Genomes, organization & Gene structure.

Medical Genetic Department

2022

Objectives

- Define genome and to recognize the differences in the genome of different species.
- To understand the Human Genome
- To understand the structure of the gene.
- To understand how the genetic code is used.

Genome

Definition

Genome: all genetic information of an organism

Genome

- The size of a genome is expressed in millions of base pairs (Mb)
- It is highly variable depending on the species:
  - VIRUSES: Thousands
  - BACTERIA: Millions
  - MAMMAL: Billions

Genome of Hepatitis B virus: 3,182 bp

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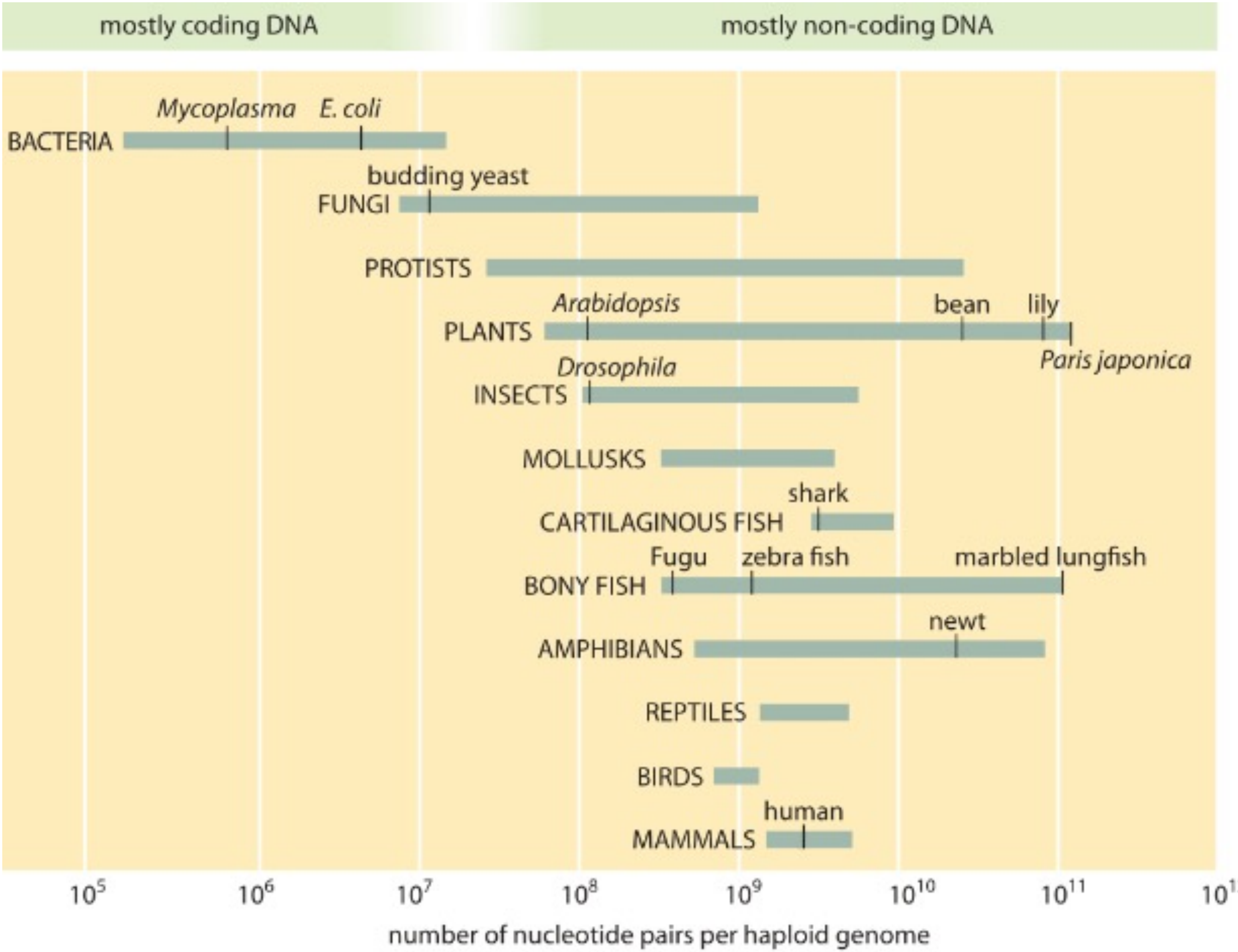
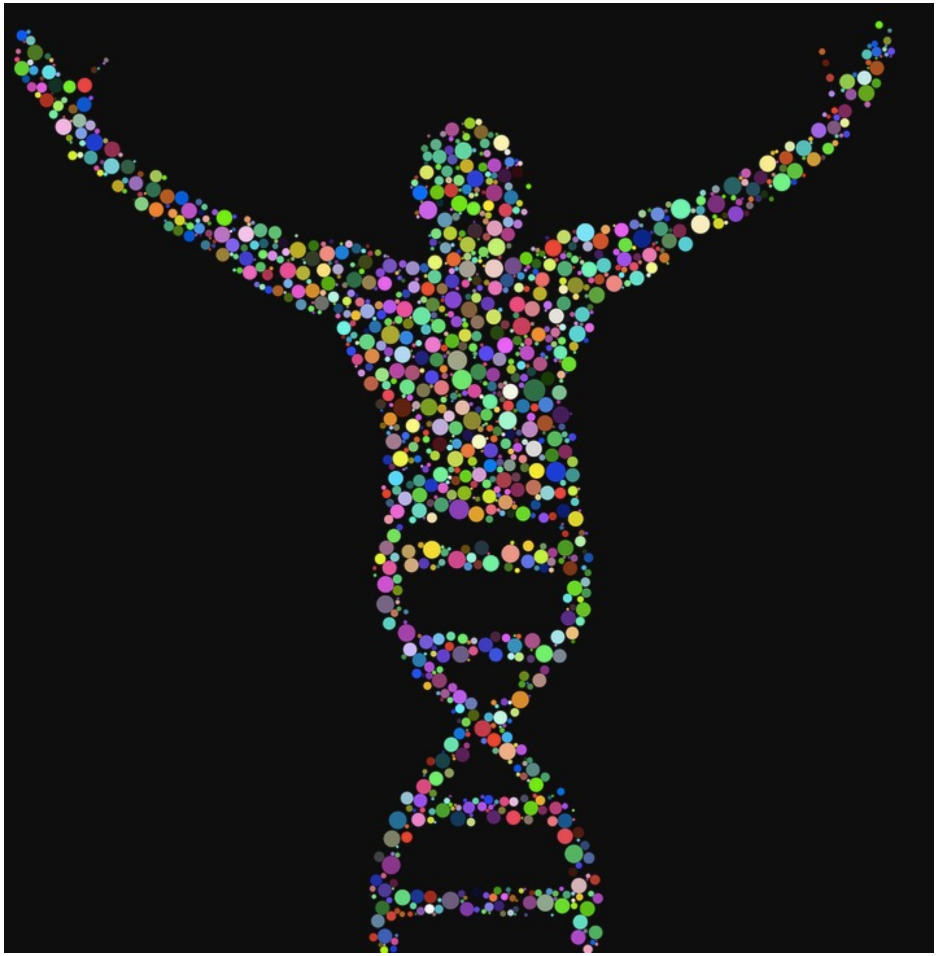
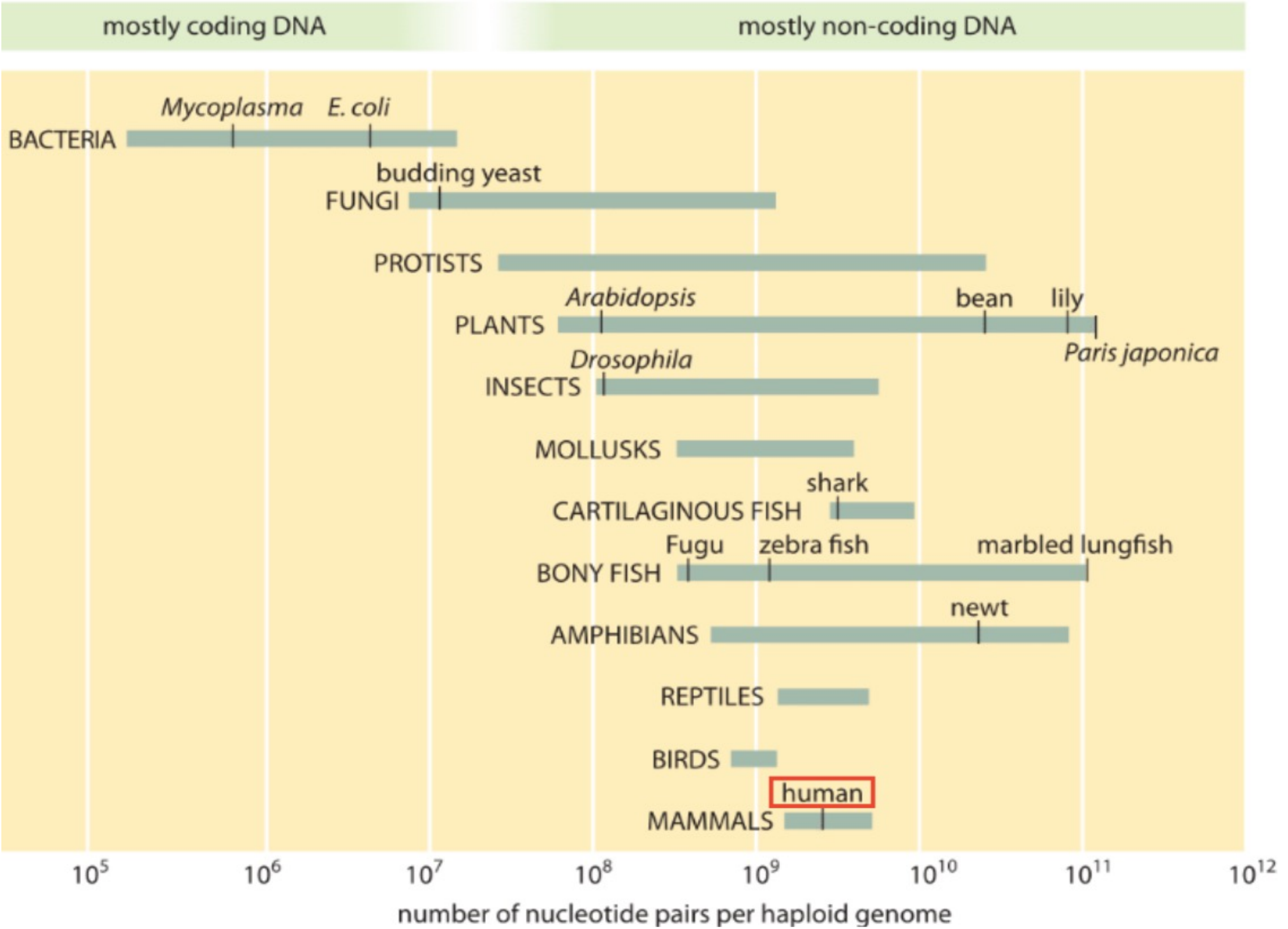


CELL BIOLOGY By the NUMBERS, 2015

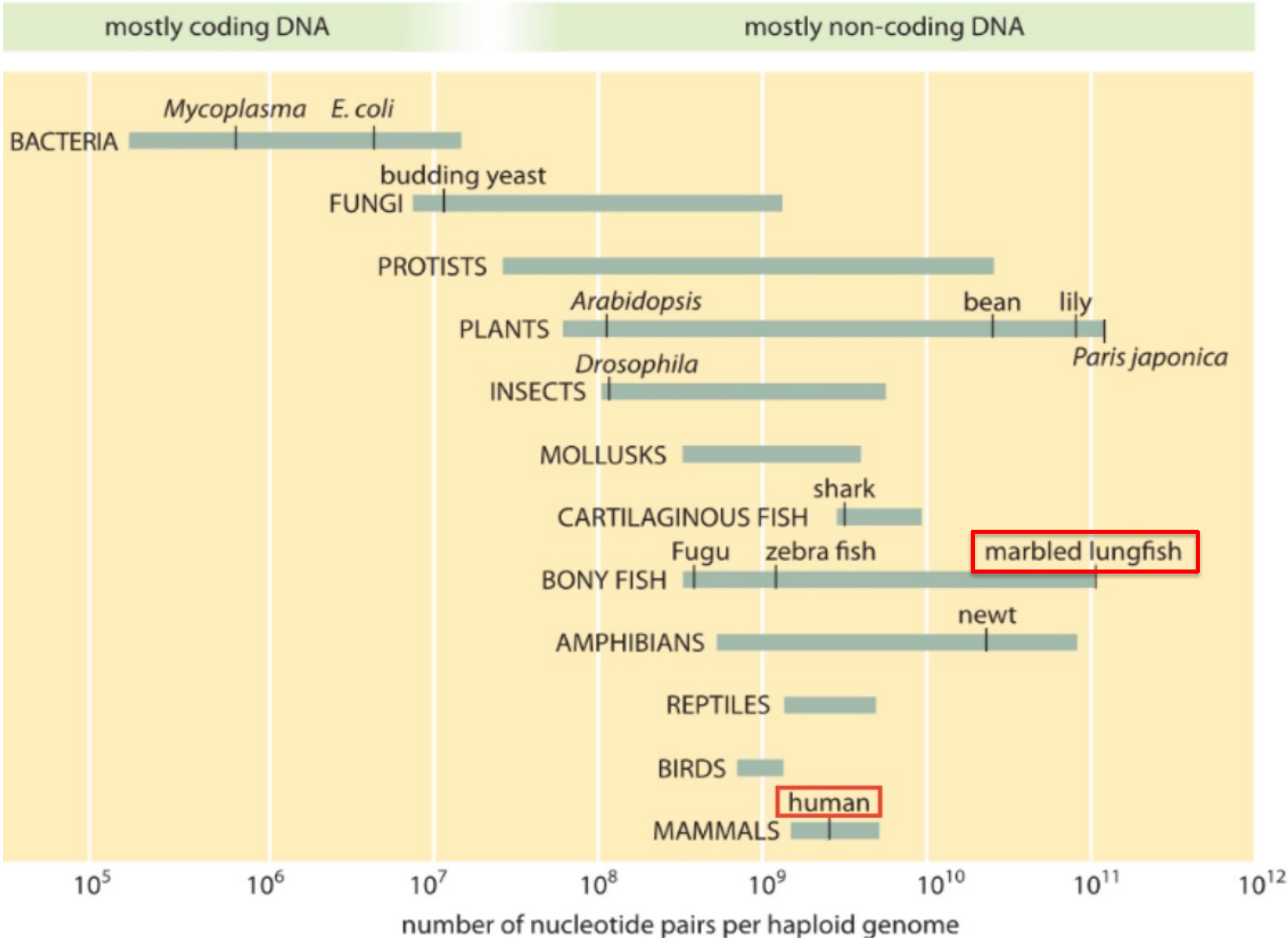
Human Genome: 3 billion DNA base pairs





CELL BIOLOGY By the NUMBERS, 2015

Marbled lungfish *130 000 Mb*



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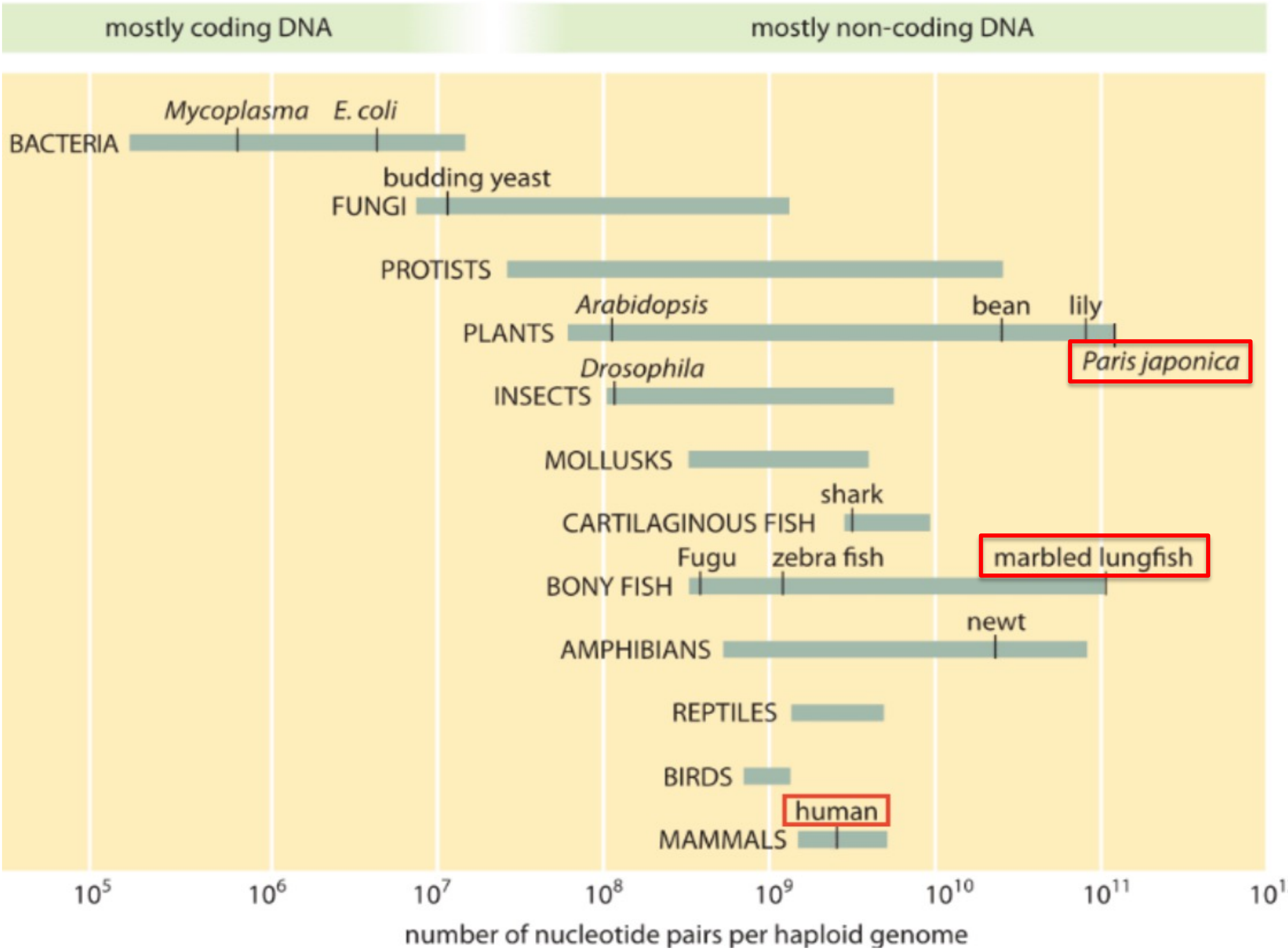
Paris Japonica 149 000 Mb

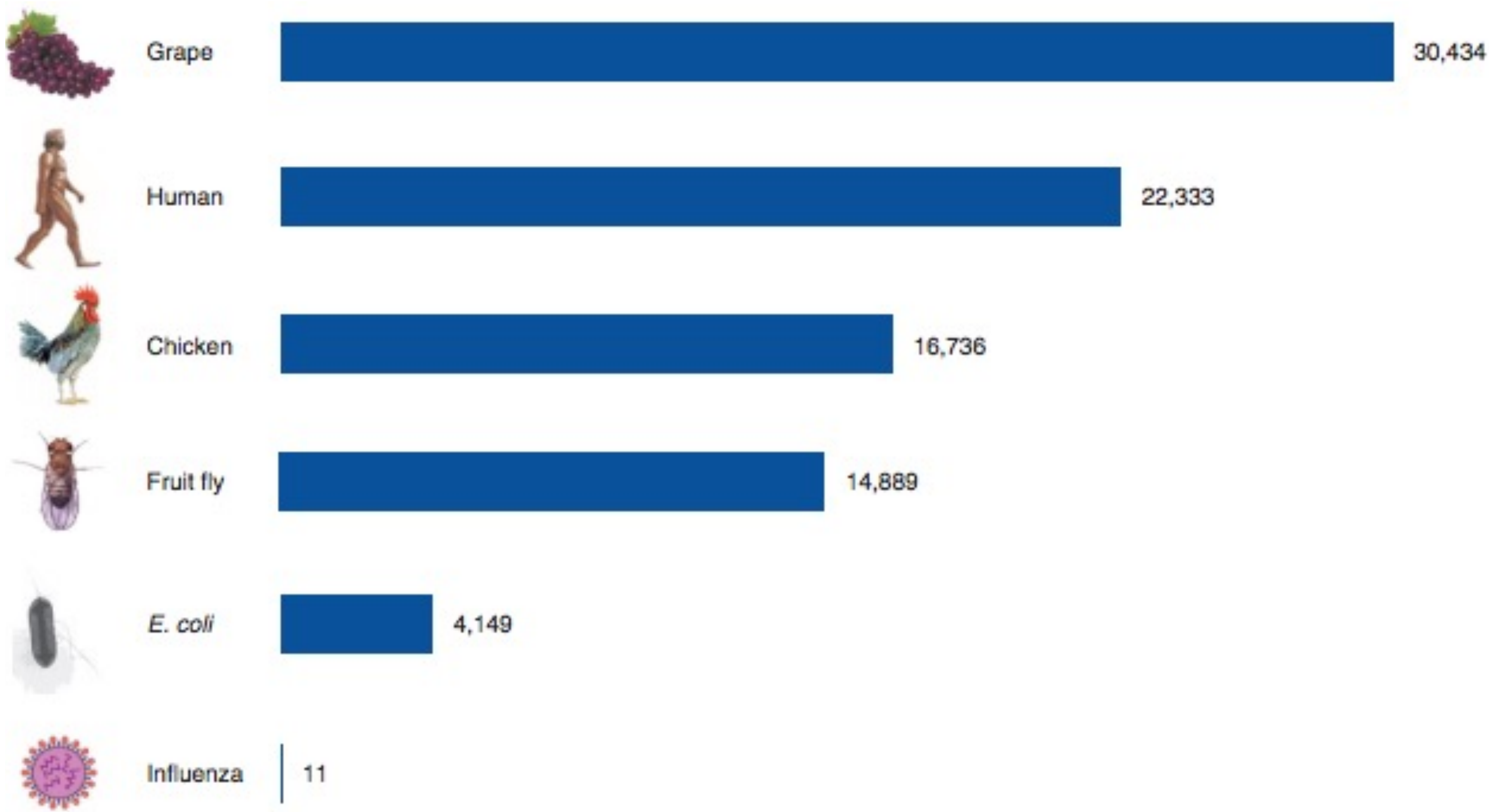


CELL BIOLOGY By the NUMBERS, 2015

Genome

- The genome contains all the instructions necessary for the development, functioning, maintenance of the integrity and reproduction of cells and the organism.
- These instructions are located in the genes.
- The number of genes varies in different species.

Gene counts in a variety of species



Pertea and Salzberg Genome Biology 2010, 11:206
http://genomebiology.com/2010/11/5/206

Shared human genome with:

- Mouse:
  - 75% are similar (16% the intron shorter than human)
  - the genes are 80% proteins same
- Chimpanzees:
  - between 98% to 95% similar (24 chromosomes)
  - the genes 99% are same
- In dogs 85% similar
- In Lily flower genome is 40 times larger

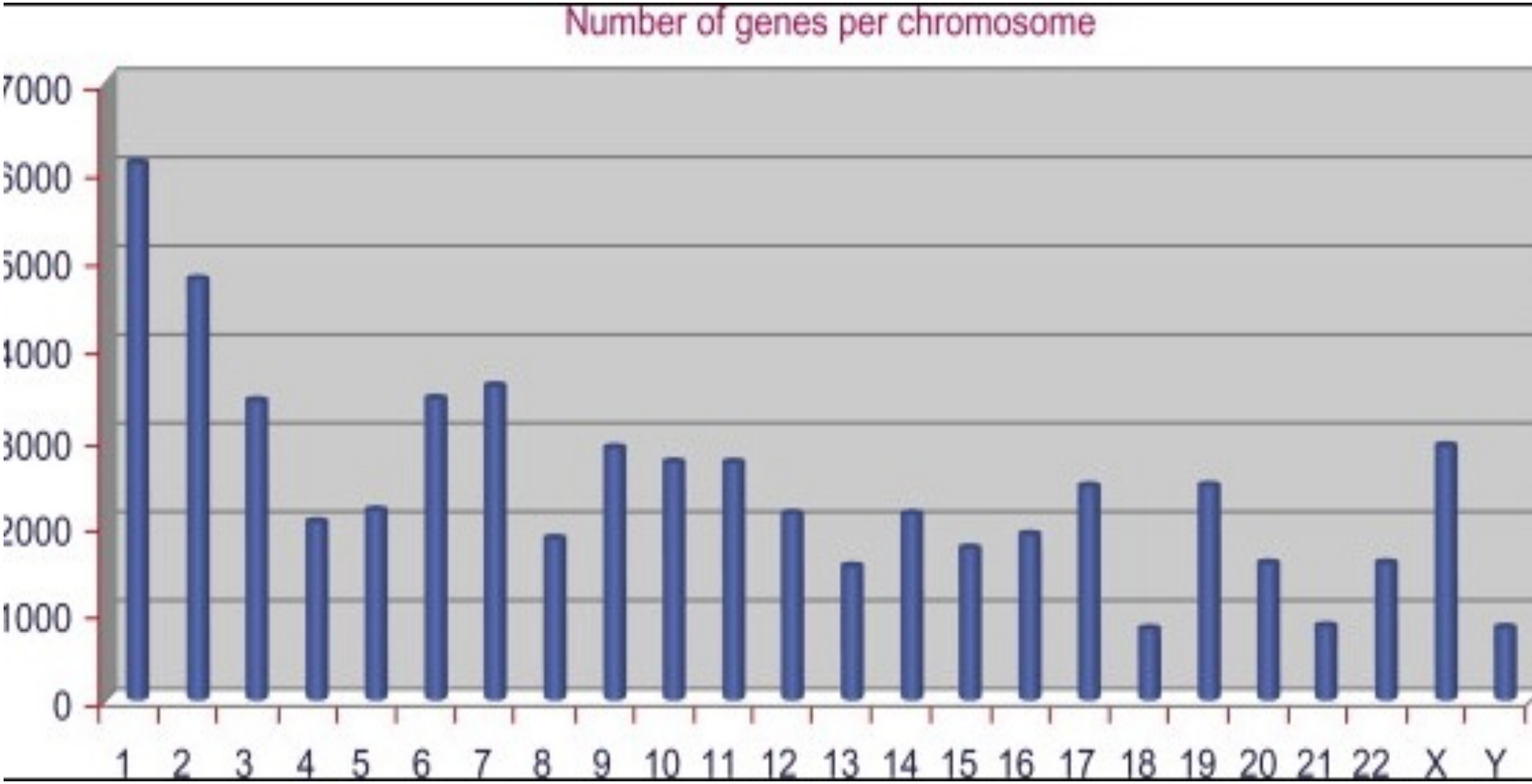
HUMAN GENOME

Human Genome

- 20,000 protein-coding genes occupying 1.2% of the genome
- Highly variable size: less than 1 kb for the SRY gene to more than 2 Mb for the DMD gene

Figure 2.13 Human Evolutionary Genetics, 2nd ed. (© Garland Science 2014)

Human Genome



Julia E. Richards, R. Scott Hawley, in The Human Genome (Third Edition), 2011

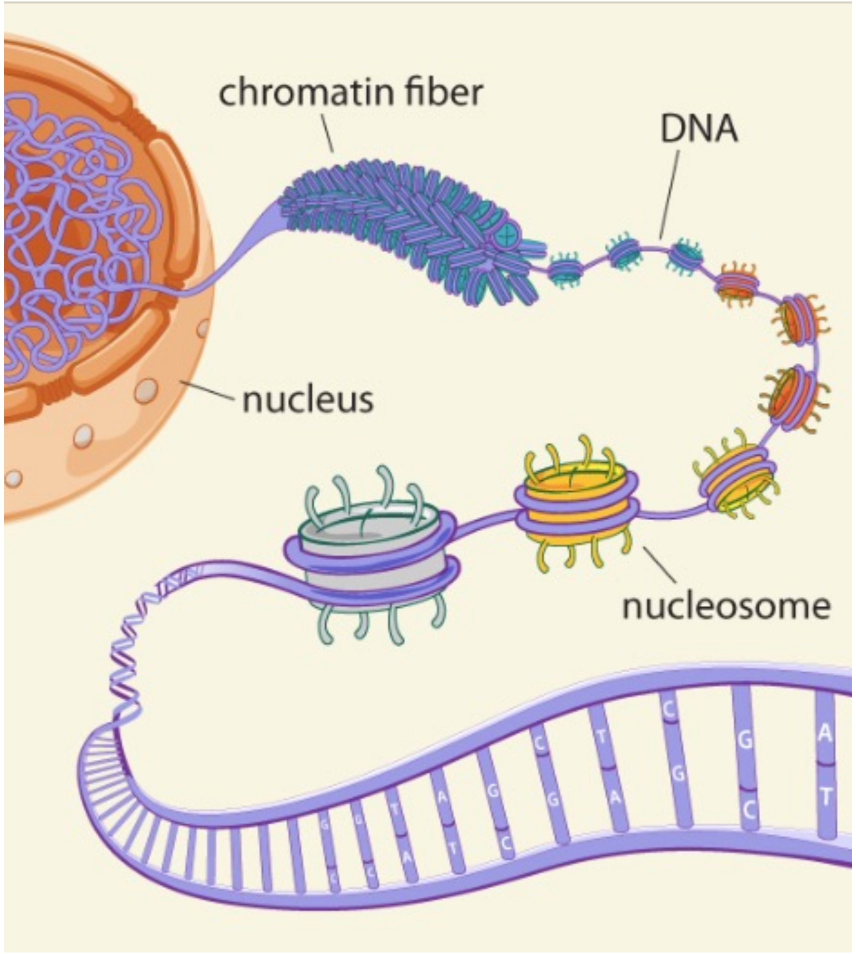
CHROMOSOME	TOTAL DNA (Mb)
1	249
2	243
3	198
4	191
5	181
6	171
7	159
8	146
9	141
10	136
11	135
12	134

CHROMOSOME	TOTAL DNA (Mb)
13	115
14	107
15	103
16	90
17	81
18	78
19	59
20	63
21	48
22	51
X	155
Y	59

Table 2.3 Genetics and Genomics in Medicine (© Garland Science 2015)

Human Genome

- Chromatin: consists of the combination of DNA and proteins



Heterochromatin:

Highly compacted chromatin with regions of untranscribed DNA

Euchromatin:

uncondensed chromatin with transcriptionally active regions

THANK YOU