



Lecture 4:

The Genome

The word and beyond

Course 485

Introduction to Genomics

AIMS

- Introduce the origins of the words “genome” and “genomics”.
- Discuss the use and misuse of OME and OMIC terms.
- Introduce the general characteristics of viruses, prokaryotes, and eukaryotes.
- Highlight similarities and differences between different genomes

Good Reads

Few references on the origins of the words
“genome” and “genomics” and some implications

Perspective

What is a gene, post-ENCODE? History and updated definition

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Comment

No place like Ome

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COMMENTARY

'Ome Sweet 'Omics-- A Genealogical Treasury of Words

By Joshua Lederberg and Alexa T. McCray



Few references on the origins of the words “genome” and “genomics” and some implications



Beer, Bethesda, and Biology: How “Genomics” Came Into Being

Over the last decade, molecular genetics has spun off a lexicon of new words that scientists, including cancer researchers, now use to describe their work. One word that has become standard fare at many cancer meetings is “genomics,” meaning the study and comparison of genomes across species.

Where did the word genomics come from? It is the brainchild of Thomas H. Roderick, Ph.D., a geneticist at the Jackson Laboratory, Bar Harbor, Maine, who dreamed up the word in 1986 as the name of the then yet-to-be-published journal *Genomics*. In a recent interview, Roderick tells the *News* the story behind the word.

News: How did you come up with the word genomics?

Roderick: In 1986, I attended a good-sized international meeting in Bethesda to discuss the feasibility of mapping the entire human genome. The meeting had adjourned for the day, and Frank Ruddle, Ph.D. [Yale University], and Victor McKusick, M.D. [The Johns Hopkins University], convened a short submeeting involving about 50 people, including myself, to discuss starting a new genome-oriented scientific journal. The journal was to be a place to include sequencing data and as well to include discovery of new genes, gene mapping, and new genetic technologies. At the end of the meeting, Frank and Victor charged us to come up with a name for the new journal.

It now was late in the evening. A few of us went out to a recommended bar near one of those big office buildings in

Bethesda. It was called the McDonald's Raw Bar [which has since been torn down]. There might have been 10 of us that night who went there and sat around drinking beer — actually a lot of beer. It was great fun.

We kept moving on the name. Some of us really wanted to name the journal, *Genome*. But the *Canadian Journal of Genetics and Cytology* had already announced their intention to change its name to “Genome,” with their first issue to appear in 1987, about the time the new journal of McKusick and Ruddle was supposed to appear. Several names were considered using “Genome” as



Dr. Thomas H. Roderick

part of the title, but it was agreed they all were too cumbersome.

So, we sat around and talked. We were into our second or third pitcher, when I proposed the word “genomics.” I don't know exactly how I came up with the word. I'm a geneticist, and it certainly isn't far from the word “genetics.” I've heard the word “genetics” since I

was in high school, so it must have played a part in the name. In fact, I'm sure it did.

I said the word to Frank Ruddle. Frank recognized it as a name that encompassed what we wanted to do. It wasn't just the objectives of the journal. It was GE-NOM-ICS. It was an activity, a new way of thinking about biology.

We adjourned that evening thinking genomics wasn't a bad name. But I didn't hear any more about it until Victor and Frank decided that was what they wanted to name the journal. Frank told me later that Victor had done some scholarly study of the word to be certain it was etymologically appropriate.

News: When you proposed the term genomics, what was the definition that was in your mind?

Roderick: Well, it certainly encompassed what the journal wanted to cover. It encompassed sequencing, mapping, and new technologies. But we felt it also had the comparative aspect of genomes of various species, their evolution, and how they related to each other. Although we didn't come up with the term “functional genomics,” we thought of the genome as a functioning whole beyond just single genes or sequences spread around a chromosome.

News: Did you ever think when you left the raw bar in Bethesda that this name would become such a big part of biology?

Roderick: No. Victor and Frank thought their proposed journal had an important set of objectives defining a specific timely mission. I thought we had a tentative name for a journal beyond just sequencing and mapping.

— Bob Kuska

PERSPECTIVES

The Wholeness in Suffix *-omics*, *-omes*, and the Word *Om*

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Eisen *GigaScience* 2012, 1:6
<http://www.gigasciencejournal.com/content/1/1/6>

(GIGA)ⁿ
SCIENCE

COMMENTARY

Open Access

Badomics words and the power and peril of the ome-meme

Jonathan A Eisen^{1,2,3,4,5*}

The genome

- The word “genome” was first proposed by a the German botanist Hans Winckler (1920).
- He wrote:

“ I propose the word **Genom** for the haploid chromosome set, which, together with the pertinent protoplasm, specifies the material foundation of the species”.

What inspired creating such a word?

The genome

- Combining gene + chromosome?
- Influenced by botany related terms such as rhizome (the entire root system)?
- Other more poetic explanations??

What is the genome?

Entire genetic material that make a species

- The word “genome” open the floodgates for thousands of OMEs!!!

OMEs are everywhere

antigenome
bacteriome
basidiome
biome
cardiome
caulome
chondriome
cladome
coelome
epigenome
erythrome
genome
geome
hadrome
histome

immunogenome
immunome
haptenome
karyome
leptome
microbiome
mnemome
mycetome
neurome
odontome
osteome
pharmacogenome
phenome
phyllome
physiome

plastidome
plerome
proteinome
proteome
psychome
regulome
rhabdome
rhizome
stereome
thallome
tracheome
transcriptome
trichome
vacuome

Listed above we present a lexicome of terms, suffixed by *-ome*, extracted from the MEDLINE database, the *OED*, and the *Web of Science*. Our aim was to select terms using the *-ome* suffix in the sense of this article. For the most part this excludes the suffixes *-tome*, *-stome*, *-some*, *-drome*. Some terms are best known as the *-omics* derivative. Today, we should assume that further derivations are no longer from Greek or Sanskrit, that the *-ome* idea is borrowed from the multitude of terms already ensconced into English or the scientific lingua franca. Most of these terms are already in print; almost all should be self-revealing; a few are conjectural. Guess which of these *-omes* were made up only just now; even for these, there may well be an **-omics.com** to match.

Some of our OMEs

Some times it seems anything can be OMEfied

EXOGEN
EPIGEN
TRANSCRIPT OME
METABOL
PROTE



What do you think about the OMEs you have encountered so far in your career?

Useful/useless?

Meaningful/meaningless?

Do you have an OME word in your mind?

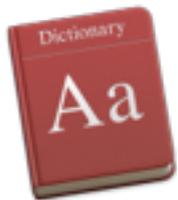
Genomics

- Thomas H. Roderick (and others) in 1986 came up with the word “**genomics**”.
- The word was chosen as a name for a new journal.
- Became a buzzword and very cool.
- Tons of OMICS out there.



What is genomics?

genomics |jē'nōmiks, -'nām-, |
pl.noun [treated as sing.] Biology
the branch of molecular biology concerned with
the structure, function, evolution, and mapping of
genomes.
ORIGIN 1980s: from *genome* '***the complete set of
genes present in an organism***' + *-ics*.



Genome organization & representation

How is the genetic code organized?

Thousands/ millions/ billions of nucleotide
basepairs

How are they organized into a genome?

Genome organization



Genome organization differ depending on:

- Genome chemical identity?
- Genome size
- Genome physical architecture
- Number of genome units
- Genome copy number
- taxonomic group (evolutionary history)

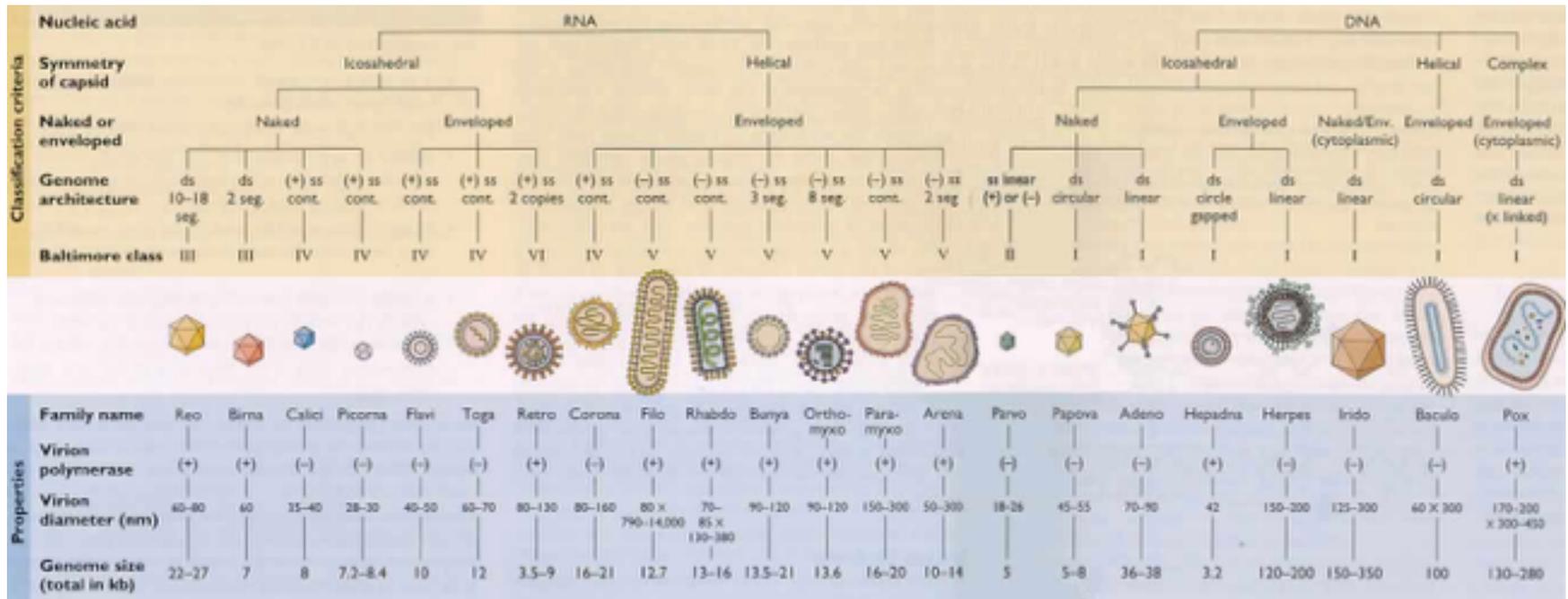
Genome chemical identity



The majority of life forms have a genetic material in the form of double stranded (ds) DNA.

Which organisms exhibit variation in the identity of their genomes?

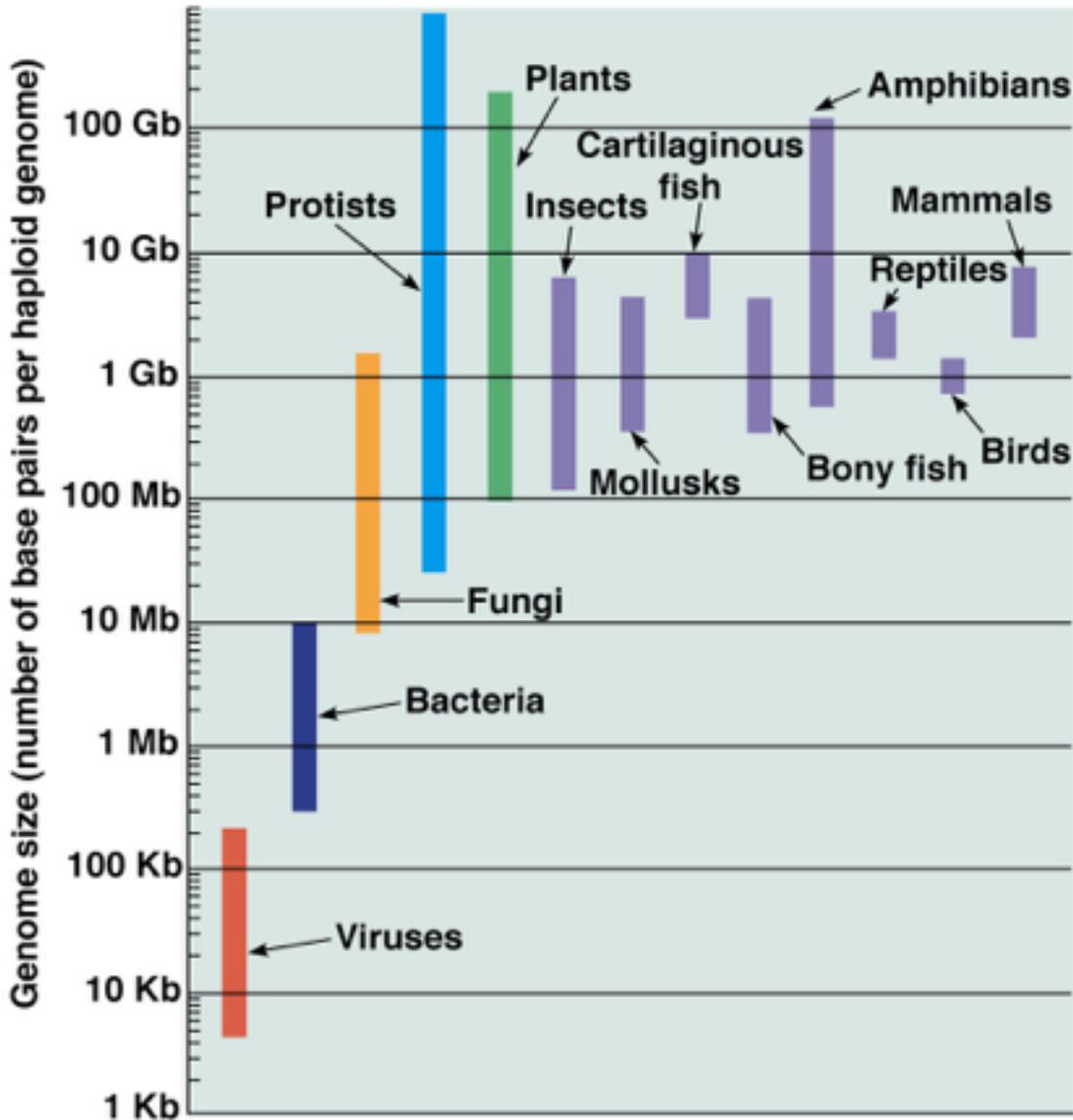
Genome chemical identity



Viruses have genomes in the form of:

- Single stranded DNA
- Single stranded RNA
- Double stranded DNA
- Double stranded RNA

Genome size



The simplicity of lower taxa is reflected in the genome size

BUT

in eukaryotes, the size of the genome is not related to organisms complexity

Genome physical architecture

The genome physical architecture can be arranged in:

- Linear unit(s)
- Circular unit(s)

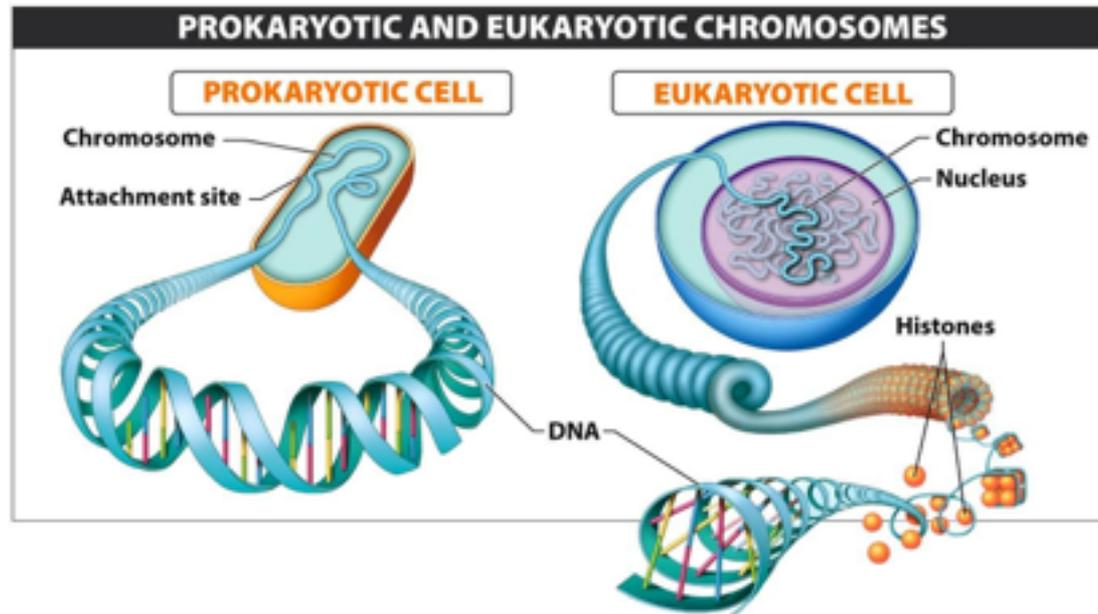


Figure 6-3
What Is Life? A Guide To Biology
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Genome physical architecture



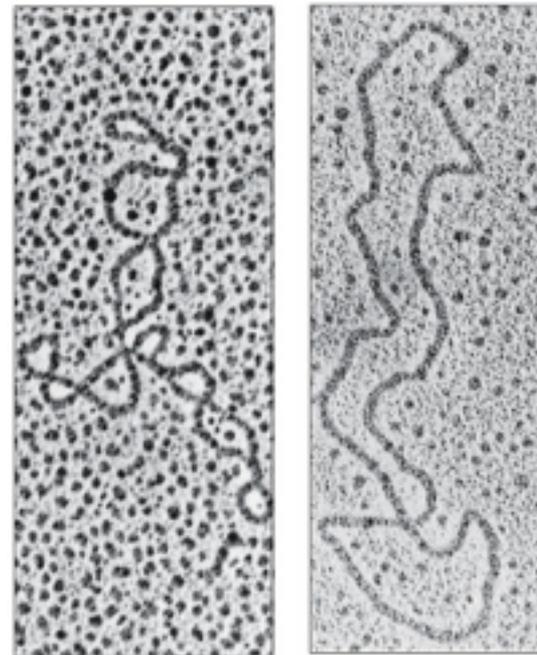
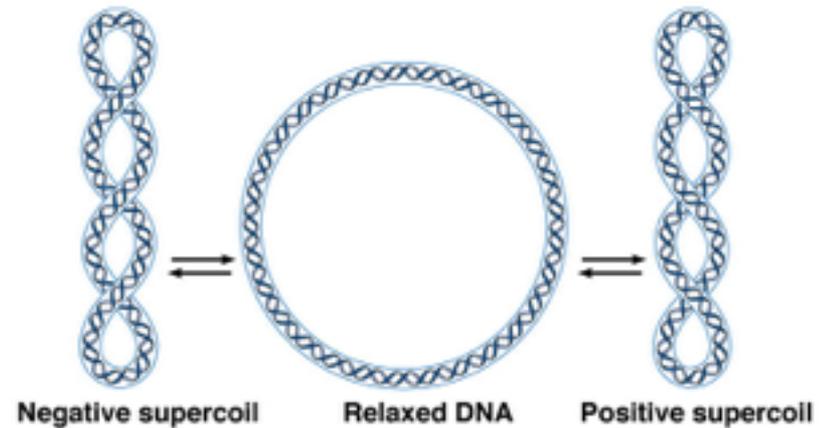
What is the advantage and disadvantages of having a circular genome?

What are the advantages and disadvantages of having a linear genome?

Genome physical architecture

Circular chromosomes:

- No ends (telomeres)
- No fear of telomere shortening
- No need for end protection.
- Suitable for reproduction by cell division.

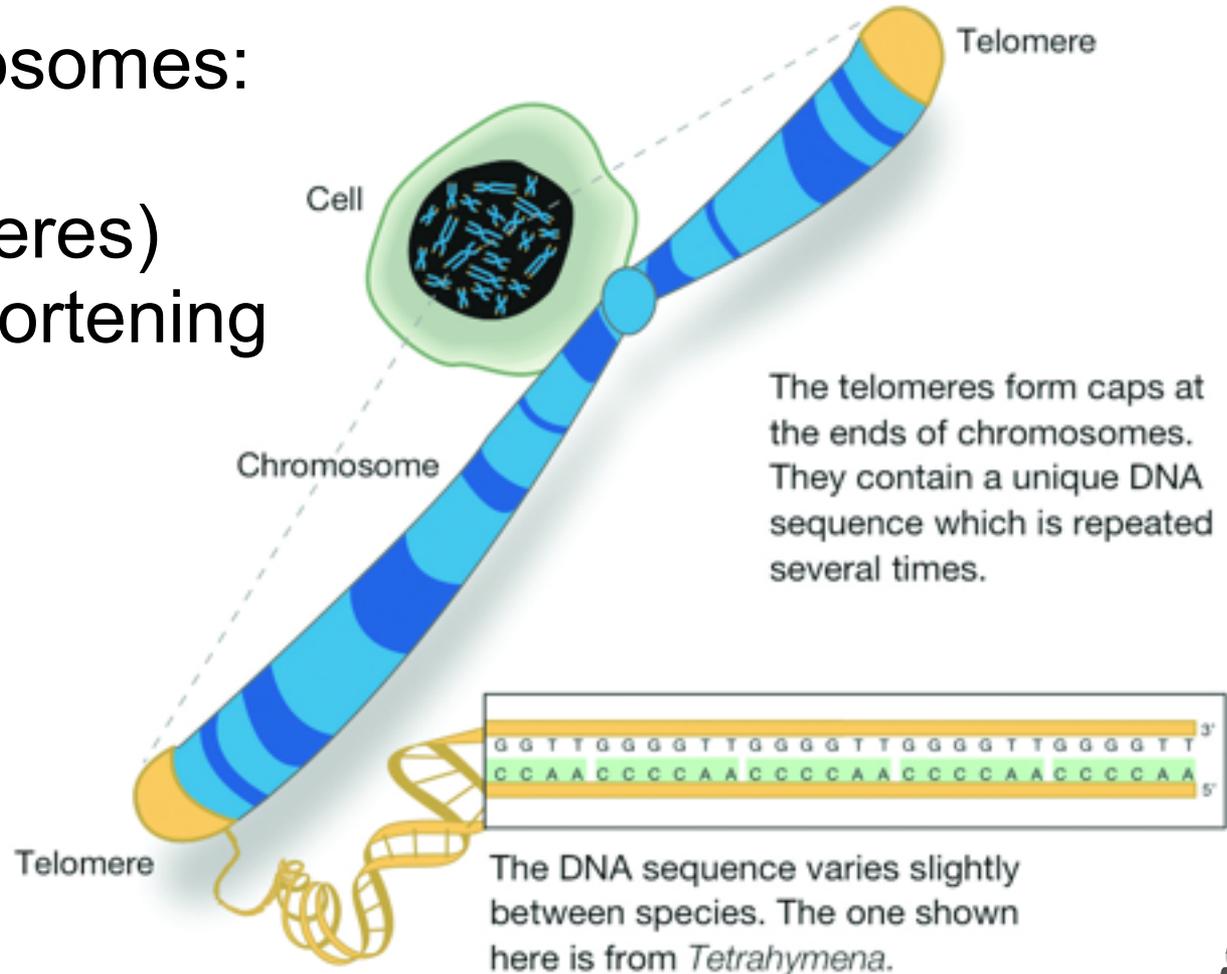


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Genome physical architecture

Linear chromosomes:

- Ends (telomeres)
- Telomere shortening
- Meiosis?



Genome physical architecture

The genome physical architecture can be arranged in:

- One unit (a single chromosome)
- Multiple units (multiple chromosomes)



Genome physical architecture

Bigger genomes are usually organized into multiple chromosomes

TABLE 7-1 Variation in Chromosome Makeup in Different Organisms

Species	Number of Chromosomes	Chromosome Copy Number	Form of Chromosome(s)	Genome Size (Mb)	
Prokaryotes					
<i>Mycoplasma genitalium</i>	1	1	Circular	0.58	
<i>Escherichia coli</i> K-12	1	1	Circular	4.6	
<i>Agrobacterium tumefaciens</i>	4	1	3 circular 1 linear	5.67	
<i>Sinorhizobium meliloti</i>	3	1	Circular	6.7	
Eukaryotes					
<i>Saccharomyces cerevisiae</i> (budding yeast)	16	1 or 2	Linear	12.1	
<i>Schizosaccharomyces pombe</i> (fission yeast)	3	1 or 2	Linear	12.5	
<i>Caenorhabditis elegans</i> (roundworm)	6	2	Linear	97	
<i>Arabidopsis thaliana</i> (weed)	5	2	Linear	125	
<i>Drosophila melanogaster</i> (fruit fly)	4	2	Linear	180	
<i>Tetrahymena thermophilus</i> (protozoa)	Micronucleus	5	2	Linear	125
	Macronucleus	225	10–10,000	Linear	
<i>Fugu rubripes</i> (fish)	22	2	Linear	393	
<i>Mus musculus</i> (mouse)	19 + X and Y	2	Linear	2,600	
<i>Homo sapiens</i>	22 + X and Y	2	Linear	3,200	



Genome physical architecture



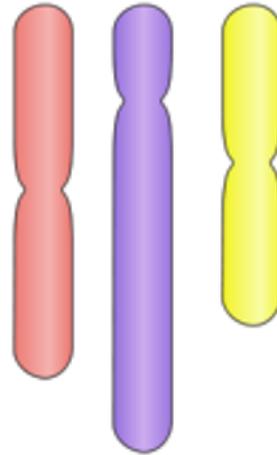
What is the advantage and disadvantages of having a single unit (chromosomes) genome?

What is the advantage and disadvantages of having multiple units (chromosomes) genome?

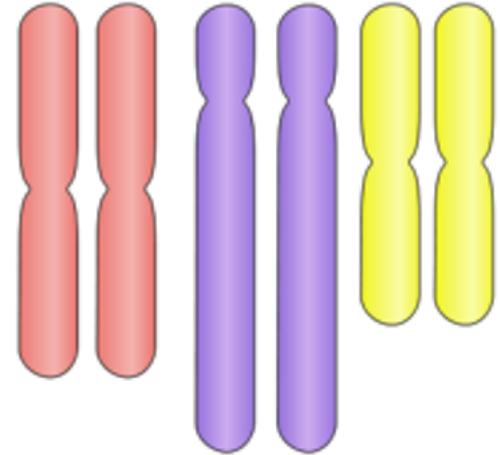
Genome physical architecture

- The number of copies of the genome within cells varies across taxa.
- What is the advantage of having multiple copies of the genome?

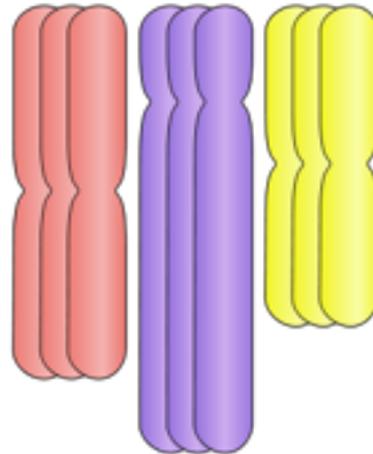
Haploid (N)



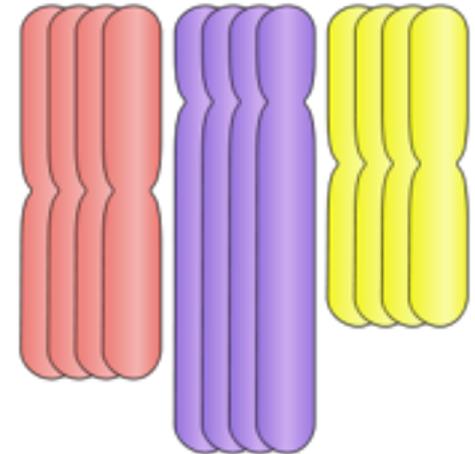
Diploid (2N)



Triploid (3N)

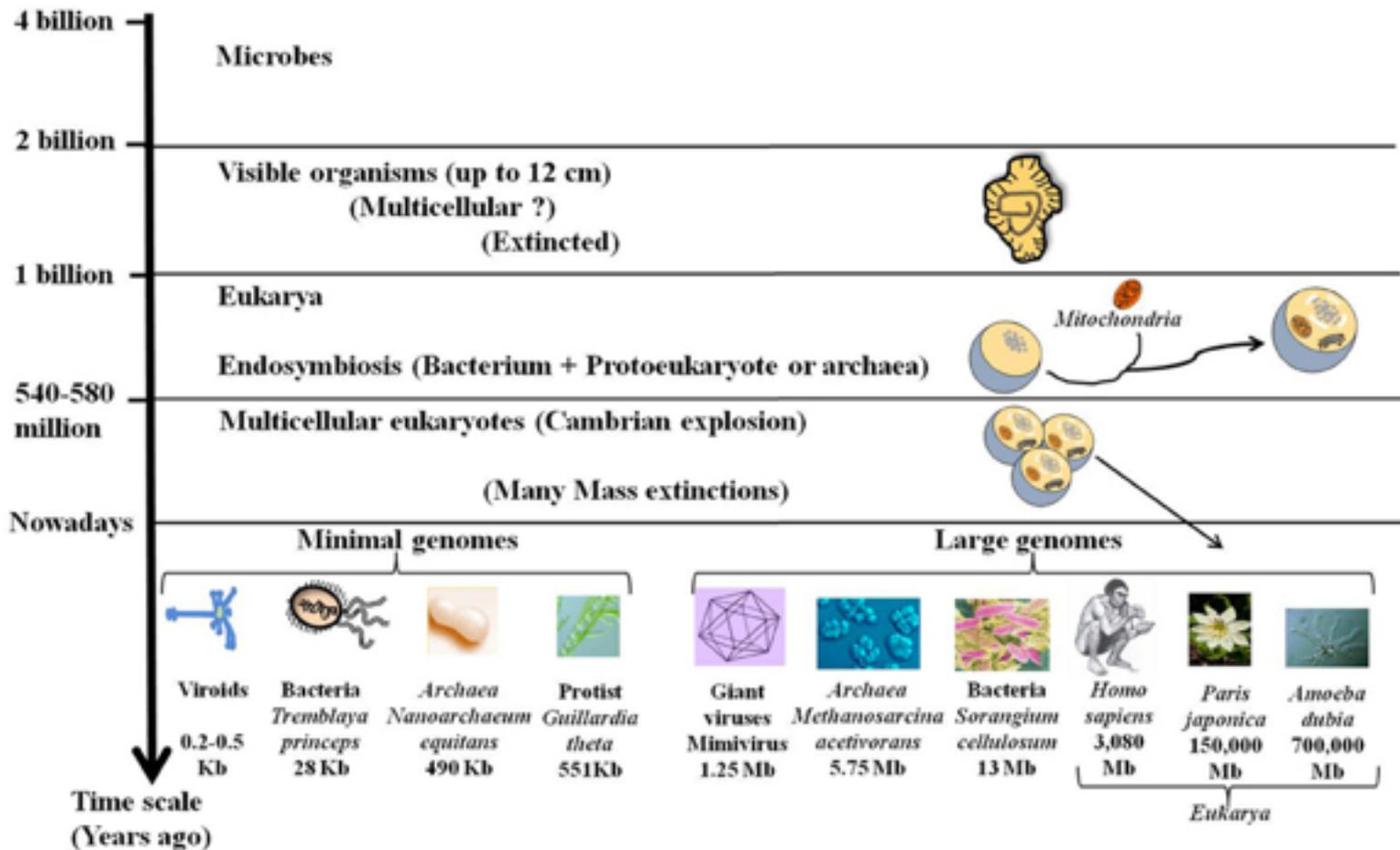


Tetraploid (4N)

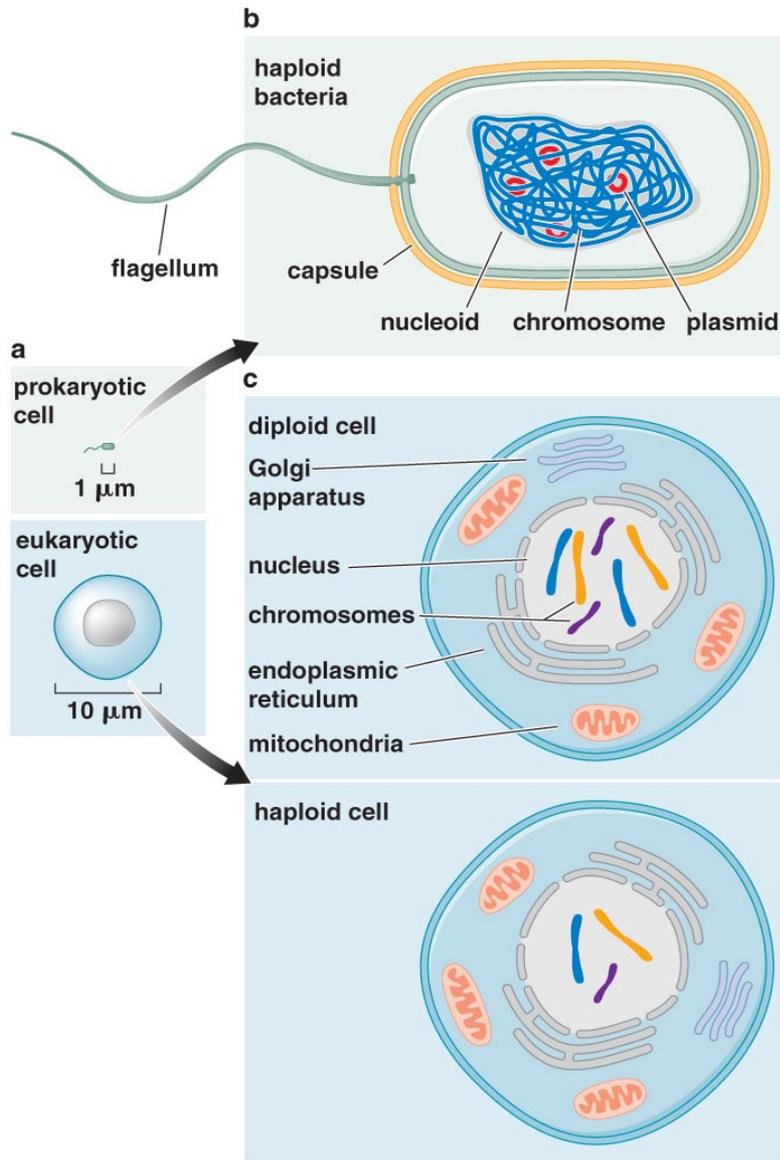


Genomes across taxa

Genomes evolved through time and genome organization evolved as well.



Genome organization



Why organizing the genome?

location location location!
Space?

How can the genetic code in a small space?

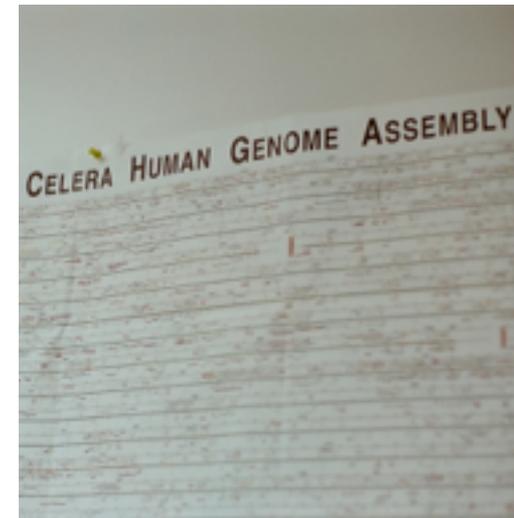
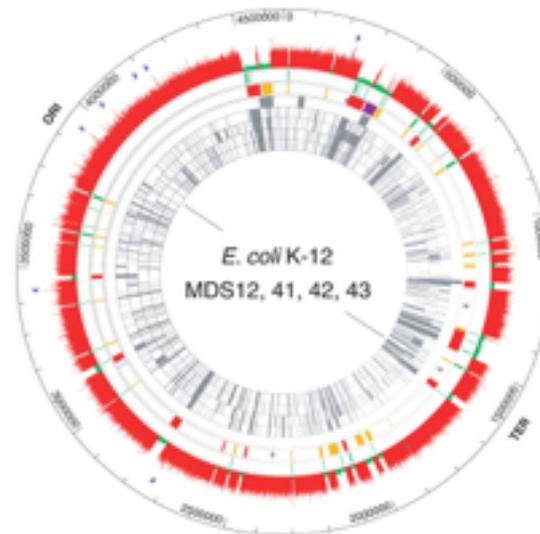
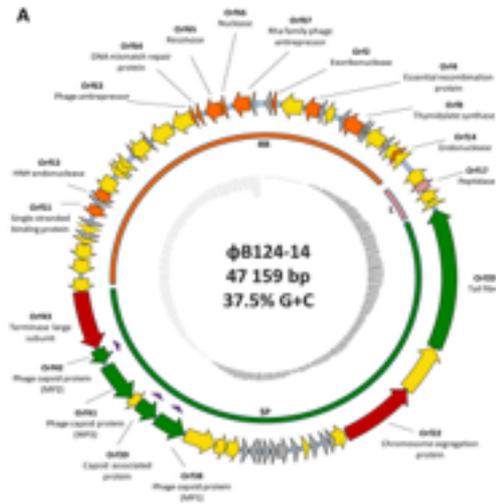
Genome organization

The characteristics of the genomes of different life forms

Viruses

Prokaryotes

Eukaryotes

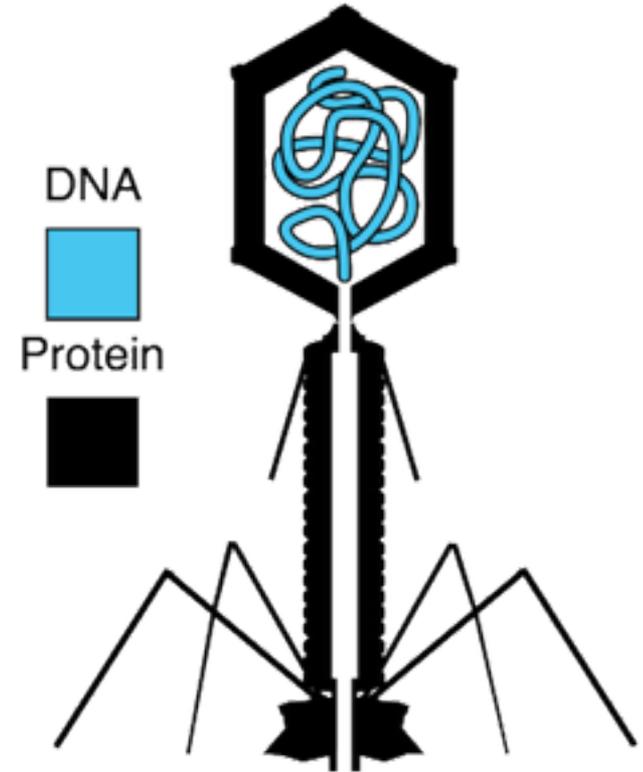


Viral genomes

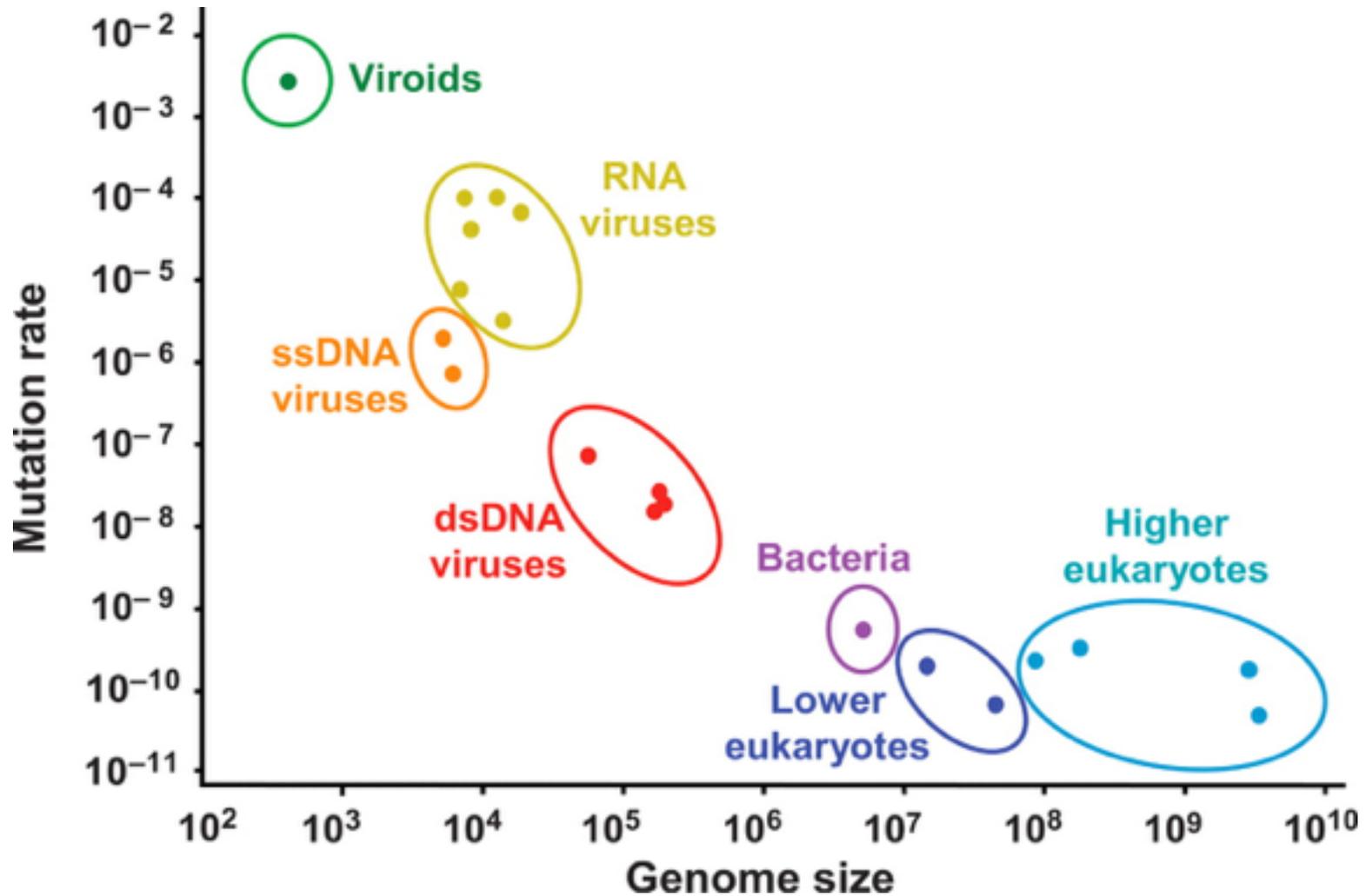
- Genome can be:
 1. single stranded DNA.
 2. double stranded DNA.
 3. single stranded RNA.
 4. double stranded RNA.
- Genome can be linear or circular.

Viral genomes

- One molecule or segmented genome (many pieces).
- Viruses with RNA genome are called retroviruses!
- Genome size 2 thousand basepairs (2Kb) – 2 million basepairs (2 Mb).
- No special organization of the genome.



Viral genomes

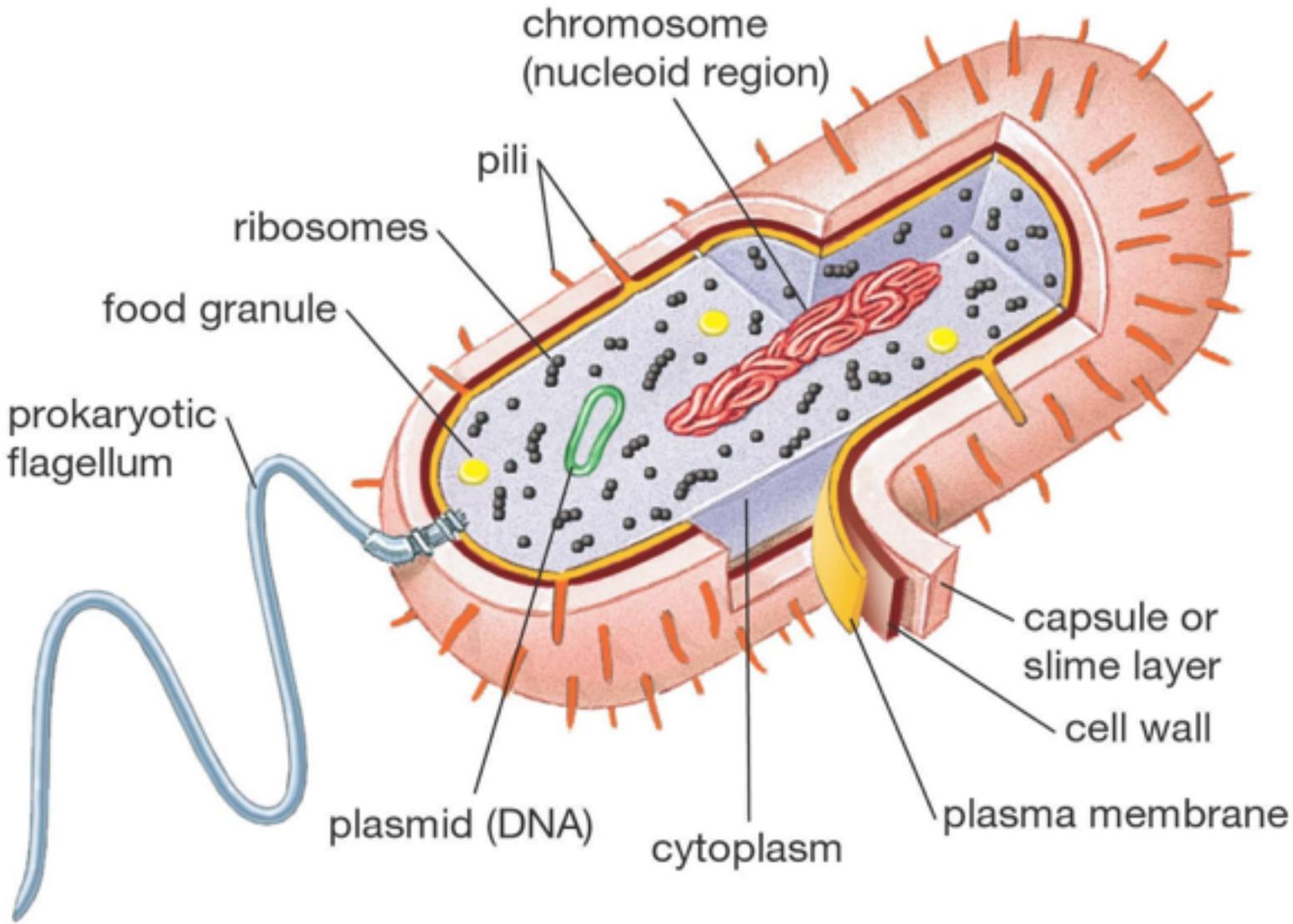


Gago S et. al., (2009). Extremely high mutation rate of a hammerhead viroid. Science 323 (5919):1308

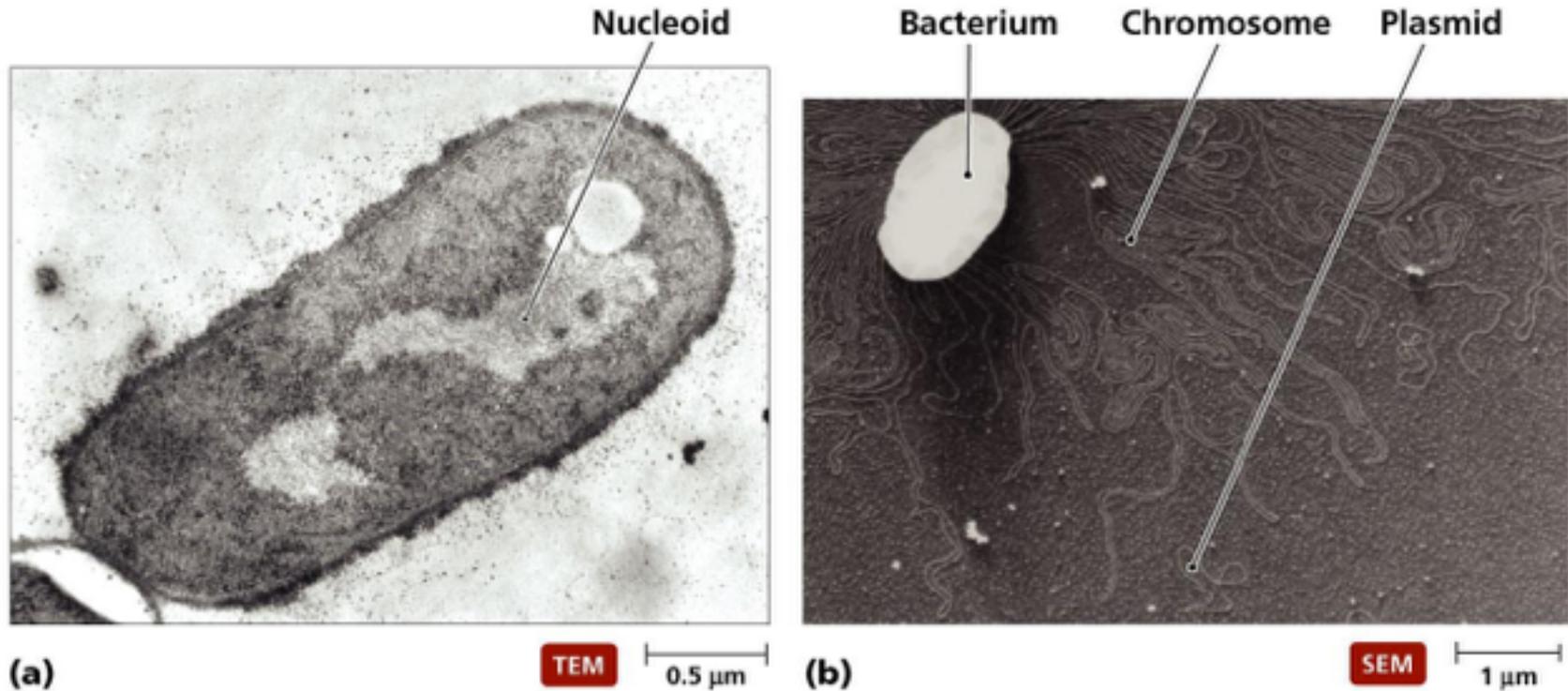
Prokaryotic genomes

- Most genomes are:
 1. Single molecule (some multiple).
 2. One circular chromosome (some linear).
 3. Double stranded DNA.
 4. Some have small additional circular DNA that can replicate independently (**plasmid**).
- Genome is organized in a structure called (**Nucleoid**).

Prokaryotic genomes



Prokaryotic genomes organization



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What are plasmids?

How big they are compared to the chromosome?

Are they independent or dependent on the chromosome?

How many copies in a single bacterium?

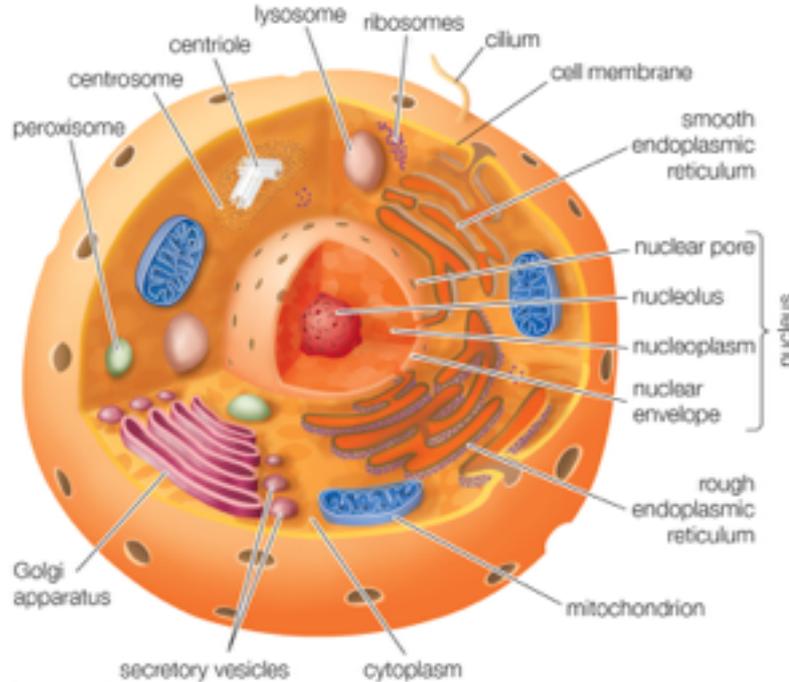
Eukaryotic genomes

Where is the eukaryotic genome located?

Eukaryotic genomes

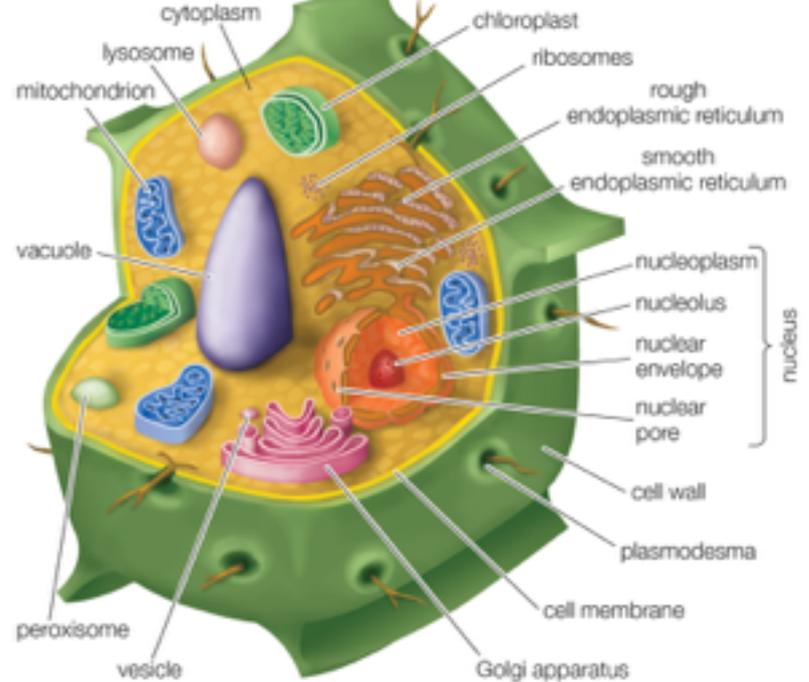
Typical animal cell and plant cell

Animal cell



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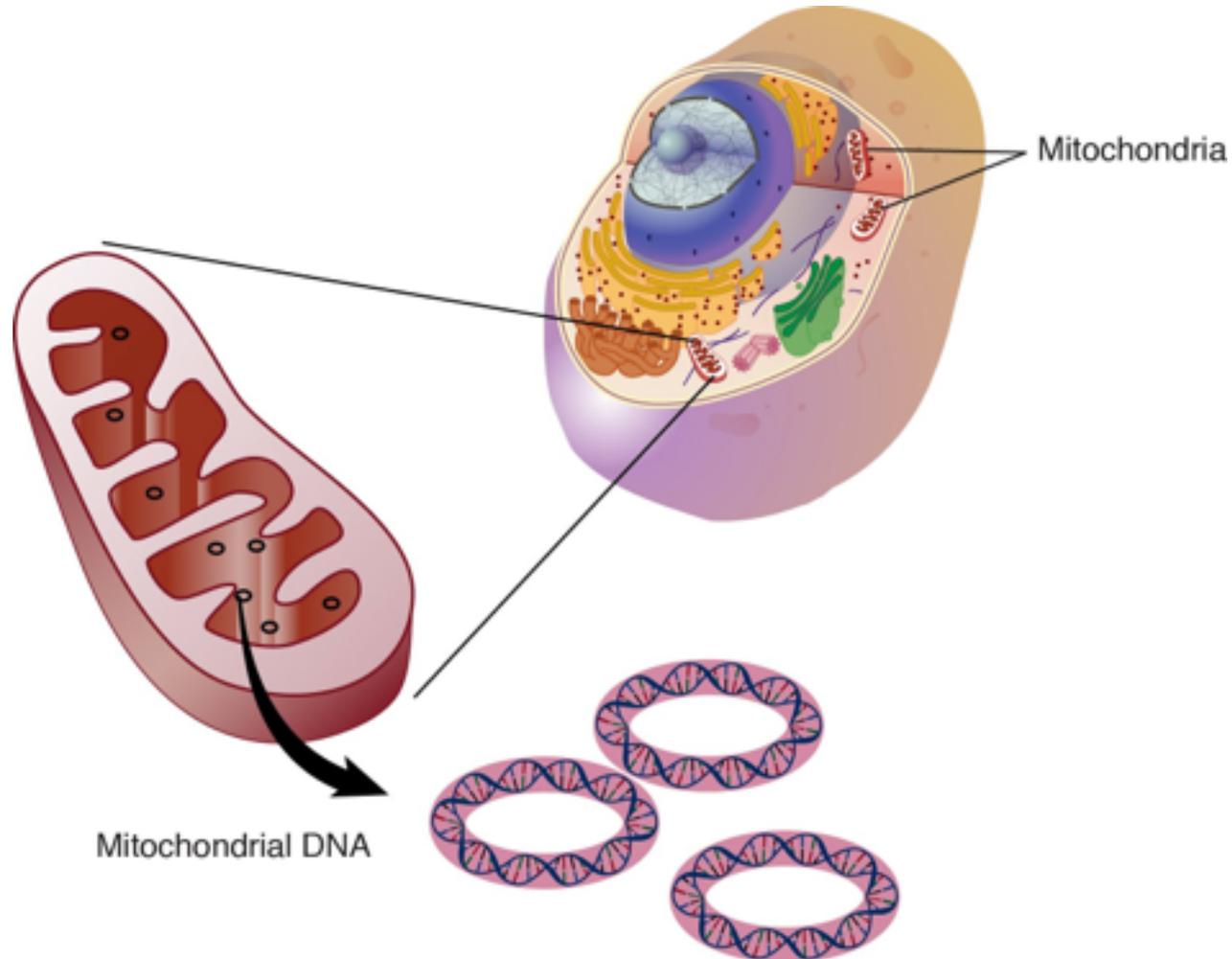
Plant cell



Nuclear genome, mitochondrial genome, chloroplast genome

Mitochondrial genome

The mitochondria has its own genetic material



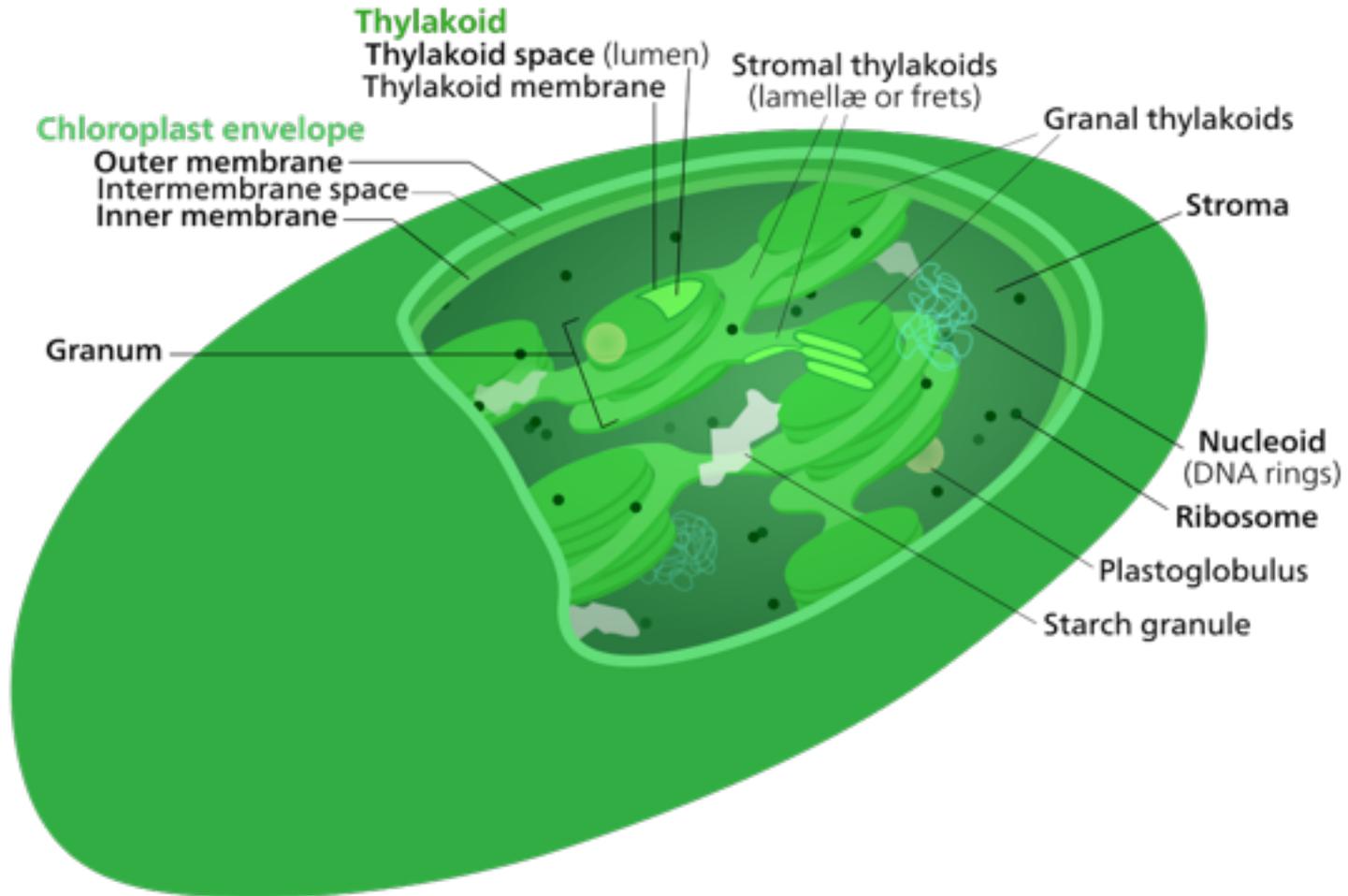


What is the function of the mitochondria?

How many copies of the genetic material a mitochondria has?

Chloroplast genome

Chloroplast has its own genetic material



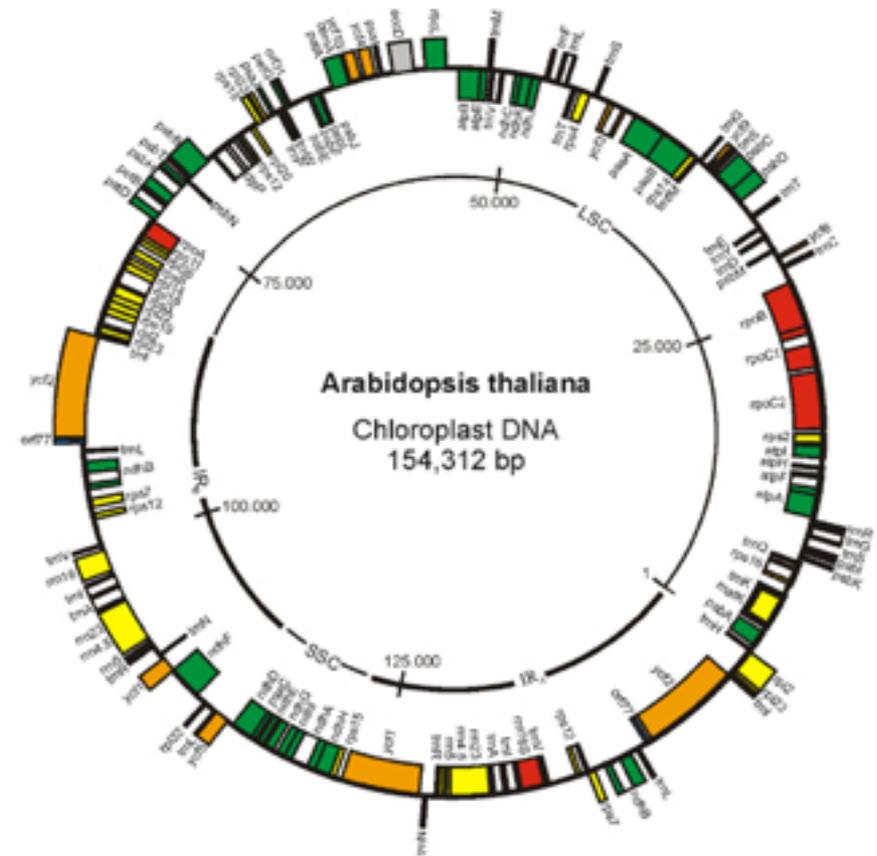


What is the function of the chloroplast?

How many copies of the genetic material a chloroplast has?

Chloroplast genome

- The chloroplast genome is a small circular one genome.
- Genome organization is similar to prokaryotic genomes.

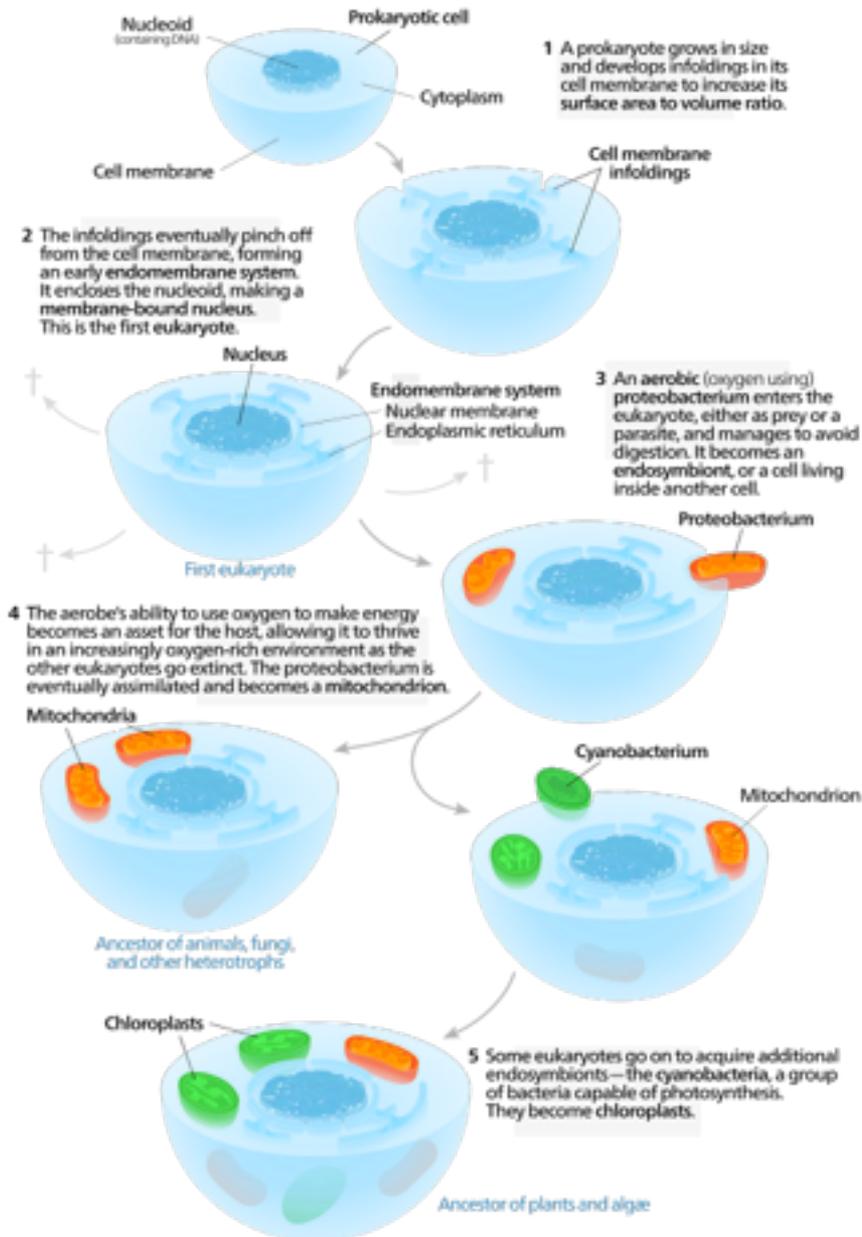


A. thaliana chloroplast DNA (inner circle: clockwise, outer: counter-clockwise). Function: transcription (red), translation (yellow), photosynthesis (green), tRNA (black), other (gray), unknown (orange). Sequence: AP000423 (see Sato et al., DNA Res 6: 283-290, 1999).



Why the mitochondria and chloroplast have similar genomes to prokaryotes?

Endosymbiosis



The mitochondrial and chloroplast are a result of an endosymbiotic relationships between various prokaryotic cells.

Aerobic proteobacterium
Cyanobacterium



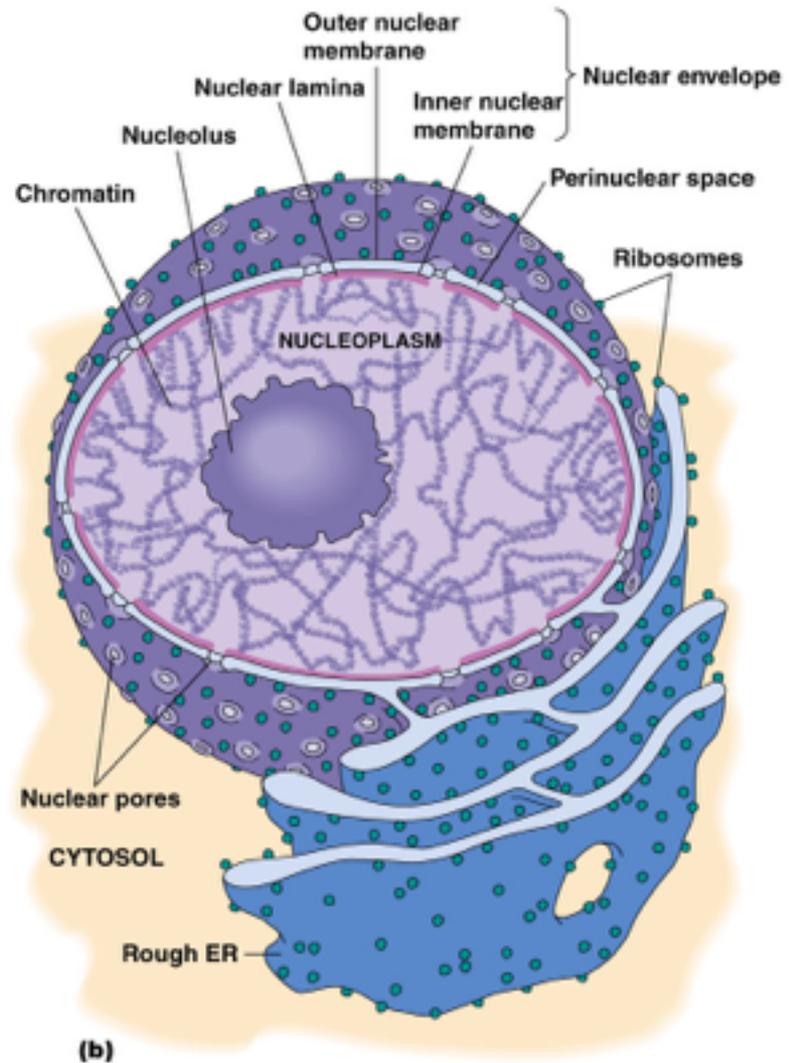
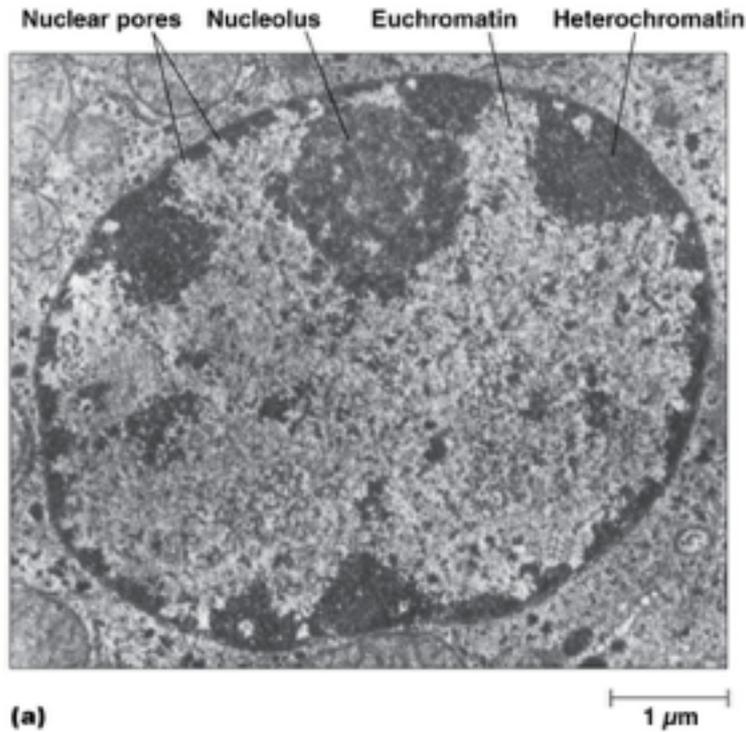
How is the mitochondrial genome organized?

How is the chloroplast genome organized?

Do plants have mitochondrial genomes?

Nuclear genome

Where is the nuclear genome located?



Nuclear genomes

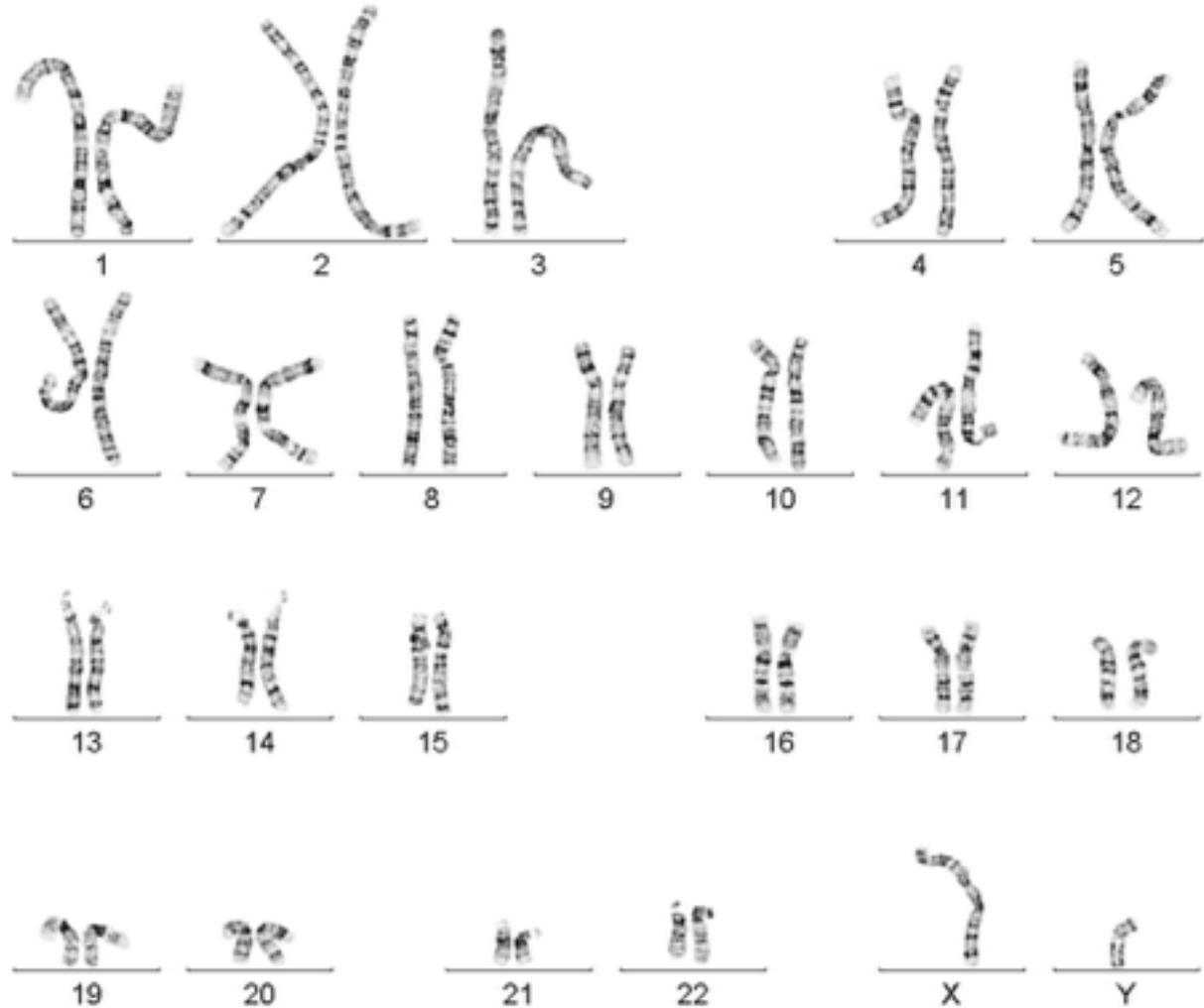
- Characteristics eukaryotic nuclear genomes are:
 1. Genome is a double stranded DNA.
 2. Genome arranged in several linear packages called **chromosomes** through interactions with several proteins.

Nuclear genomes

- The chromosomes in their most condensed form are called metaphase chromosomes.
- The entire genome represented by metaphase chromosomes is called **karyotype**.
- The number of sets of chromosomes in a given eukaryotic cell is referred to as the level of ploidy.

What is your level of ploidy?

The genome packaged into chromosomes



Karyotype

The genome packaged into chromosomes

Eukaryotic nuclear chromosomes are linear with different chromosome copy numbers

TABLE 7-1 Variation in Chromosome Makeup in Different Organisms

Species	Number of Chromosomes	Chromosome Copy Number	Form of Chromosome(s)	Genome Size (Mb)
Prokaryotes				
<i>Mycoplasma genitalium</i>	1	1	Circular	0.58
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<i>Agrobacterium tumefaciens</i>	4	1	3 circular 1 linear	5.67
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Eukaryotes				
<i>Saccharomyces cerevisiae</i> (budding yeast)	16	1 or 2	Linear	12.1
<i>Schizosaccharomyces pombe</i> (fission yeast)	3	1 or 2	Linear	12.5
<i>Caenorhabditis elegans</i> (roundworm)	6	2	Linear	97
<i>Arabidopsis thaliana</i> (weed)	5	2	Linear	125
<i>Drosophila melanogaster</i> (fruit fly)	4	2	Linear	180
<i>Tetrahymena thermophilus</i> (protozoa)	5 225	2 10–10,000	Linear Linear	125
<i>Fugu rubripes</i> (fish)	22	2	Linear	393
<i>Mus musculus</i> (mouse)	19 + X and Y	2	Linear	2,600
<i>Homo sapiens</i>	22 + X and Y	2	Linear	3,200

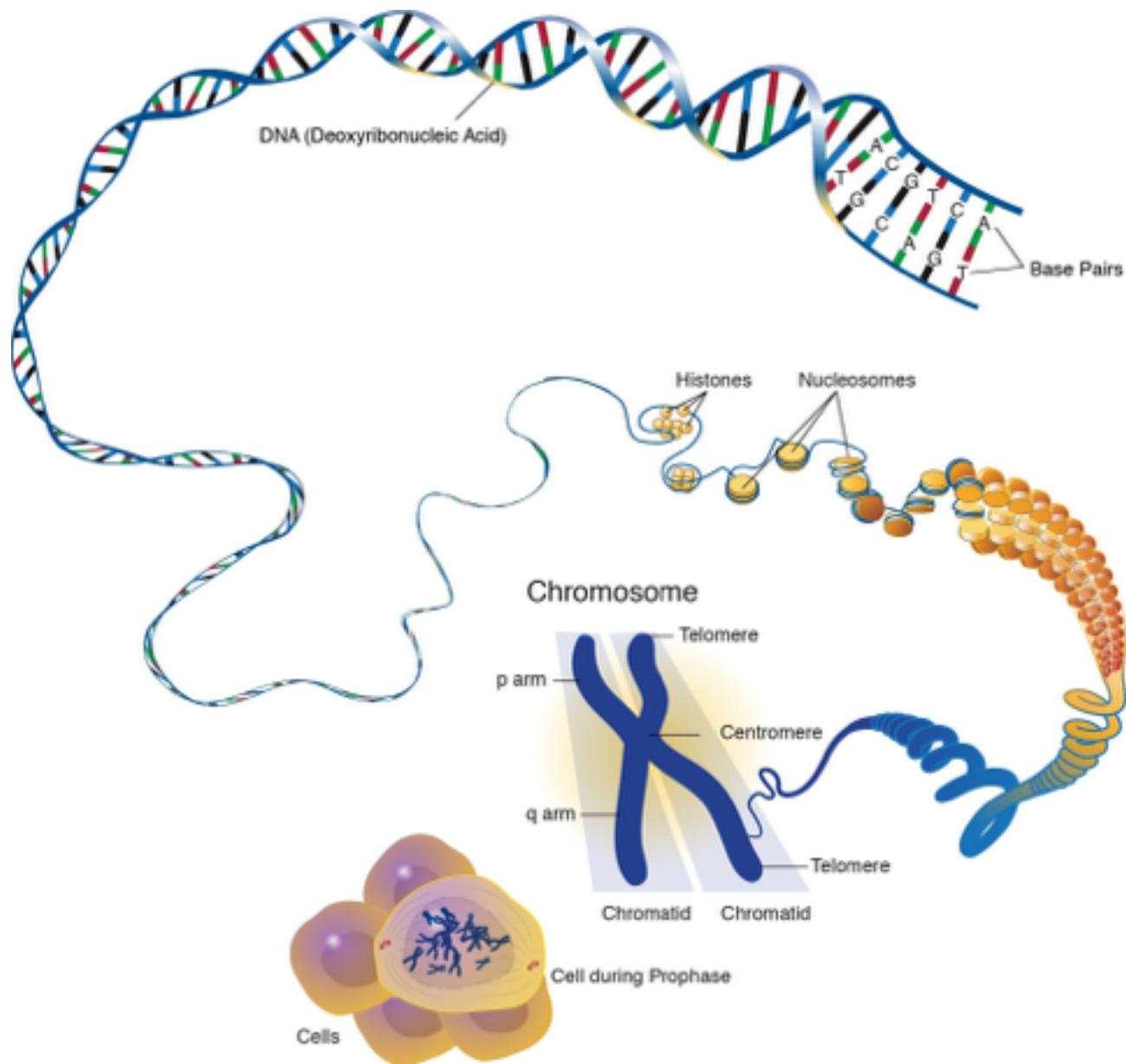




How is the nuclear genome organized and packaged?

Do you remember from last lecture?

Eukaryotic genome packaging





To think about

How can we start exploring genomes?

Can we study the whole thing at once?

Expectations

- You enjoy knowing some of the history of the words.
- You understand the general characteristics of viral, prokaryotic and eukaryotic genomes.

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