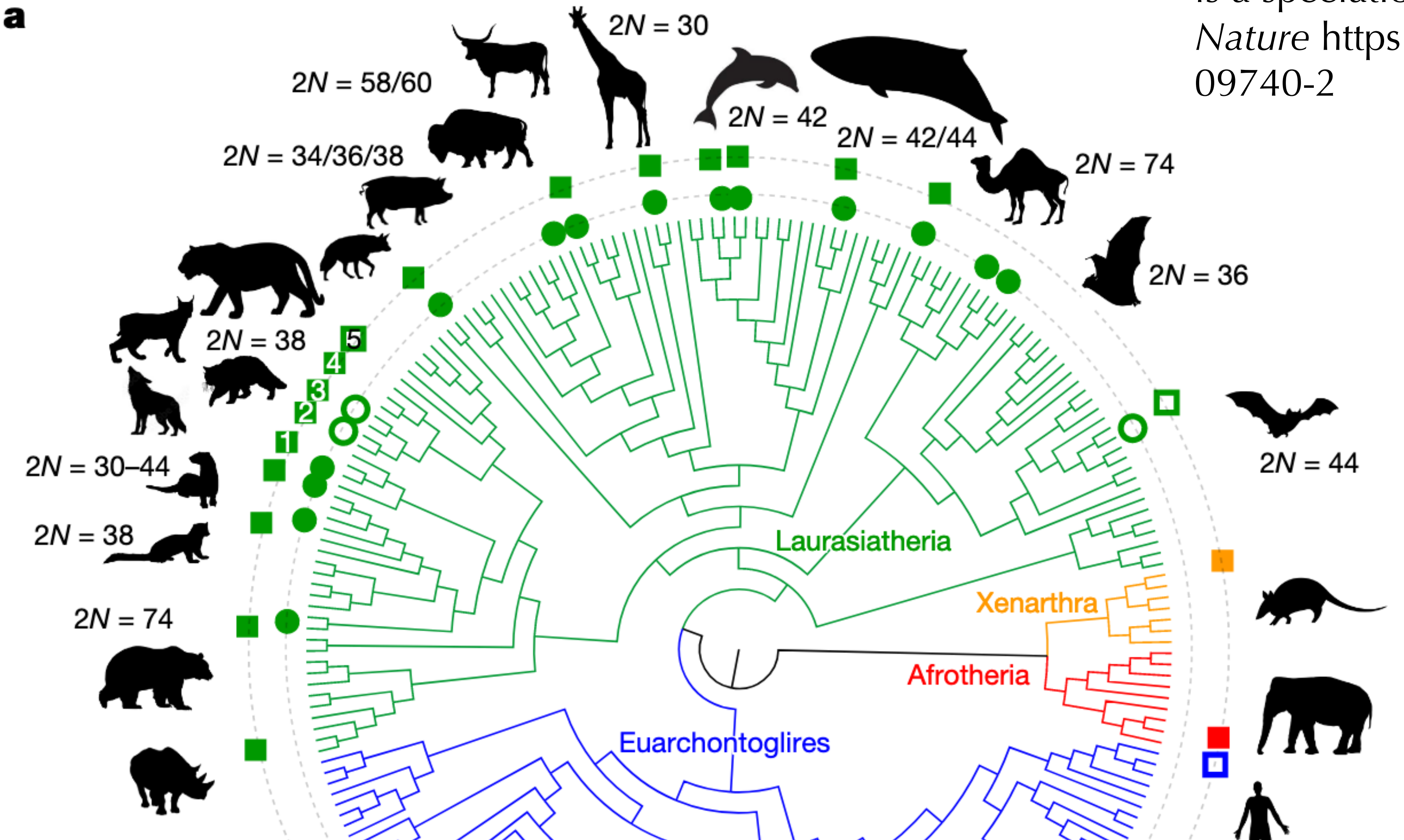
Nicole M. Foley, Richard G. Rasulis, Zoya Wani, Mayra N. Mendoza Cerna, Henrique V. Figueiró, Klaus Peter Koepfli, Terje Raudsepp & William J. Murphy. 2025. “An ancient recombination desert is a speciation supergene in placental mammals” *Nature* https://doi.org/10.1038/s41586-025-09740-2



# Class focus area: Genomics and a speciation desert

EEB603: Brian O’Meara

All quotes and images from the above paper unless otherwise noted

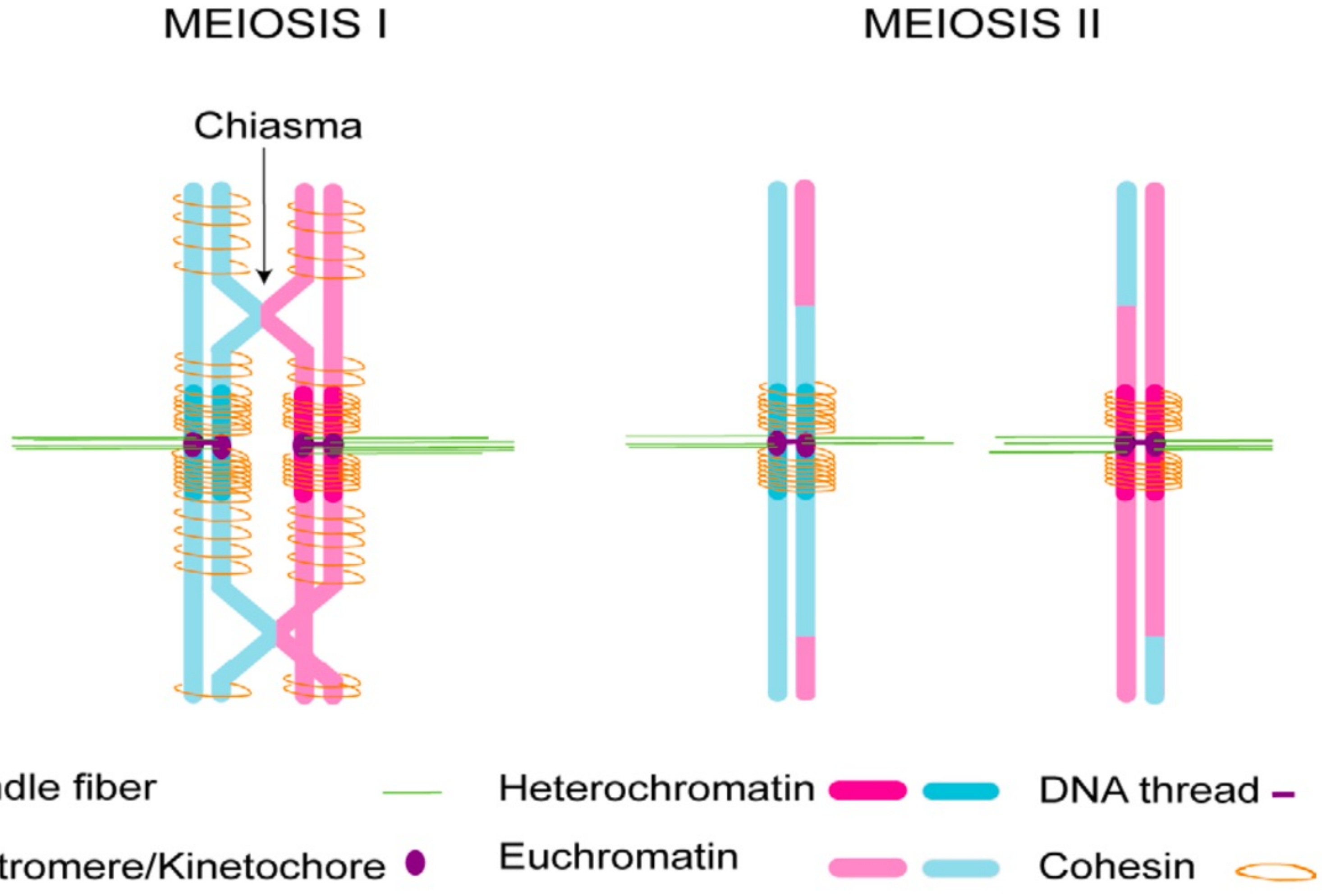


**Figure 1. Chromosome connections in meiosis.** Kinetochores attach homologous chromosomes to opposite halves of the spindle. Homologs are held together by chiasmata, in which recombinant chromatids cross each other. Sisters are held together by cohesins and possibly by catenation of centromeric DNA threads, which have been observed in human mitosis. Cohesion is released in two steps: on chromosome arms to resolve chiasmata and separate homologs in the first meiotic division; and around centromeres to separate sisters in the second meiotic division. doi:10.1371/journal.pbio.1000326.g001

Talbert & Henikoff (2010)

Randall Munroe,
https://xkcd.com/1838/

Also see: https://www.explainxkcd.com/wiki/index.php/1838:_Machine_Learning

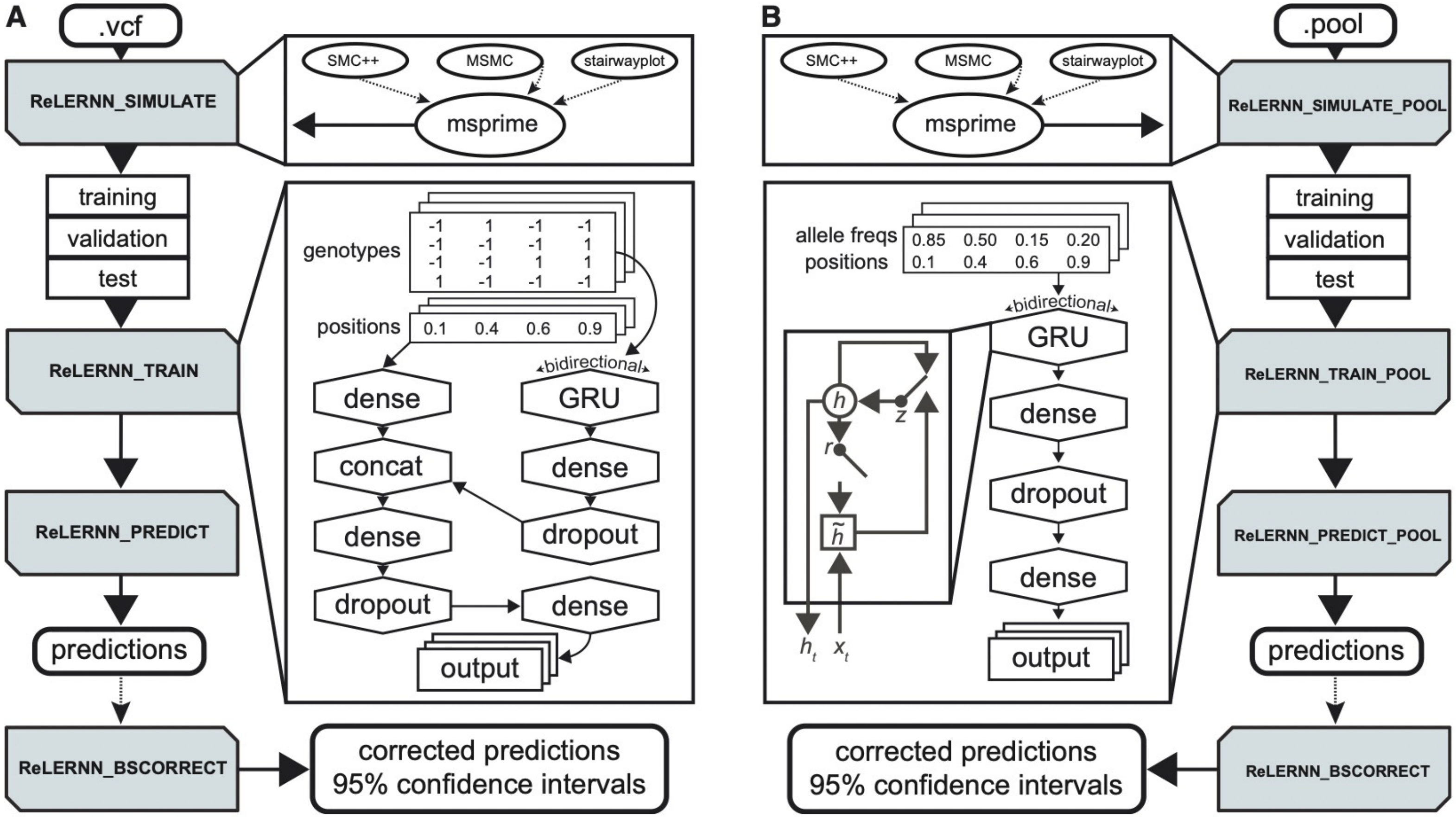
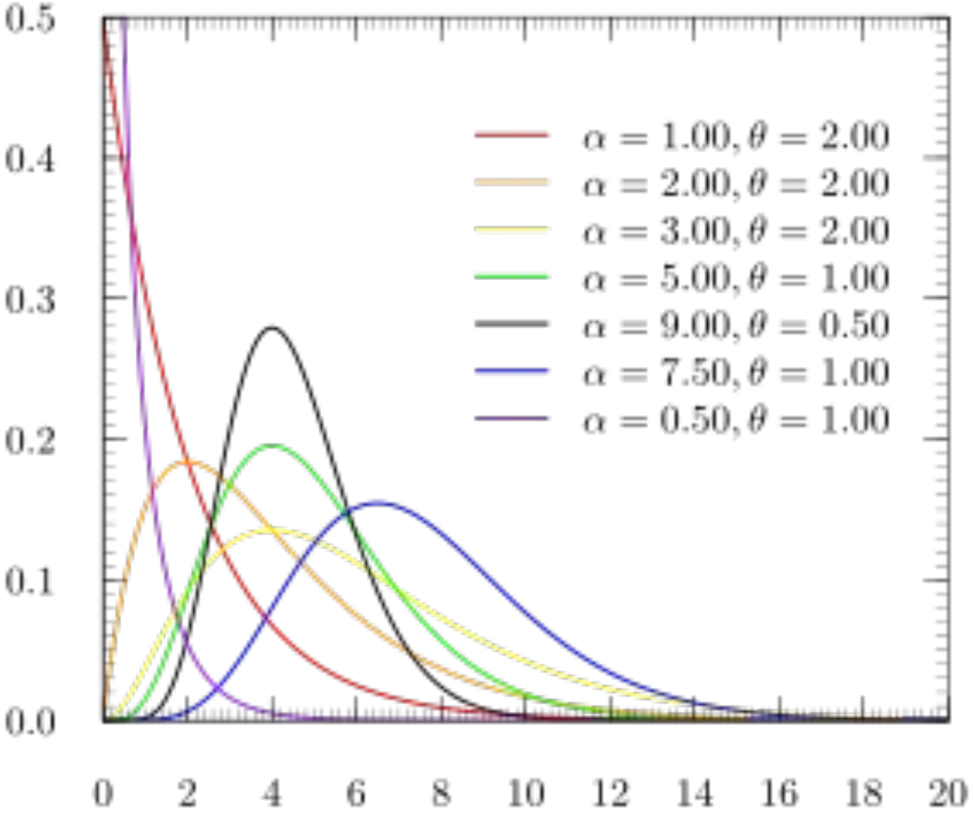


**FIG. 1.** A cartoon depicting a typical workflow using ReLERNN's four modules (shaded boxes) for (*A*) individually sequenced genomes or (*B*) pooled sequences. ReLERNN can optionally (dotted lines) utilize output from stairwayplot, SMC++, and MSMC to simulate under a demographic history with msprime. Training inlays show the network architectures used, with the GRU inlay in (*B*) depicting the gated connections within each hidden unit. Here, $r$, $z$, $h_t$, and $\tilde{h}_t$ are the reset gate, update gate, activation, and candidate activation, respectively (Cho et al. 2014). The genotype matrix encodes alleles as reference (−1), alternative (1), or padded/missing data (0; not shown). Variant positions are encoded along the real number line (0–1).

Adrion, Galloway, and Kern (2020) https://doi.org/10.1093/molbev/msaa038



Wikipedia gamma distribution

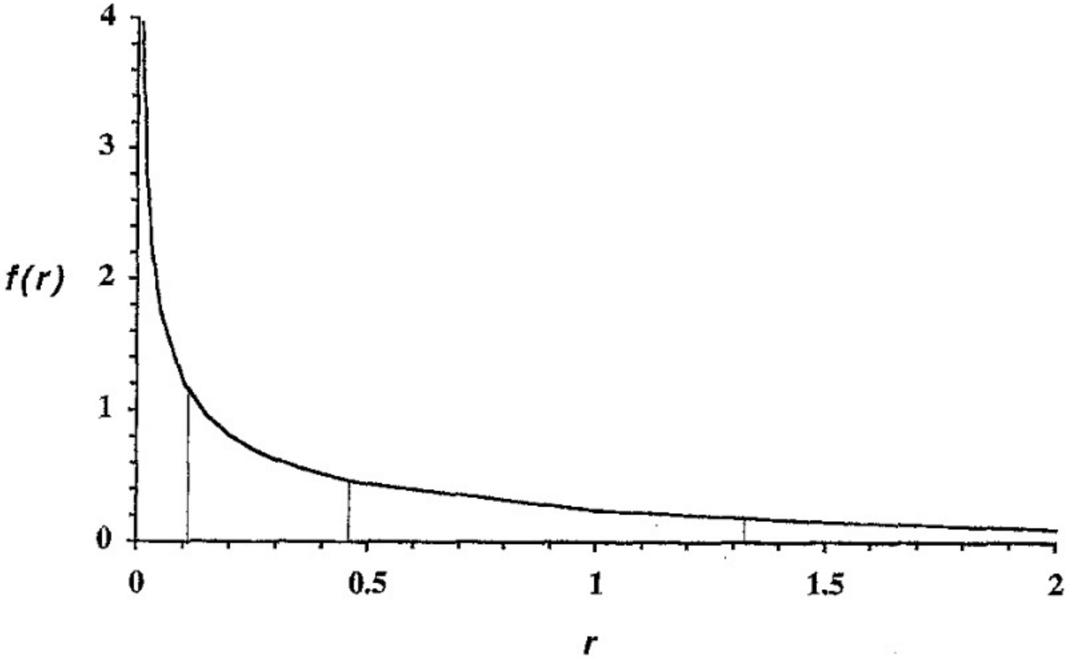
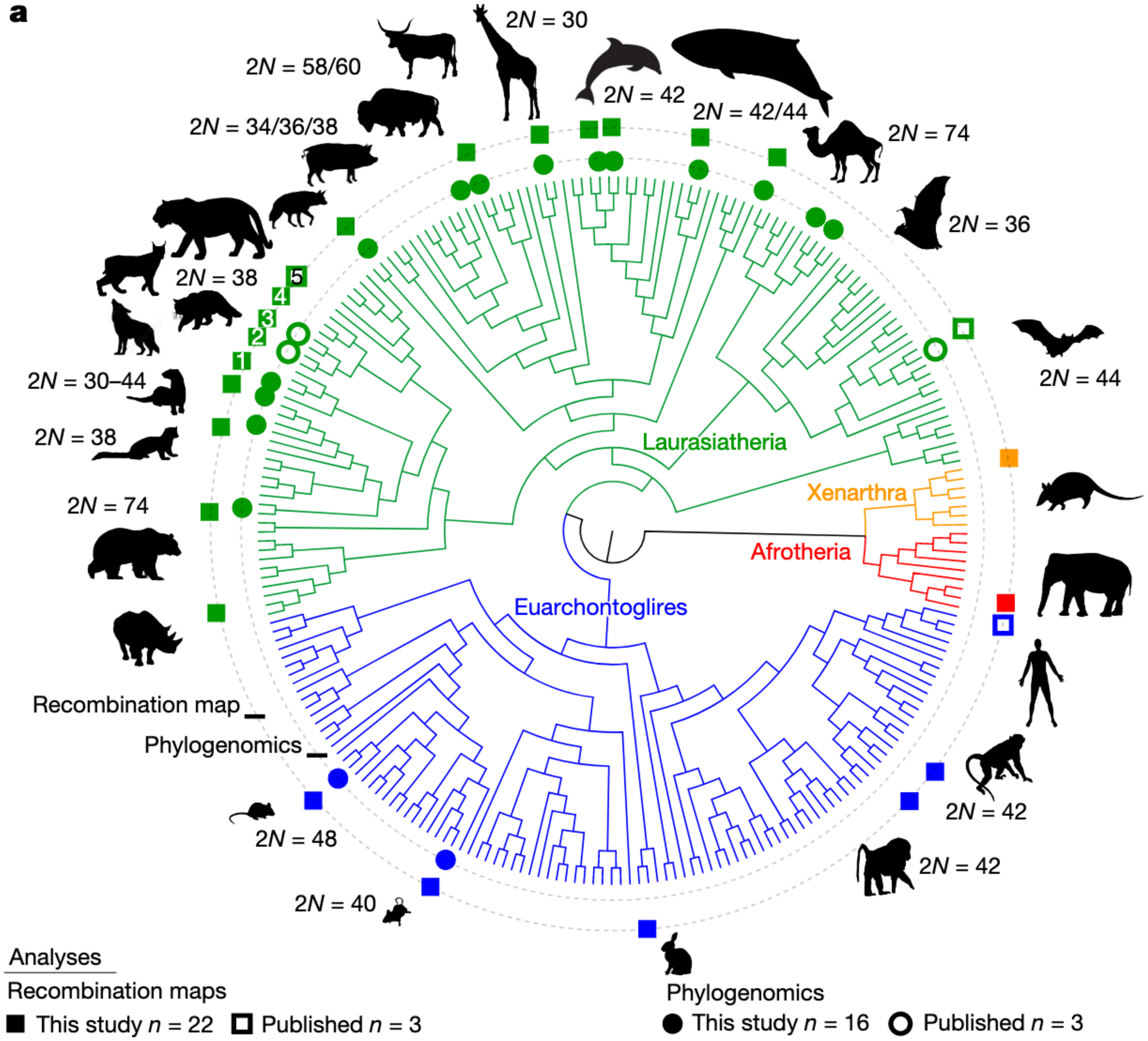
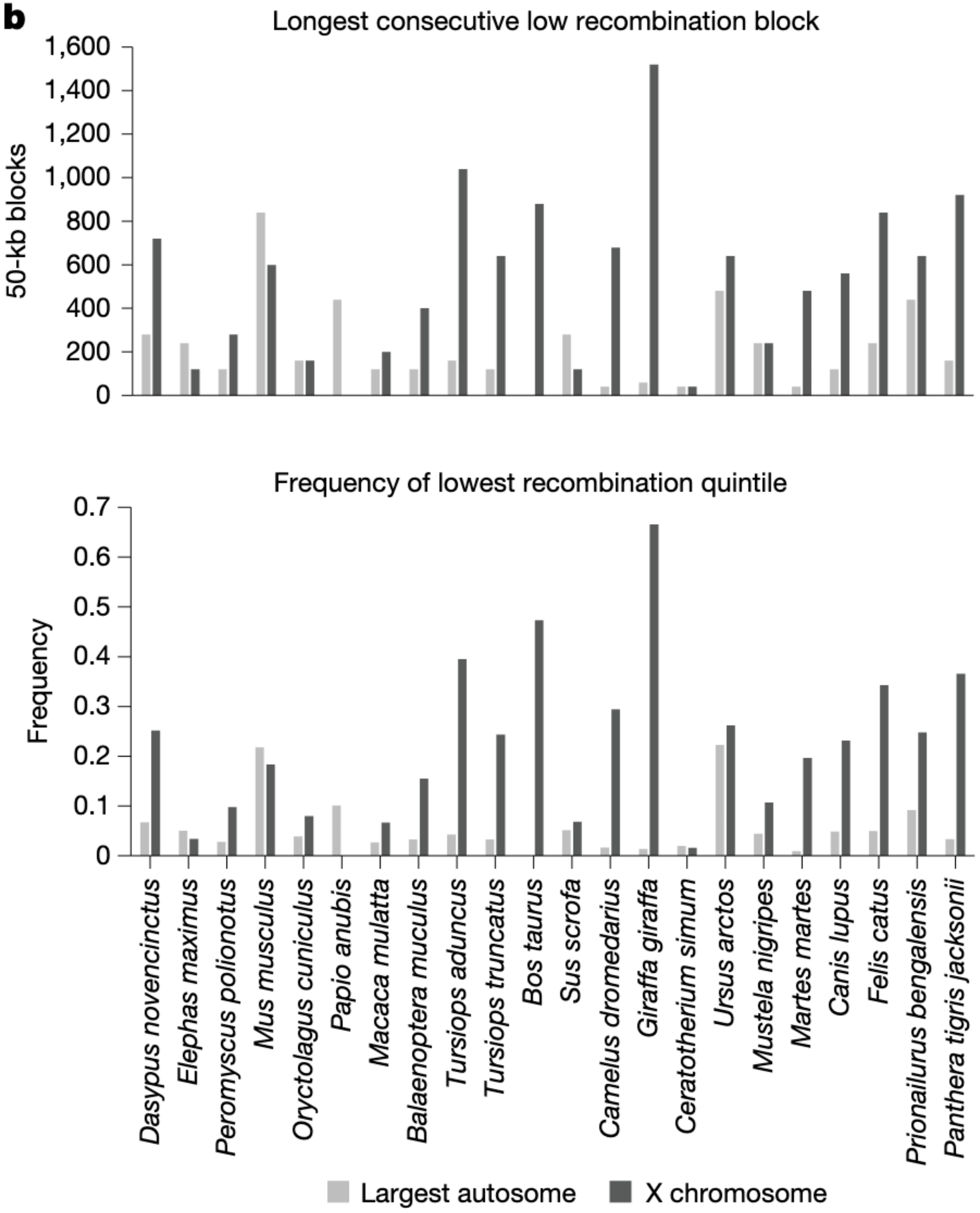
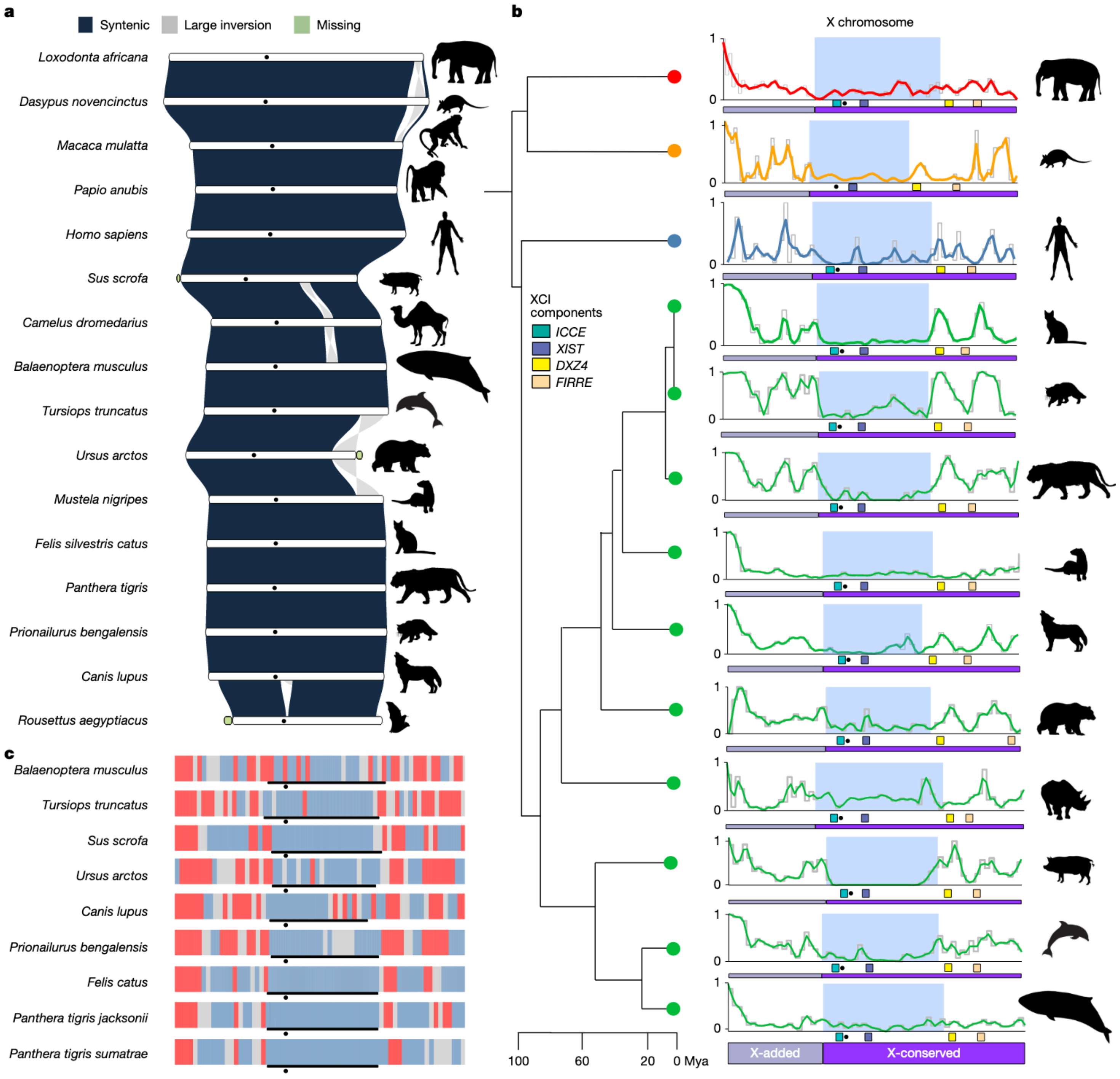


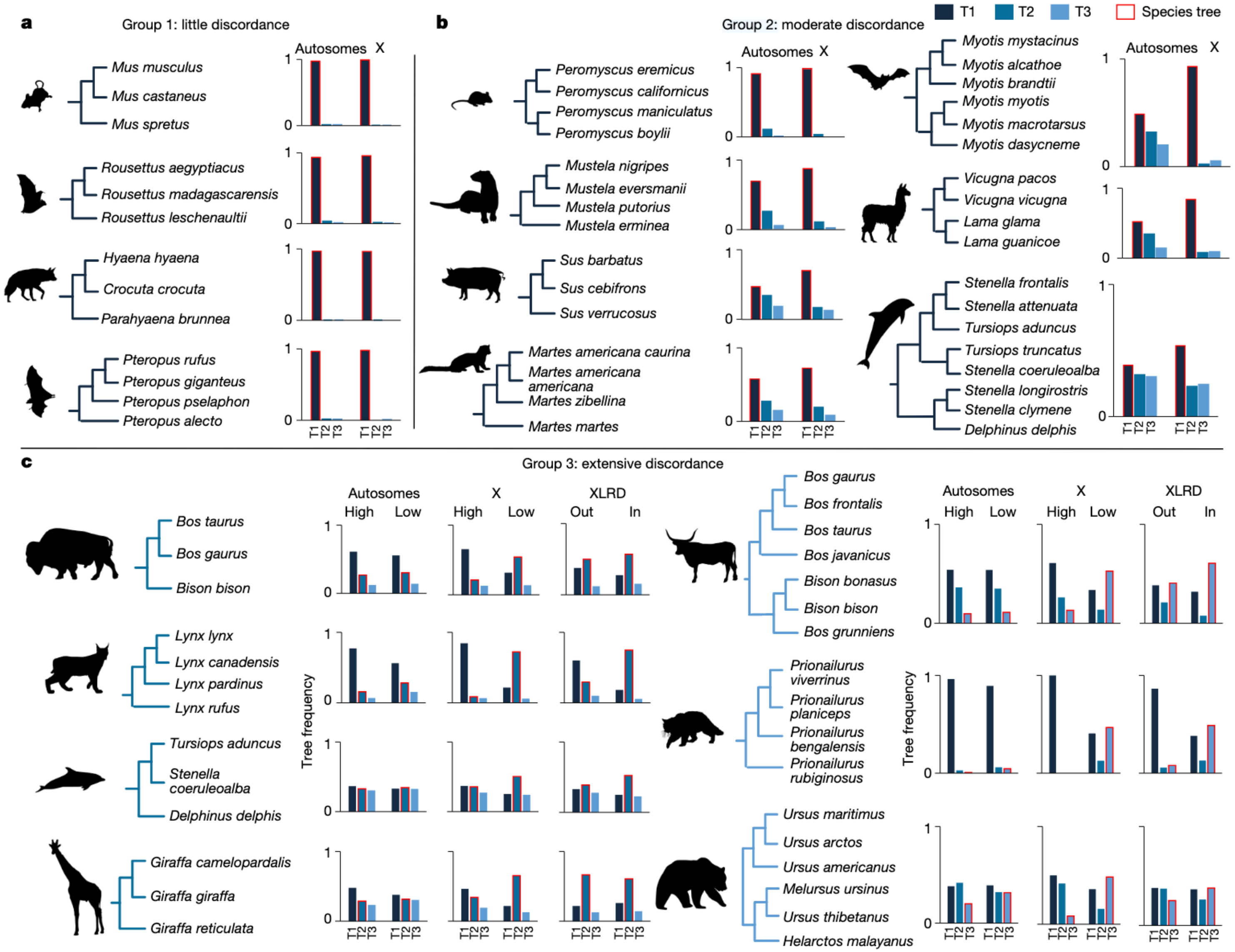
**Fig. 1.** Discrete approximation to the gamma distribution $G(\alpha,\beta)$, with $\alpha = \beta = {}^1/_2$. Four categories are used to approximate the continuous distribution, with equal probability for each category. The three boundaries are 0.1015, 0.4549, and 1.3233, which are the percentage points corresponding to $p = {}^1/_4, {}^2/_4, {}^3/_4$. The means of the four categories are 0.0334, 0.2519, 0.8203, 2.8944. The medians are 0.0247, 0.2389, 0.7870, 2.3535, and these are scaled to get 0.0291, 0.2807, 0.9248, and 2.7654, so that the mean of the discrete distribution is one.

Yang 1994 “Maximum Likelihood Phylogenetic Estimation from DNA Sequences with Variable Rates over Sites: Approximate Methods” J Molecular Evolution 39:306-314

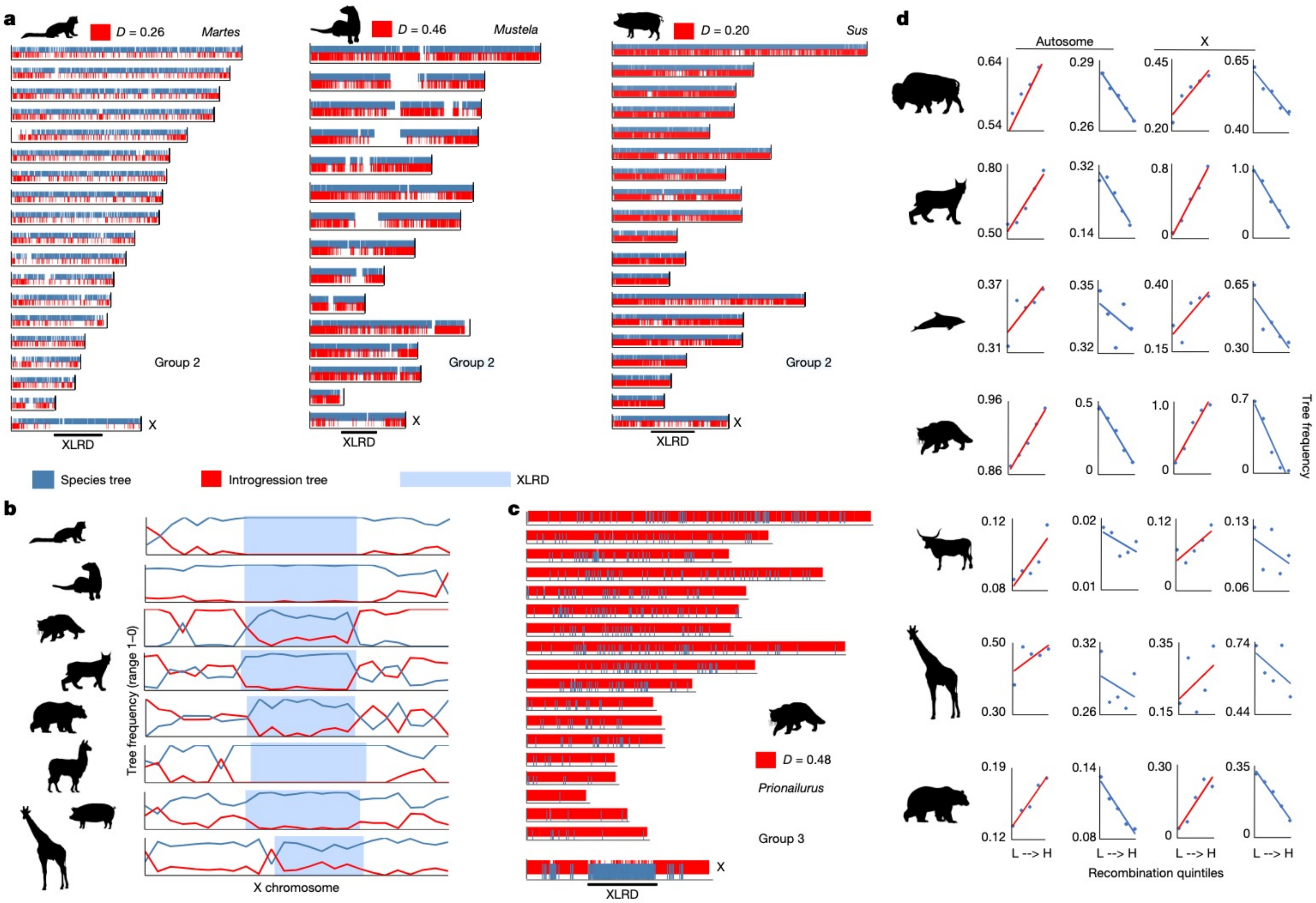

a
2N = 58/60
2N = 30
2N = 42
2N = 42/44
2N = 74
2N = 34/36/38
2N = 36
2N = 38
5
4
3
2
1
2N = 44
2N = 30–44
2N = 38
Laurasiatheria
Xenarthra
2N = 74
Afrotheria
Euarchontoglires
Recombination map
Phylogenomics
2N = 42
2N = 48
2N = 42
2N = 40
Analyses
Recombination maps
■ This study n = 22 □ Published n = 3
Phylogenomics
● This study n = 16 ○ Published n = 3

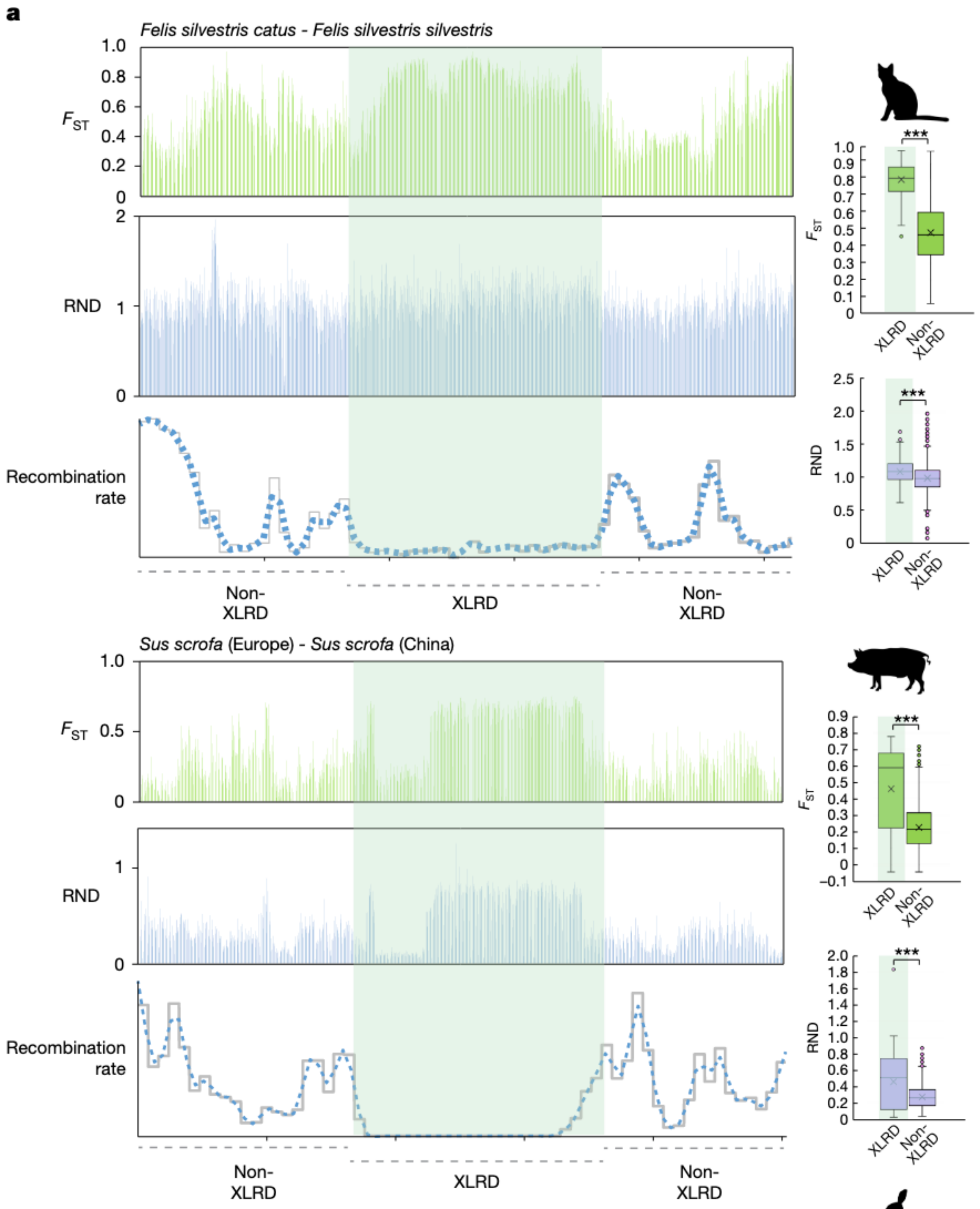

b
Longest consecutive low recombination block
50-kb blocks
1,600
1,400
1,200
1,000
800
600
400
200
0
Frequency of lowest recombination quintile
Frequency
0.7
0.6
0.5
0.4
0.3
0.2
0.1
0
Dasypus novencinctus
Elephas maximus
Peromyscus polionotus
Mus musculus
Oryctolagus cuniculus
Papio anubis
Macaca mulatta
Balaenoptera muculus
Tursiops aduncus
Tursiops truncatus
Bos taurus
Sus scrofa
Camelus dromedarius
Giraffa giraffa
Ceratotherium simum
Ursus arctos
Mustela nigripes
Martes martes
Canis lupus
Felis catus
Prionailurus bengalensis
Panthera tigris jacksonii
Largest autosome
X chromosome

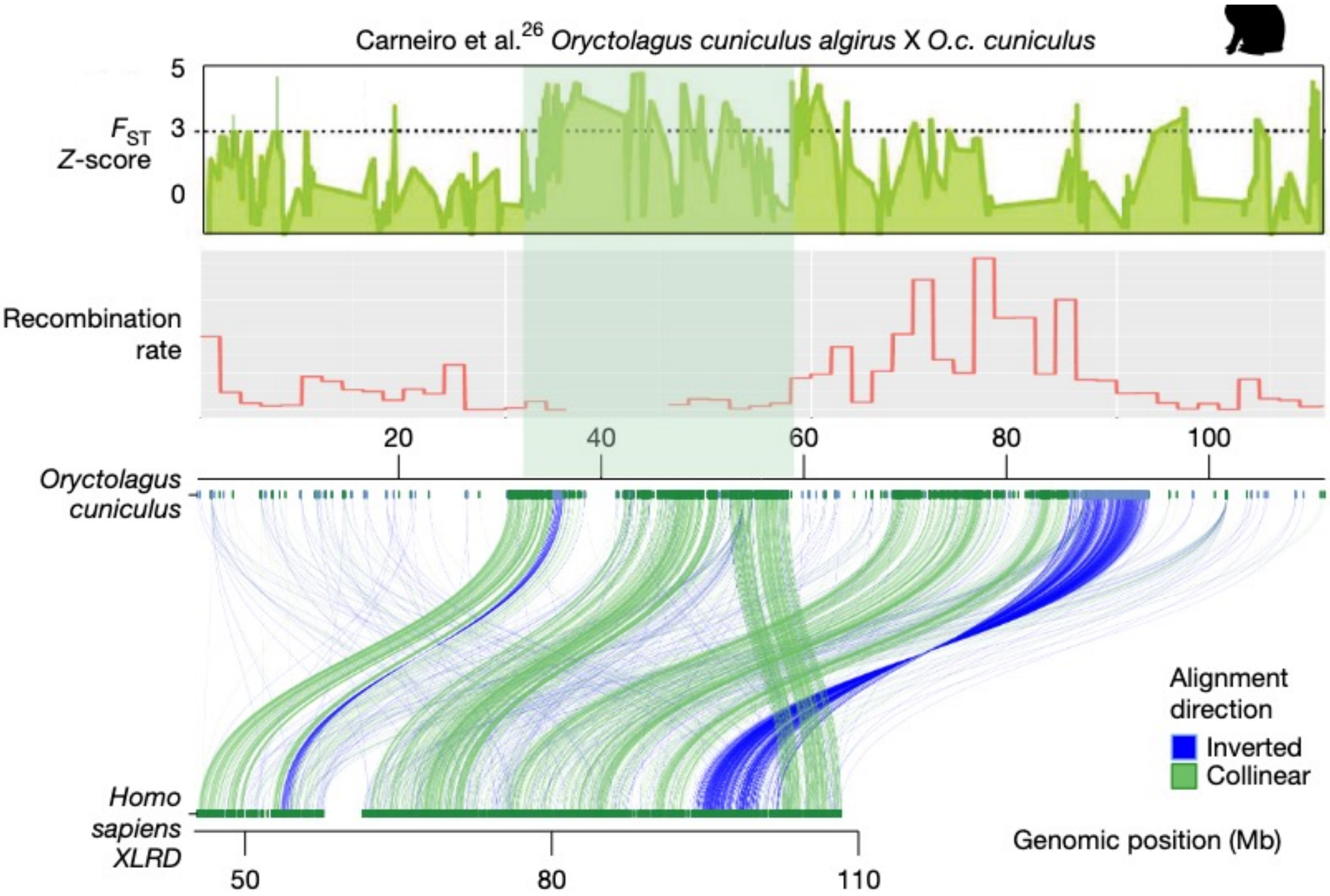

a
Syntenic
Large inversion
Missing
Loxodonta africana
Dasypus novencinctus
Macaca mulatta
Papio anubis
Homo sapiens
Sus scrofa
Camelus dromedarius
Balaenoptera musculus
Tursiops truncatus
Ursus arctos
Mustela nigripes
Felis silvestris catus
Panthera tigris
Prionailurus bengalensis
Canis lupus
Rousettus aegyptiacus
b
X chromosome
1
0
XCI components
ICCE
XIST
DXZ4
FIRRE
100
60
20
0 Mya
X-added
X-conserved
c
Balaenoptera musculus
Tursiops truncatus
Sus scrofa
Ursus arctos
Canis lupus
Prionailurus bengalensis
Felis catus
Panthera tigris jacksonii
Panthera tigris sumatrae



T1
T2
T3
Species tree
a
Group 1: little discordance
Autosomes X
Mus musculus
Mus castaneus
Mus spretus
Rousettus aegyptiacus
Rousettus madagascarensis
Rousettus leschenaultii
Hyaena hyaena
Crocuta crocuta
Parahyaena brunnea
Pteropus rufus
Pteropus giganteus
Pteropus pselaphon
Pteropus alecto
b
Group 2: moderate discordance
Peromyscus eremicus
Peromyscus californicus
Peromyscus maniculatus
Peromyscus boylii
Mustela nigripes
Mustela eversmanii
Mustela putorius
Mustela erminea
Sus barbatus
Sus cebifrons
Sus verrucosus
Martes americana caurina
Martes americana americana
Martes zibellina
Martes martes
Myotis mystacinus
Myotis alcathoe
Myotis brandtii
Myotis myotis
Myotis macrotarsus
Myotis dasycneme
Vicugna pacos
Vicugna vicugna
Lama glama
Lama guanicoe
Stenella frontalis
Stenella attenuata
Tursiops aduncus
Tursiops truncatus
Stenella coeruleoalba
Stenella longirostris
Stenella clymene
Delphinus delphis
c
Group 3: extensive discordance
Autosomes
High Low
X
High Low
XLRD
Out In
Bos taurus
Bos gaurus
Bison bison
Lynx lynx
Lynx canadensis
Lynx pardinus
Lynx rufus
Tursiops aduncus
Stenella coeruleoalba
Delphinus delphis
Giraffa camelopardalis
Giraffa giraffa
Giraffa reticulata
Tree frequency
Bos gaurus
Bos frontalis
Bos taurus
Bos javanicus
Bison bonasus
Bison bison
Bos grunniens
Prionailurus viverrinus
Prionailurus planiceps
Prionailurus bengalensis
Prionailurus rubiginosus
Ursus maritimus
Ursus arctos
Ursus americanus
Melursus ursinus
Ursus thibetanus
Helarctos malayanus
0
1
T1 T2 T3


a
D = 0.26
Martes
D = 0.46
Mustela
D = 0.20
Sus
Group 2
Group 2
Group 2
X
X
X
XLRD
XLRD
XLRD
Species tree
Introgression tree
XLRD
b
Tree frequency (range 1–0)
X chromosome
c
D = 0.48
Prionailurus
Group 3
X
XLRD
d
Autosome
X
0.64
0.54
0.29
0.26
0.45
0.20
0.65
0.40
0.80
0.50
0.32
0.14
0.8
0
1.0
0
0.37
0.31
0.35
0.32
0.40
0.15
0.65
0.30
0.96
0.86
0.5
0
1.0
0
0.7
0
0.12
0.08
0.02
0.01
0.12
0
0.13
0.06
0.50
0.30
0.32
0.26
0.35
0.15
0.74
0.44
0.19
0.12
0.14
0.08
0.30
0
0.35
0
Tree frequency
L --> H
L --> H
L --> H
L --> H
Recombination quintiles


a
Felis silvestris catus - Felis silvestris silvestris
$F_{ST}$
1.0
0.8
0.6
0.4
0.2
0
RND
2
1
0
Recombination rate
Non-XLRD
XLRD
Non-XLRD
***
$F_{ST}$
XLRD
Non-XLRD
***
RND
Sus scrofa (Europe) - Sus scrofa (China)
$F_{ST}$
1.0
0.5
0
RND
1
0
Recombination rate
Non-XLRD
XLRD
Non-XLRD
***
$F_{ST}$
XLRD
Non-XLRD
***
RND
XLRD
Non-XLRD


Carneiro et al.[26] Oryctolagus cuniculus algirus X O.c. cuniculus
5
3
0
FST Z-score
Recombination rate
20
40
60
80
100
Oryctolagus cuniculus
Homo sapiens XLRD
50
80
110
Alignment direction
Inverted
Collinear
Genomic position (Mb)

- Deep learning algorithm?
- Why just X, why not Y?
- Syntenic alignments? Haldane's rule? Chromosome enrichment? Dosage compensation?
- Is there evidence this occurs beyond mammals?
- Also how uncertain are we about some of the realtionships within mammals? I know the broad (Afrotheria, Laurasiathera, etc.) seem stable from what I have seen, but how much uncertanty do we have?
- It makes me wonder which region is so well-conserved in plants (chloroplast?), given that they don't exhibit much sexual isolation