FACULTY OF ENGINEERING &TECHNOLOGY
DEPARTMENT OF BIOTECHNOLOGY

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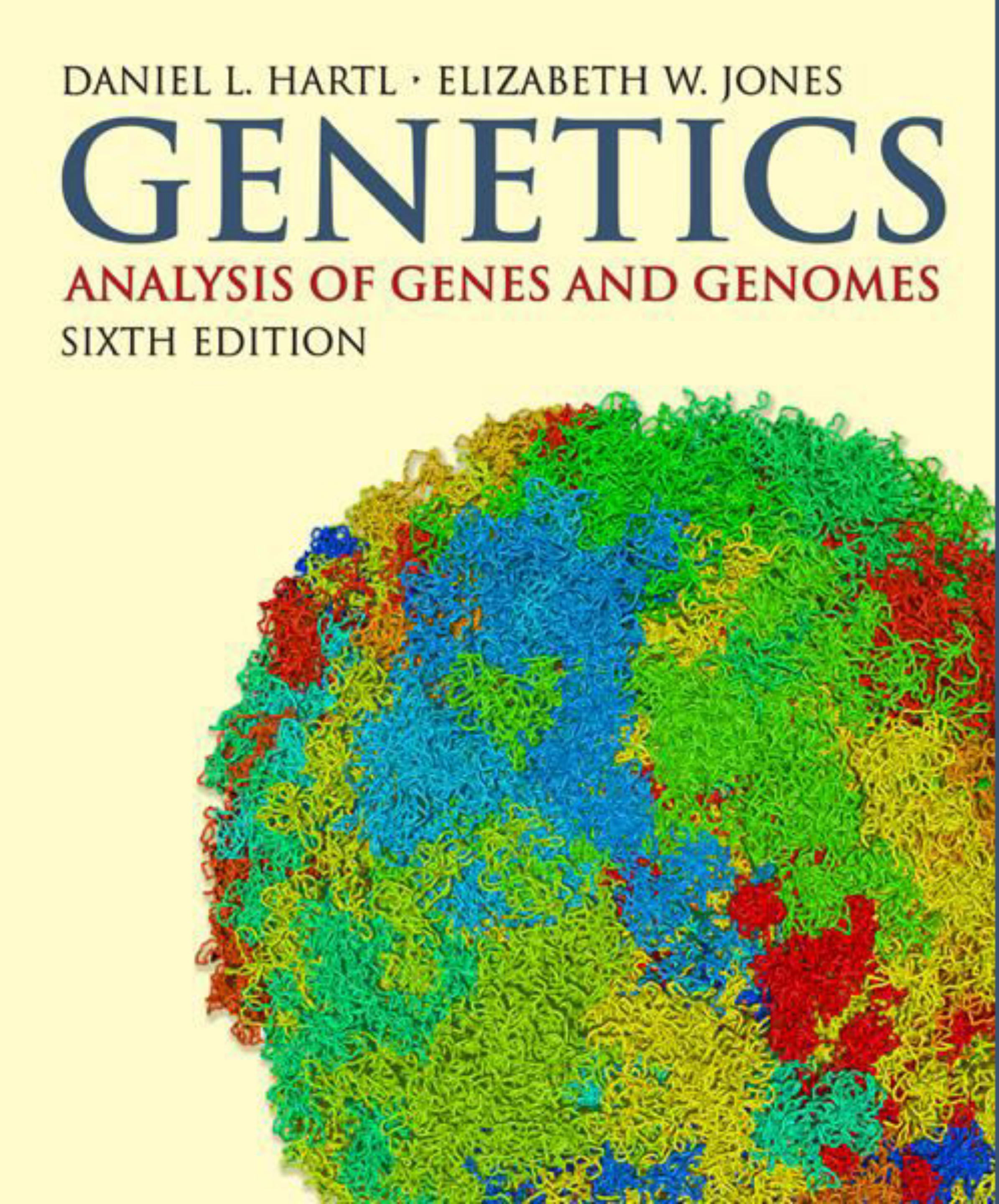
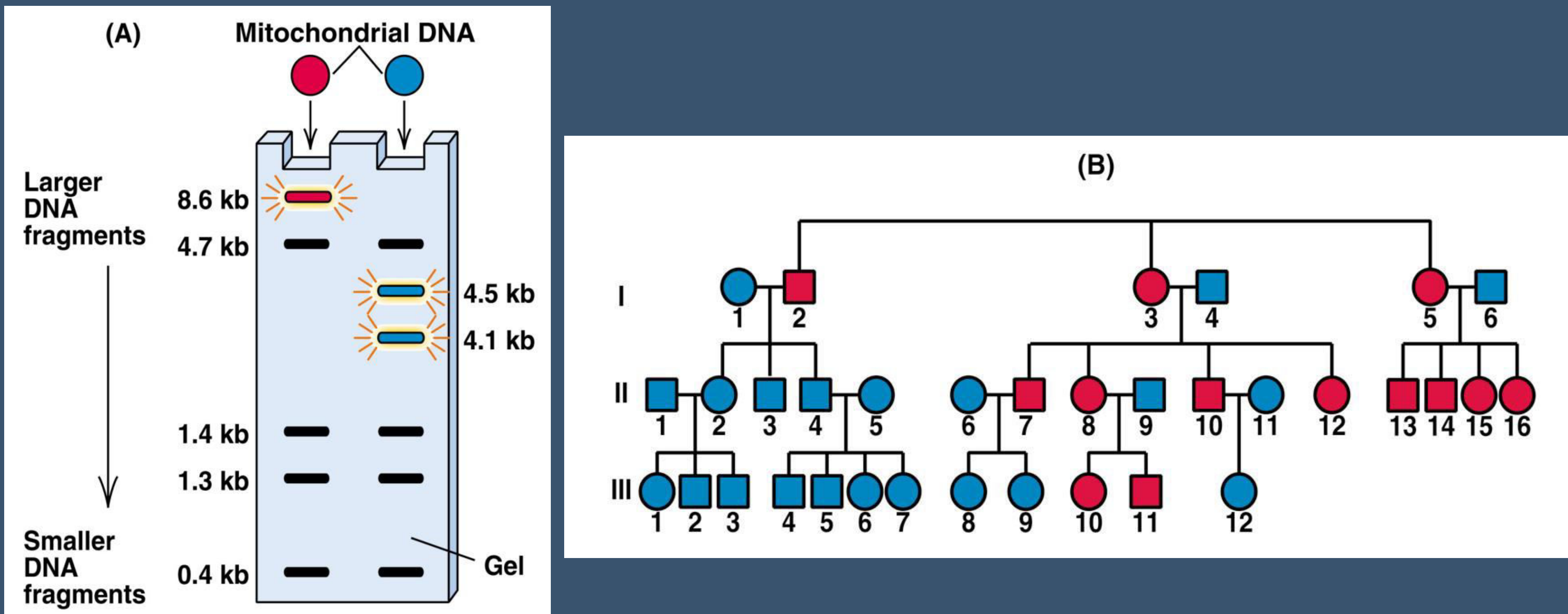


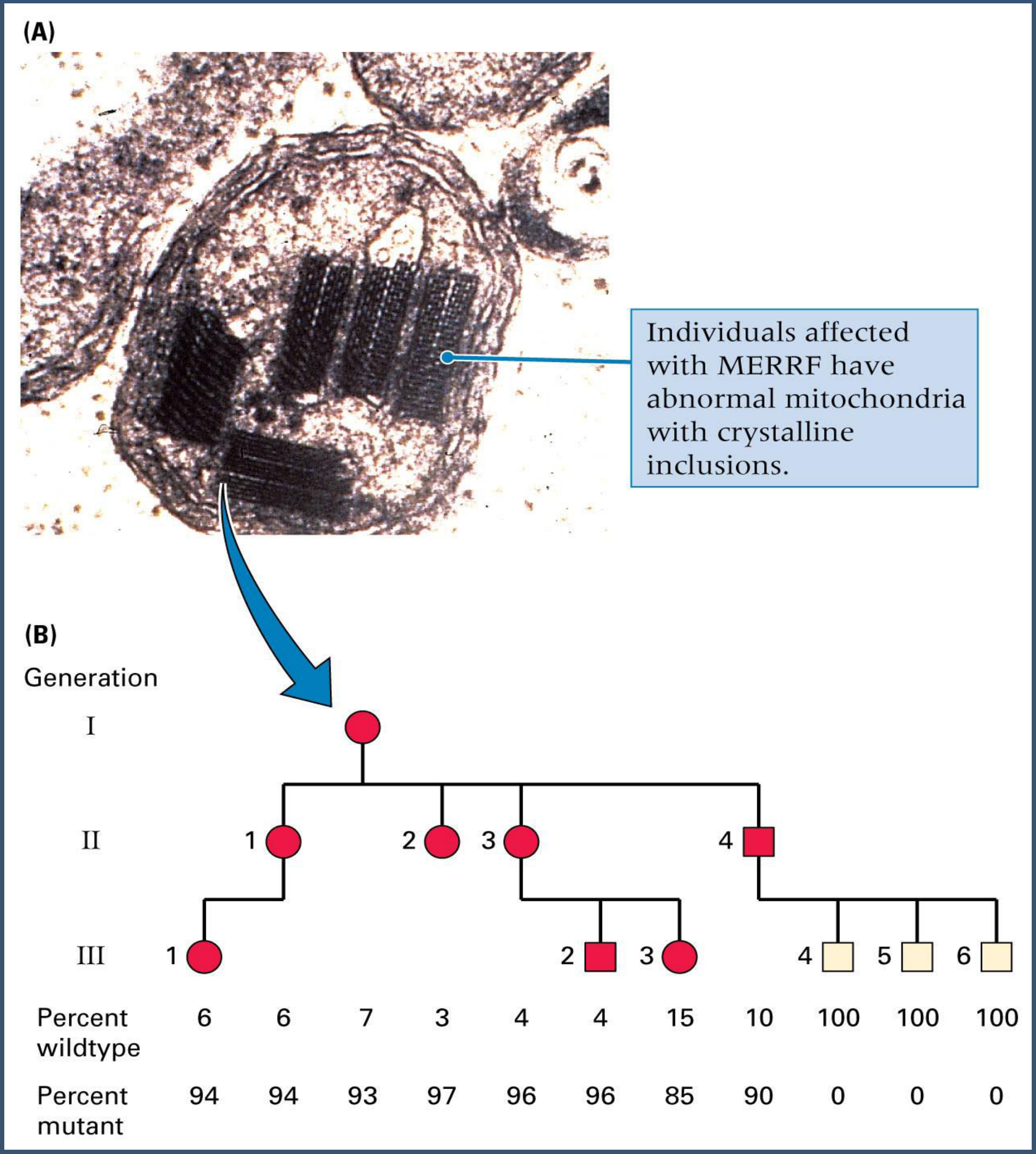
UNIT-1

Mitochondrial DNA and Extranuclear Inheritance

For cytoplasmically inherited genes, all progeny have the mother's genotype and phenotype



For cytoplasmically inherited genes, the father's genes are not passed onto progeny



Gene imprinting

- Maternal effects result from gene products inherited from the mother, in the egg cytoplasm.
- Gene imprinting – alleles of genes function differently depending on whether they come from the paternal or maternal parent. Methylation of DNA is thought to be responsible.

Cytoplasmic Organelles

- Eukaryote cells contain mitochondria (plants, fungi, and animals) and chloroplasts (plants only).
- Both have genomes organized as a single circular chromosome.
- (This is very similar to bacterial genomes.)

Mitochondria

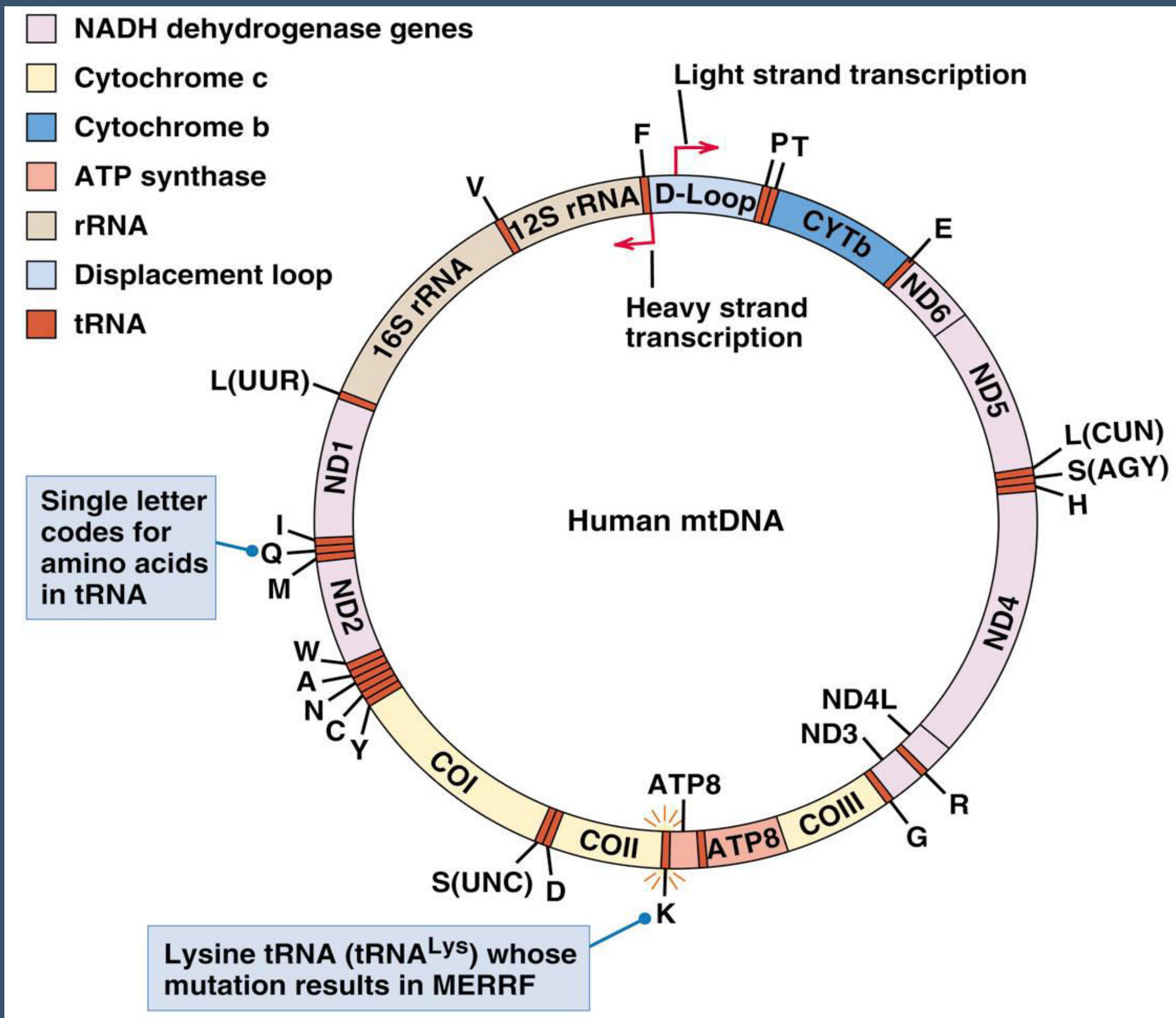
- **Animal mitochondrial genomes are 13-18 kb in size.**
- **Fungal mitochondrial genomes are ~75 kb.**
- **Higher plant mitochondrial genomes are 300-500 kb.**
- **Each mitochondrion has 5-20 copies of the mitochondrial chromosomes.**

Mitochondria

- **Human cells have a range of numbers of mitochondria:**
  - **Liver cells have 1000 mitochondria per cell.**
  - **Skin cells have 100.**
  - **Egg cells have up to 10 million.**

Human mitochondria have 37 genes.

Genes and regulatory sites in the human mitochondrial DNA

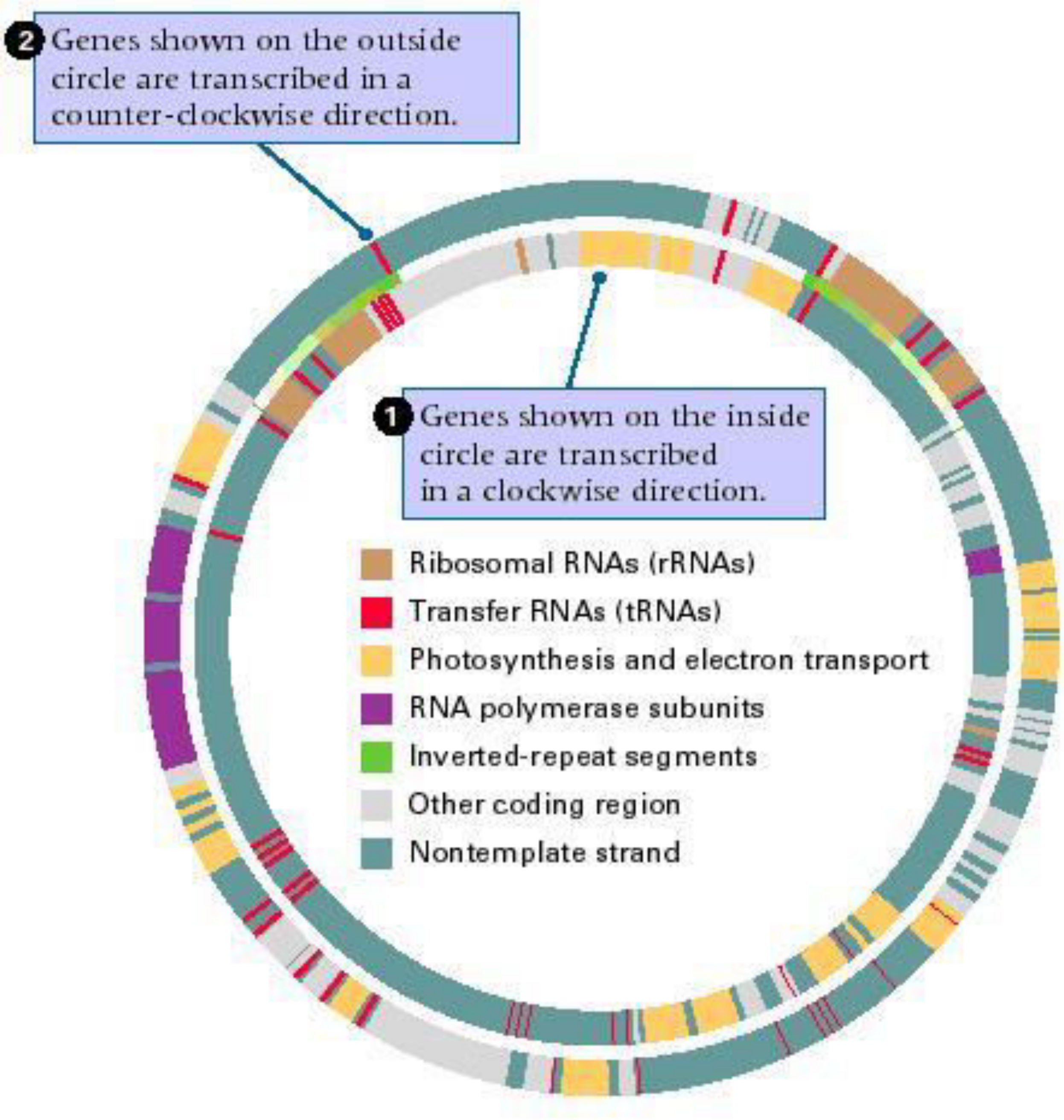


Sites of leftward and rightward transcription start are shown as red arrows. The site named D-loop at ~12 O'clock is the origin of DNA replication.

Chloroplasts

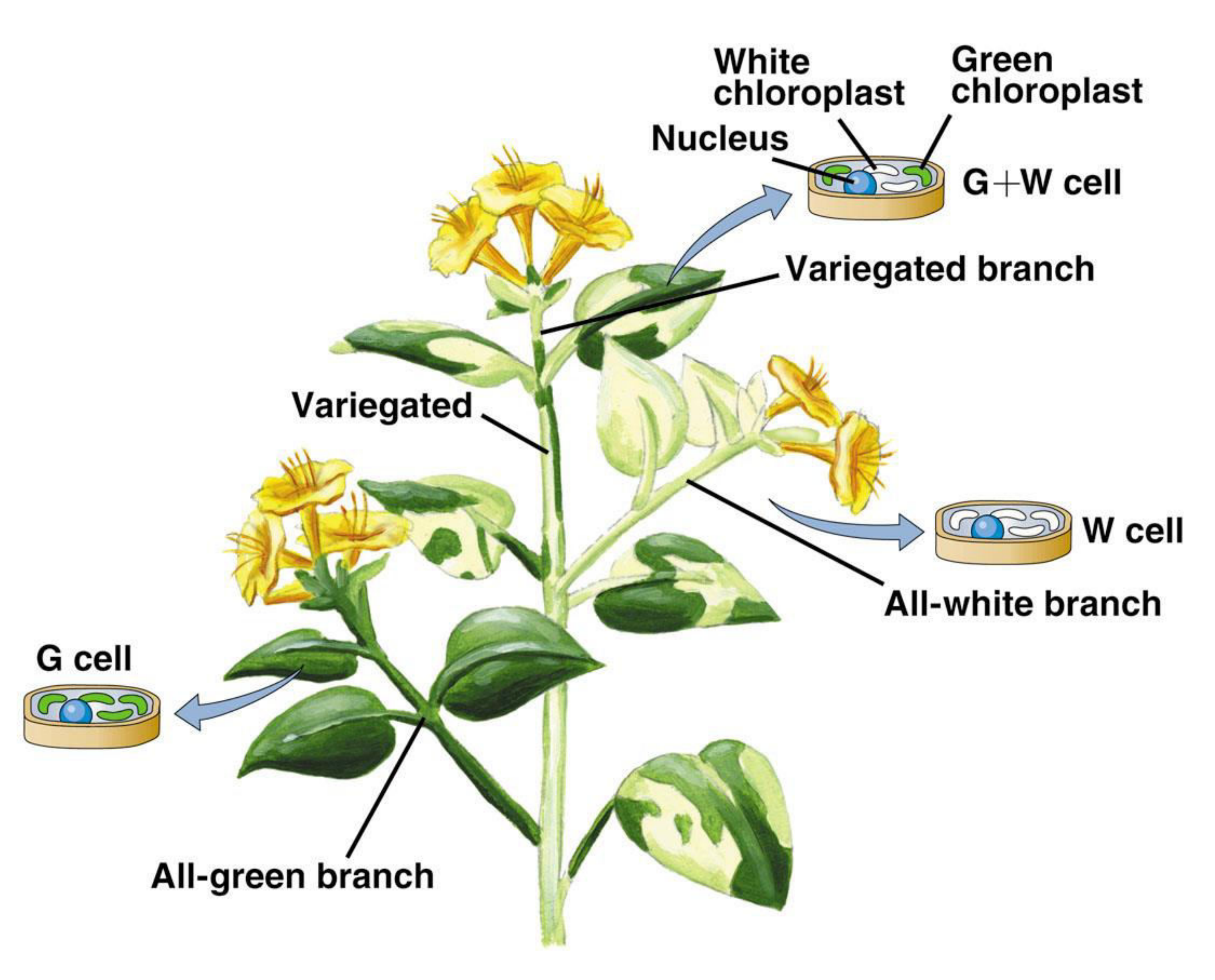
- **Chloroplast genomes are 130-150 kb in size.**
- **Chloroplasts have more genes than mitochondria (110 vs. 37). Most genes are involved in photosynthesis.**
- **Corn has 20-40 chloroplasts per cell, with each chloroplast having 20-40 chromosomes (can make up 15% of DNA)**

Genes in liverwort chloroplast DNA



Leaf variegation caused by different chloroplast genotypes

Chloroplast DNA based cytoplasmic inheritance of leaf color in Four O'clock (*Mirabilis*) plant



Inheritance of leaf variegation in *Mirabilis*

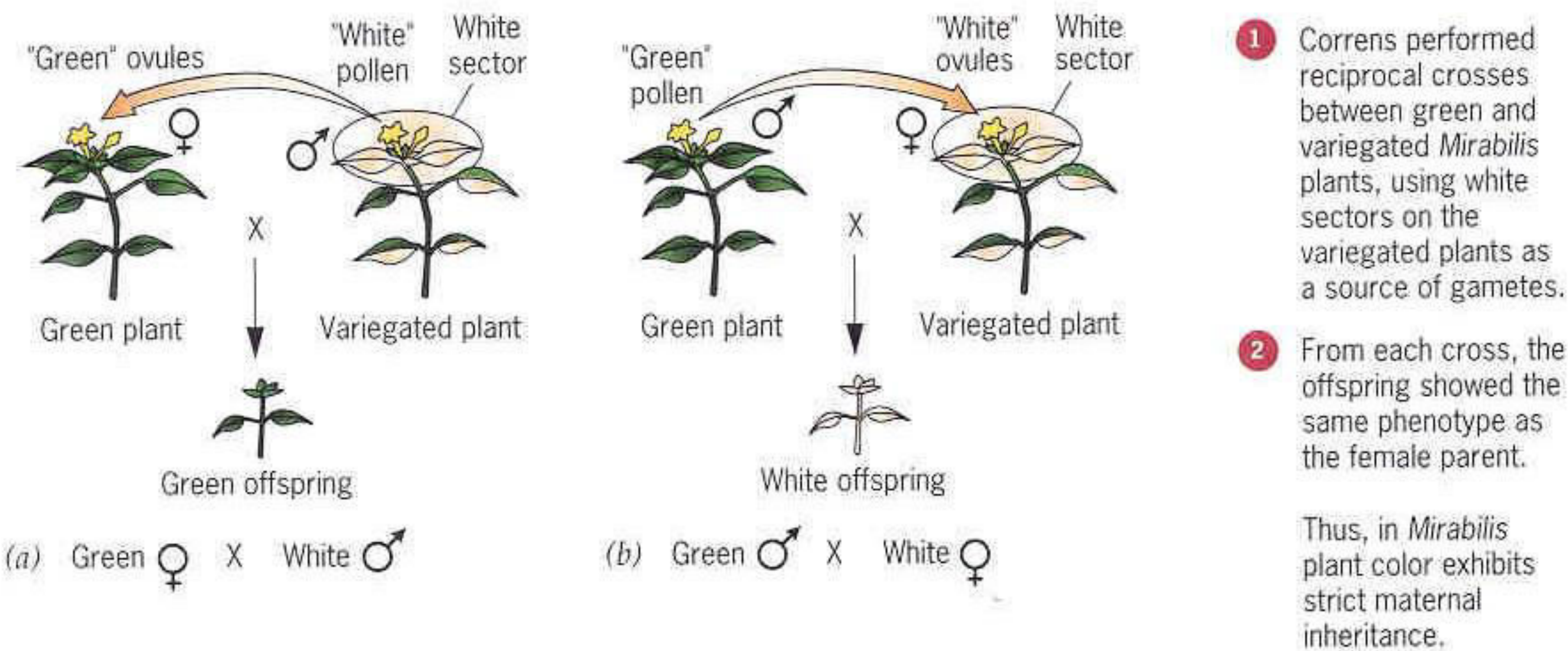


Figure 19.4 Correns' experiments on the inheritance of leaf variegation in *Mirabilis*. (*a*) Green female × white male (on a variegating plant). (*b*) Green male × white female (on a variegating plant).

Chloroplast sorting during mitosis

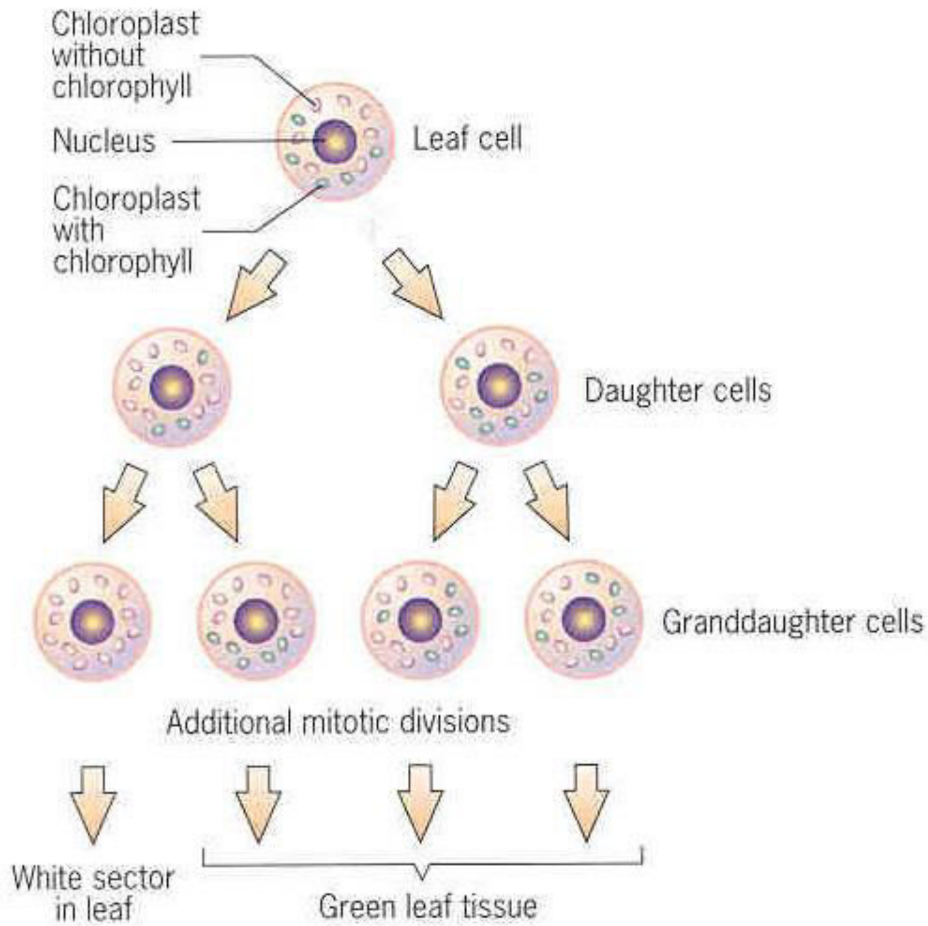
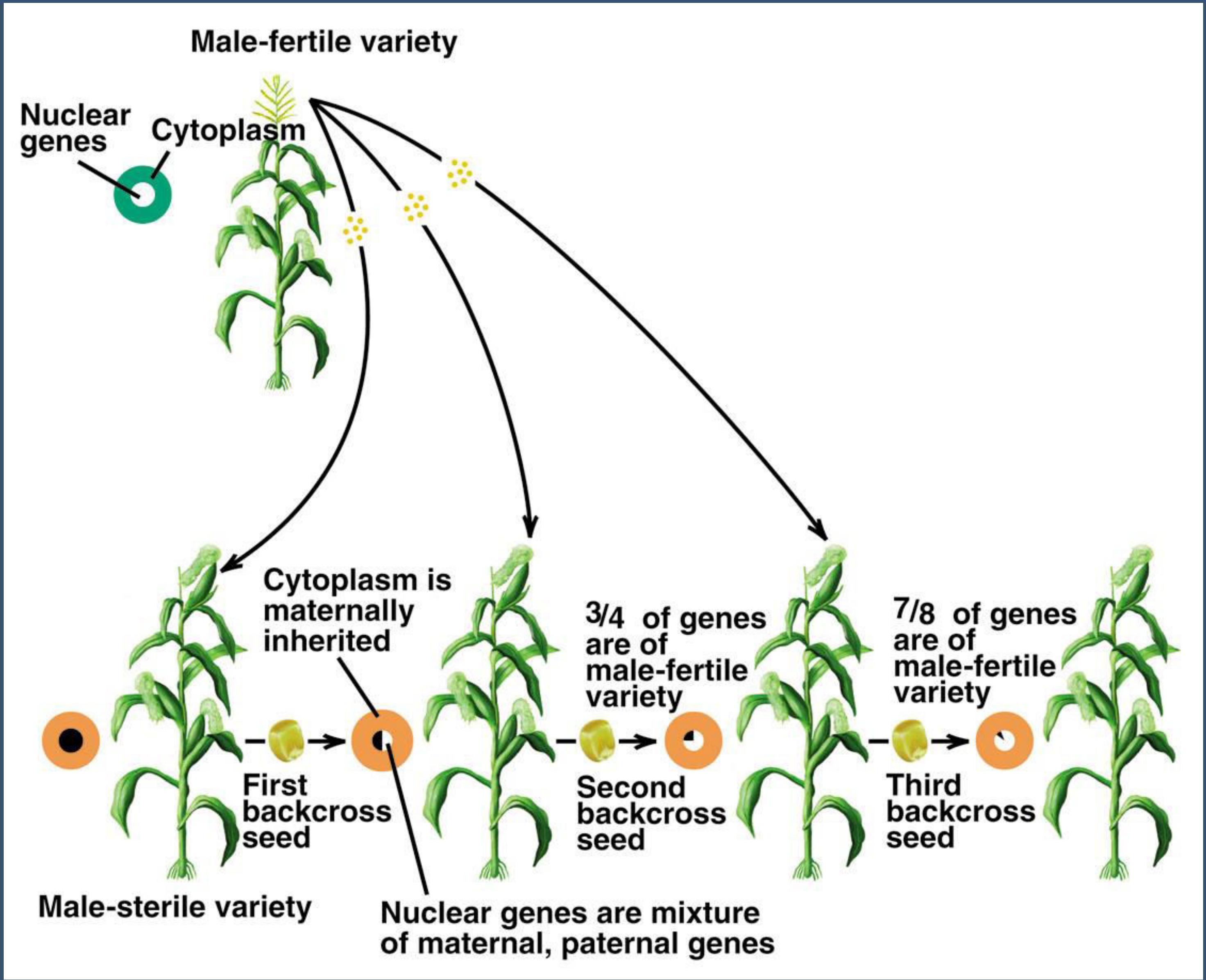


Figure 19.3 Chloroplast sorting during mitosis. The irregular distribution of chloroplasts during cell division may produce a cell that lacks chloroplasts capable of making chlorophyll. Through further divisions, such a cell will produce a white sector of tissue in an otherwise green leaf.

Use of cytoplasmic male sterility in corn breeding



Organellar genes interact with nuclear genes

- CMS in corn:

Cytoplasm	Nuclear genes	Phenotype
CMS-T	*Rf1*/+ *Rf2*/+	viable pollen
CMS-T	+/+ *Rf2*/+	male sterile
CMS-T	Rf1/+ +/+	male sterile

Mitochondria have a mutation that kills pollen.

1970 Southern corn blight epidemic – $1 billion.

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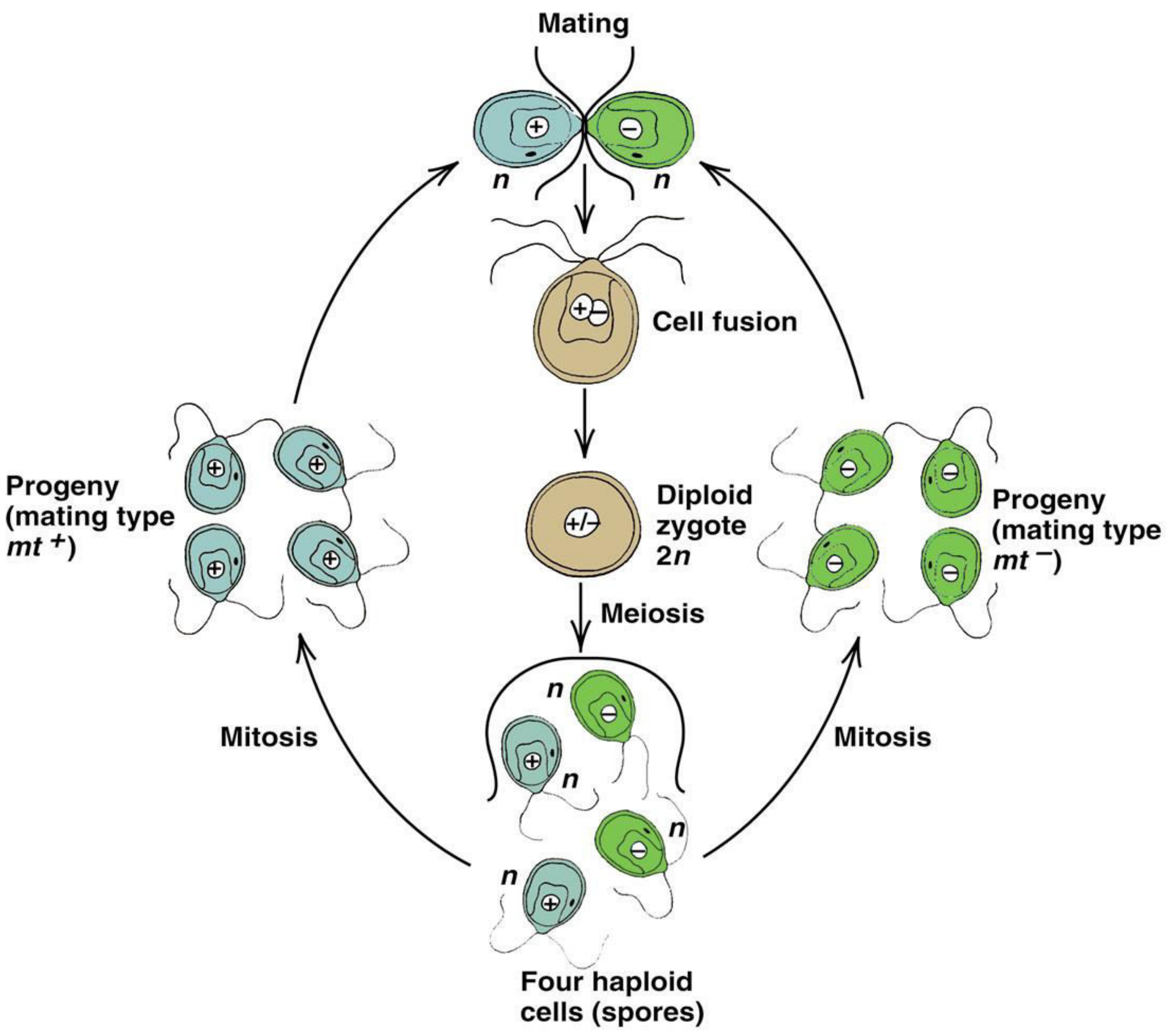
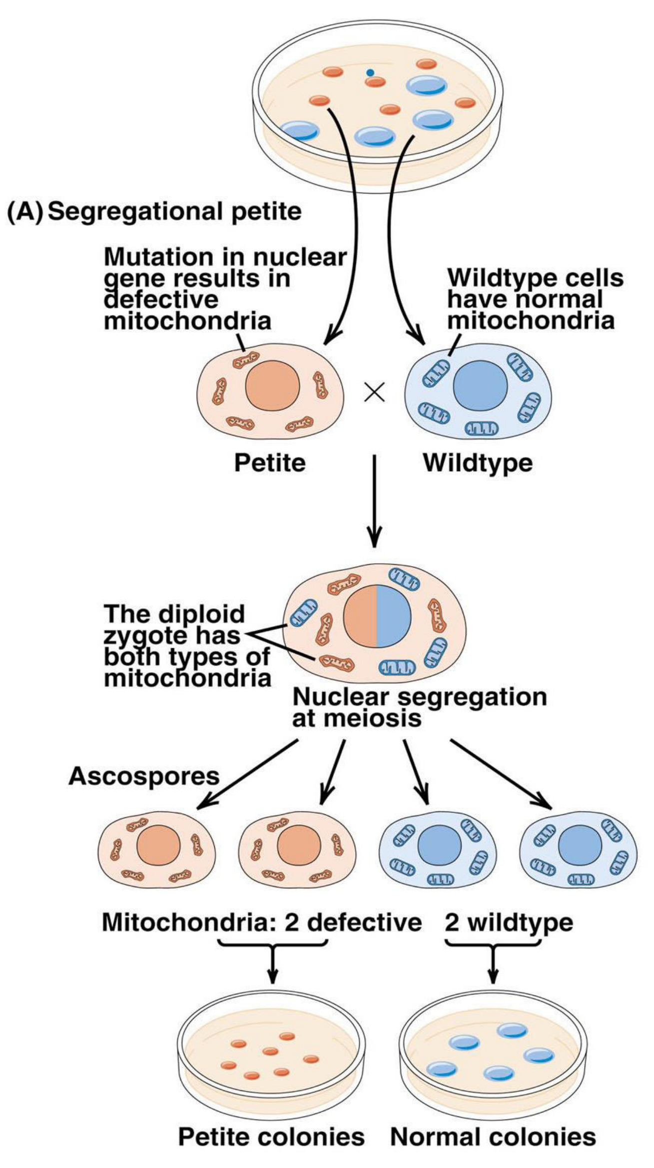
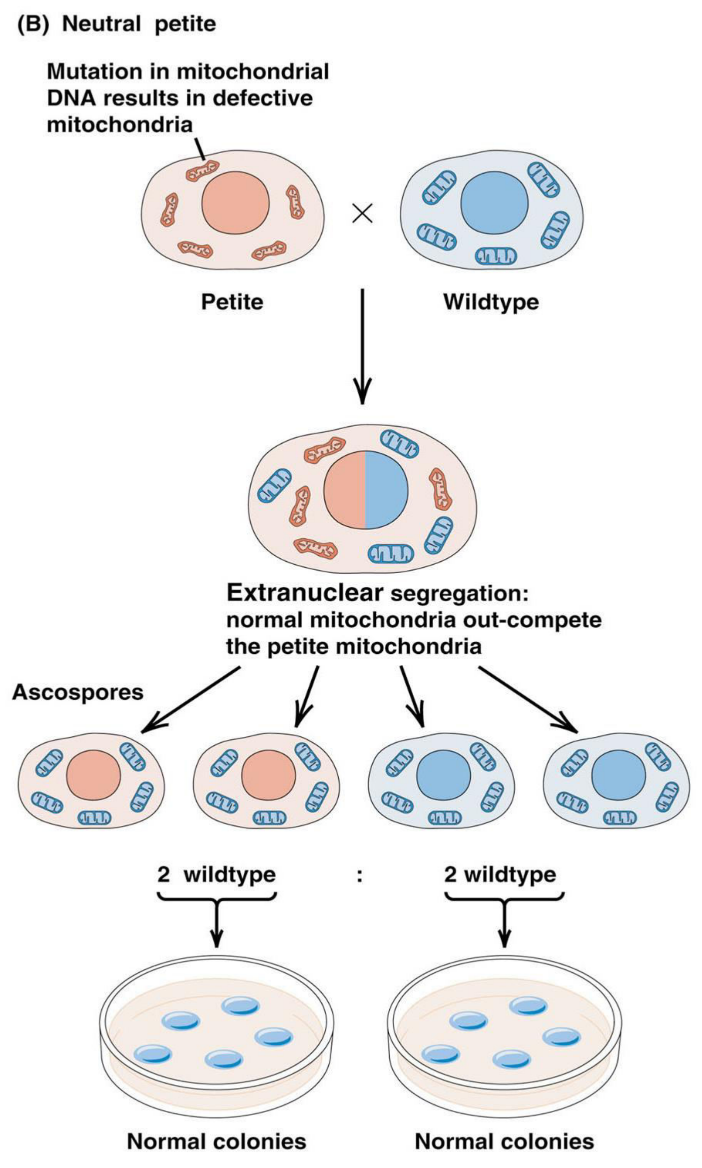
The life cycle of the unicellular green alga *Chlamydomonas*



Only one chloroplast is present per cell. Antibiotic resistance comes from mt+ cell. Mitochondria come from mt- cell. Heteroplasmic cells allow recombination between DNA in the two choloroplasts

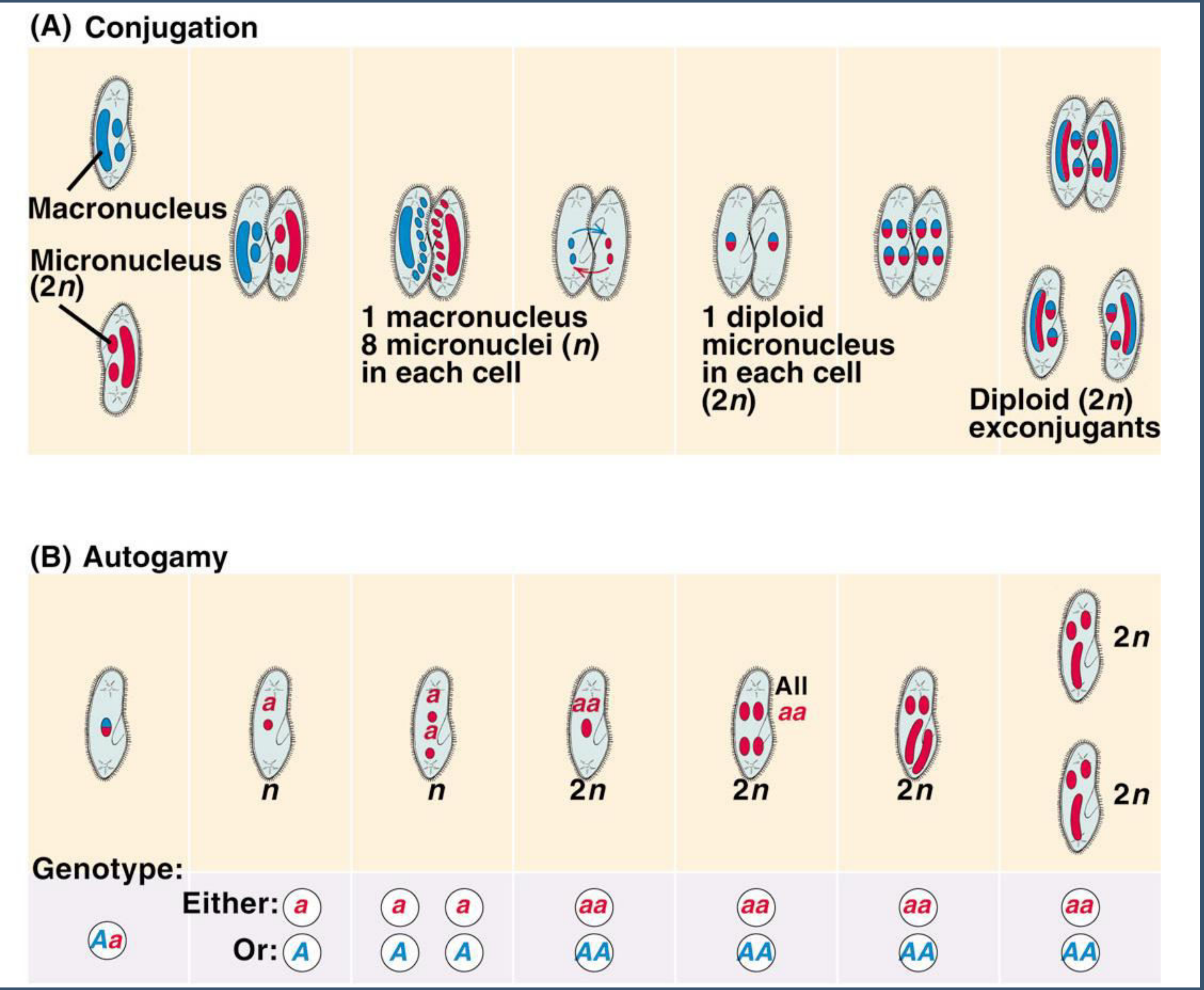
Inheritance of petite mutations in *Saccharomyces*





There are 2 kinds of petite mutations. Segregational petites are due to a mutation in the nuclear DNA. Neutral petites are due to a mutation in the mitochondrial DNA.

The life cycle of the protozoan *Paramecium* showing steps in conjugation and autogamy



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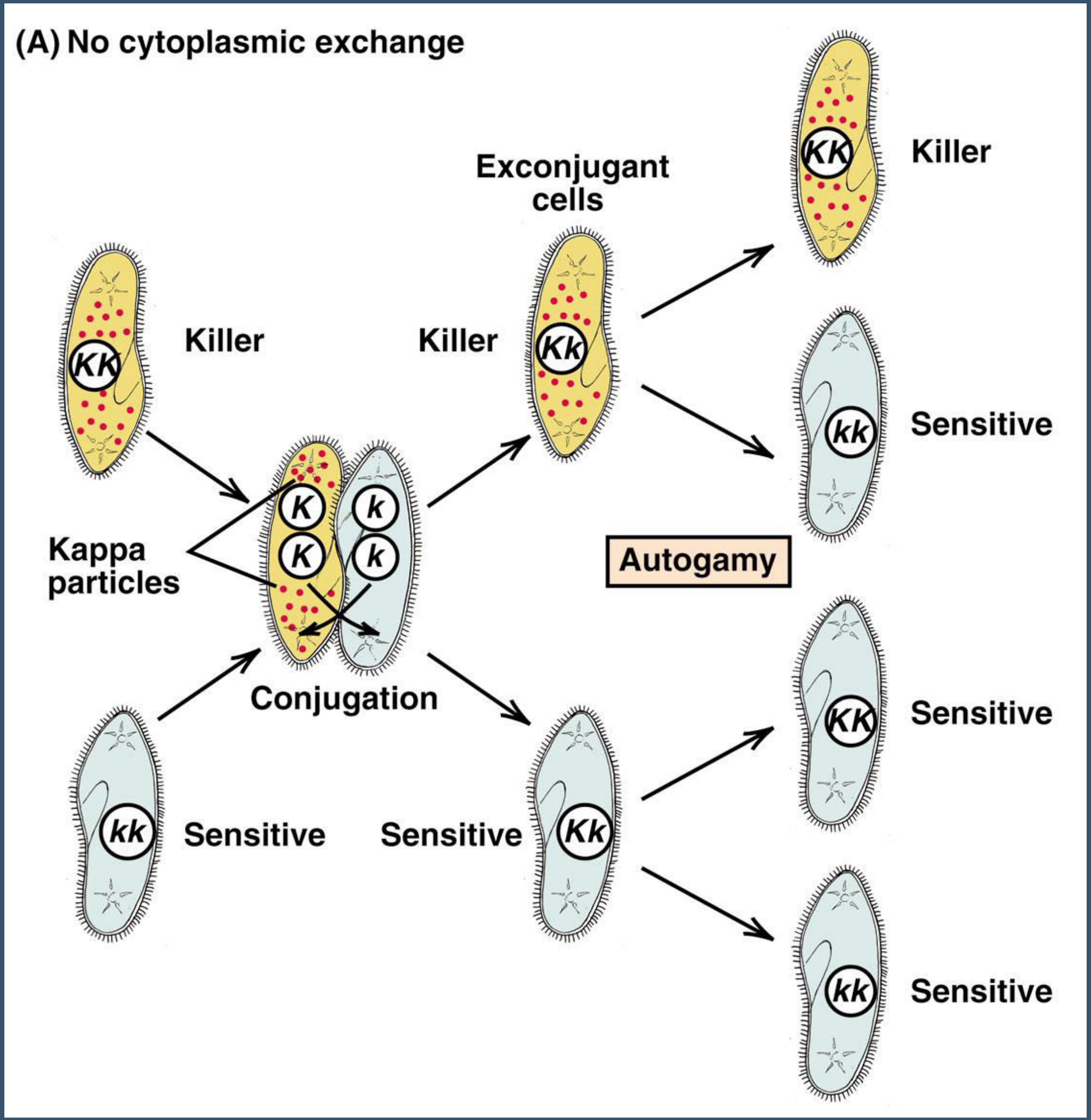
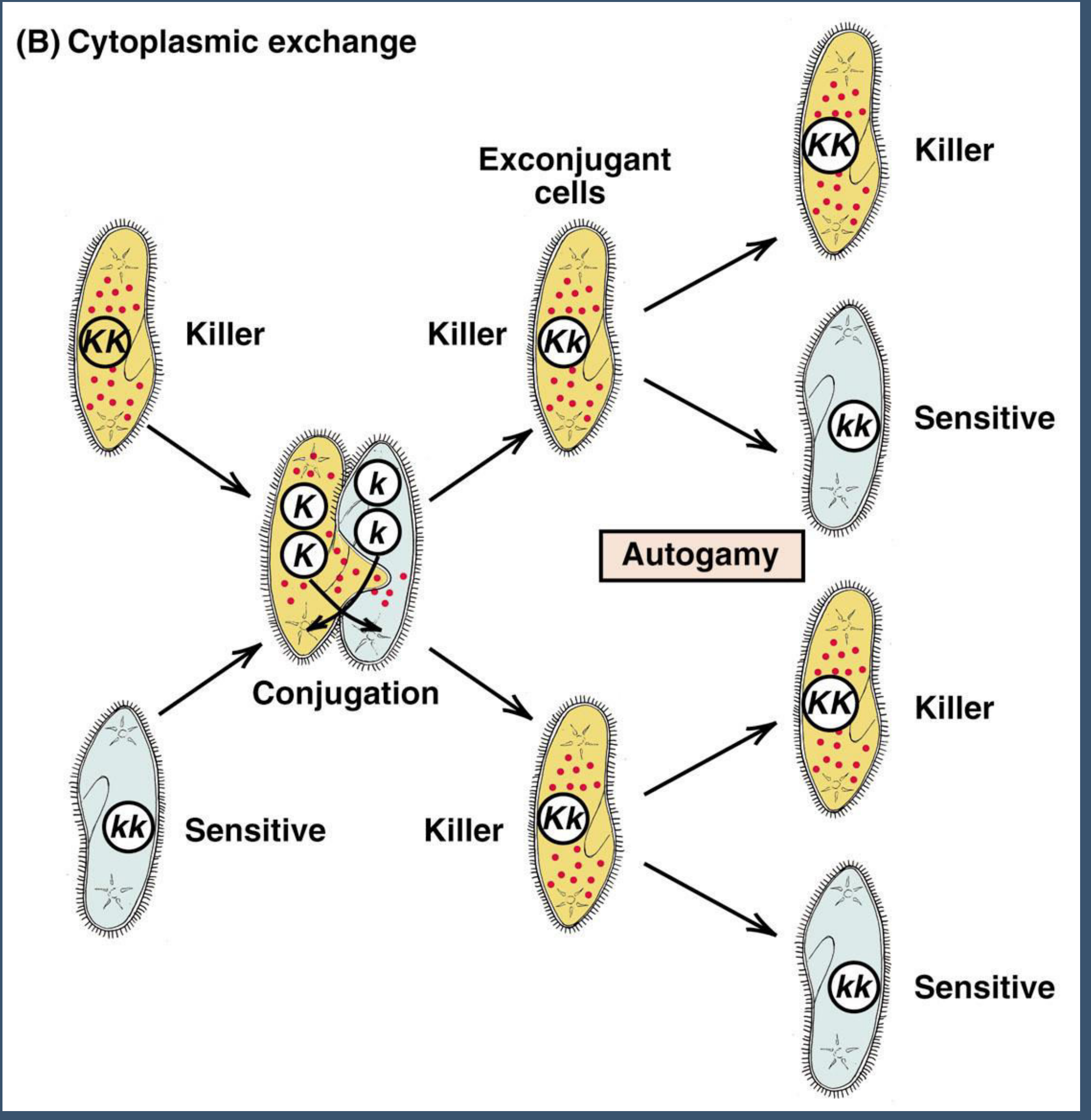
Inheritance of the killer phenotype in *Paramecium* during conjugation **without** cytoplasmic exchange



Inheritance of the killer phenotype in *Paramecium* during conjugation with cytoplasmic exchange



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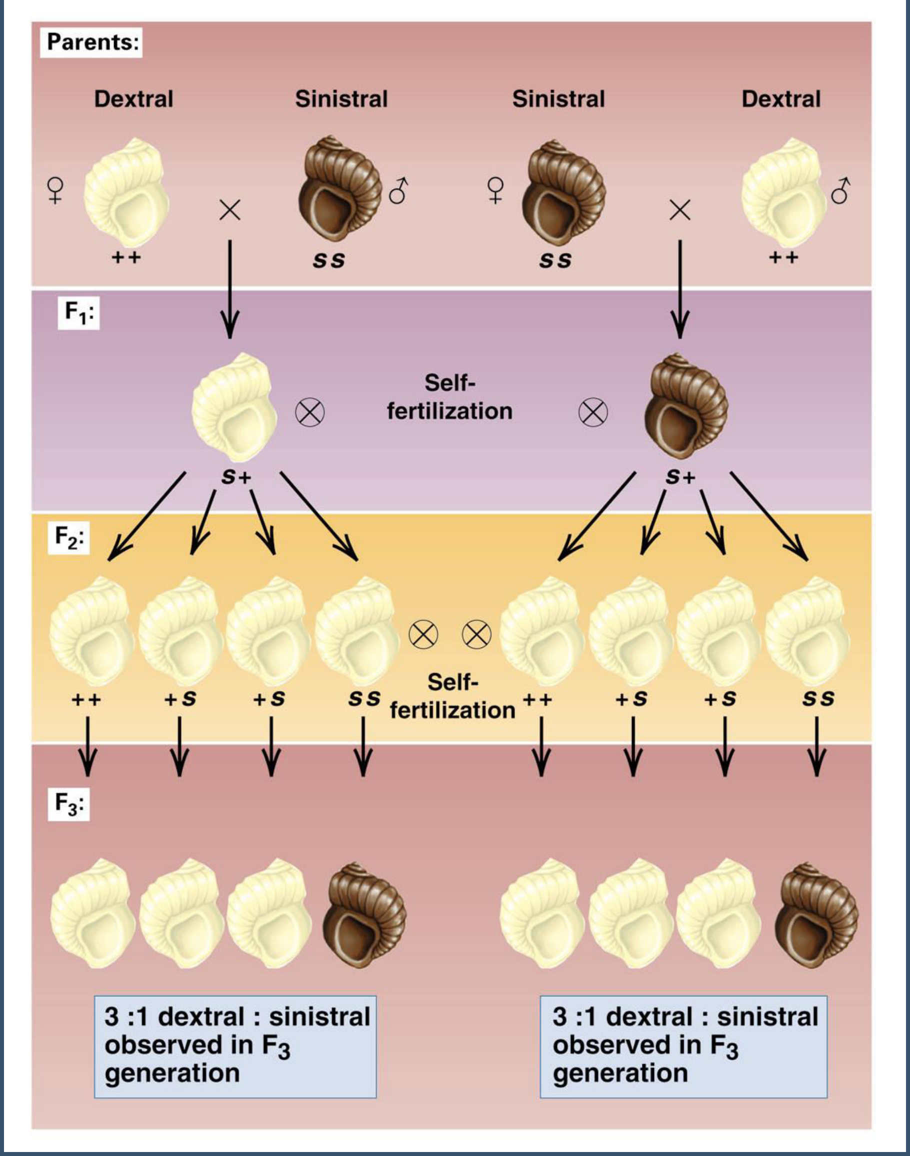
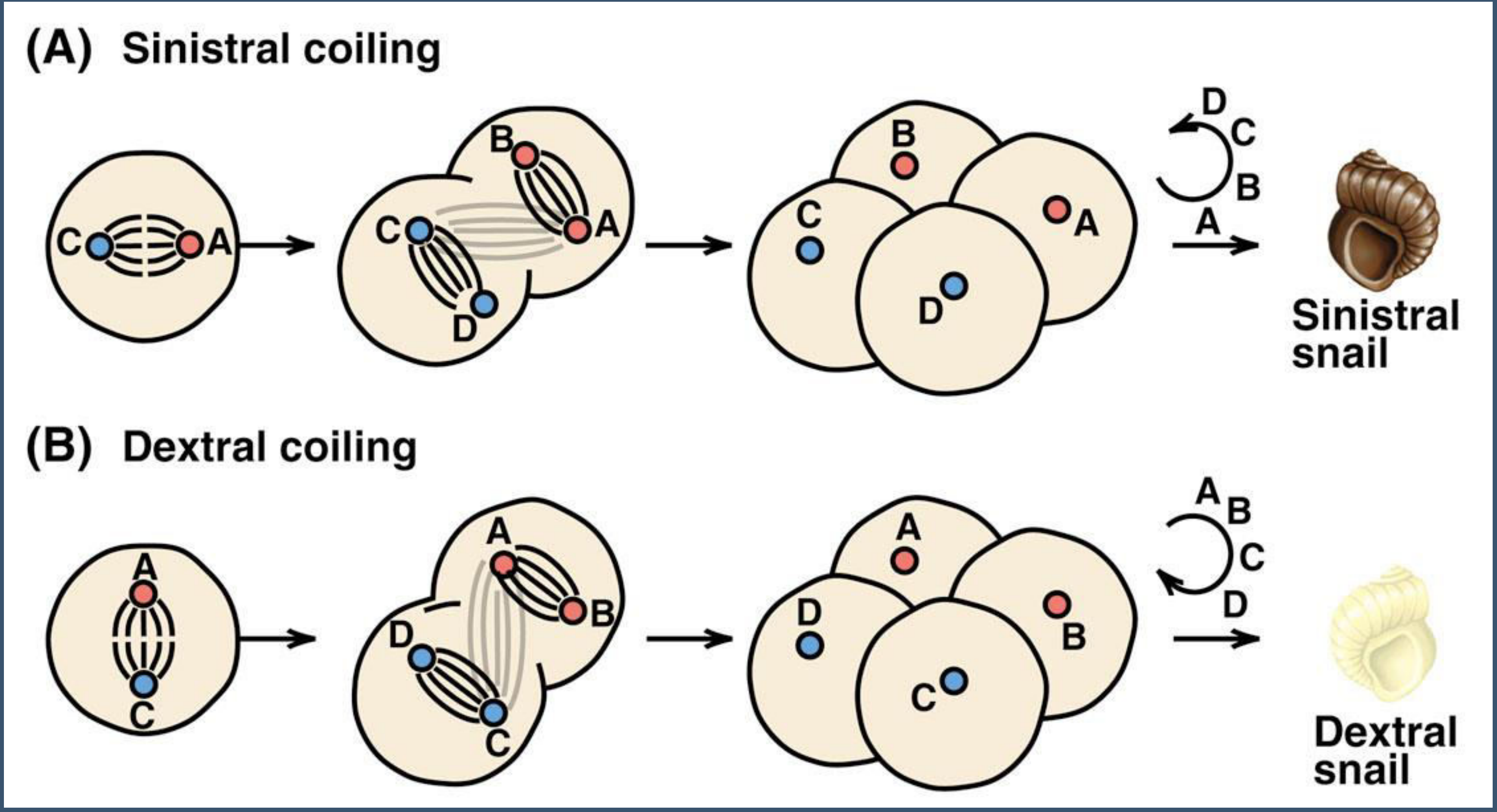
Maternal effect during the inheritance of shell coiling in the snail *Limnaea*



The direction of shell coiling is controlled by the mother's **nuclear** genotype

The direction of shell coiling in *Limnaea* is due to a maternal developmental gene which controls the orientation of the spindle in the second mitotic division of the zygote



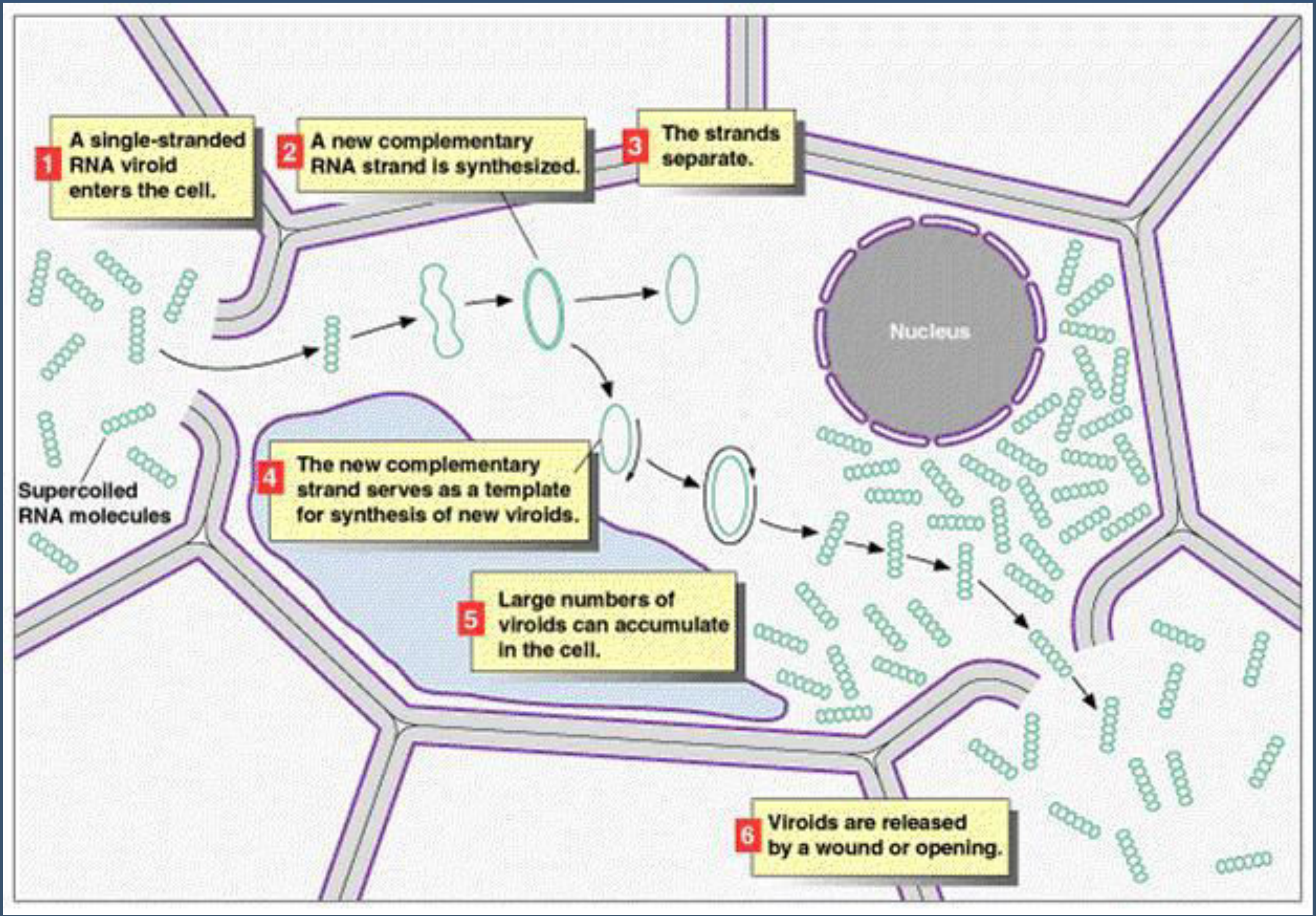
Mitochondria and chloroplasts originated as symbionts

- **Endosymbiont theory:**
  - **Both organelles reproduce by fission and have genomes remarkably like bacteria and algae.**
  - **Organelle DNA synthesis is not regulated like nuclear DNA (occurs at all stages of cell cycle).**

Infectious inheritance

- Viruses can survive in the cytoplasm or in the nuclear genome of the host.
- HIV can appear to be inherited maternally, but infection occurs *in utero*.
- Prions are infectious proteins.
- Viroids are infectious RNA particles.
  - Contain no genes, but are small circular nucleic acid molecules.
  - Potato Spindle Tuber Viroid.

Viroid life cycle



If you were asked to name the organelle that contains DNA, what would you say? If you said the nucleus, you'd definitely get full points, but the nucleus is not the only source of DNA in most cells.

Instead, DNA is also found in the mitochondria present in most plant and animals cells, as well as in the chloroplasts of plant cells. Here, we'll explore how mitochondrial and chloroplast DNA are inherited.

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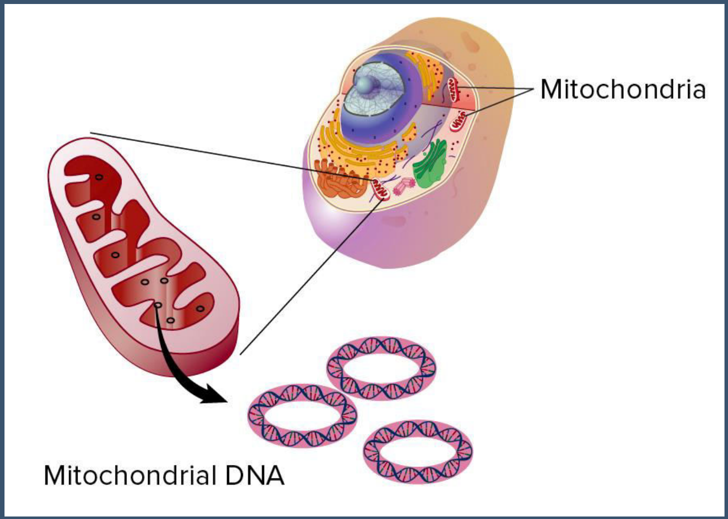
Mitochondrial and chloroplast DNA

The DNA molecules found in mitochondria and chloroplasts are small and circular, much like the DNA of a typical bacterium. There are usually many copies of DNA in a single mitochondrion or chloroplasts.



Similarities between the DNA of mitochondria and chloroplasts and the DNA of bacteria are an important line of evidence supporting the endosymbiont theory, which suggests that mitochondria and chloroplasts originated as free-living prokaryotic cells.

How is non-nuclear DNA inherited?

Here are some ways that mitochondrial and chloroplast DNA differ from the DNA found in the nucleus:

High copy number. A mitochondrion or chloroplast has multiple copies of its DNA, and a typical cell has many mitochondria (and, in the case of a plant cell, chloroplasts). As a result, cells usually have many copies – often thousands – of mitochondrial and chloroplast DNA. *[Are the copies identical within a single organelle?]*

Random segregation. Mitochondria and chloroplasts (and the genes they carry) are randomly distributed to daughter cells during mitosis and meiosis. When the cell divides, the organelles that happen to be on opposite sides of the cleavage furrow or cell plate will end up in different daughter cells^33cubed.

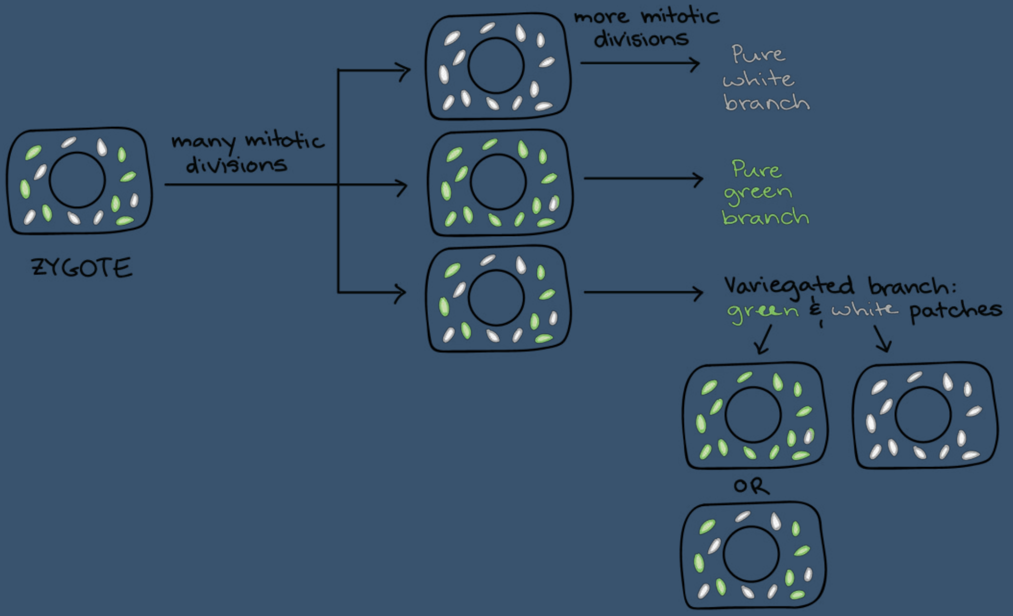
Single-parent inheritance. Non-nuclear DNA is often inherited uniparentally, meaning that offspring get DNA only from the male or the female parent, not both^44start superscript, 4, end superscript. In humans, for example, children get mitochondrial DNA from their mother (but not their father).

Chloroplast inheritance: Early experiments

At the turn of the 20th century, Carl Correns, a German botanist, did a series of genetic experiments using four o'clock plants (*Mirabilis jalapa*). We now know that his work demonstrated how chloroplast DNA is passed on from cell to cell and from parent to offspring—though Correns himself didn't know it at the time^55start superscript, 5, end superscript!

Explaining Correns' results

How can the idea of chloroplast inheritance make variegated plants variegated? Let's follow a zygote (1-celled embryo) with mixture of chloroplasts inherited from the egg cell. Some of the chloroplasts are green, while others are white. As the zygote undergoes many rounds of mitosis to form an embryo and then a plant, the chloroplasts also divide and are distributed randomly to daughter cells at each division.



Over the many cell divisions, some cells will end up with a pure set of normal chloroplasts, making green patches). Others will get a pure set of nonfunctional chloroplasts (making white patches). Others yet will have a mix of normal and nonfunctional chloroplasts, producing green patches that may give rise to pure green or pure white sectors^77start superscript, 7, end superscript.

What about the maternal pattern of inheritance? Plants make germ cells late in development, converting cells at the tip of a branch into gamete-producing cells. A branch that’s pure green will make egg cells with green chloroplasts that give rise to pure green offspring. Similarly, a branch that's pure white will make egg cells that contain only white chloroplasts and will give rise to pure white offspring.

If a branch is variegated, it has a mixture of cells, some with only functional chloroplasts, some with only nonfunctional chloroplasts, and some with a mixture of chloroplasts. All three of these cell types may give rise to egg cells, leading to the green offspring, white offspring, and variegated offspring in unpredictable ratios^{6,7}6,7start superscript, 6, comma, 7, end superscript.

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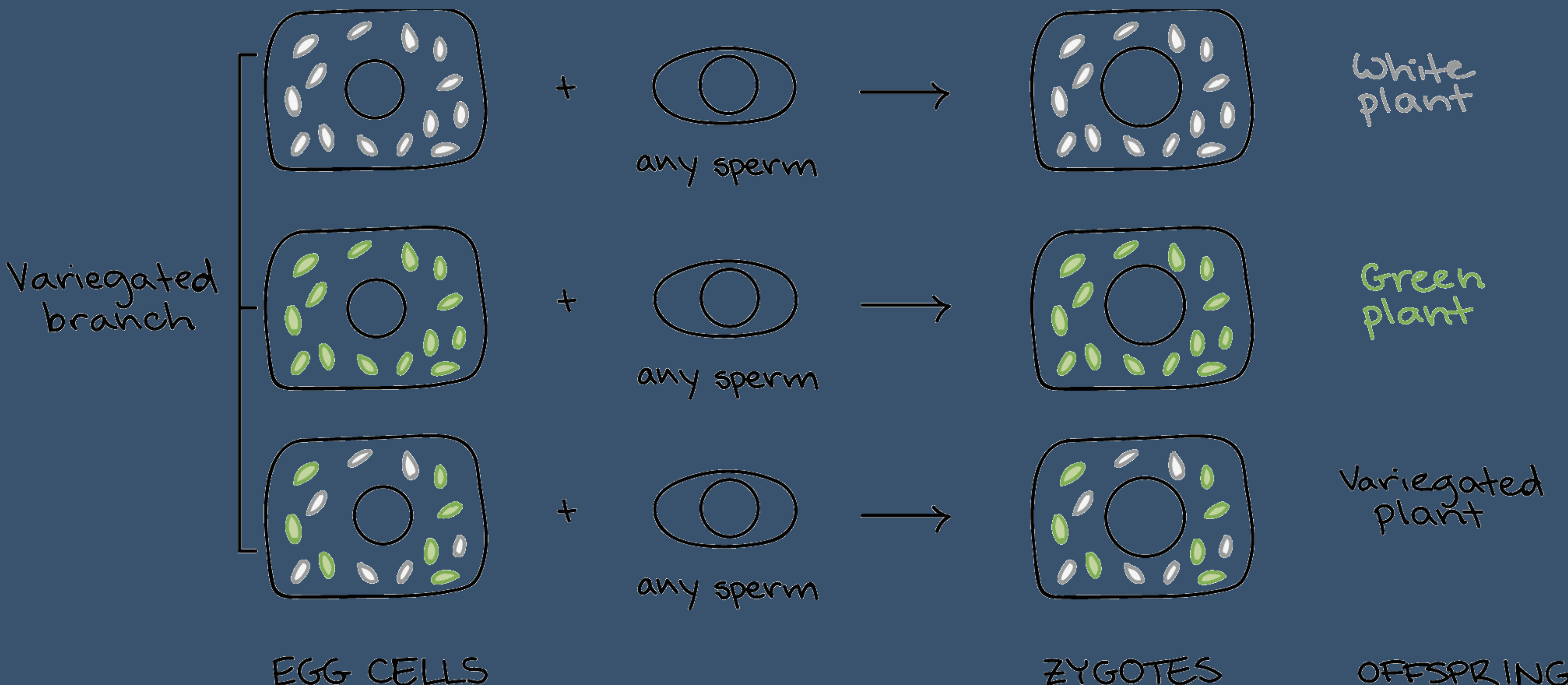
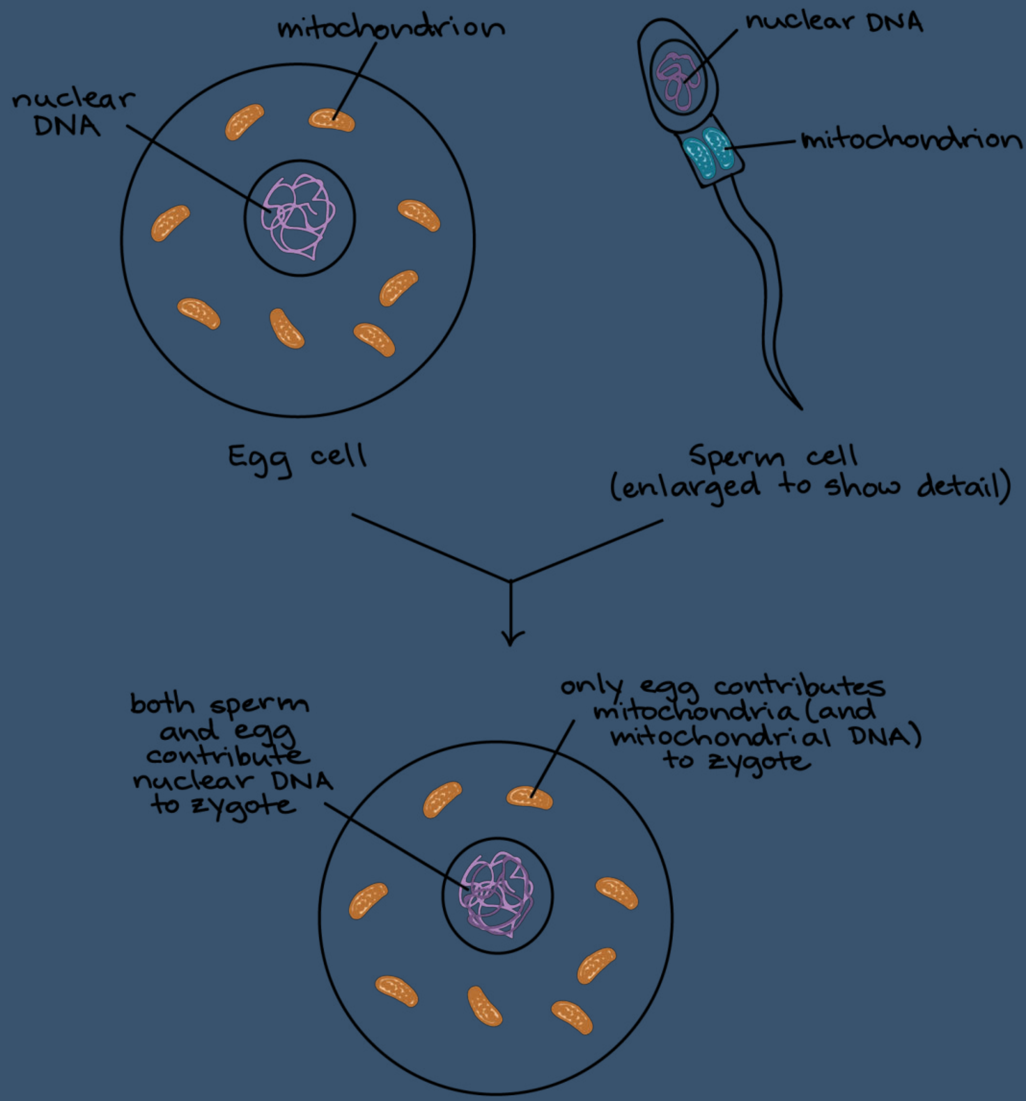

Variegated branch
+
any sperm
+
any sperm
+
any sperm
White plant
Green plant
Variegated plant
EGG CELLS
ZYGOTES
OFFSPRING

Mitochondrial inheritance

Mitochondria, like chloroplasts, tend to be inherited from just one parent or the other (or at least, to be unequally inherited from the two parents)^44start superscript, 4, end superscript. In the case of humans, it is the mother who contributes mitochondria to the zygote, or one-celled embryo, by way of the egg's cytoplasm. Sperm do contain mitochondria, but they are not usually inherited by the zygote. There has been a reported case of paternal inheritance of mitochondria in a human, but this is extremely rare^88start superscript, 8, end superscript.

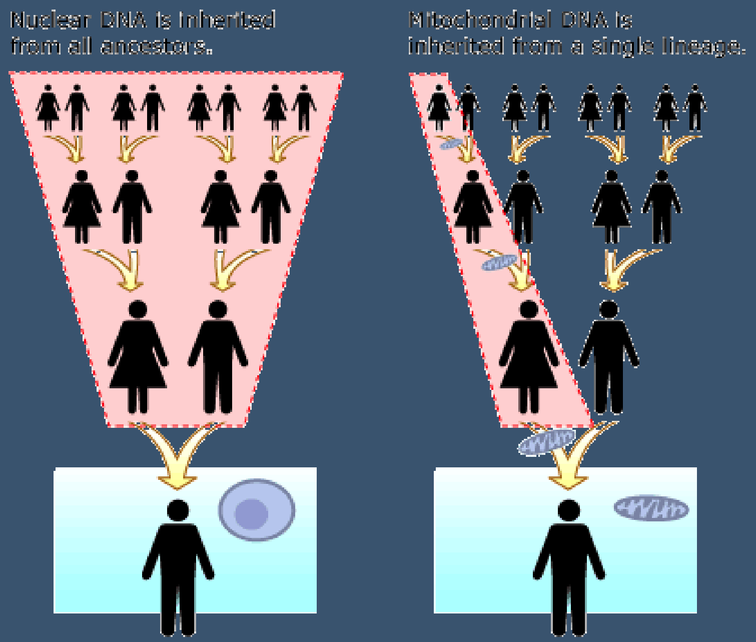

mitochondrion
nuclear DNA
Egg cell
nuclear DNA
mitochondrion
Sperm cell
(enlarged to show detail)
both sperm and egg contribute nuclear DNA to zygote
only egg contributes mitochondria (and mitochondrial DNA) to zygote

Maternal inheritance of mitochondria in humans

Because mitochondria are inherited from a person's mother, they provide a way to trace matrilineal ancestry (line of descent through an unbroken chain of female ancestors).

To understand how mitochondria connect you to your mother's mother's foremothers, consider where your mitochondria came from. They were received from your mother, in the cytoplasm of the egg cell that gave rise to you. Where did your mother get her own mitochondria? From *her* mother, that is, your maternal grandmother^99start superscript, 9, end superscript.

If you keep asking this question, you can walk backward in time through your family tree, following your matrilineal ancestors and tracing the transmission route of your mitochondrial DNA.



As shown in the diagram above, the inheritance pattern of mitochondrial DNA is different from that of nuclear DNA. A person's nuclear DNA is a "patchwork" of segments inherited from many different ancestors, while a person's mitochondrial DNA is inherited through a single, unbroken line of female ancestors^{9,10}9,10start superscript, 9, comma, 10, end