Genetic Divergence of Humans, Chimpanzees, & Gorillas Along the X & Y Chromosomes

Motivation

Published: 17 May 2006

Genetic evidence for complex speciation of humans and chimpanzees

Nick Patterson, Daniel J. Richter, Sante Gnerre, Eric S. Lander & David Reich

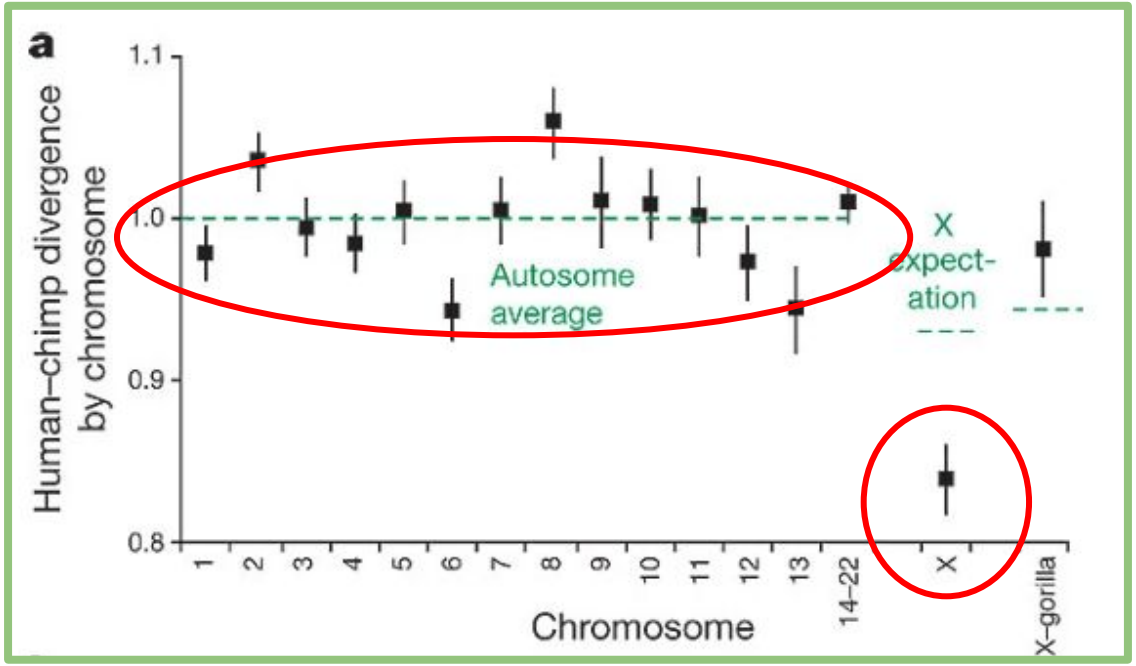
Nature **441**, 1103–1108 (2006) | Cite this article

7029 Accesses | **390** Citations | **130** Altmetric | Metrics

Abstract

The genetic divergence time between two species varies substantially across the genome, conveying important information about the timing and process of speciation. Here we develop a framework for studying this variation and apply it to about 20 million base pairs of aligned sequence from humans, chimpanzees, gorillas and more distantly related primates.

Motivation


a
Human–chimp divergence by chromosome
1.1
1.0
0.9
0.8
Autosome average
X expect-ation
1
2
3
4
5
6
7
8
9
10
11
12
13
14–22
X
X–gorilla
Chromosome

Goal

- Recreate Patterson et al.’s analysis using updated genome assemblies
- Look at both chimp and gorilla divergence
- Look across both the X and Y chromosomes
- Evaluate Patterson et al.’s conclusions about human-chimp speciation

Dataset

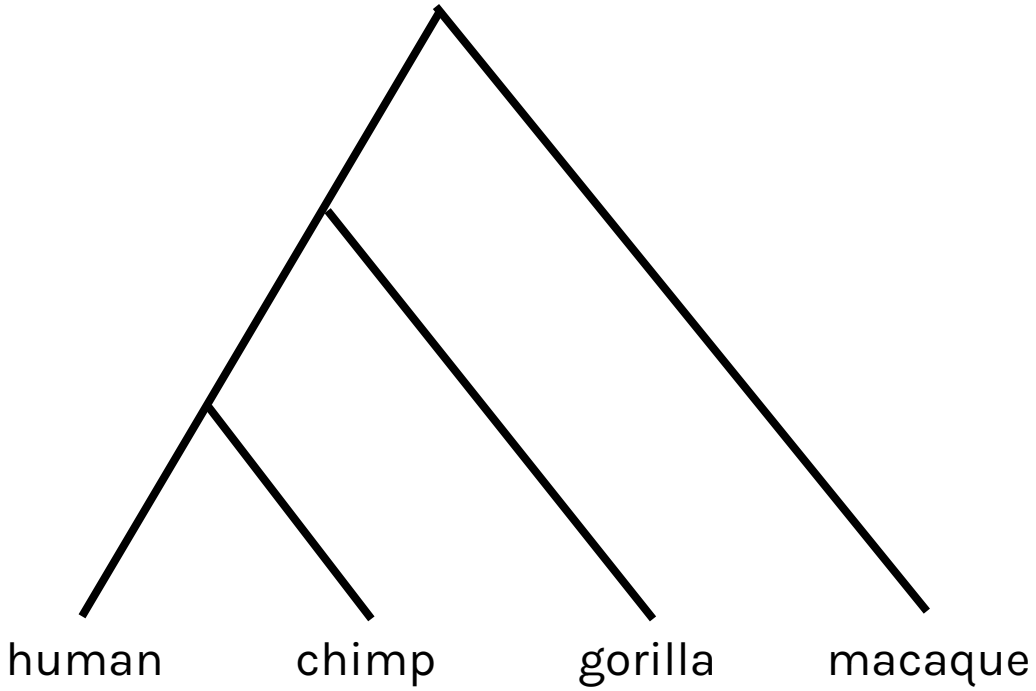
Human: hg19, Feb. 2009

Chimp: panTro6, Jan. 2018

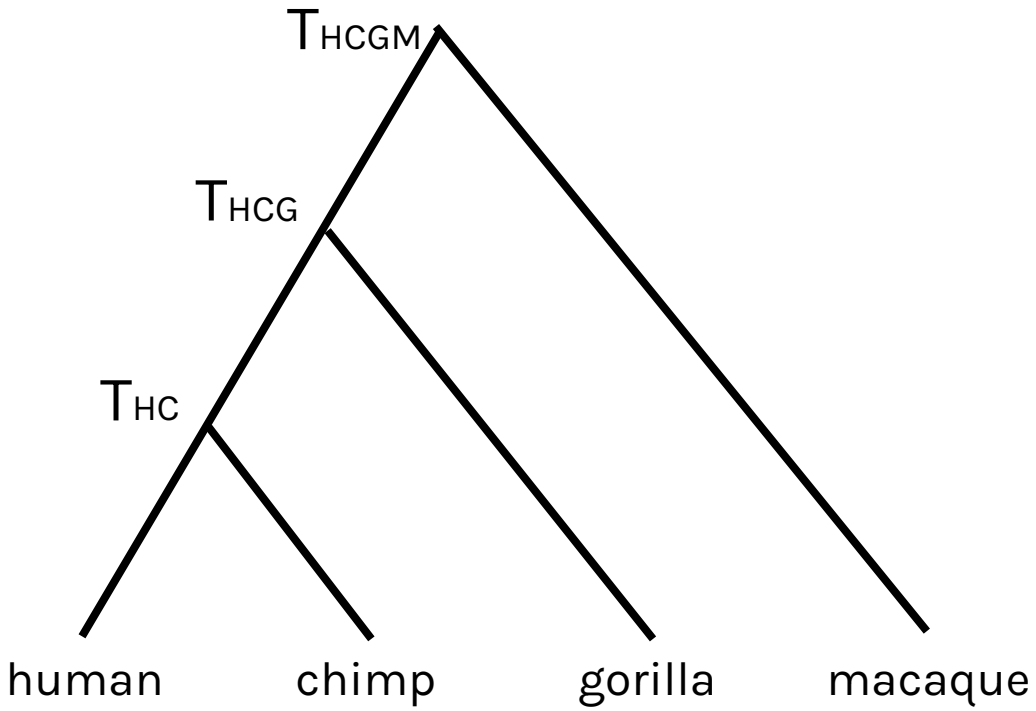
Gorilla: gorGor5, Mar. 2016

Macaque: rheMac10, Feb 2019

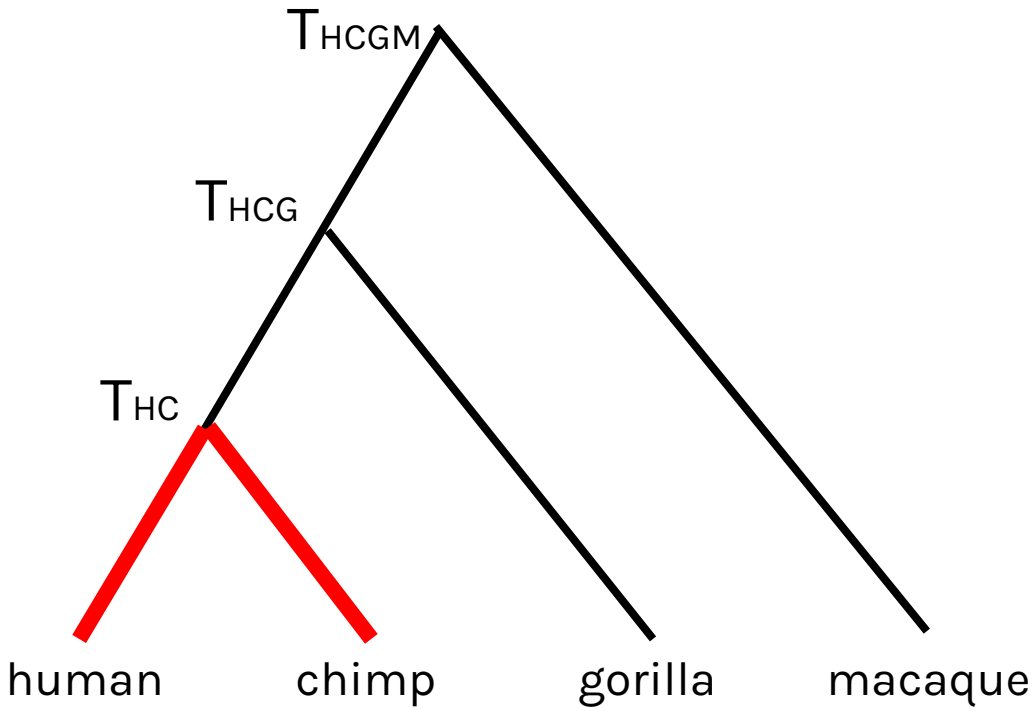
Species relationships



Species relationships



Species relationships



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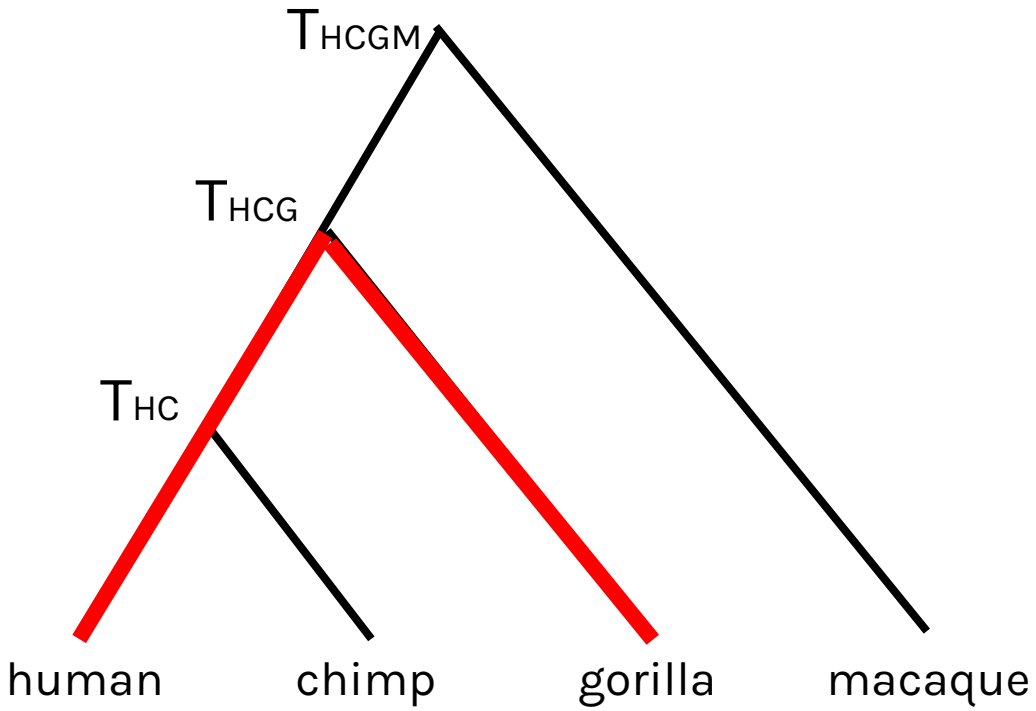
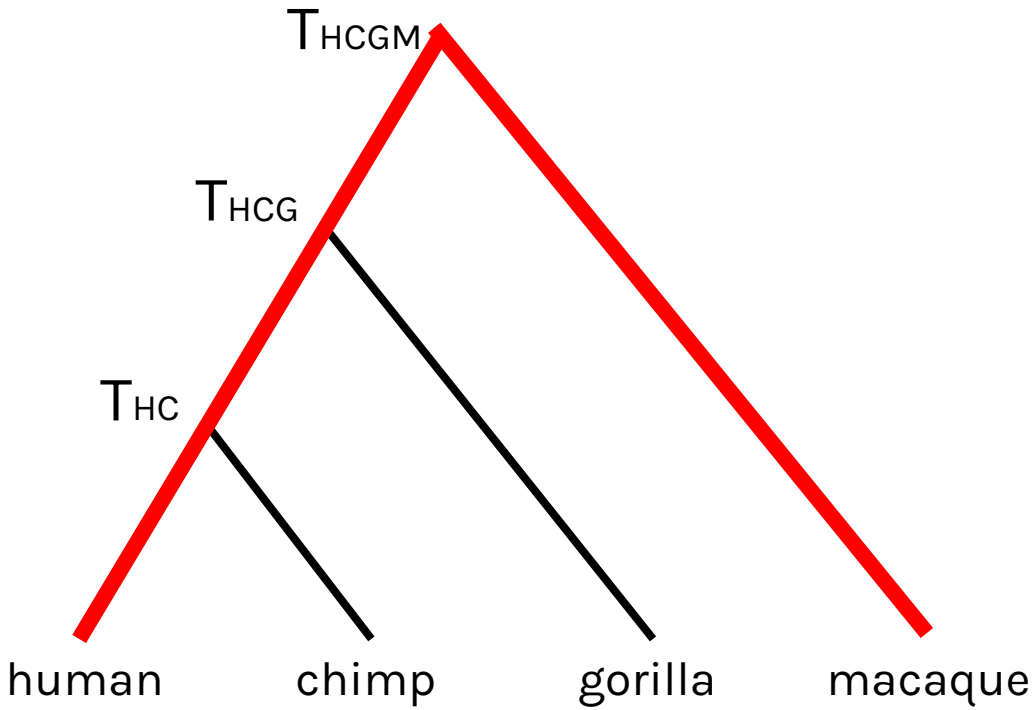


Species relationships

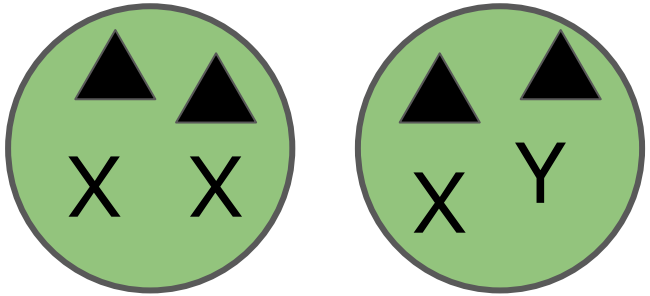


Calculating divergence

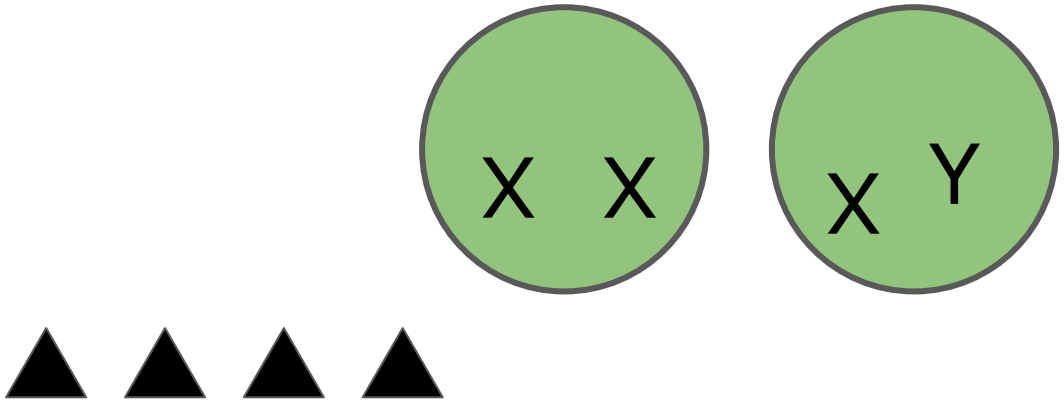
1. Remove sites with missing data
2. Align the genomes (human-chimp, human-gorilla, human-macaque)
3. Compute the number of nucleotide differences
4. Calculate divergence

   divergence = differences / sites
5. Normalize the human-chimp and human-macaque divergence values by the human-macaque value
6. Scale so autosomal divergence = 1

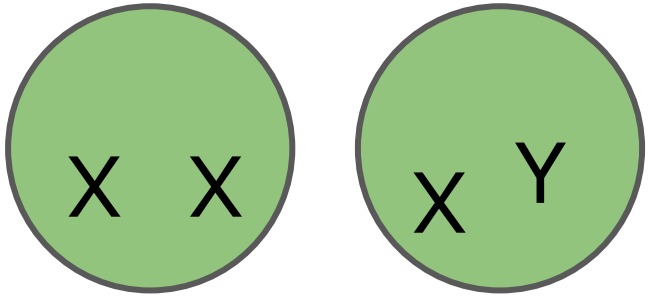
X and Y expectations



X and Y expectations

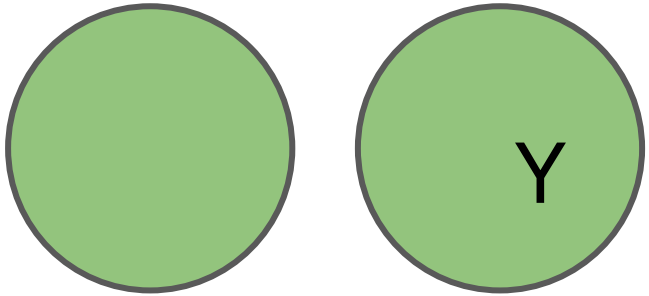


X and Y expectations



Autosome divergence = 1

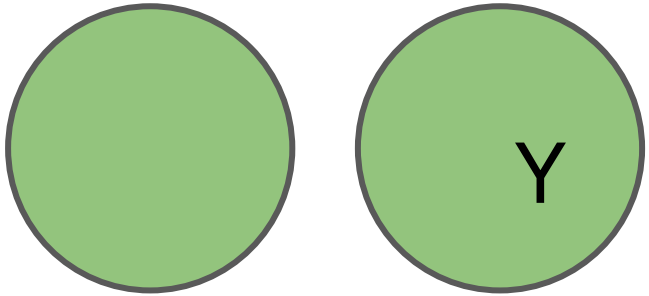
X and Y expectations



Autosome divergence = 1

X X X

X and Y expectations

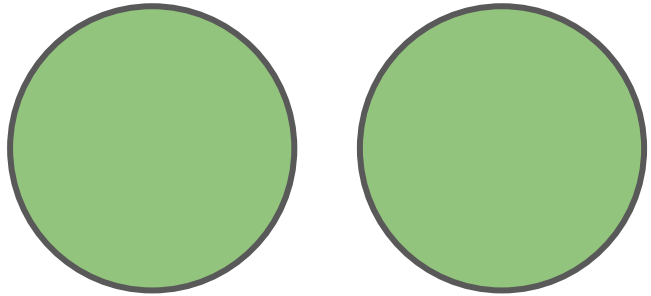


Autosome divergence = 1

X X X

X divergence expectation = 0.75

X and Y expectations



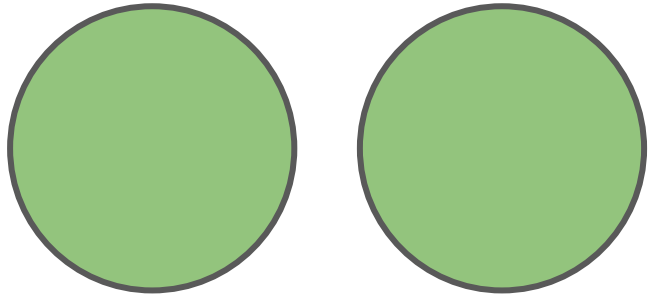
Y

Autosome divergence = 1

X X X

X divergence expectation = 0.75

X and Y expectations



Autosome divergence = 1

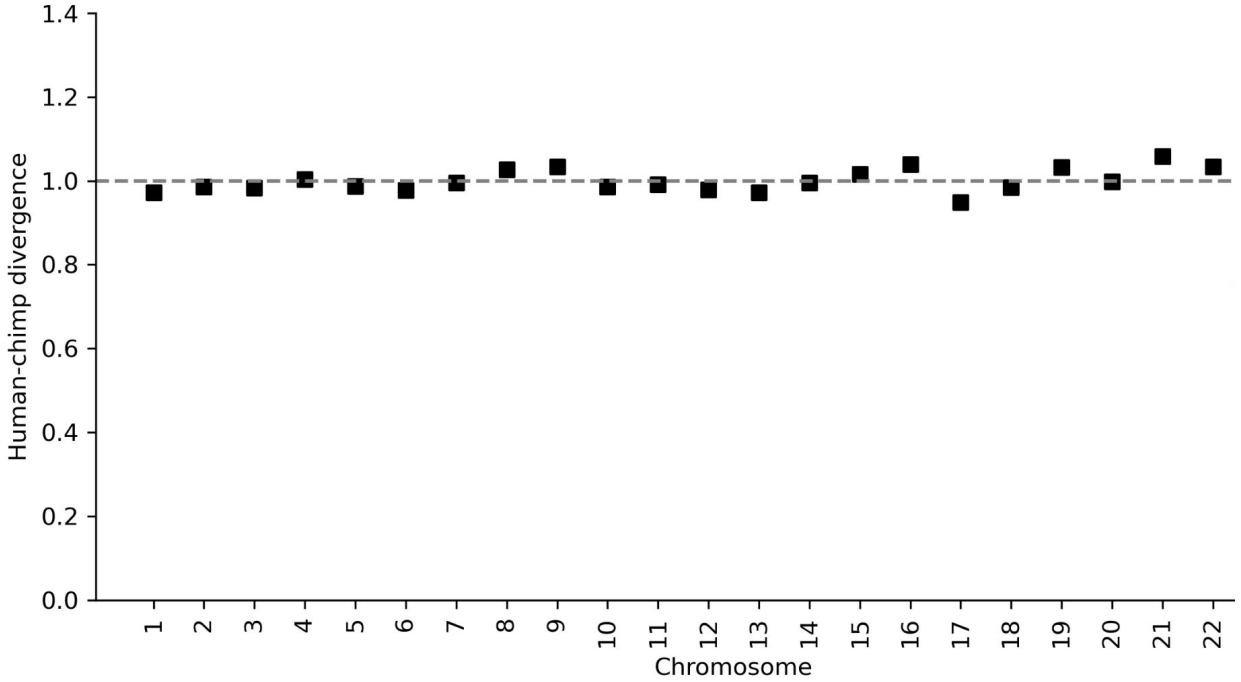
Y

Y divergence expectation = 0.25

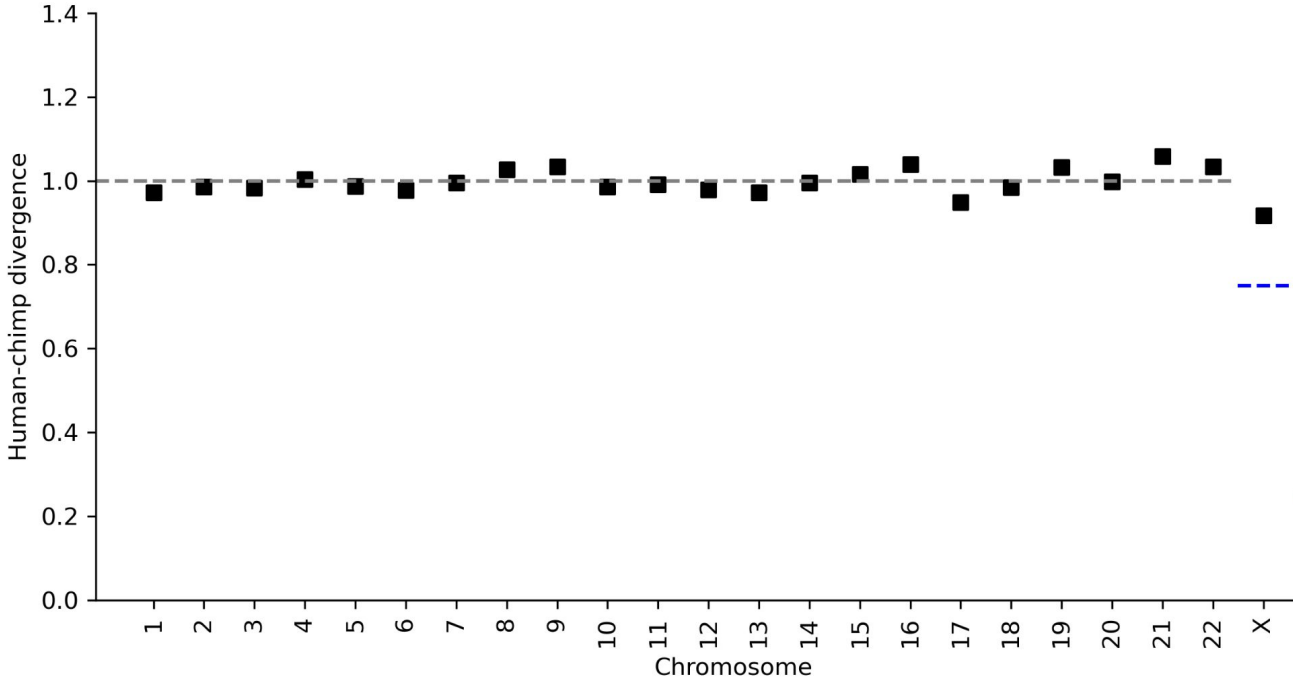
X X X

X divergence expectation = 0.75

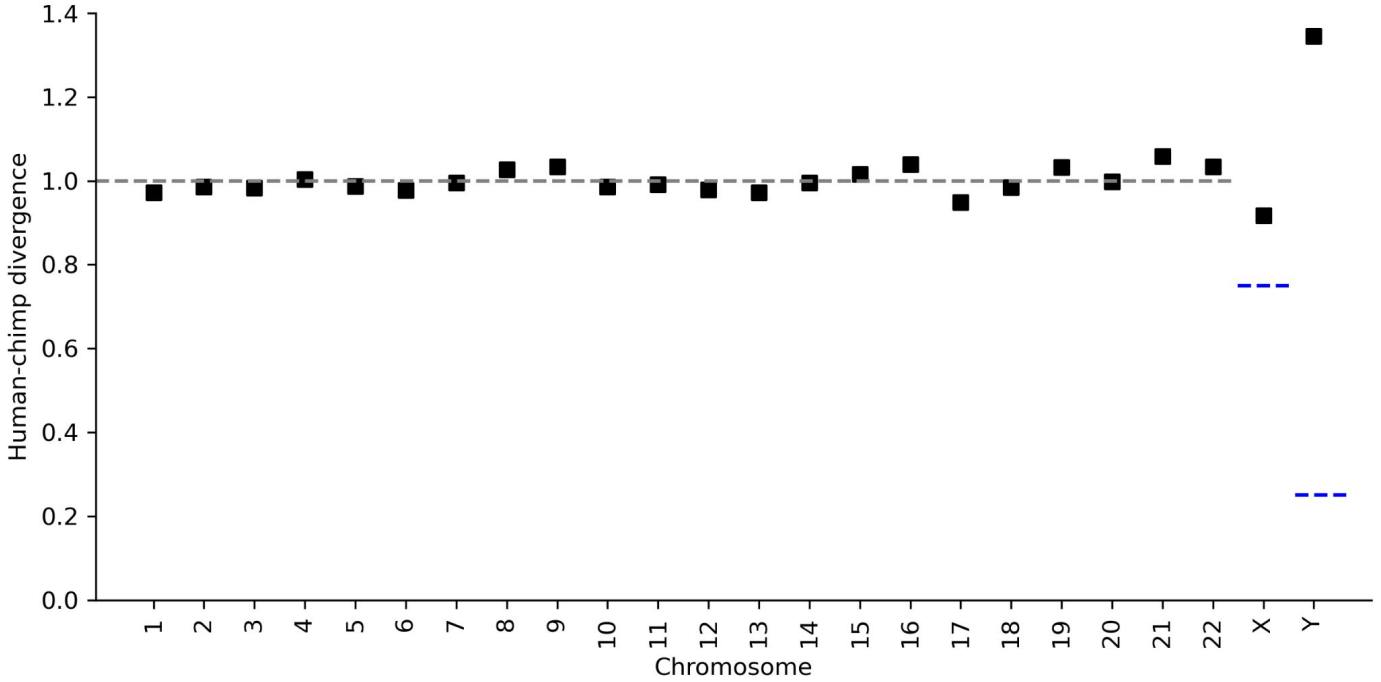
Human-chimp divergence, autosomes vs. X vs. Y



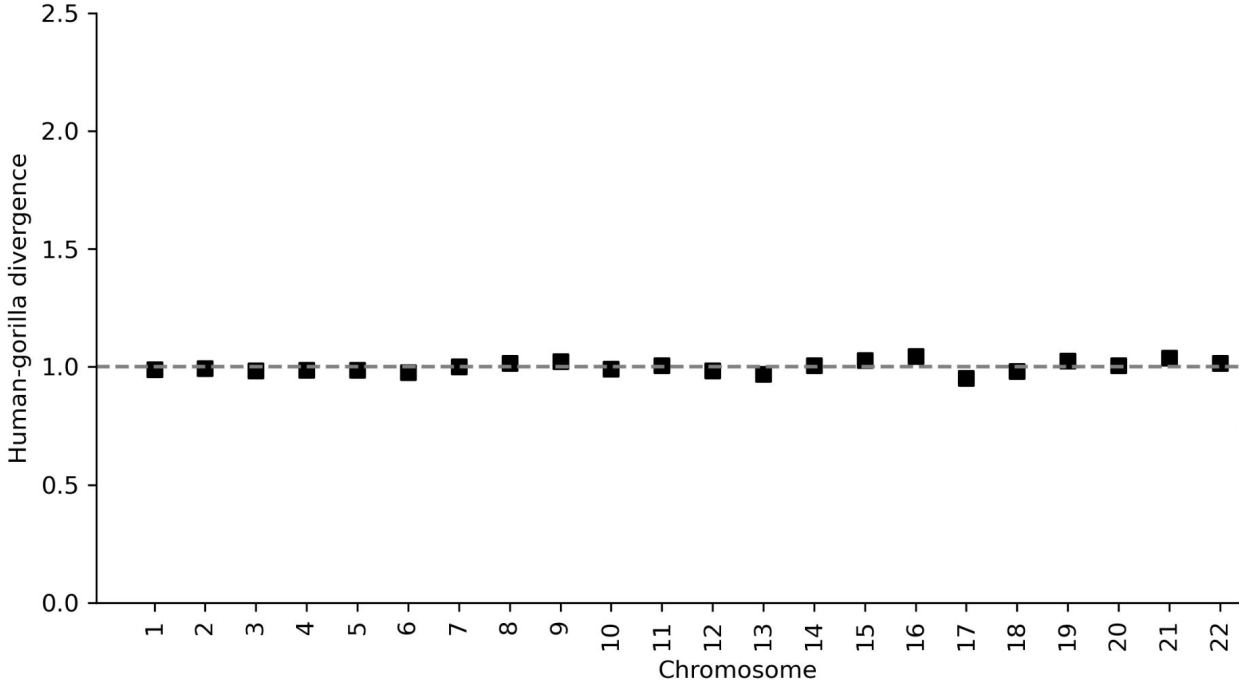
Human-chimp divergence, autosomes vs. X vs. Y



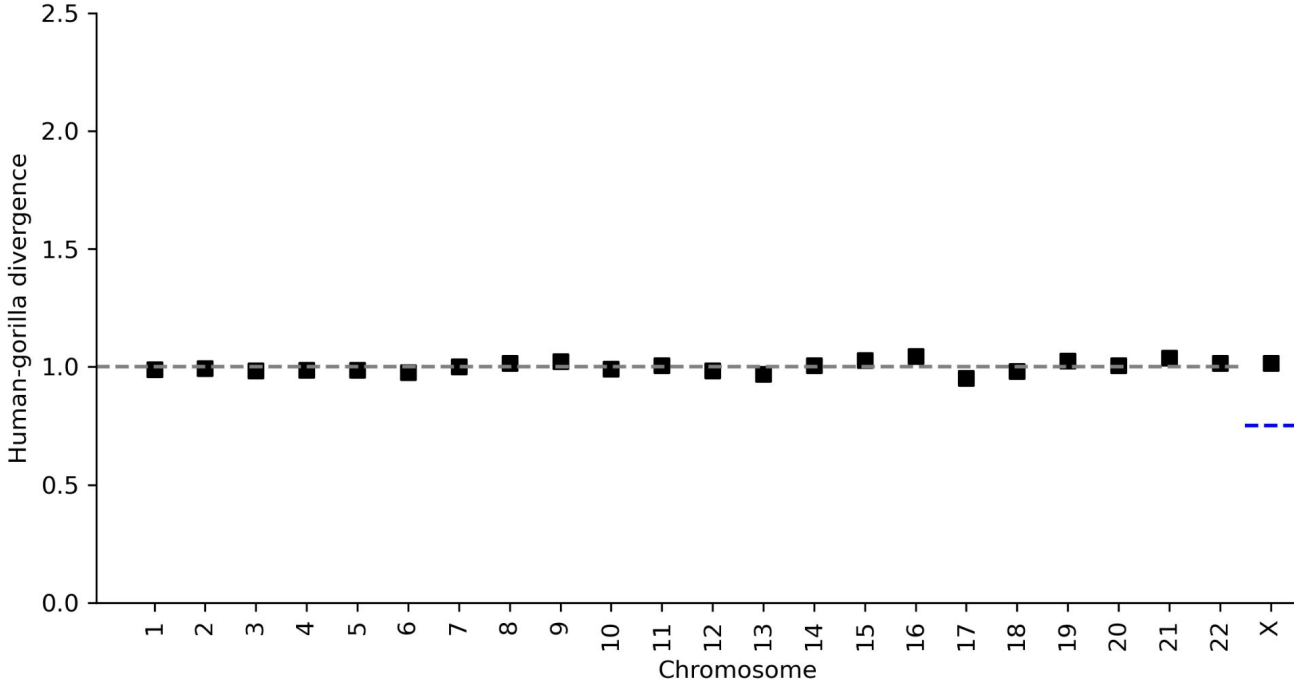
Human-chimp divergence, autosomes vs. X vs. Y



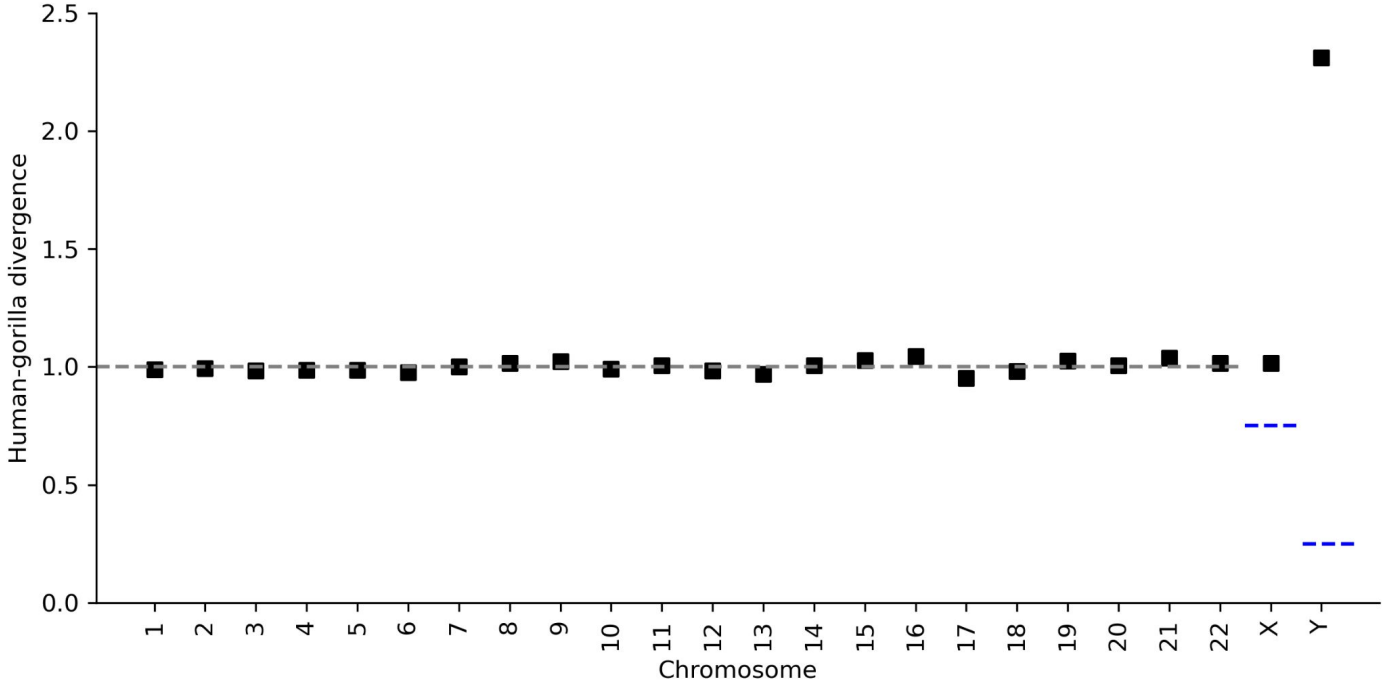
Human-gorilla divergence, autosomes vs. X vs. Y



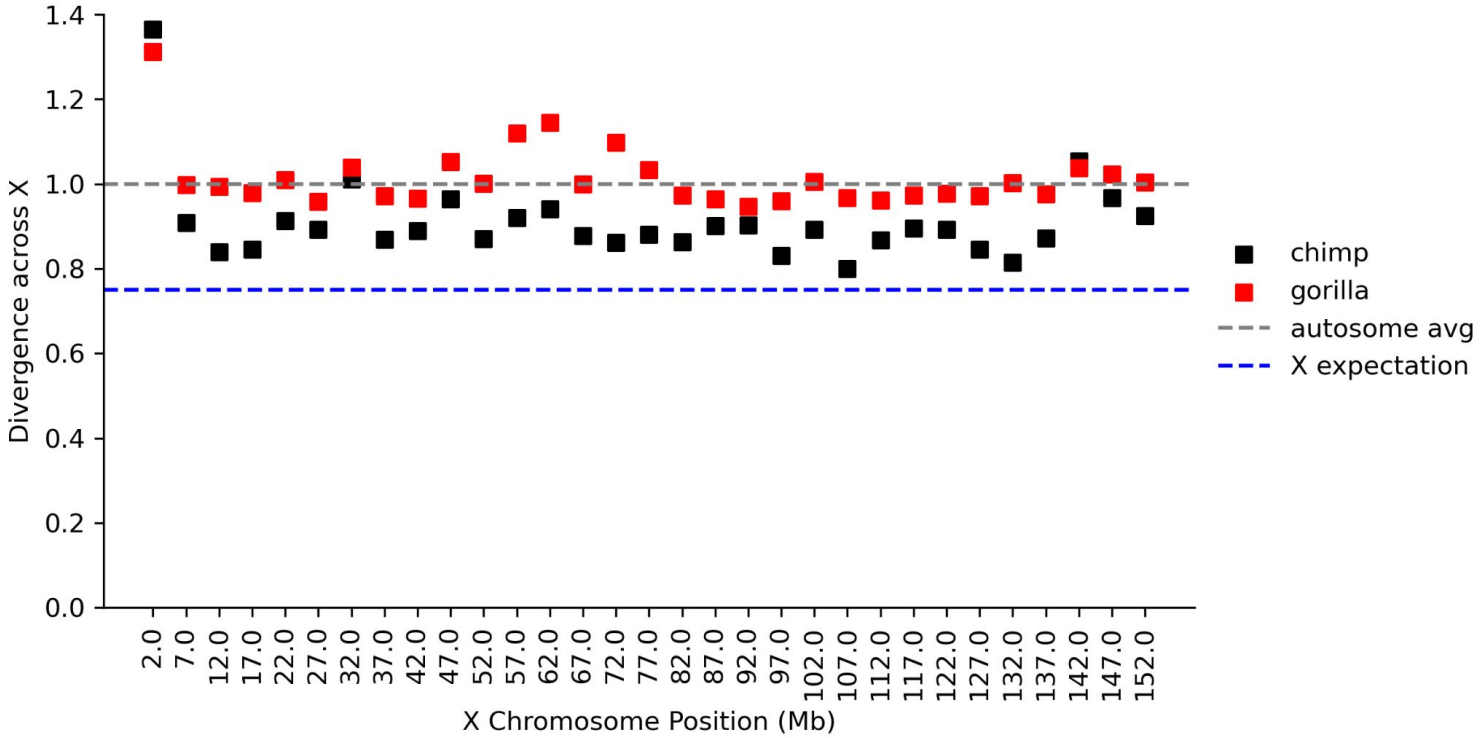
Human-gorilla divergence, autosomes vs. X vs. Y



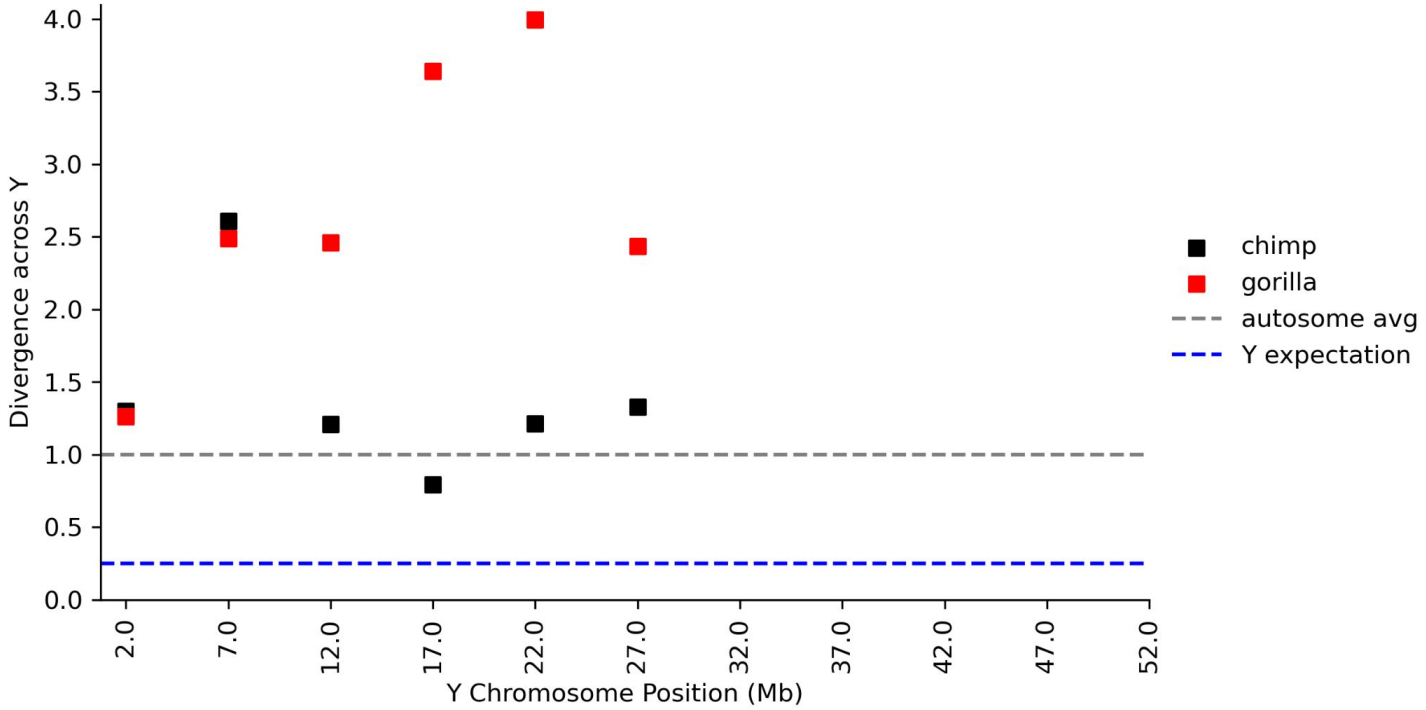
Human-gorilla divergence, autosomes vs. X vs. Y



Divergence across X chromosome



Divergence across Y chromosome



Conclusions and next steps

- Human-chimp and human-gorilla divergences are greater than expected, especially for X chromosome
- Divergence values are greater than expected in almost every window along the X and Y chromosomes
- More data is needed to explore further
- Ideally compare to a less-distant outgroup

THANK YOU!

Questions?