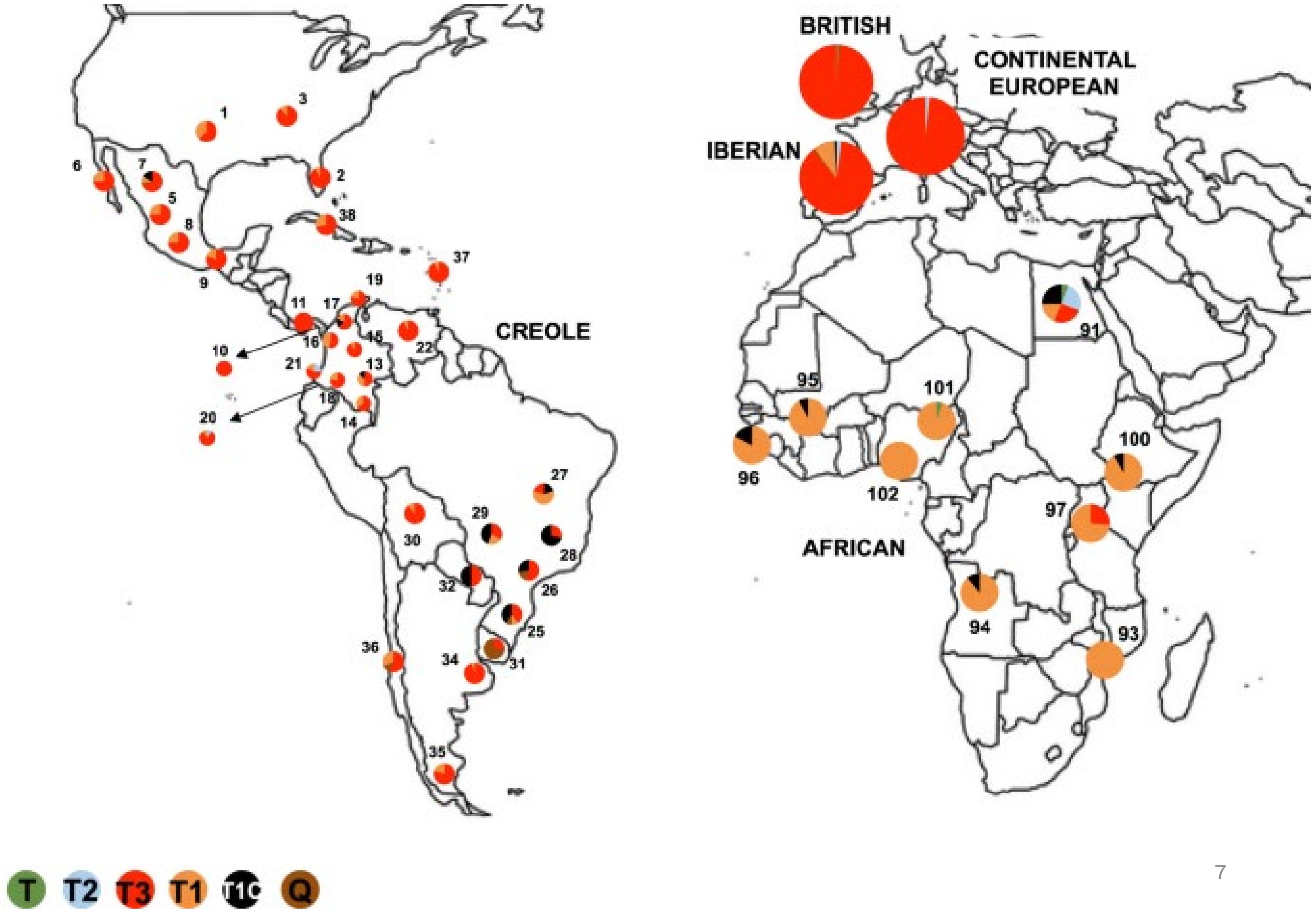
ACTIVITIES & IMPACT

4,658 animals from 114 cattle breeds distributed worldwide, including 1,480 animals from 40 Creole breeds.

GENETIC ANCESTRY OF CREOLE CATTLE

African & Iberian ancestry

T1c mitochondrial lineages are rare in Iberia but common in Africa and are well represented in Creoles from **Brazil and Colombia**, lending support to a direct African influence.



GENETIC ANCESTRY OF CREOLE CATTLE

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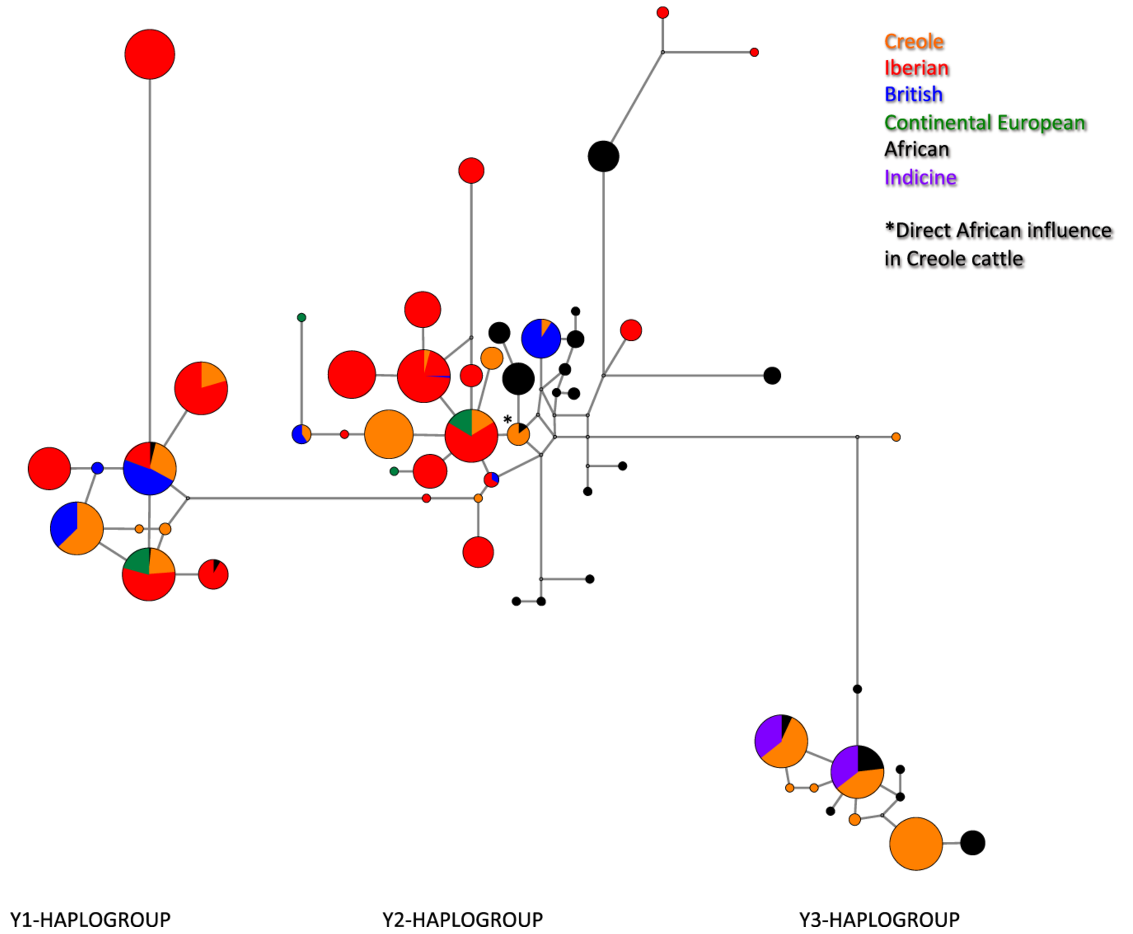


ELSEVIER

Animal

Volume 12, Issue 10, 2018, Pages 2017-2026

Dissection of ancestral genetic contributions to Creole goat populations

N. Sevane[1], O. Cortés[1], L.T. Gama[2], A. Martínez[3 4], P. Zaragoza[5], M. Amills[6], D.O. Bedotti[7], C. Bruno de Sousa[8], J. Cañon[1], S. Dunner[1], C. Ginja[9], M.R. Lanari[10], V. Landi[3 4], P. Sponenberg[11], J.V. Delgado[3], The BioGoat Consortium

CONBIAND

CONSERVACION de la BIODIVERSIDAD de los ANIMALES DOMESTICOS LOCALES

BIOGOAT
CONSORTIUM

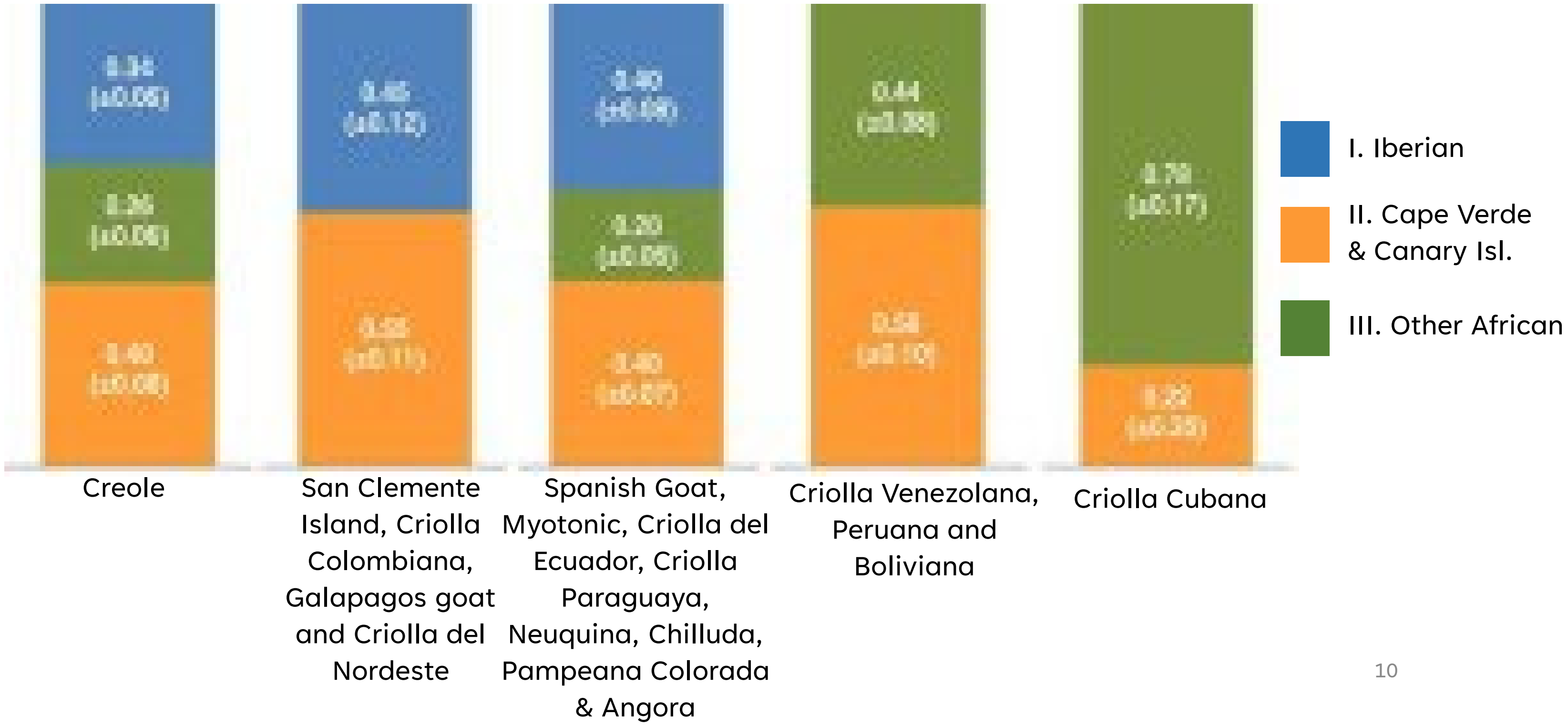
ACTIVITIES & IMPACT

2,395 goats from 71 breeds from diverse geographic origins, including 786 and 441 goats from 23 Creole and 13 African populations.

GENETIC ANCESTRY OF CREOLE GOATS

African & Iberian ancestry

The admixture estimates indicated that the Creole goat populations, including Brazilian breeds, had average ancestral contributions of 40%, 26% and 34% from Clusters I, II and III, respectively.



BIOPIG
CONSORTIUM

Heredity

Original Article | Published: 30 March 2016

Conservation priorities of Iberoamerican pig breeds and their ancestors based on microsatellite information

O Cortés, A M Martinez, J Cañon, N Sevane, L T Gama, C Ginja, V Landi, P Zaragoza, N Carolino, A Vicente, P Sponenberg & J V Delgado for the BioPig Consortium

Heredity **117**, 14–24 (2016) | Cite this article

ACTIVITIES & IMPACT

1,670 samples belonging to 45 populations/breeds, including 605 creole pigs from 17 populations.

GENETIC DIVERSITY OF CREOLE PIGS

Conservation priorities

The Core Set methodologies highly prioritized Criollo pig breeds: Cr. Boliviano, Cr. Pacifico, Cr. Cubano and Cr. Guadalupe. However, **weighing the between- and within-breed components** with FST and 1-FST, respectively, resulted in higher contributions of Iberian breeds.

BREED GROUP	Phylogenetic Group	N	He	Ho	Mean Number of Alleles
Iberian	Mediterranean	435	0,63	0,48	10.5
	Celtic	347	0.69	0.49	9.9
Criollo	CA	255	0.70	0.57	12.1
	CB	267	0.68	0.56	11.7
	CC	83	0.69	0.58	9.3
Commercial	Duroc	50	0.56	0.55	4.0
	other	120	0.69	0.59	7.4
Wild boar		113	0.65	0.55	9.0
Total	All	1670	0.66±0.05	0.55±0.04	9.2±3.5

CA: Mulefoot, Pampa Rocha, Caracolero Wet, Criollo Boliviano, Criollo El Salvador, Criollo Pacifico, Caracolero Dry
CB: Red Wattle Hog, Criollo Cubano, Pelon Mexicano, Zungo, New Mexico, Criollo Venezolano, Criollo Ecuatoriano
CC: Guinea Hog, Criollo Guadalupe, Sanpedreño

CONBIAND
CONSERVACION de la BIODIVERSIDAD
de los ANIMALES DOMESTICOS LOCALES

BIOHORSE
CONSORTIUM

Journal of
Animal Breeding and Genetics

ORIGINAL ARTICLE

The legacy of Columbus in American horse populations assessed by microsatellite markers

O. Cortés, S. Dunner, L.T. Gama, A.M. Martínez, J.V. Delgado, C. Ginja, L.M. Jiménez, J. Jordana, C. Luis, M.M. Oom, D.P. Sponenberg, P. Zaragoza, Biohorse Consortium, J.L. Vega-Pla

First published: 14 February 2017 | https://doi.org/10.1111/jbg.12255 | Citations: 22

Biohorse Consortium: The list of Biohorse Consortium members are given in the Acknowledgment section (http://biohorse.jimdo.com).

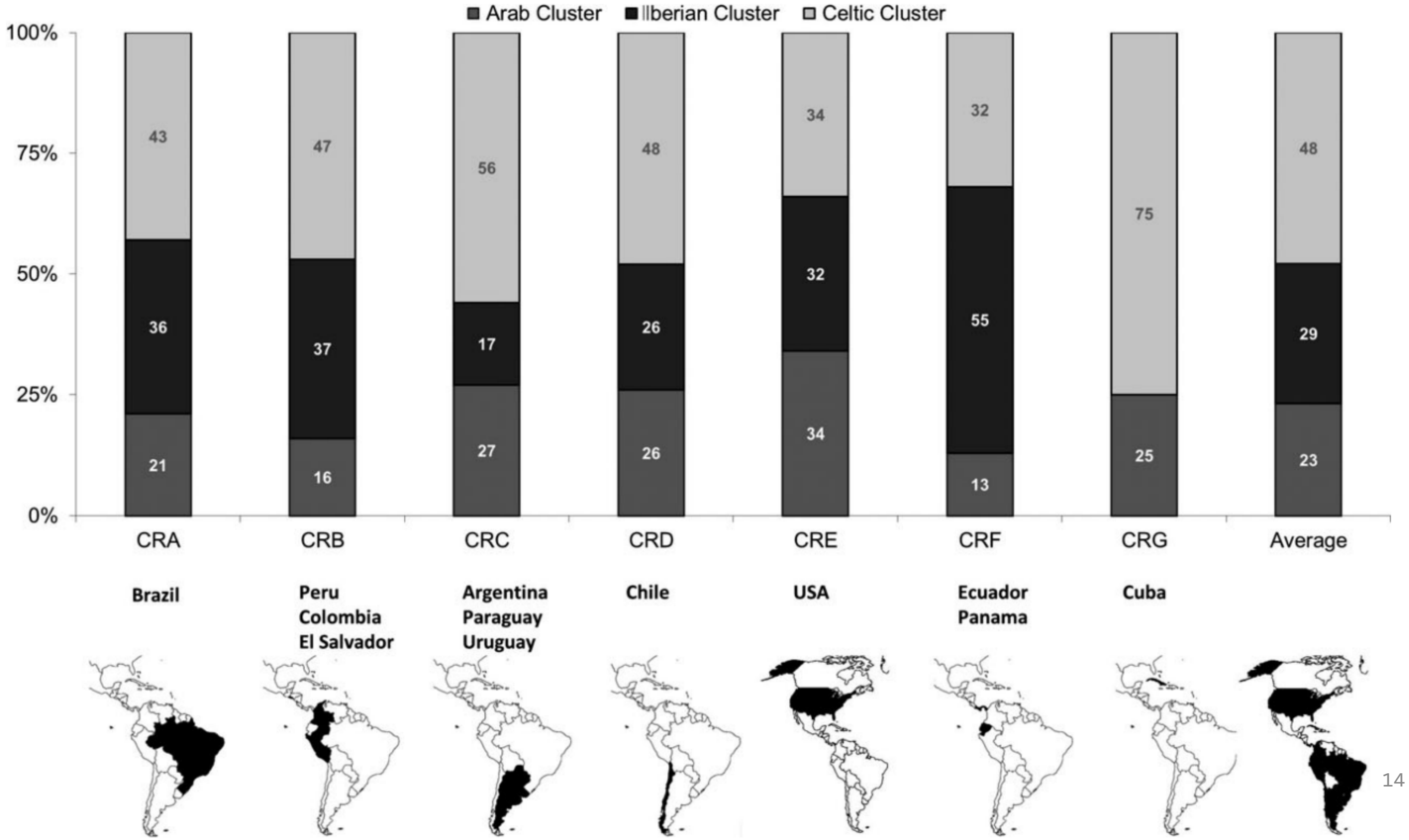
ACTIVITIES & IMPACT

2,385 samples belonging to 50 horse breeds, including 27 Creole populations.

GENETIC ANCESTRY OF CREOLE HORSES

Iberian ancestry

For the Criollo breeds, the **estimated genetic contribution from other breeds** was ~50%, 30% and 20% for the Celtic, Iberian and Arab-Thoroughbred groups, respectively. **Hotspots of genetic diversity** are observed in populations from Colombia, Ecuador, Brazil, Paraguay and the western USA.



BIOVIS
CONSORTIUM

ACTIVITIES & IMPACT

1,913 samples belonging to 41 Iberian and 6 transboundary commercial sheep breeds.

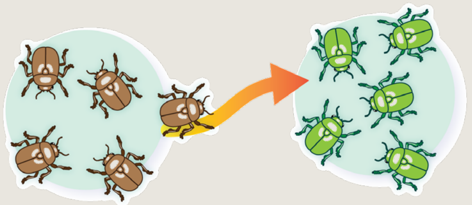
WE HAVE A GOOD KNOWLEDGE OF THE GENETIC DIVERSITY & POPULATION STRUCTURE OF CREOLE LIVESTOCK

OMICS’ APPROACHES FOR CONSERVATION

Genetic drift

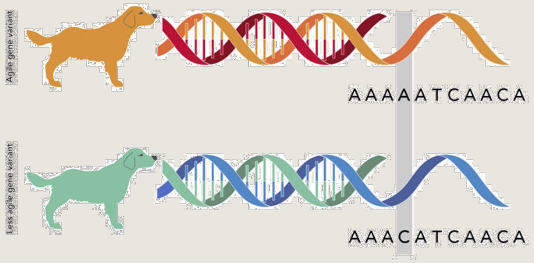
some individuals may **randomly** produce more offspring (and genes, of course!)

Gene flow & Crossbreeding



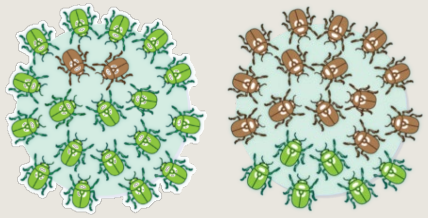
Genetic variants introduced into a population can be an important source of **genetic variability**.

Mutation & Selection



New source of genetic variation. Beneficial non-synonymous mutations can increase frequency in the population – positive selection.

Evolution



These two generations of the same population are genetically different. **Populations evolve NOT individuals!**

https://evolution.berkeley.edu/sampling-error-and-evolution/

OMICS’ APPROACHES FOR CONSERVATION

High-throughput sequencing

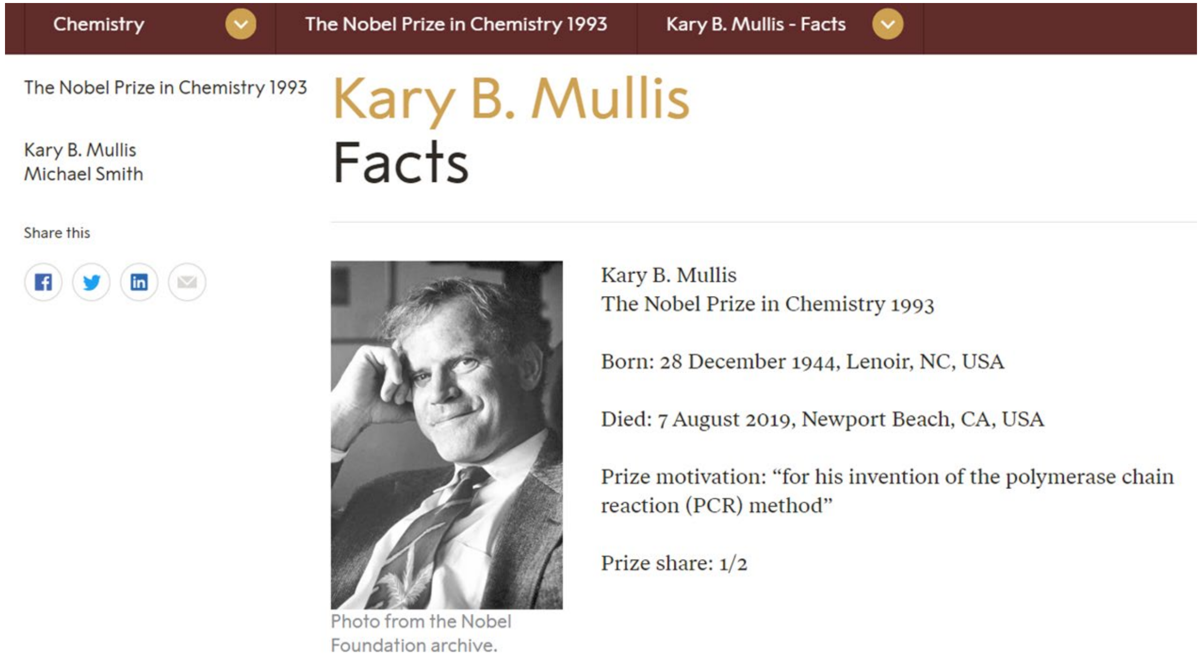
Unprecedented high-resolution when studying origins and admixture events; selection for ancestral and novel variants – identify loci that underwent positive selection.

Ultimately, allow us to:

- Disclose genome-wide patterns of diversity associated with the formation of local breeds;
- Understand the directions and degree of gene flow between populations from different geographical locations;
- Investigate selection and animal breeding/improvement.

Polymerase Chain Reaction – PCR

PCR was invented in 1983 by American biochemist Kary Mullis at Cetus Corporation. Mullis and biochemist Michael Smith, who had developed other essential ways of manipulating DNA, were jointly awarded the Nobel Prize in Chemistry in 1993.



Kary B. Mullis
Facts

Kary B. Mullis
The Nobel Prize in Chemistry 1993

Born: 28 December 1944, Lenoir, NC, USA

Died: 7 August 2019, Newport Beach, CA, USA

Prize motivation: “for his invention of the polymerase chain reaction (PCR) method”

Prize share: 1/2

Photo from the Nobel Foundation archive.

https://www.nobelprize.org/prizes/chemistry/1993/mullis/facts/

HUMAN GENOME – WORLD’S LARGEST COLLABORATIVE BIOLOGICAL PROJECT!

Released in February 2001, “complete” genomes were produced by Celera (privately funded by Celera Genomics, Craig Venter, and the publicly funded Human Genome Project.

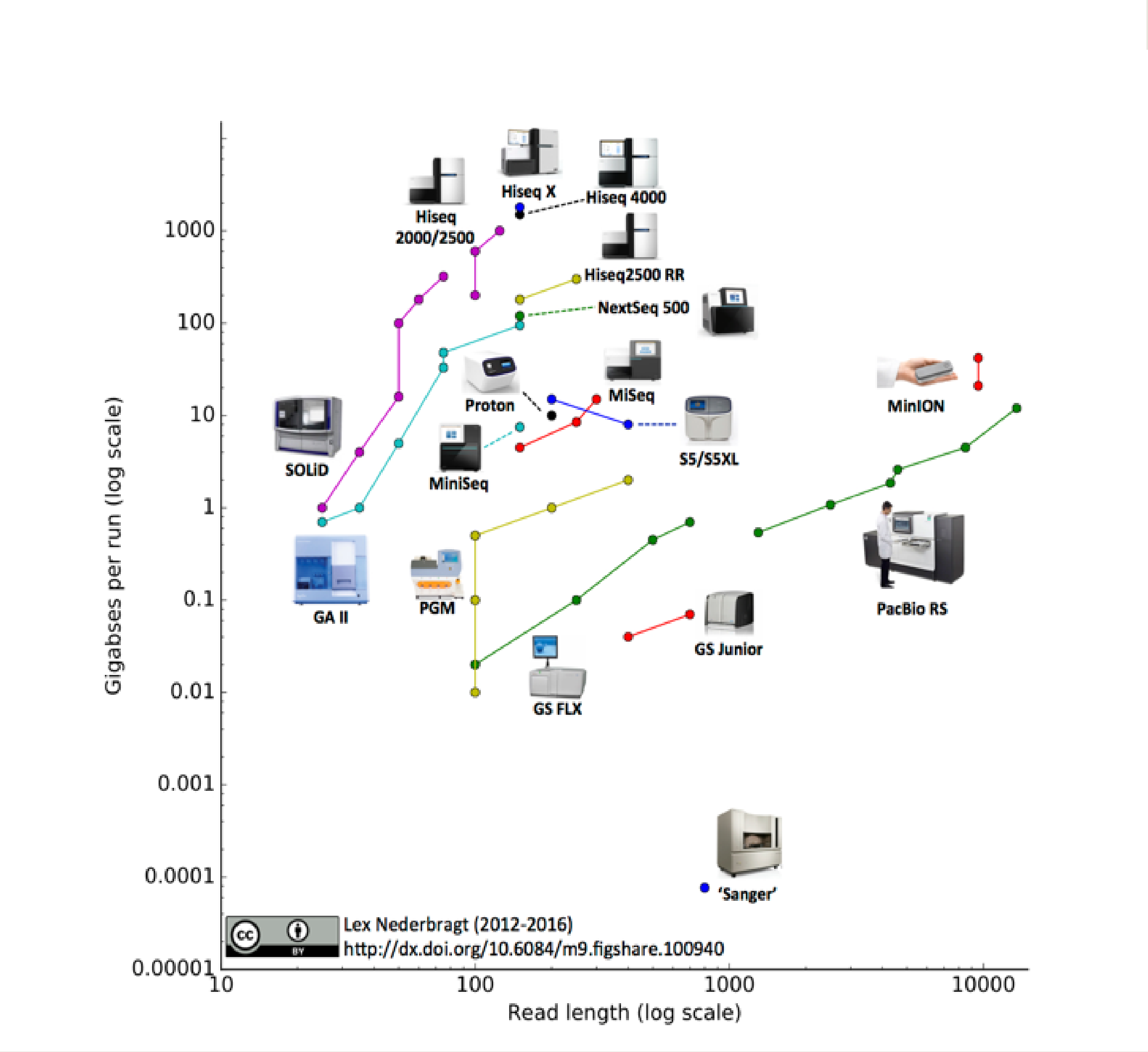
3 billion base pairs!

https://www.genome.gov/about-genomics/educational-resources/fact-sheets/human-genome-project

OMICS' APPROACHES

High-throughput sequencing

- The NovaSeq 6000 (Illumina) system (short reads) can sequence up to 48 genomes in ~2 days with **high coverage**. Allows for **multiplexing** to maximize the number of samples per flow cell.
- PacBio's ability to generate long reads enables easier **genome assembly** and increases **accuracy**.
- MinION (Oxford nanopore) the only **portable, real-time** devices for (long) DNA and RNA sequencing.



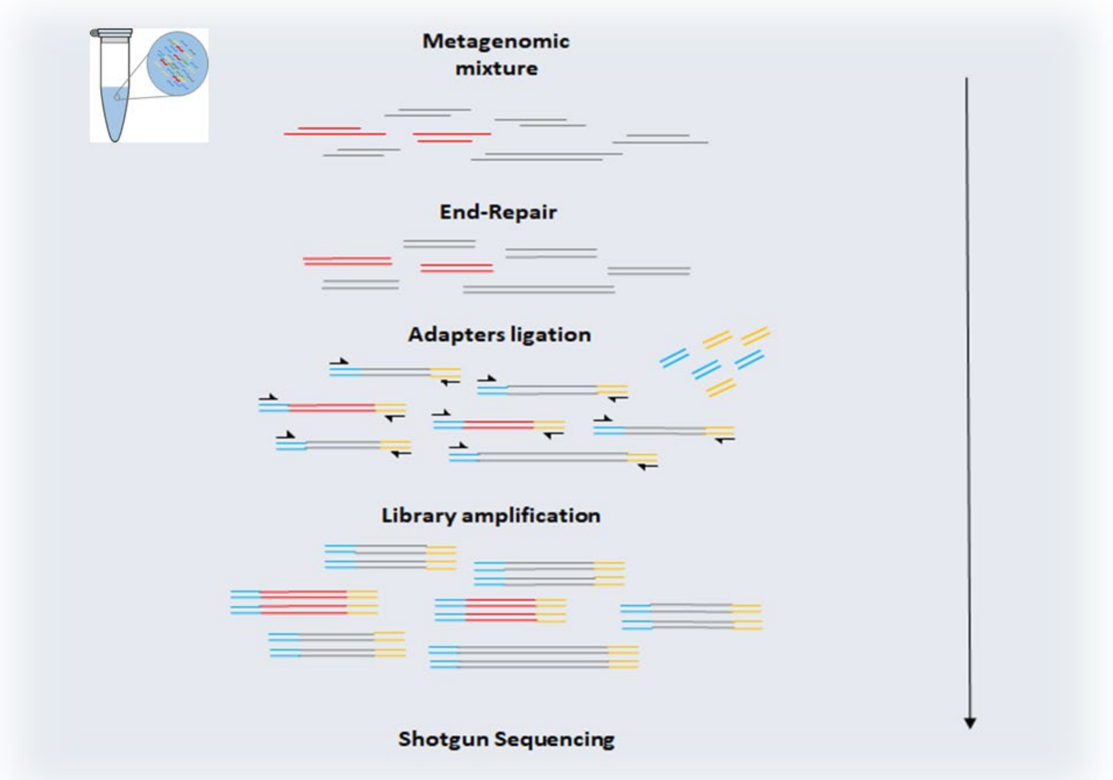
Lex Nederbragt (2012-2016)
http://dx.doi.org/10.6084/m9.figshare.100940

OMICS' APPROACHES

Shotgun sequencing

The method involves randomly breaking up the genome into small DNA fragments that are sequenced individually. A computer program looks for overlaps in the DNA sequences, using them to reassemble the fragments in their correct order to reconstitute an organism's genome.

https://www.genome.gov/genetics-glossary/Shotgun-Sequencing

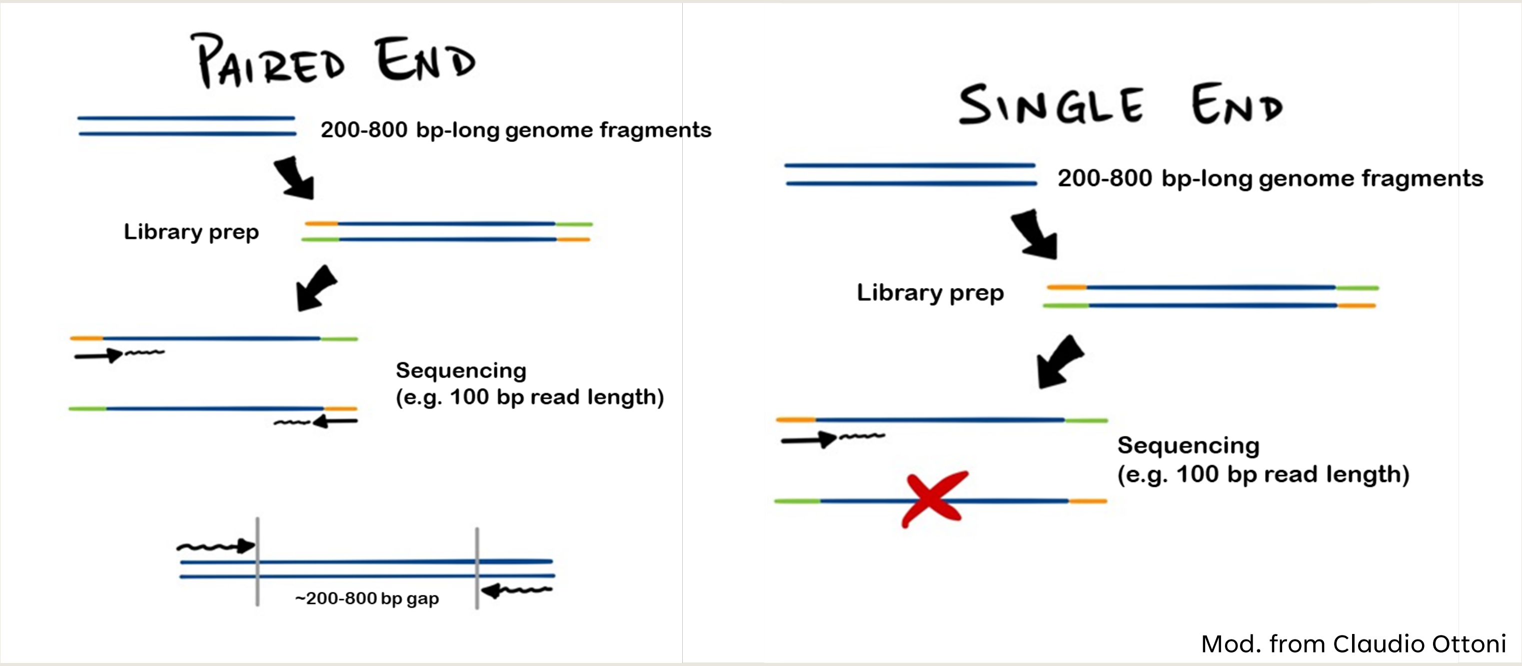


Mod. from Stoneking & Krause 2011 and Metzker 2010 Nat Rev Genet

OMICS' APPROACHES

Shotgun sequencing

Paired-end sequencing allows users to sequence both ends of a fragment, thus more accurate read alignment.



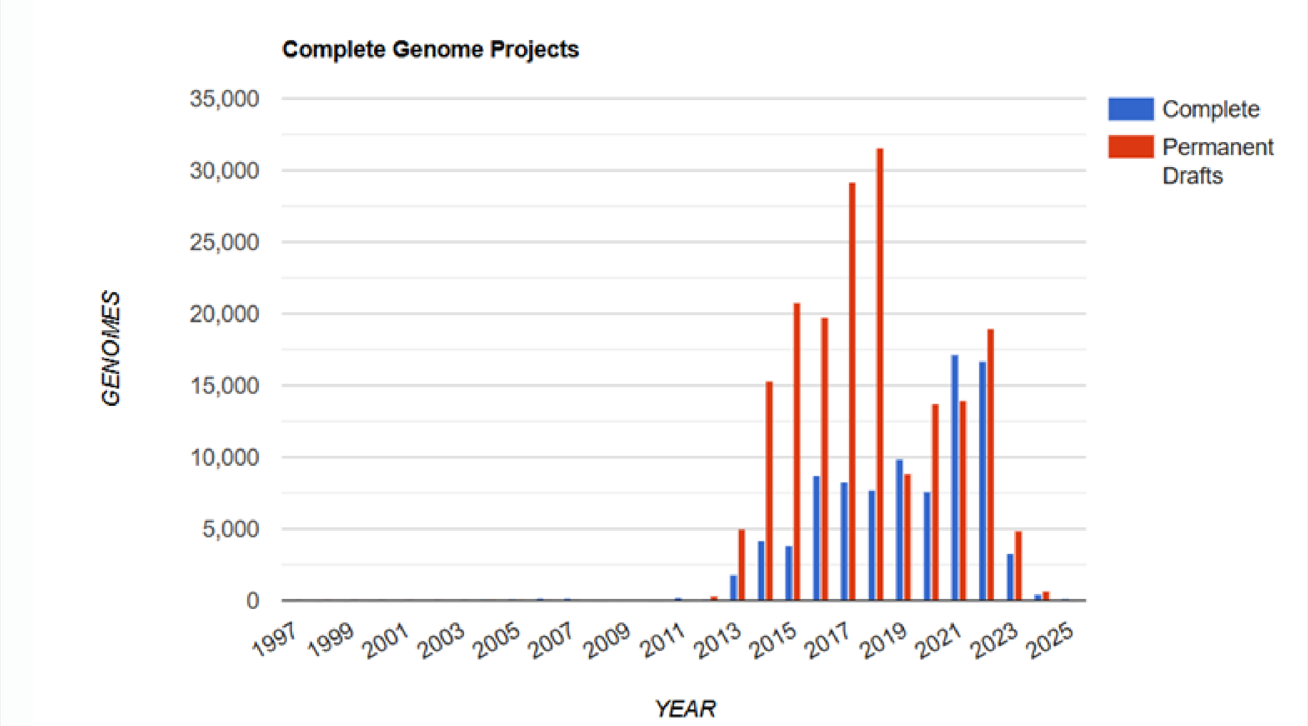
Mod. from Claudio Ottoni

https://www.illumina.com/science/technology/next-generation-sequencing/plan-experiments/paired-end-vs-single-read.html

OMICS' APPROACHES

Whole-genome sequencing

"Complete" genomes have increased, but draft genomes remain to be completed.

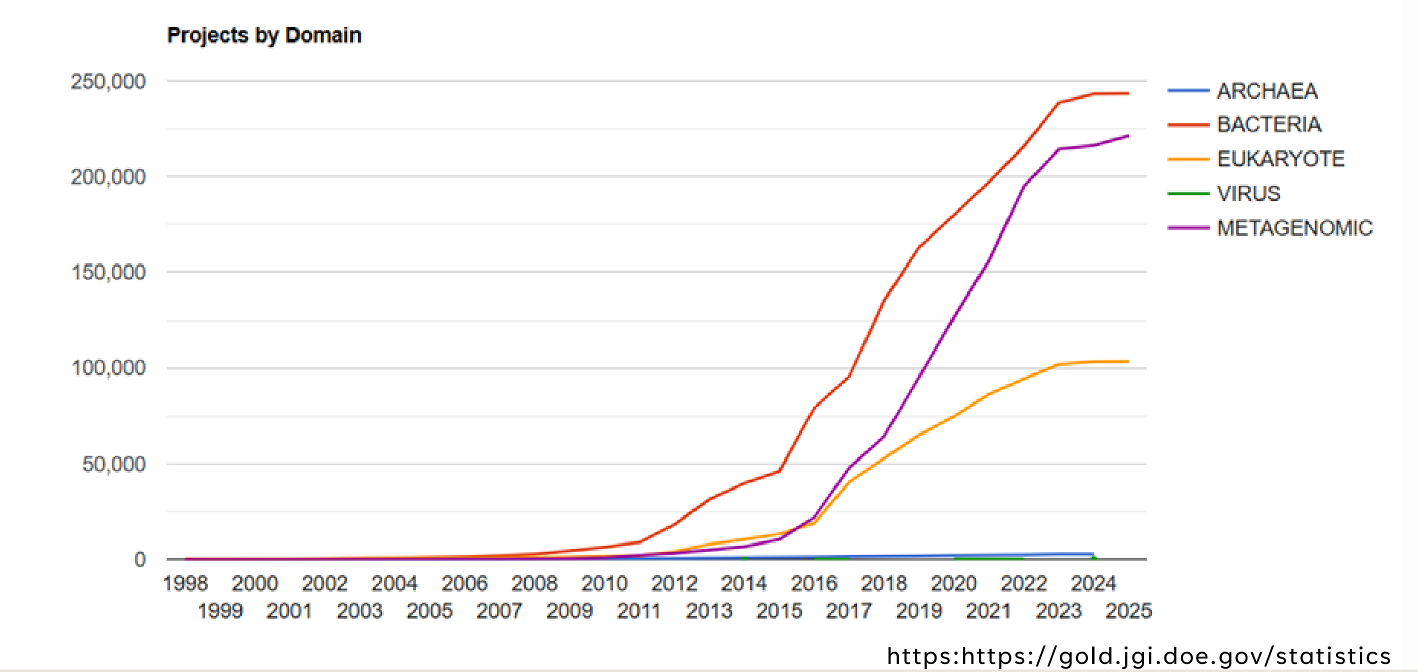


https:https://gold.jgi.doe.gov/statistics

OMICS' APPROACHES

Whole-genome sequencing

Since 2010, a huge increase in genome sequencing projects across taxa.

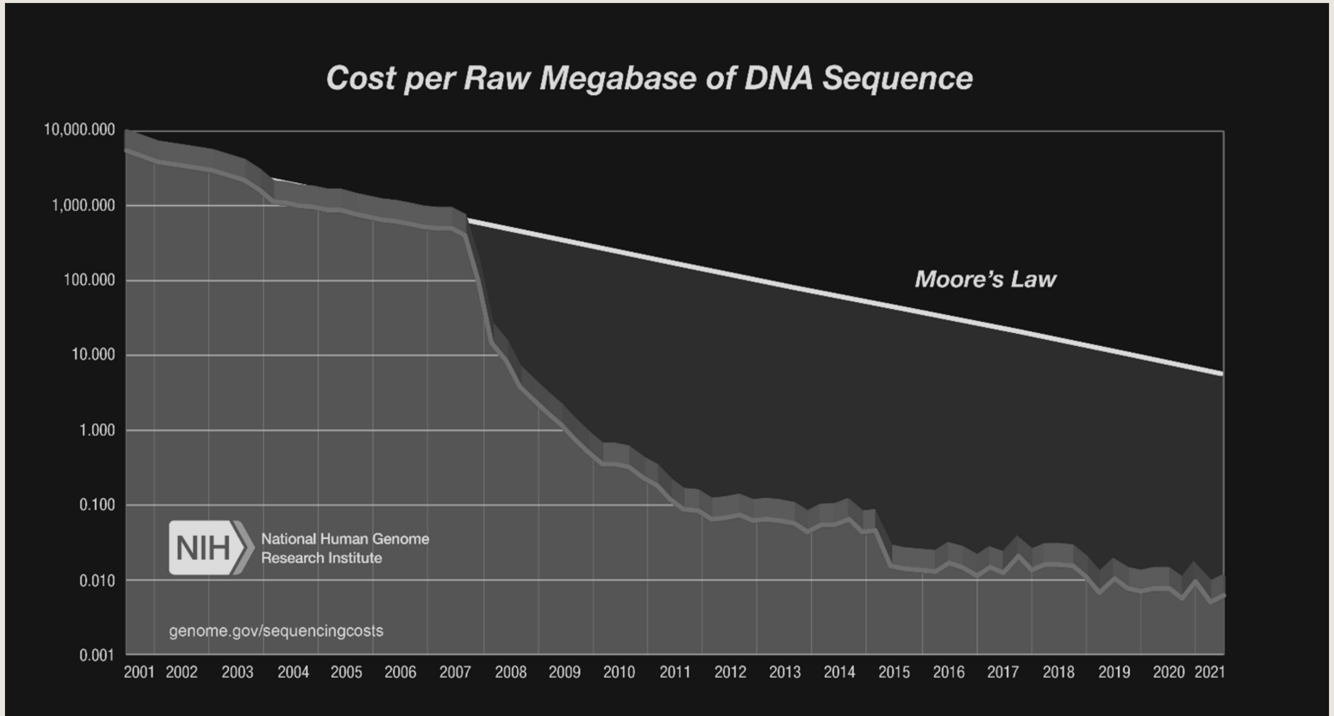


https:https://gold.jgi.doe.gov/statistics

OMICS’ APPROACHES

Whole-genome sequencing

Significant decrease in sequencing costs.



https://www.genome.gov/sequencingcostsdata/

OMICS' APPROACHES

Whole-genome sequencing

- Large amount of information available
- Reading quality improved
- Less time-consuming & lower costs for lab processing
- More time-consuming & increased complexity in data analysis
- Need to check sequences’ authenticity

OMICS' APPROACHES

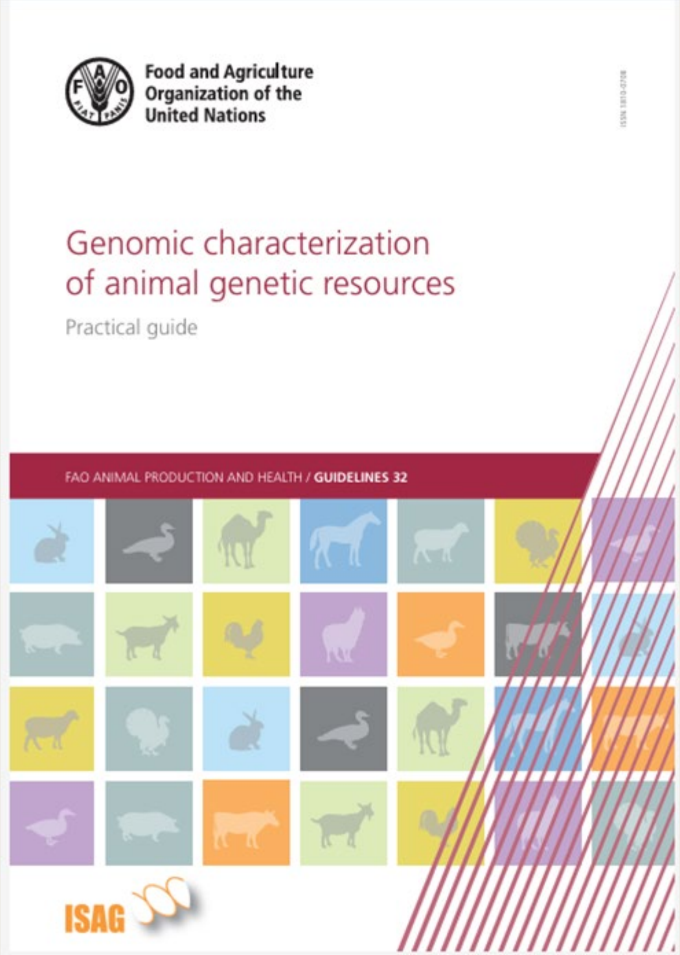
Several applications

- **Genome sequencing**: whole-genome shotgun sequencing is the dominant methodology
- **Transcriptome sequencing** (RNAseq): gene expression analysis with substantial advantages over microarray analysis
- **Protein/DNA/RNA interaction-seq**: ChIP-seq (sequence of DNA bound to proteins, e.g. chromatins etc.) and other methods
- **Epigenetic modifications**: bisulfite sequencing: 5-methylcytosine (5mC) and 5-hydrossymethylcytosine (5hmC)

GENOMIC CHARACTERIZATION OF ANIMAL GENETIC RESOURCES

FAO & ISAG practical guidelines

Advancements in the sequencing of genomes and related genotyping methods have created opportunities for gathering much more information on the molecular level than ever possible, at a faster rate, and for exponentially decreased costs. The technical capacity for **the full application of genomic tools may represent a greater obstacle to their utilization than the expenses involved**.



Food and Agriculture Organization of the United Nations

Genomic characterization of animal genetic resources

Practical guide

FAO ANIMAL PRODUCTION AND HEALTH / **GUIDELINES 32**

ISAG

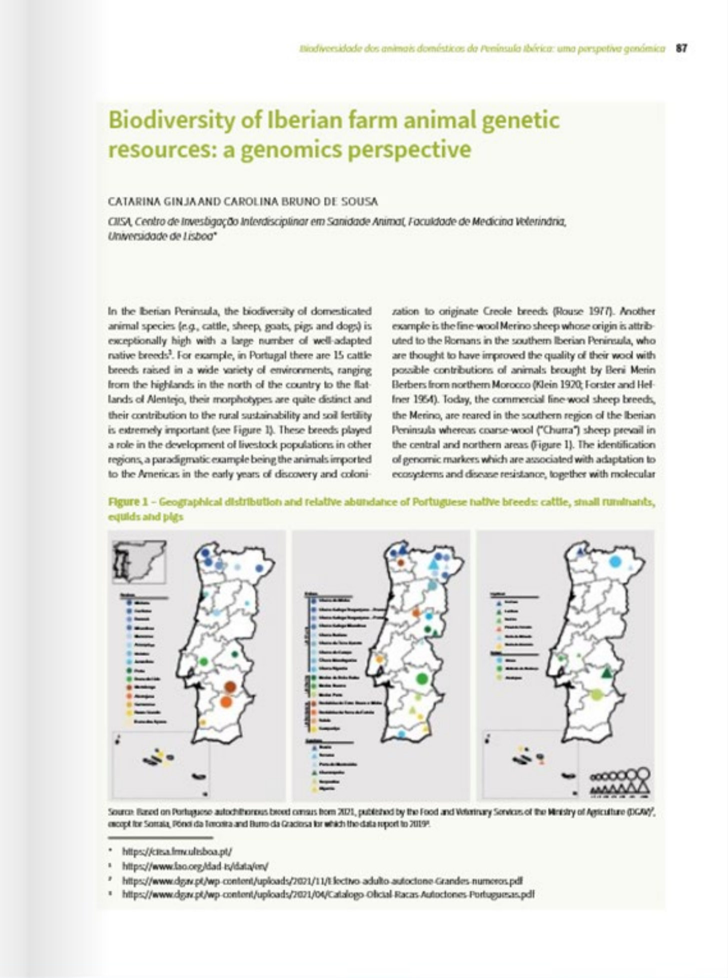
https://doi.org/10.4060/cc3079en

GENOMIC CHARACTERIZATION OF ANIMAL GENETIC RESOURCES

Cultivar special title

Focused on the work of Portuguese team of the REZGEN-IBA, provides an **overview of the recent technological advancements and its applications for the genomic characterization of animal genetic resources**.

Available online for free in English and Portuguese with a glossary of genomic terms.



Biodiversidade dos animais domésticos da Península Ibérica: uma perspetiva genómica **87**

Biodiversity of Iberian farm animal genetic resources: a genomics perspective

CATARINA GINJA AND CAROLINA BRUNO DE SOUSA

CIISA, Centro de Investigação Interdisciplinar em Sanidade Animal, Faculdade de Medicina Veterinária, Universidade de Lisboa[*]

In the Iberian Peninsula, the biodiversity of domesticated animal species (e.g., cattle, sheep, goats, pigs and dogs) is exceptionally high with a large number of well-adapted native breeds[1]. For example, in Portugal there are 15 cattle breeds raised in a wide variety of environments, ranging from the highlands in the north of the country to the flatlands of Alentejo, their morphotypes are quite distinct and their contribution to the rural sustainability and soil fertility is extremely important (see Figure 1). These breeds played a role in the development of livestock populations in other regions, a paradigmatic example being the animals imported to the Americas in the early years of discovery and colonization to originate Creole breeds (Rouse 1977). Another example is the fine-wool Merino sheep whose origin is attributed to the Romans in the southern Iberian Peninsula, who are thought to have improved the quality of their wool with possible contributions of animals brought by Beni Merin Berbers from northern Morocco (Klein 1920; Forster and Helfner 1964). Today, the commercial fine-wool sheep breeds, the Merino, are reared in the southern region of the Iberian Peninsula whereas coarse-wool ("Churra") sheep prevail in the central and northern areas (Figure 1). The identification of genomic markers which are associated with adaptation to ecosystems and disease resistance, together with molecular

Figure 1 – Geographical distribution and relative abundance of Portuguese native breeds: cattle, small ruminants, equids and pigs

Source: Based on Portuguese autochthonous breed census from 2021, published by the Food and Veterinary Services of the Ministry of Agriculture (DGAV)[2], except for Sorraia, Pónei da Terceira and Burro da Graciosa for which the data report to 2019[3].

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[*] https://ciisa.fmv.ulisboa.pt/

[1] https://www.fao.org/dad-is/data/en/

[2] https://www.dgav.pt/wp-content/uploads/2021/11/Efectivo-adulto-autoctone-Grandes-numeros.pdf

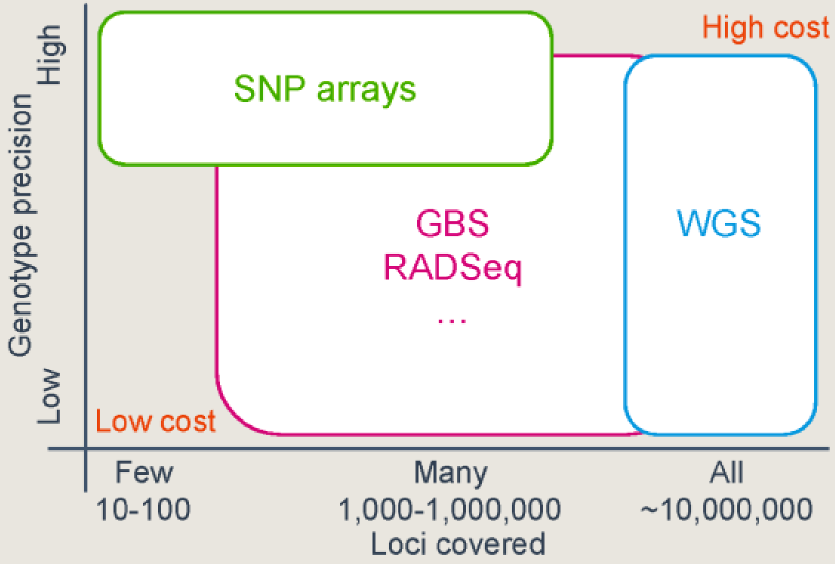
[3] https://www.dgav.pt/wp-content/uploads/2021/04/Catalogo-Oficial-Racas-Autoctones-Portuguesas.pdf

https://www.gpp.pt/images/GPP/O_que_disponibilizamos/Publicacoes/CULTIVAR_30/78/

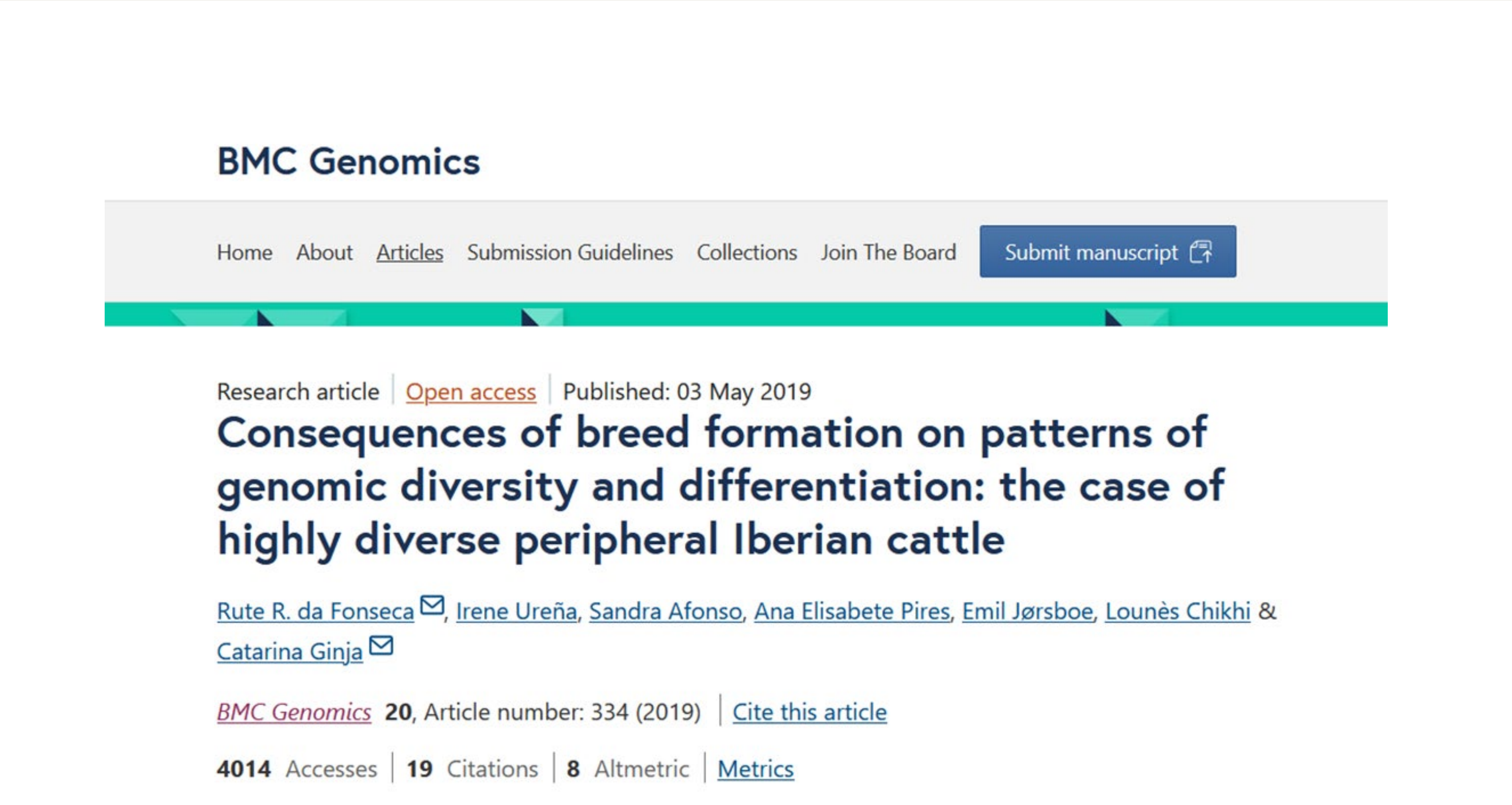
OMICS' APPROACHES

SNPs vs whole-genome-sequencing

The reduction in the cost of **genomic sequencing** facilitates the development of biomarkers for adaptive and productive traits of interest, with a focus on local resources, allowing us to circumvent the **bias associated with the use of SNPs identified in commercial breeds**.



Whole-Genome Sequencing



BMC Genomics

Research article | Open access | Published: 03 May 2019

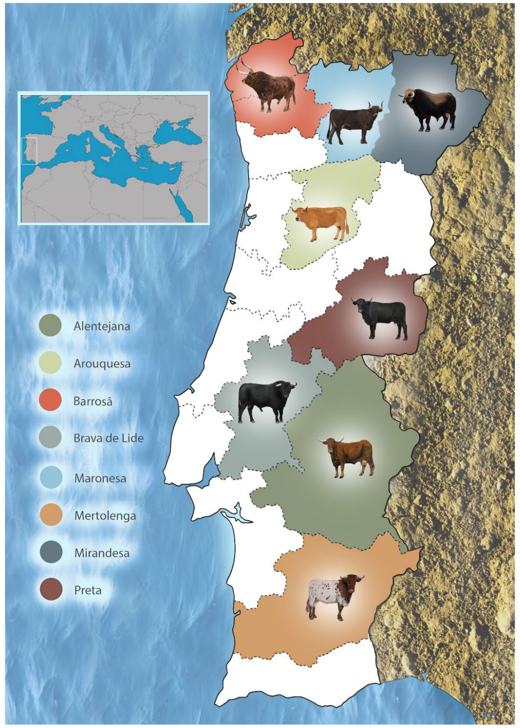
Consequences of breed formation on patterns of genomic diversity and differentiation: the case of highly diverse peripheral Iberian cattle

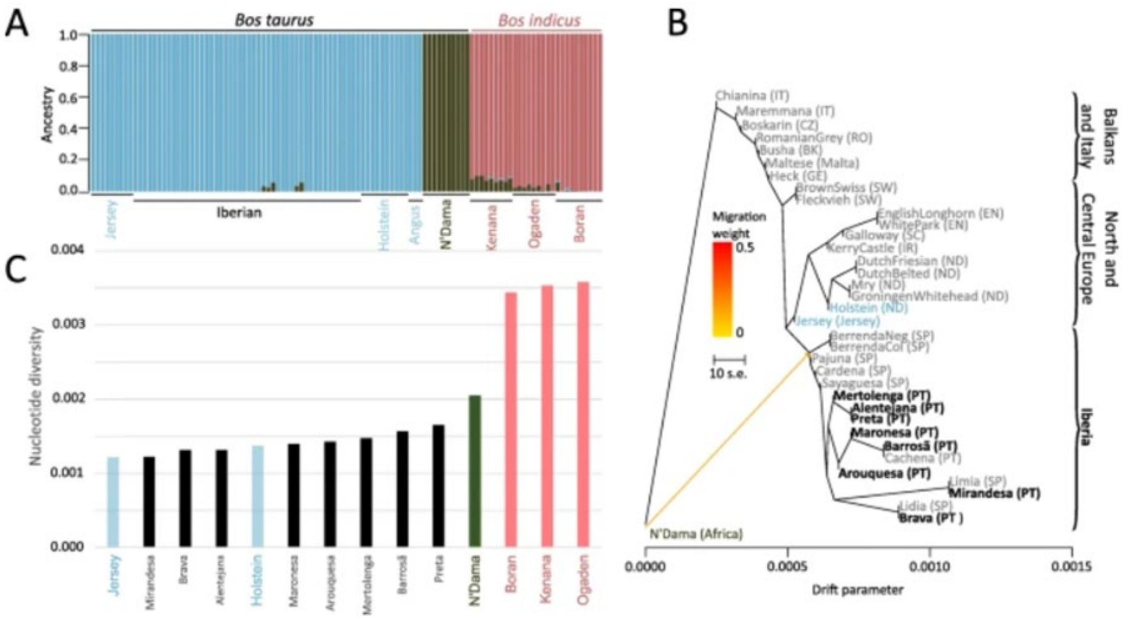
Rute R. da Fonseca, Irene Ureña, Sandra Afonso, Ana Elisabete Pires, Emil Jørsboe, Lounès Chikhi & Catarina Ginja

BMC Genomics **20**, Article number: 334 (2019) | Cite this article

4014 Accesses | **19** Citations | **8** Altmetric | Metrics

Low coverage: 1.4X-2.3X (2x100bp)
48 animals from 8 Iberian cattle breeds
~1 million SNPs



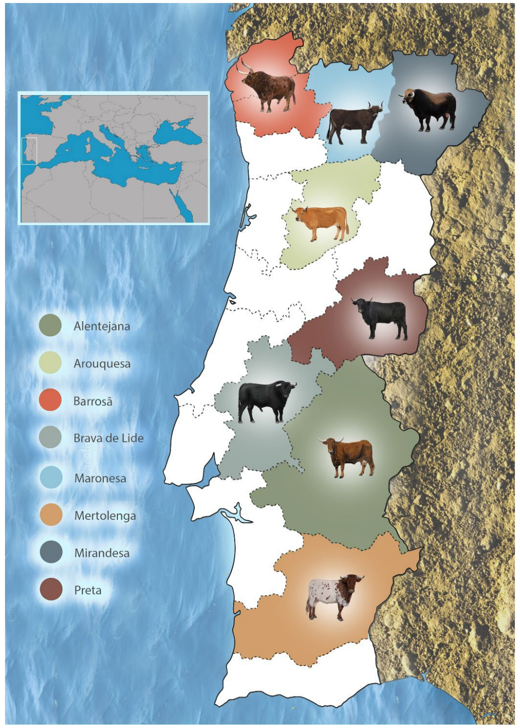


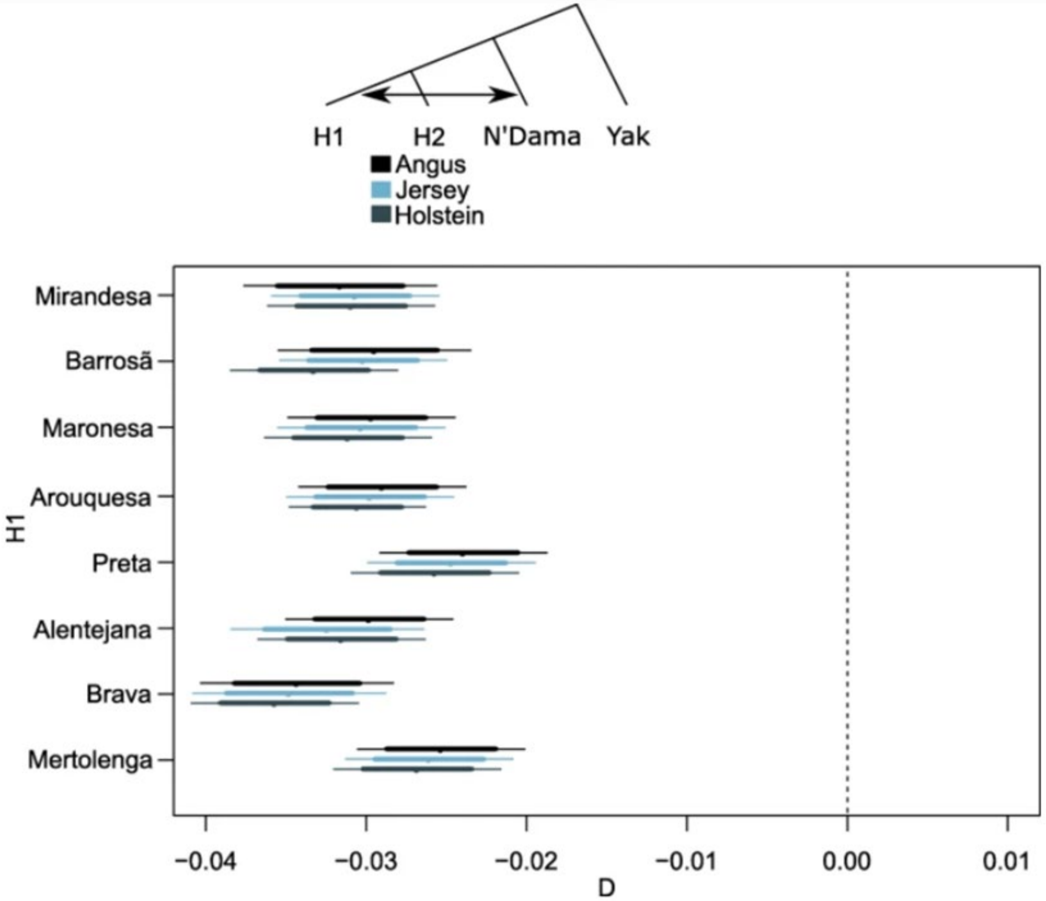
a Population structure using 108 individuals at K = 3 clearly divides the European taurine (blue), African taurine (green) and African indicine (pink) ancestries. **b** Treemix maximum likelihood tree depicting the relationships between taurine cattle breeds (grey: Illumina BovineHD SNP data; black: whole genome data). **c** Nucleotide diversity in taurine and indicine breeds (Iberian breed names in black)

Whole-Genome Sequencing

- High genomic diversity
- Well-structured
- African influence

Whole-Genome Sequencing

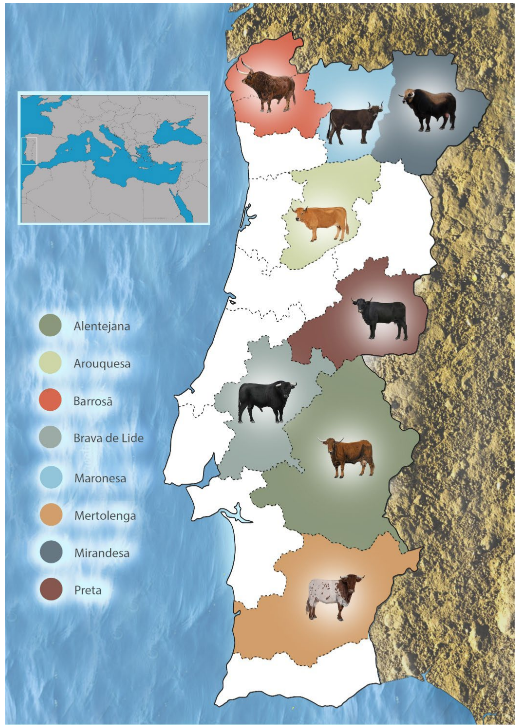


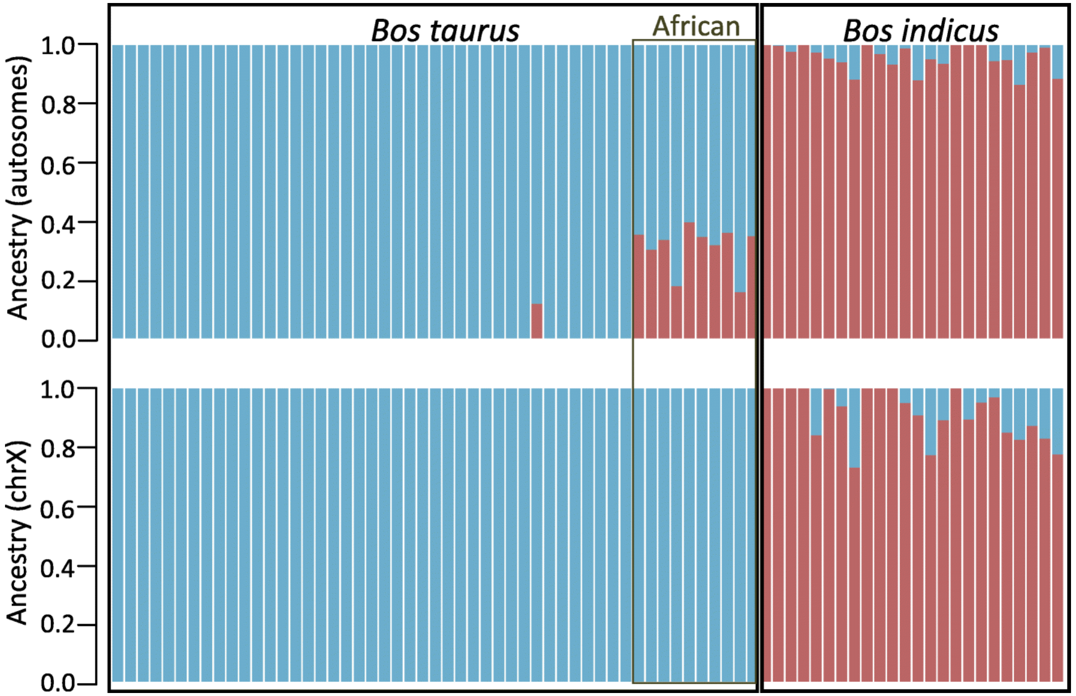


D-statistics determined using genome-wide autosomal data. Negative values indicate an excess of derived alleles shared by the breeds in H1 (denoted in the y-axis) and the African N'Dama breed in comparison with European taurine breeds (H2)

Significant **allele sharing** (gene flow) among Iberian and African taurine cattle.

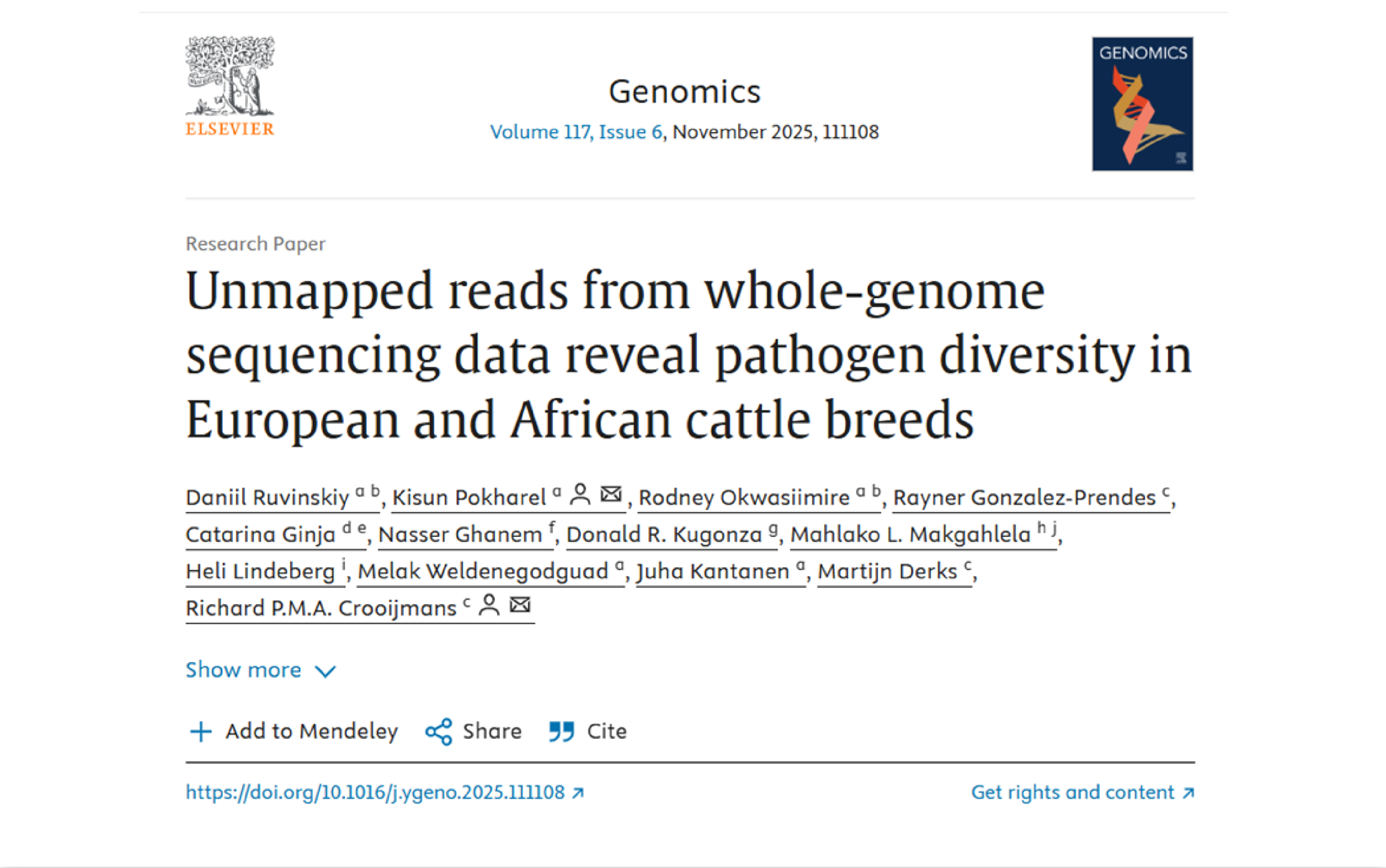
Whole-Genome Sequencing





Population structure at K = 2 determined using the female individuals only. The indicine contribution to African taurine (N'Dama) is not observed in sex chromosome X (bottom) compared to the autosomes (top)

Extensive **male-biased gene flow** within taurine and within indicine cattle.



Genomics
Volume 117, Issue 6, November 2025, 111108

Research Paper

Unmapped reads from whole-genome sequencing data reveal pathogen diversity in European and African cattle breeds

Daniil Ruvinskiy [a b], Kisun Pokharel [a], Rodney Okwasiimire [a b], Rayner Gonzalez-Prendes [c], Catarina Ginja [d e], Nasser Ghanem [f], Donald R. Kugonza [g], Mahlako L. Makgahlela [h j], Heli Lindeberg [i], Melak Weldenegodguad [a], Juha Kantanen [a], Martijn Derks [c], Richard P.M.A. Crooijmans [c]

https://doi.org/10.1016/j.ygeno.2025.111108

Whole-Genome Sequencing

- Unmapped sequence reads' analysis of blood reveals **signatures of disease** occurrence.
- Pathogen prevalence in cattle declines from the Southern to Northern Hemisphere.

Article | Open access | Published: 27 September 2024

Adipose gene expression profiles in Northern Finncattle, Mirandesa cattle, Yakutian cattle and commercial Holstein cattle

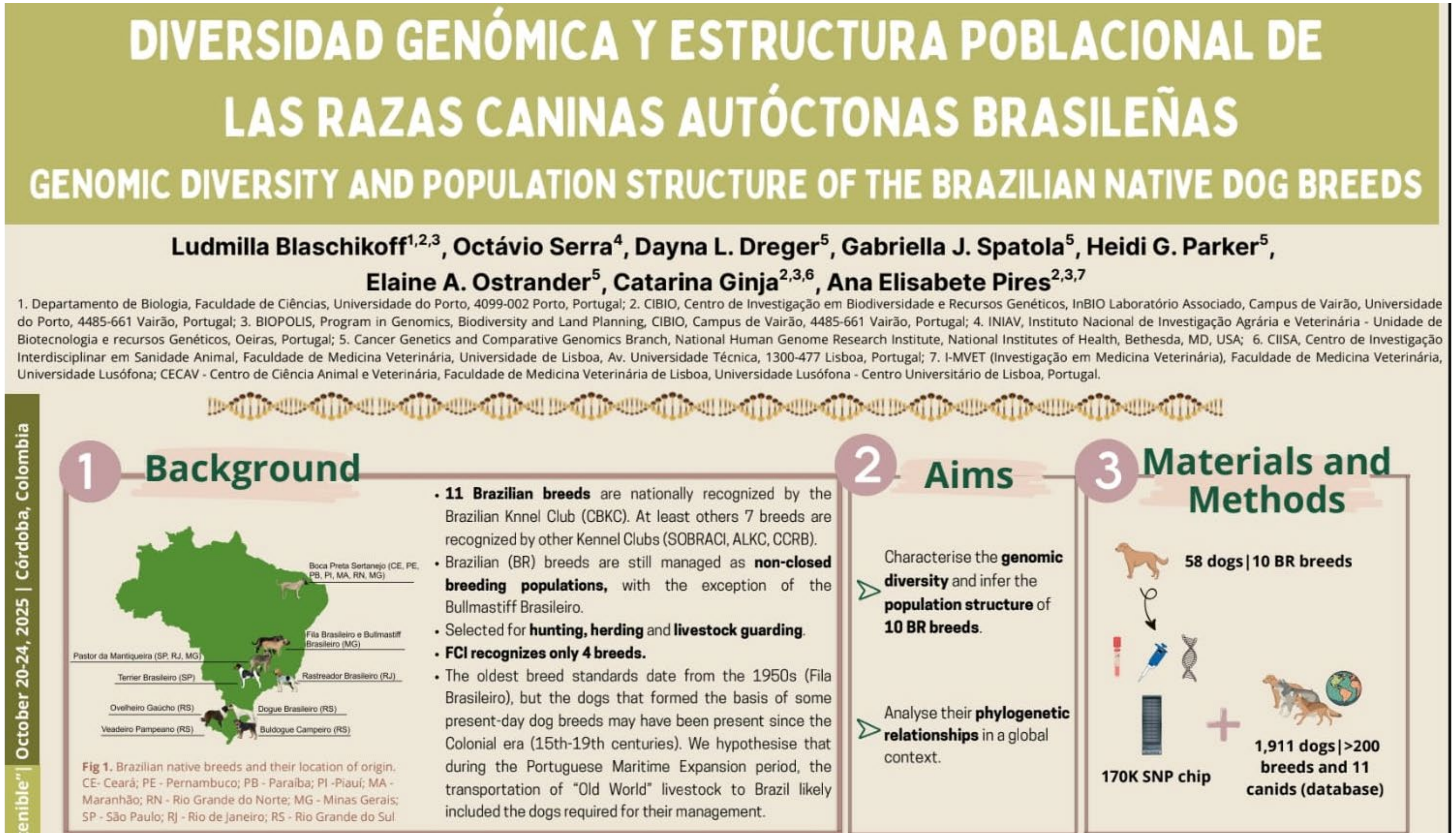
Daniil Ruvinskiy, Andreia Amaral, Melak Weldenegodguad, Innokentyi Ammosov, Mervi Honkatukia, Heli Lindeberg, Jaana Peippo, Ruslan Popov, Päivi Soppela, Florian Stammler, Pekka Uimari, Catarina Ginja, Juha Kantanen & Kisun Pokharel

Scientific Reports **14**, Article number: 22216 (2024) | Cite this article

2310 Accesses | **2** Citations | Metrics

RNA Sequencing

- Genes upregulated in the metacarpal adipose tissue of Yakutian cattle were associated with **energy metabolism** and **response to cold temperatures**.
- In Mirandesa cattle, the upregulated genes in perirenal adipose tissue were related **to immune response** and **inflammation**



DIVERSIDAD GENÓMICA Y ESTRUCTURA POBLACIONAL DE LAS RAZAS CANINAS AUTÓCTONAS BRASILEÑAS

GENOMIC DIVERSITY AND POPULATION STRUCTURE OF THE BRAZILIAN NATIVE DOG BREEDS

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1 Background

Fig 1. Brazilian native breeds and their location of origin. CE- Ceará; PE - Pernambuco; PB - Paraíba; PI -Piauí; MA - Maranhão; RN - Rio Grande do Norte; MG - Minas Gerais; SP - São Paulo; RJ - Rio de Janeiro; RS - Rio Grande do Sul

- **11 Brazilian breeds** are nationally recognized by the Brazilian Knnel Club (CBKC). At least others 7 breeds are recognized by other Kennel Clubs (SOBRACI, ALKC, CCRB).
- Brazilian (BR) breeds are still managed as **non-closed breeding populations,** with the exception of the Bullmastiff Brasileiro.
- Selected for **hunting, herding** and **livestock guarding**.
- **FCI recognizes only 4 breeds.**
- The oldest breed standards date from the 1950s (Fila Brasileiro), but the dogs that formed the basis of some present-day dog breeds may have been present since the Colonial era (15th-19th centuries). We hypothesise that during the Portuguese Maritime Expansion period, the transportation of "Old World" livestock to Brazil likely included the dogs required for their management.

2 Aims

ᐅ Characterise the **genomic diversity** and infer the **population structure** of **10 BR breeds**.

ᐅ Analyse their **phylogenetic relationships** in a global context.

3 Materials and Methods

58 dogs | 10 BR breeds

170K SNP chip

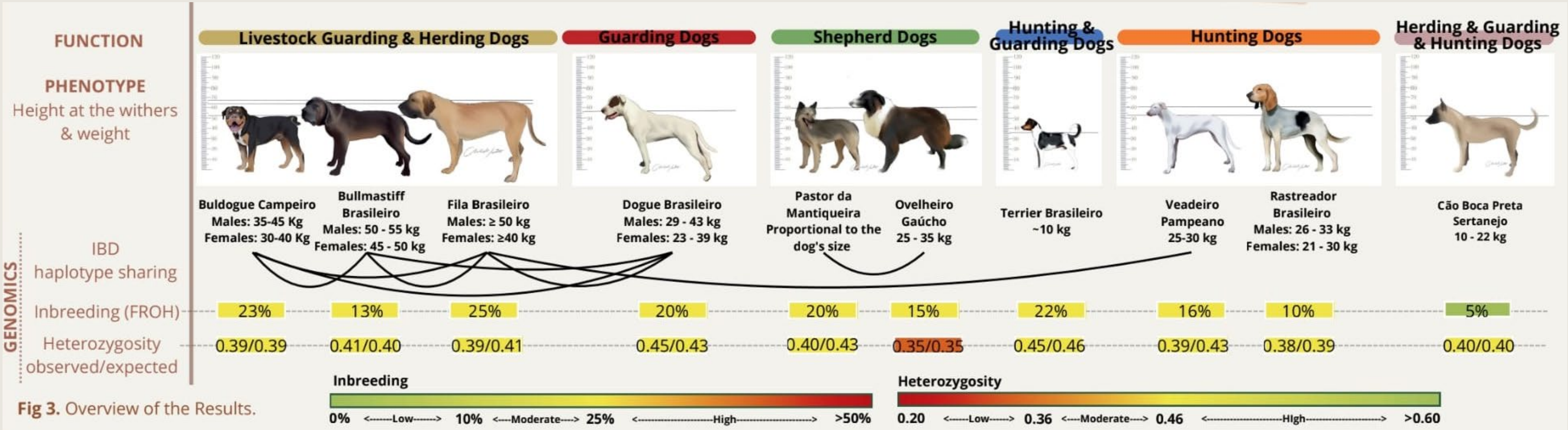
\+

1,911 dogs | >200 breeds and 11 canids (database)

SNP Genotyping

60 samples from 9 local dog breeds and 1 landrace population genotyped for ~135 K SNPs

SNP GENOTYPING



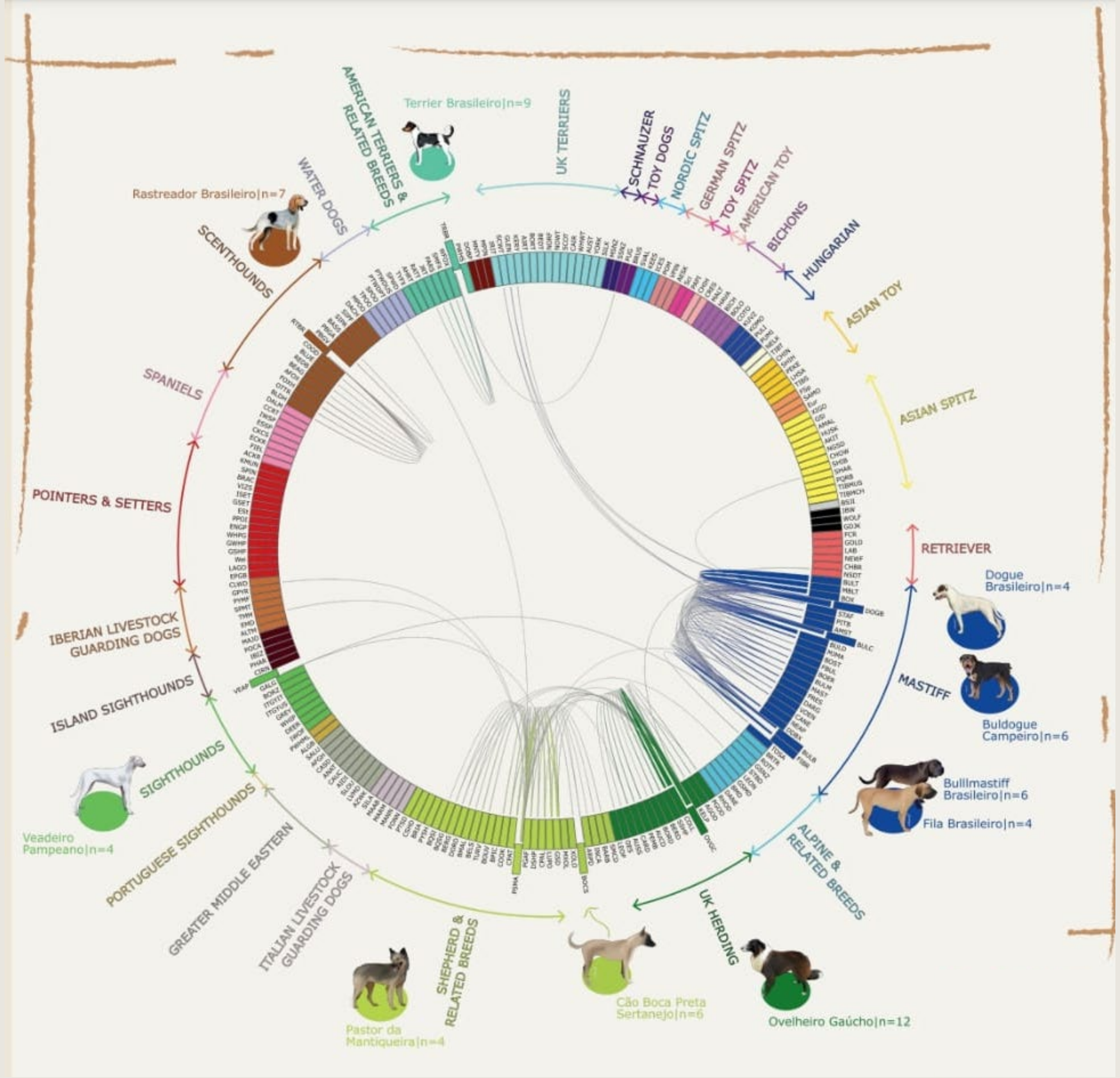
FUNCTION	PHENOTYPE Height at the withers & weight	Inbreeding (FROH)	Heterozygosity observed/expected
Livestock Guarding & Herding Dogs	Buldogue Campeiro Males: 35-45 Kg Females: 30-40 Kg	23%	0.39/0.39
	Bullmastiff Brasileiro Males: 50 - 55 kg Females: 45 - 50 kg	13%	0.41/0.40
	Fila Brasileiro Males: ≥ 50 kg Females: ≥40 kg	25%	0.39/0.41
Guarding Dogs	Dogue Brasileiro Males: 29 - 43 kg Females: 23 - 39 kg	20%	0.45/0.43
Shepherd Dogs	Pastor da Mantiqueira Proportional to the dog's size	20%	0.40/0.43
	Ovelheiro Gaúcho 25 - 35 kg	15%	0.35/0.35
Hunting & Guarding Dogs	Terrier Brasileiro ~10 kg	22%	0.45/0.46
Hunting Dogs	Veadeiro Pampeano 25-30 kg	16%	0.39/0.43
	Rastreador Brasileiro Males: 26 - 33 kg Females: 21 - 30 kg	10%	0.38/0.39
Herding & Guarding & Hunting Dogs	Cão Boca Preta Sertanejo 10 - 22 kg	5%	0.40/0.40

Fig 3. Overview of the Results.

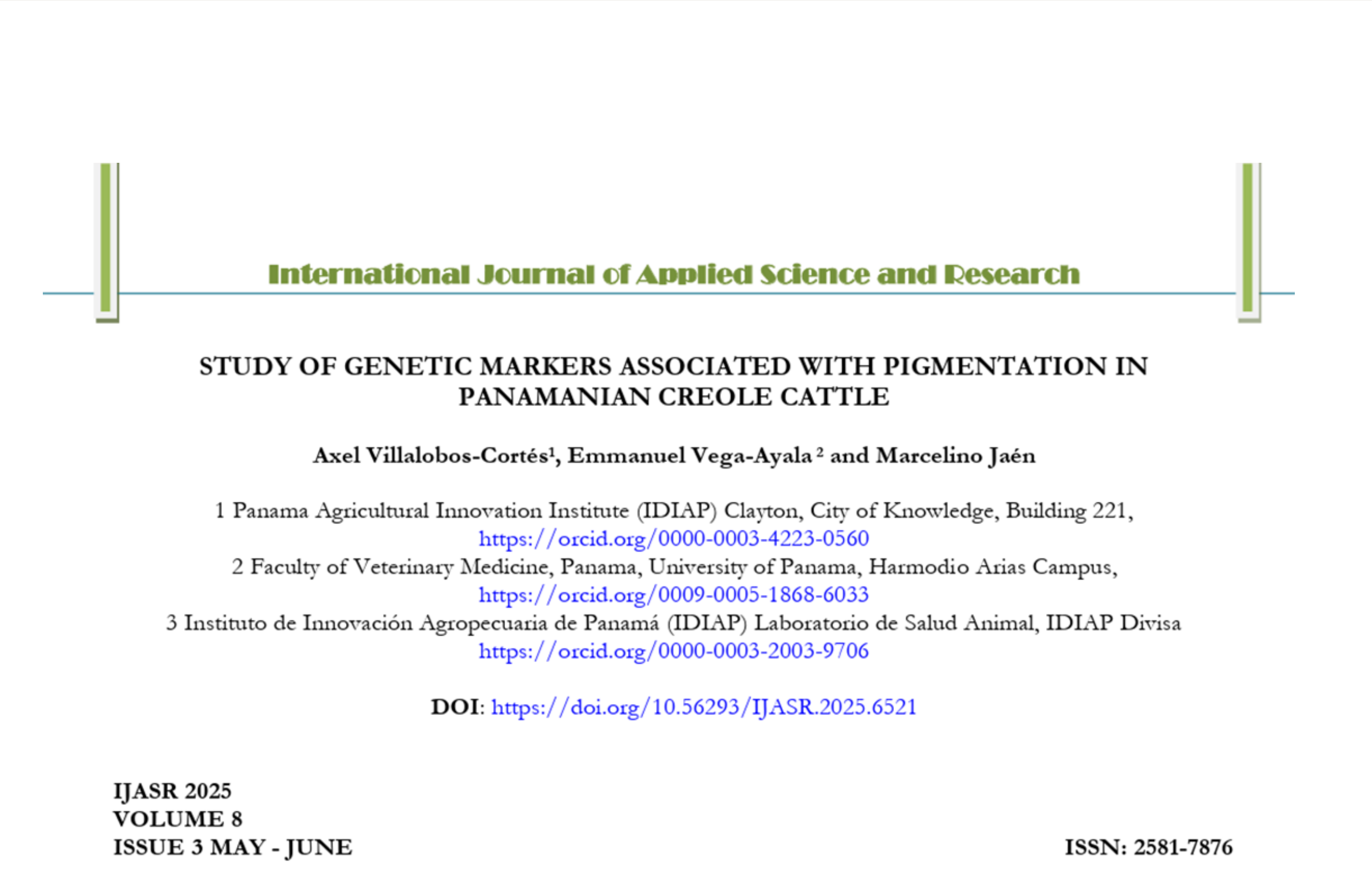
Population genomics – a 1st glimpse of the diversity, levels of inbreeding, and population structure of local dog breeds from Brazil

SNP GENOTYPING

Provide a context for the local breeds from Brazil within the phylogeny of worldwide dogs



SNP Genotyping



International Journal of Applied Science and Research

STUDY OF GENETIC MARKERS ASSOCIATED WITH PIGMENTATION IN PANAMANIAN CREOLE CATTLE

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34 cattle samples belonging to Guaymi and Guabala breeds genotyped with the Affymetrix 10 K SNP array.

ACTIVITIES & IMPACT

- Genomic characterization of the Guaymi and Guabala breeds

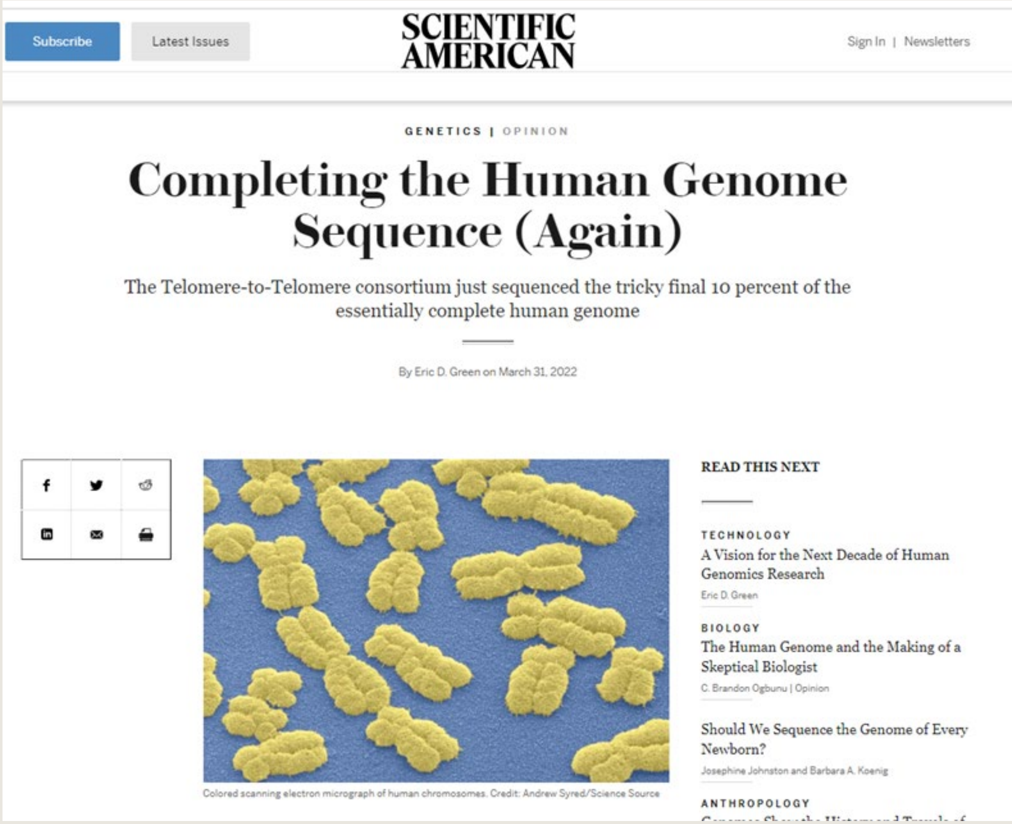
INSTITUTO DE INNOVACIÓN AGROPECUARIA DE PANAMÁ

Asociación de Criadores de Criollos de Panamá

ACTIVITIES & IMPACT

The 2030 Agenda for Sustainable Development

Human genome – world’s largest collaborative biological project!



SCIENTIFIC AMERICAN

GENETICS | OPINION

Completing the Human Genome Sequence (Again)

The Telomere-to-Telomere consortium just sequenced the tricky final 10 percent of the essentially complete human genome

By Eric D. Green on March 31, 2022

Colored scanning electron micrograph of human chromosomes. Credit: Andrew Syred/Science Source

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ANTHROPOLOGY

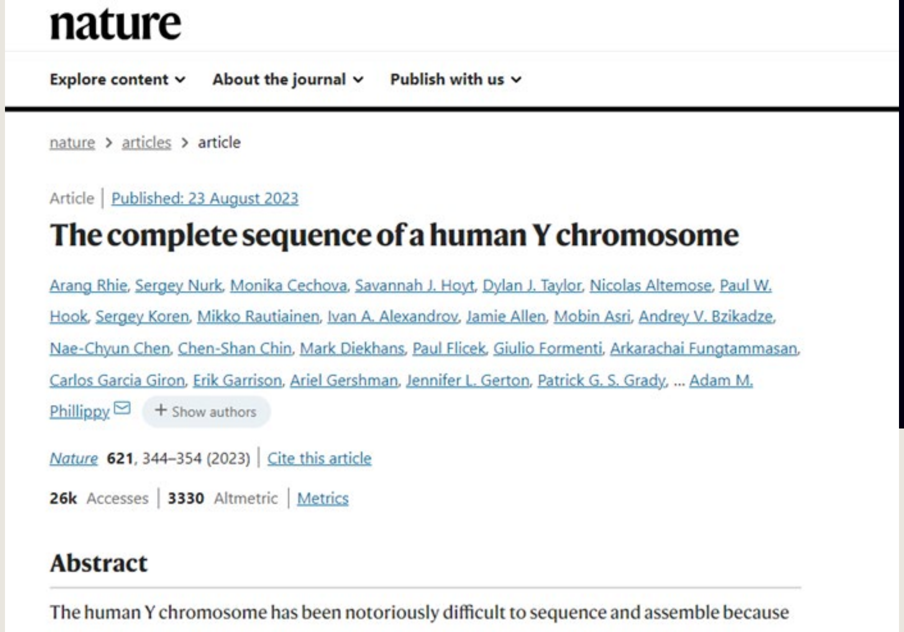
~35,000 genes

2% of the DNA corresponds to coding genes
98% is “garbage”

On March 31, 2022, the Telomere-to-Telomere (T2T) consortium announced that had filled in the remaining gaps and produced the first truly complete human genome sequence.

https://www.genome.gov/about-genomics/educational-resources/fact-sheets/human-genome-project

Human genome – chromosome Y



Article | Published: 23 August 2023

The complete sequence of a human Y chromosome

Arang Rhie, Sergey Nurk, Monika Cechova, Savannah J. Hoyt, Dylan J. Taylor, Nicolas Altemose, Paul W. Hook, Sergey Koren, Mikko Rautiainen, Ivan A. Alexandrov, Jamie Allen, Mobin Asri, Andrey V. Bzikadze, Nae-Chyun Chen, Chen-Shan Chin, Mark Diekhans, Paul Flicek, Giulio Formenti, Arkarachai Fungtammasan, Carlos Garcia Giron, Erik Garrison, Ariel Gershman, Jennifer L. Gerton, Patrick G. S. Grady, ... Adam M. Phillippy

Nature **621**, 344–354 (2023) | Cite this article

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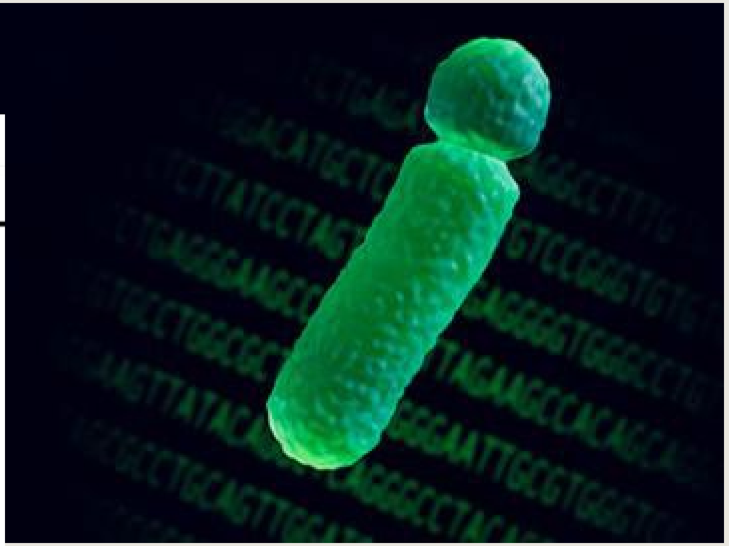
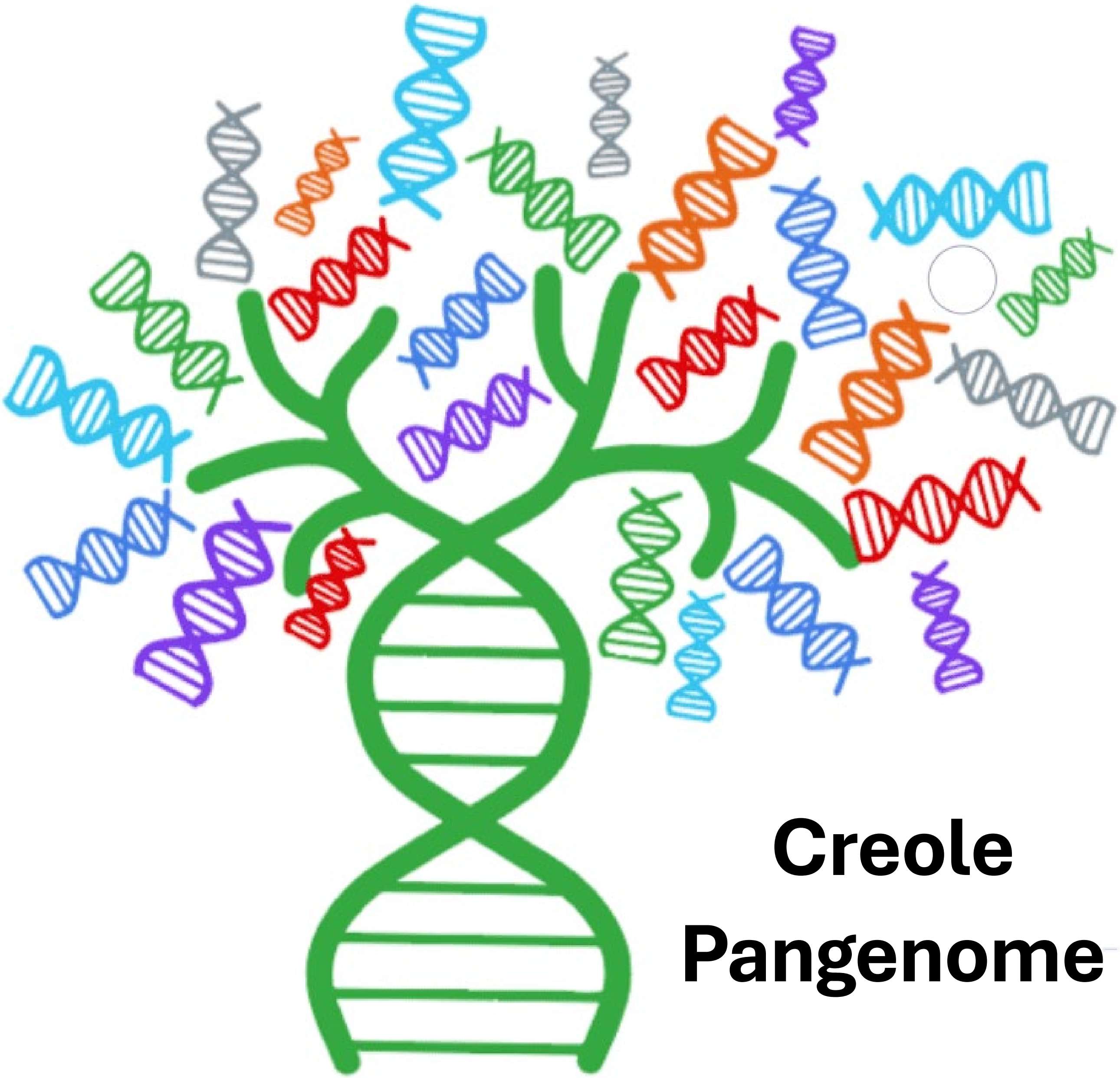
Abstract

The human Y chromosome has been notoriously difficult to sequence and assemble because



Released in August 2023, the Y chromosome is unusually repetitive. Assembling sequencing data is like trying to read a long book cut into strips. About 30 million letters of the Y chromosome are repetitive sequences.



Creole Pangenome

Image credits: humanpangenome.org

A collection of genome sequences from Creole animals of diverse well-adapted populations to **capture the breadth of genomic variation across domestic animal species from Latin America**. Use these data as “reference” to **investigate selection, demographic processes and differentiation**.

The identification of the genetic basis of adaptive characteristics lacks information at the genome and transcriptome levels.

Funding

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