# Extranuclear Inheritance

Organelle Heredity Involves DNA in Chloroplasts and Mitochondria

• Some observations indicate an apparent **extranuclear influence on the phenotype.**

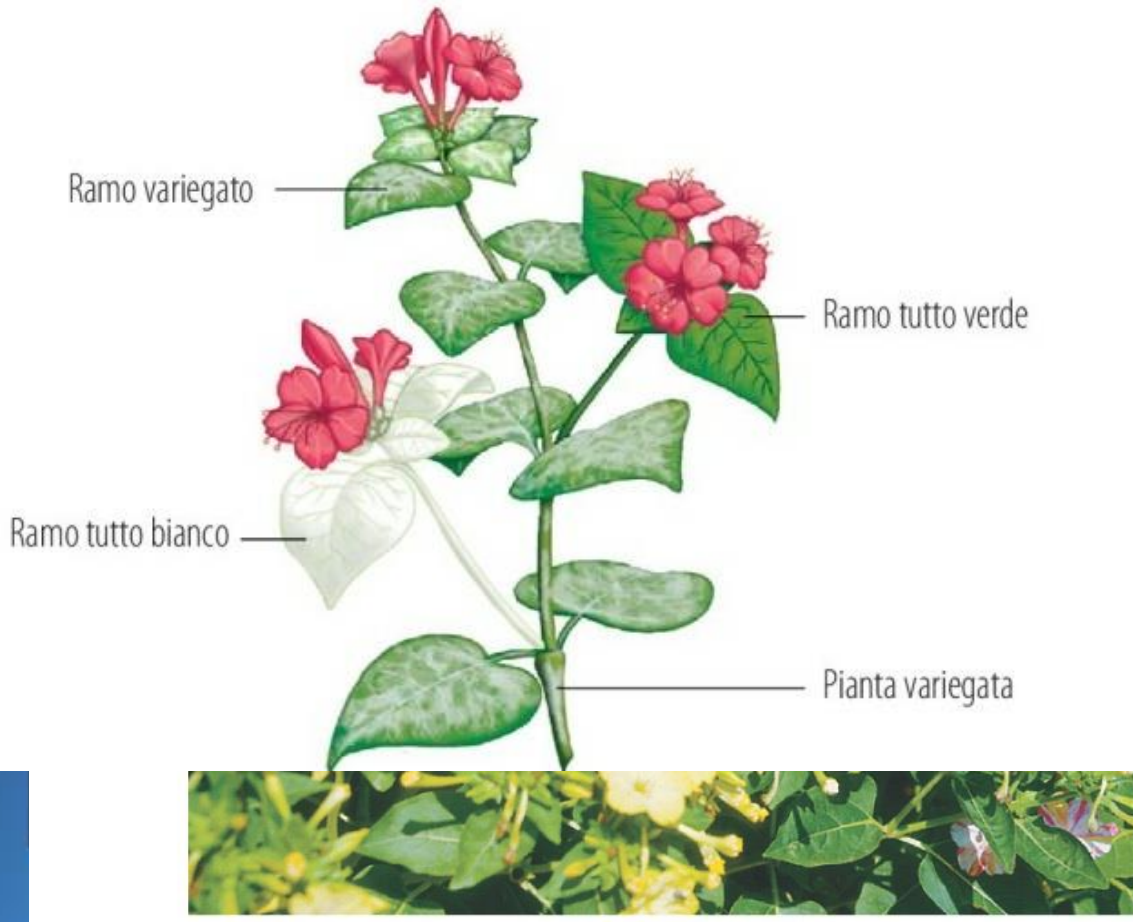
•With the discovery of DNA in mitochondria and chloroplasts, extranuclear inheritance is now recognized as an important aspect of genetics.

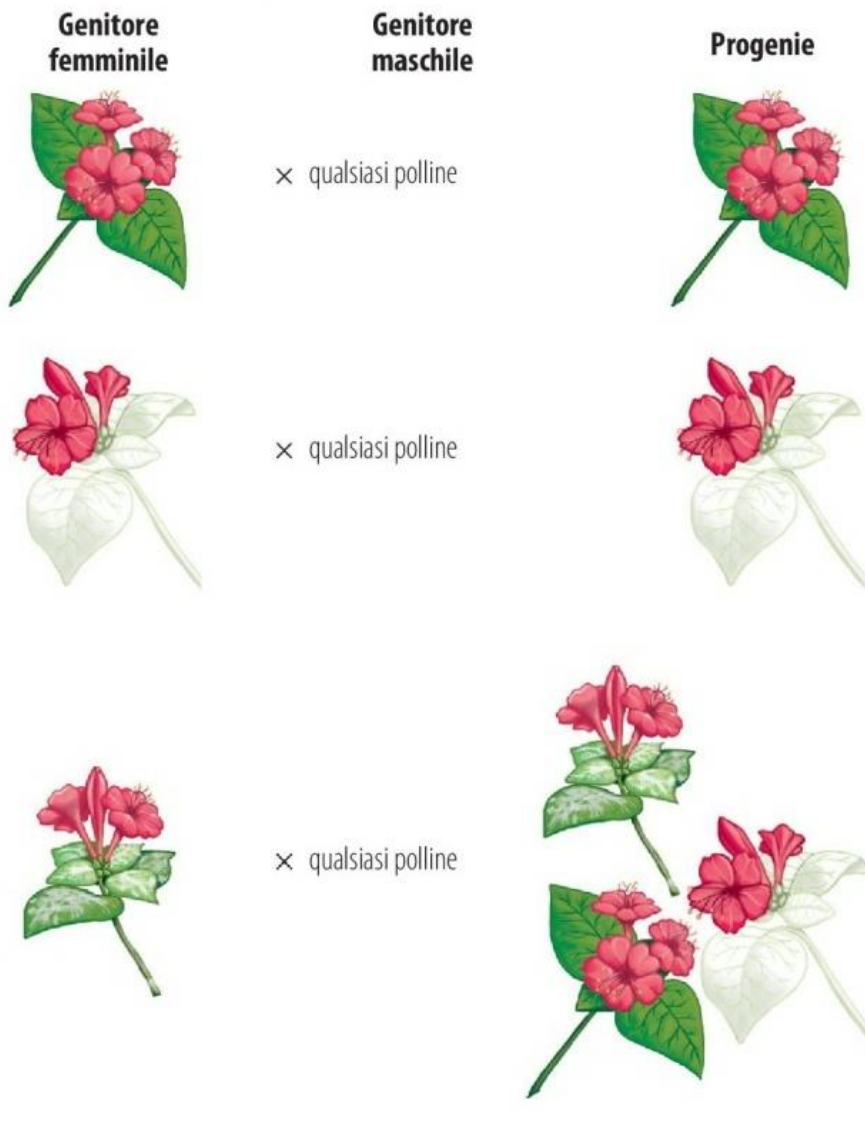
1 - One variety of extranuclear inheritance is **organelle heredity**, in which DNA contained in mitochondria or chloroplasts determines certain phenotypic characteristics of the offspring.

2 - A second type third variety involves the maternal effect on the phenotype, whereby **nuclear gene products** are stored in the egg and then transmitted to offspring from the female parent in the cytoplasm of the ovule.

Nel 1908, Erwin Baur e Carl Correns, lavorando indipendentemente l'uno dall'altro, il primo su *Pelargonium* (gerani) e il secondo su *Mirabilis jalapa (la* bella di notte), scoprirono entrambi l'ereditarieta non-mendeliana.

| | Location of Ovule | | |
|---|---|---|---|
| *Source of Pollen* | White branch | Green branch | Variegated branch |
| **White branch** | White | Green | White, green, or variegated |
| **Green branch** | White | Green | White, green, or variegated |
| **Variegated branch** | White | Green | White, green, or variegated |

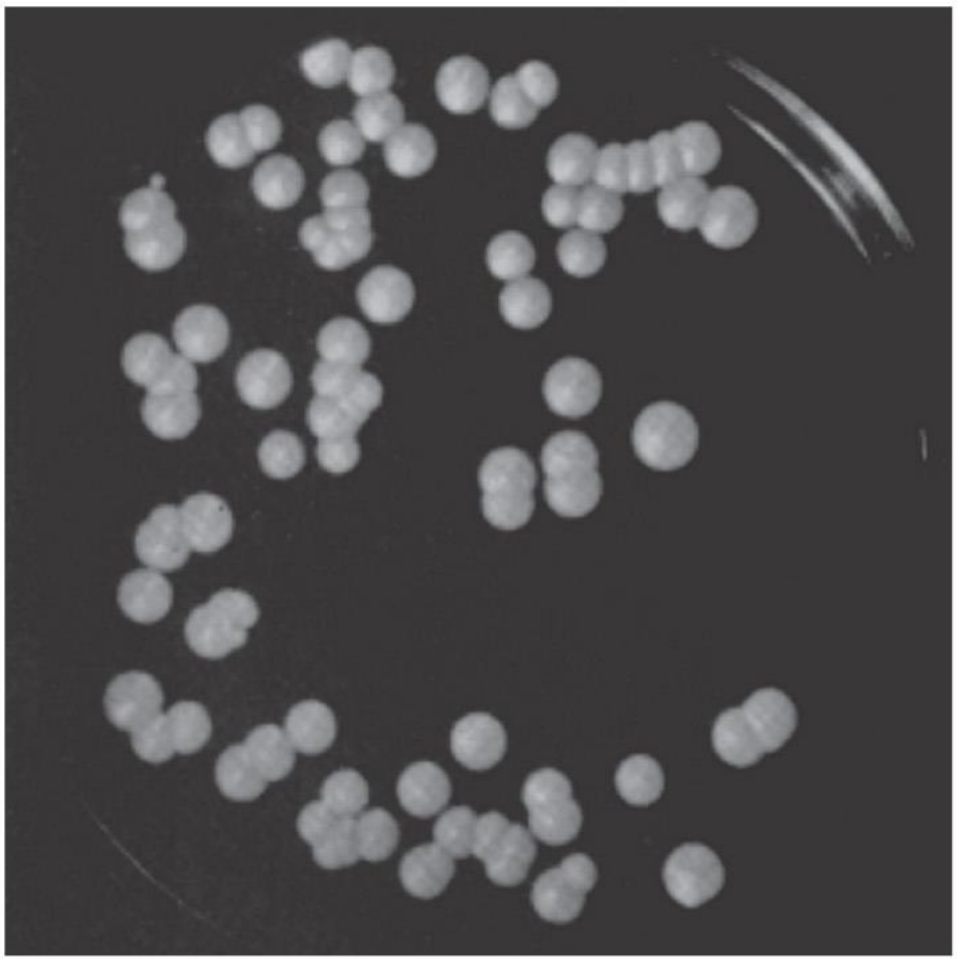


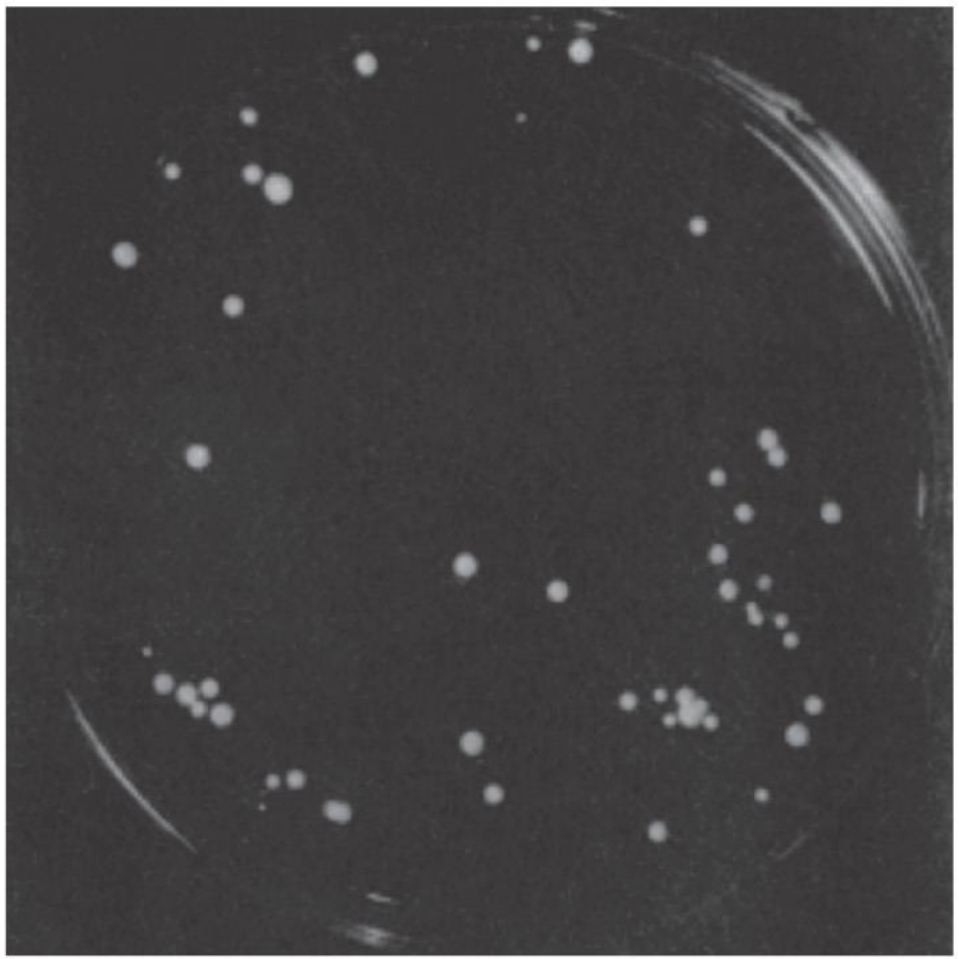
Negli anni Cinquanta, diverse decine di anni

dopo che Correns e Baur avevano descritto le loro osservazioni relative all'ereditarieta non-mendeliana nelle piante, Yasutane Chiba e colleghi suggerirono che i mitocondri e i cloroplasti contenessero un proprio genoma.

# Organelle Heredity Involves DNA in Chloroplasts and Mitochondria

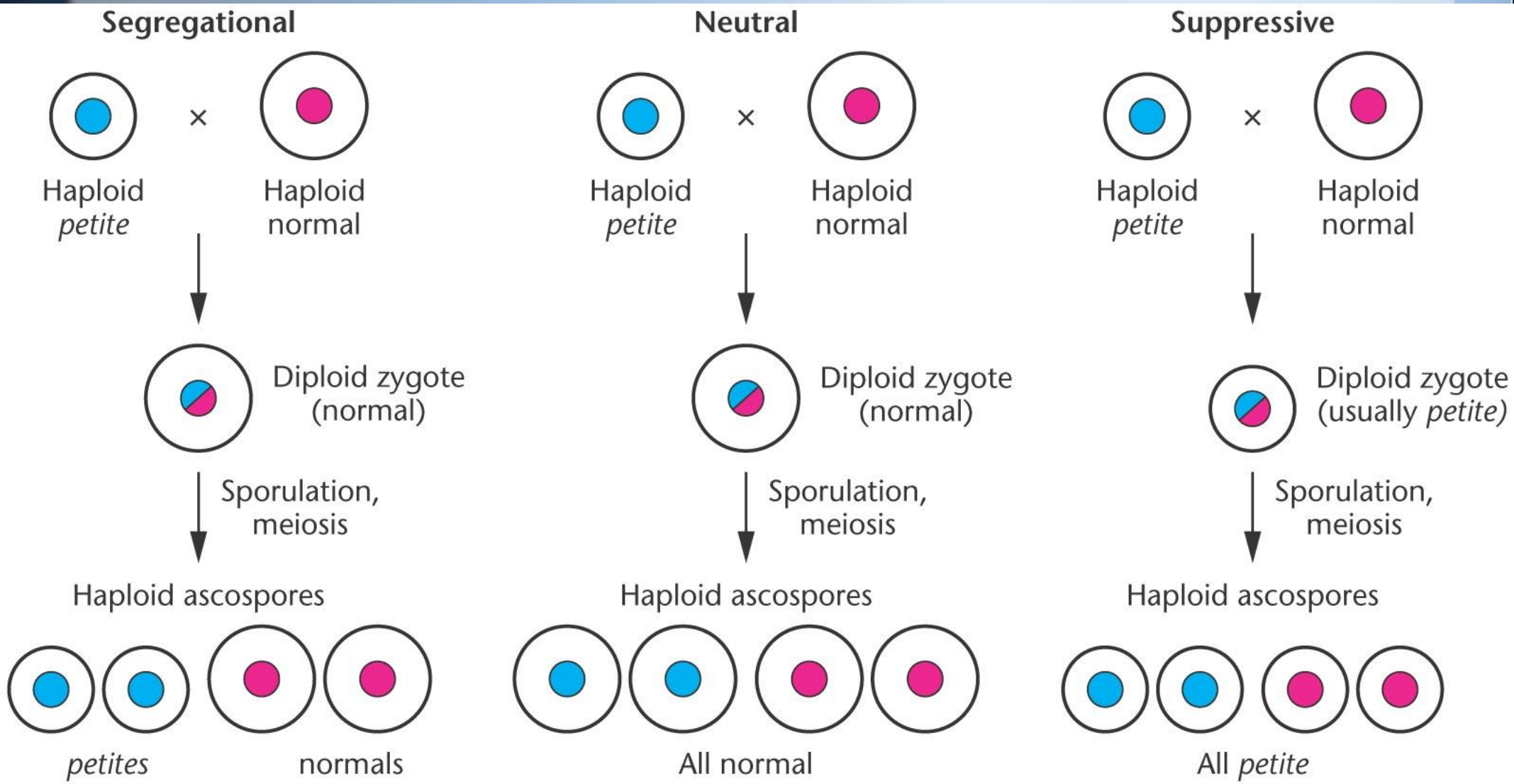


Normal colonies



Petite colonies

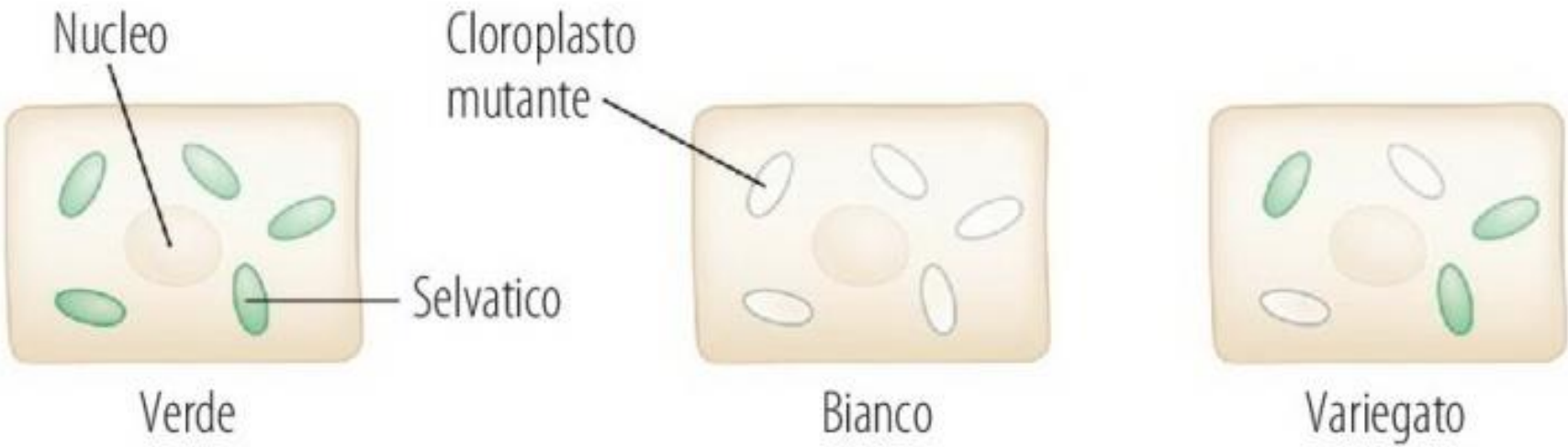
# Ereditarietà biparentale in *Saccharomyces cerevisiae*



i termini ***omozigote ed eterozigote non possono essere*** applicati agli alleli dei geni dei genomi degli organelli.

Omoplasmico
Eteroplasmico

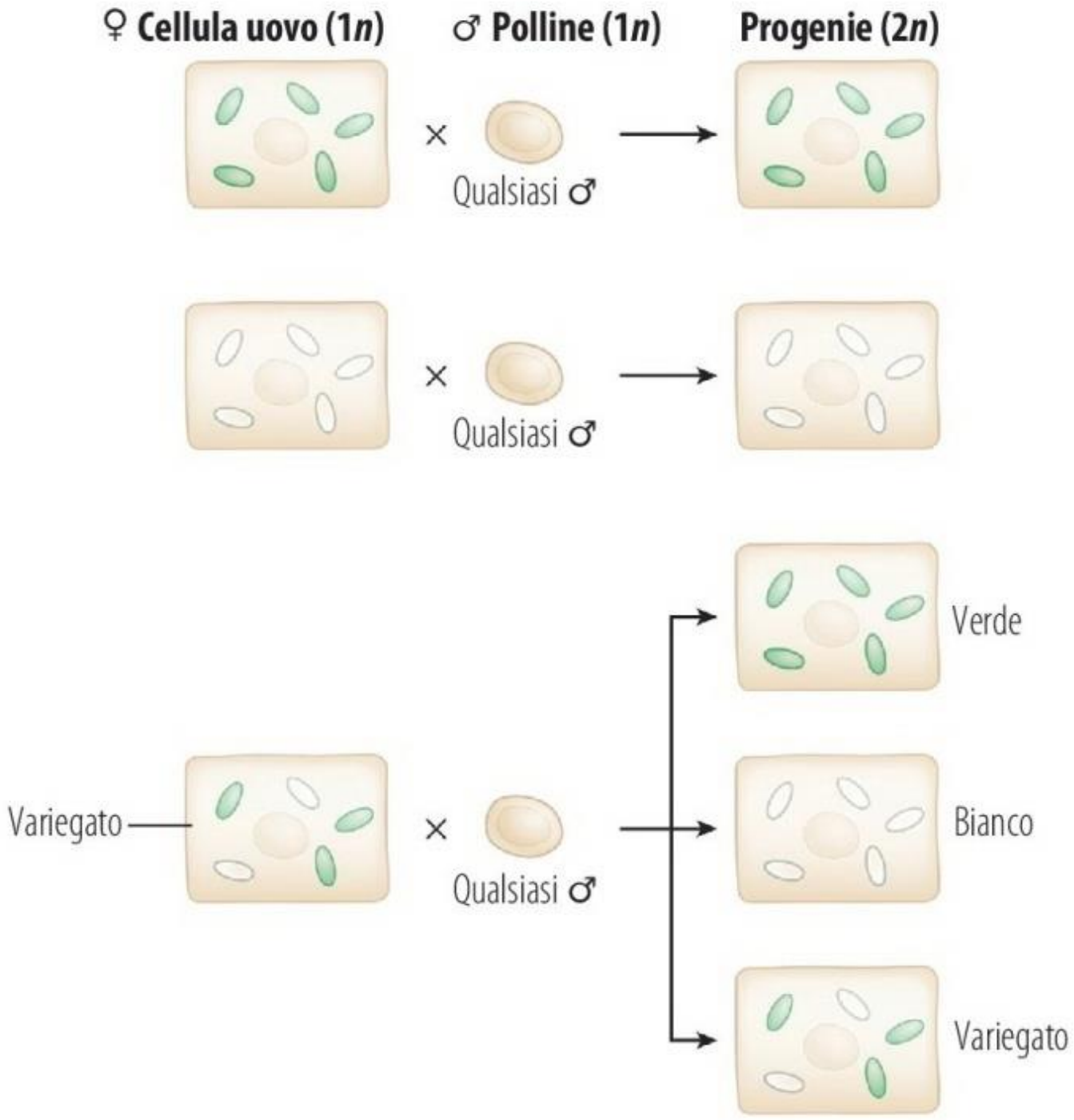
(**a**) Cellule omoplasmiche ed eteroplasmiche.



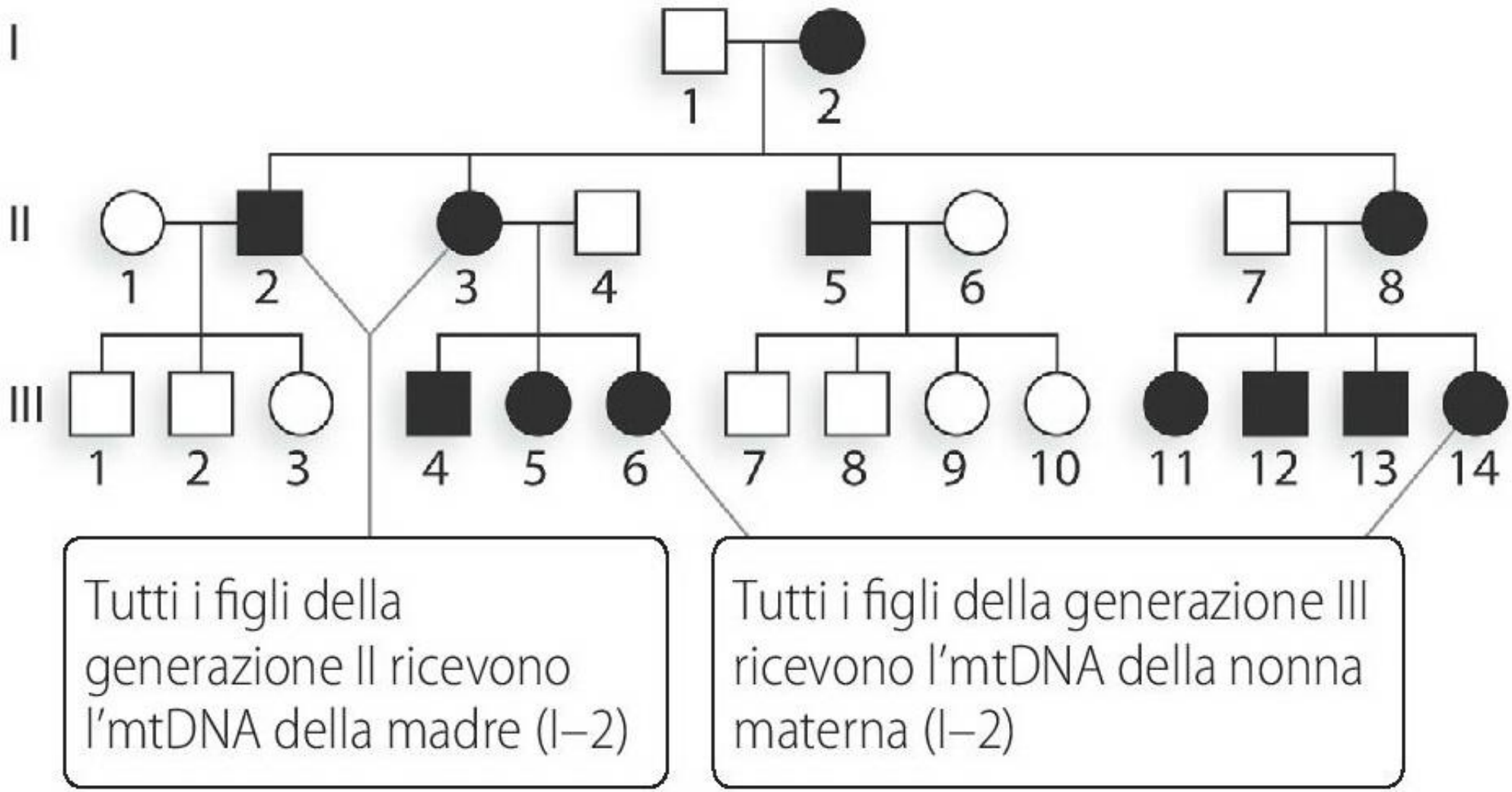

Le cellule omoplasmiche presentano organelli con genotipo identico

Le cellule eteroplasmiche contengono una miscela di alleli

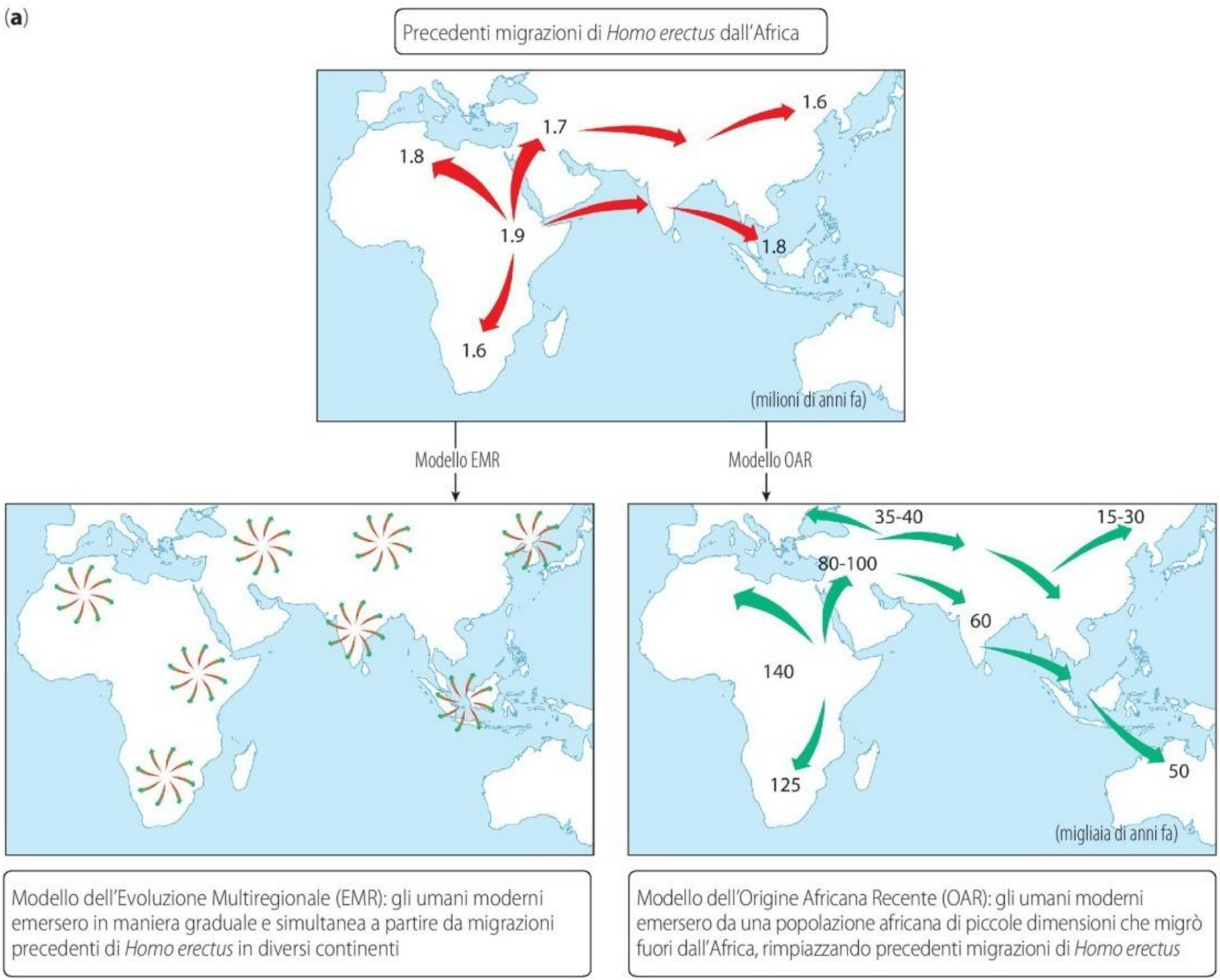
(**b**) Il fenotipo della progenie dipende esclusivamente dal genotipo del genitore materno.



# genotipizzazione



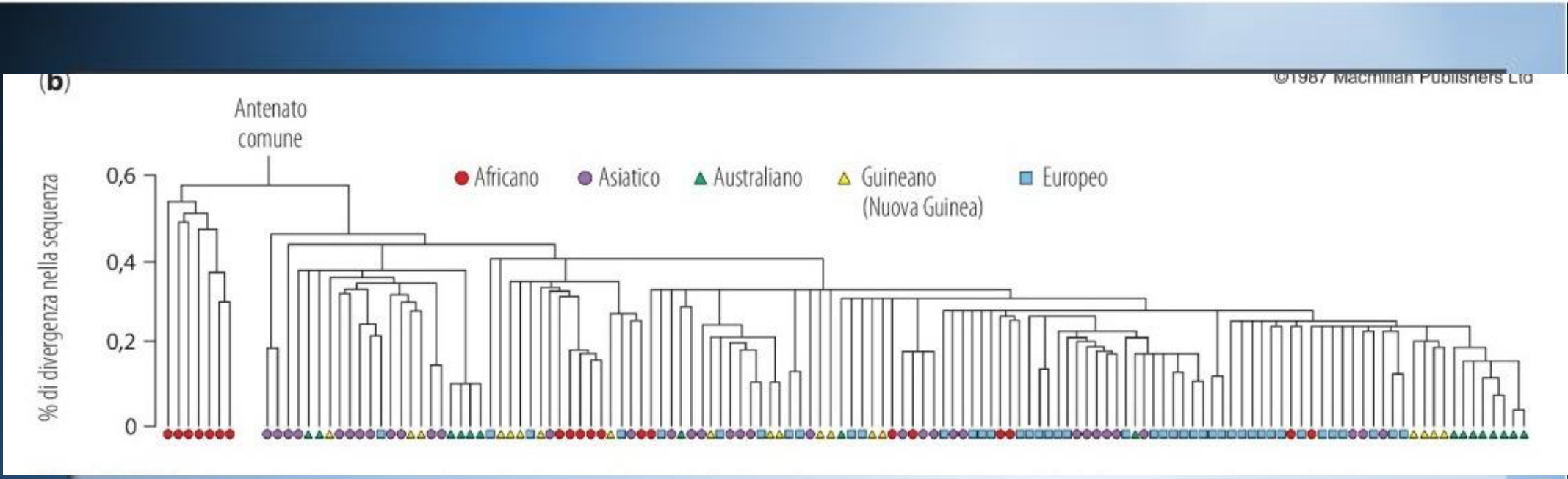
# L'Eva mitocondriale.




Modello dell'Evoluzione Multiregionale (EMR): gli umani moderni emersero in maniera graduale e simultanea a partire da migrazioni precedenti di *Homo erectus* in diversi continenti

Modello dell'Origine Africana Recente (OAR): gli umani moderni emersero da una popolazione africana di piccole dimensioni che migrò fuori dall'Africa, rimpiazzando precedenti migrazioni di *Homo erectus*

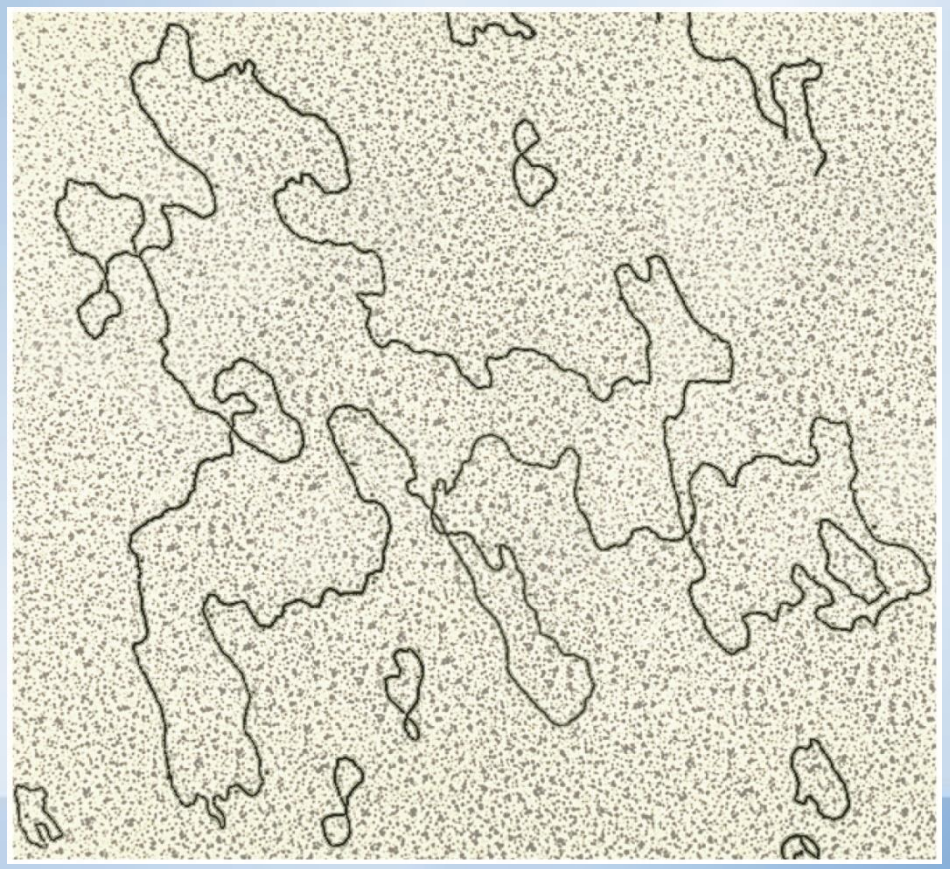
# L'Eva mitocondriale.




L'mtDNA di tutti gli umani viventi discende da una donna o da un gruppo di donne che viveva nell'Africa Orientale circa 120.000-200.000 anni fa.

# Knowledge of Mitochondrial and Chloroplast DNA Helps Explain Organelle Heredity

## Organelle DNA and the Endosymbiotic Theory

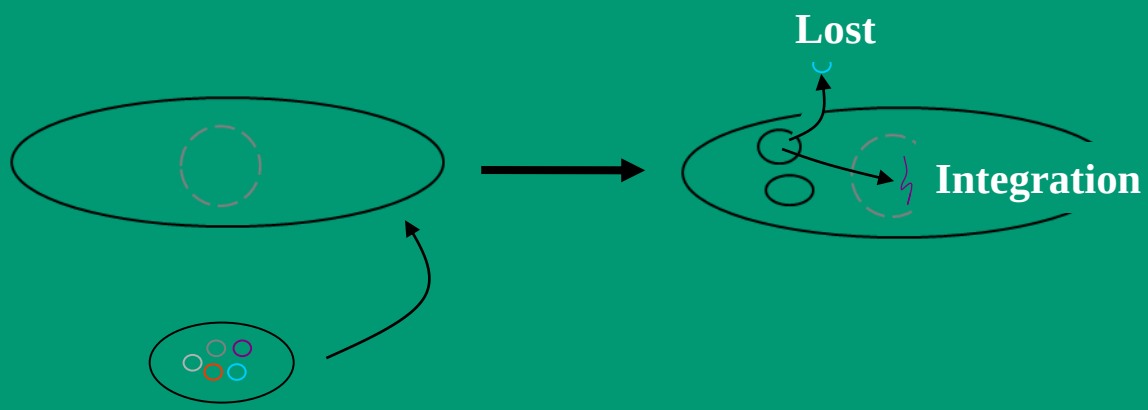


• The endosymbiotic theory states that mitochondria and chloroplasts arose independently about 2 billion years ago from free-living bacteria that possessed the abilities now attributed to these organelles—aerobic respiration and photosynthesis, respectively.

# Origin of extranuclear genes

## Generally believed

'Endosymbionts'




## Most modern eukaryotic cells

Fully dependent on the organelle genes for their normal function

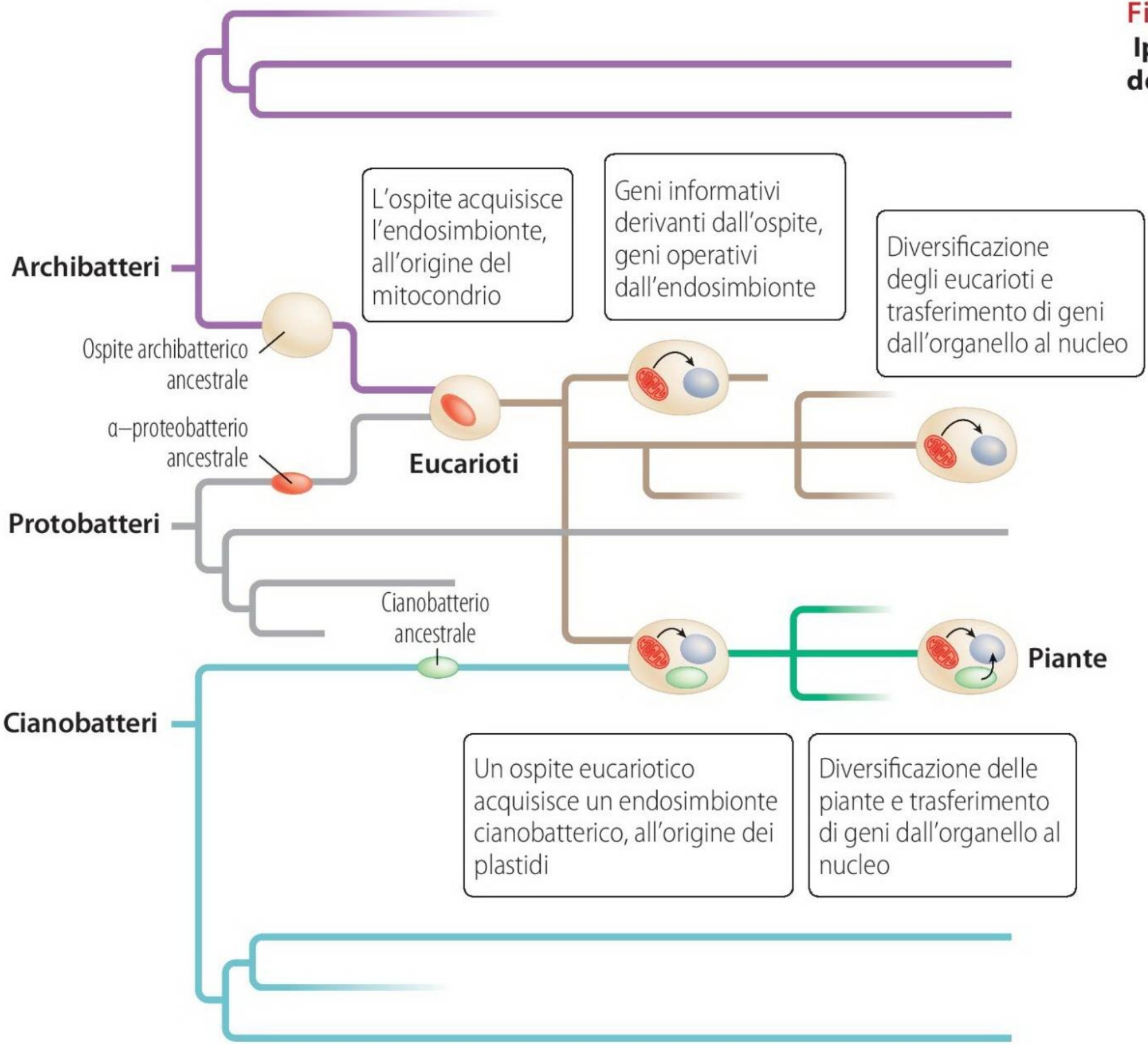
cf.) The yeast, '*Saccharomyces cerevisiae*'

Without mitochondria ⟶ obtain energy from fermentation

• These bacteria were engulfed by larger eukaryotic cells, and a beneficial symbiotic relationship developed.

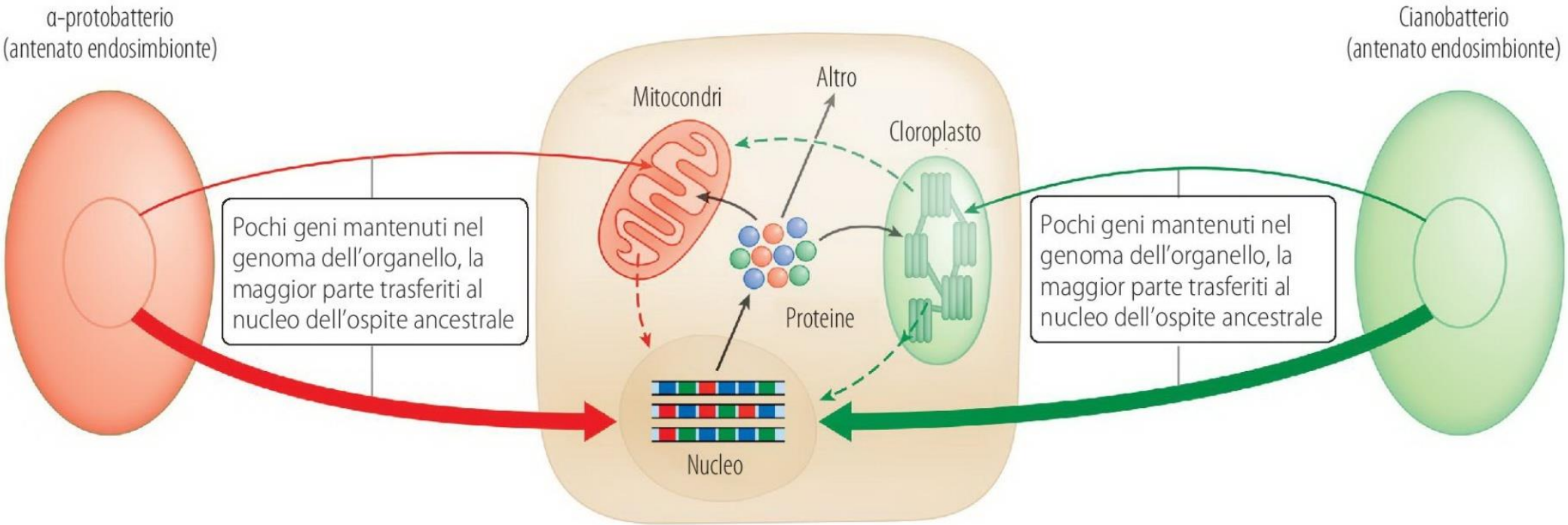
Chloroplast DNA ranges from 100 to 225 kb in length, and the genes carried on the DNA encode products involved in photosynthesis and translation.

**Figura 17.21**
**Ipotesi sull'evoluzione degli eucarioti.**



Il trasferimento di materiale genetico dagli organelli al nucleo e tra gli organelli continua nelle specie ancora esistenti (frecce tratteggiate rosse e verdi)

Le proteine codificate da geni originariamente derivati da genomi endosimbionti possono essere utilizzate per altre funzioni nella cellula ospite




Le proteine importate negli organelli possono essere codificate da geni originariamente derivati da endosimbionti (in rosso e verde) oppure originariamente derivate dal genoma dell'ospite (in blu)

• Animal mitochondrial DNA (mtDNA) is smaller than the DNA in chloroplasts, and introns are absent. Most of the protein encoding genes are located on a single strand.

## TABLE 9.1

# THE SIZE OF MTDNA IN DIFFERENT ORGANISMS

| Organisms | Size (kb) |
|---|---|
| Human | 16.6 |
| Mouse | 16.2 |
| *Xenopus* (frog) | 18.4 |
| *Drosophila* (fruit fly) | 18.4 |
| *Saccharomyces* (yeast) | 75.0 |
| *Pisum sativum* (pea) | 110.0 |
| *Arabidopsis* (mustard plant) | 367.0 |

# How many copies?

Nuclear chromosome : 1 copy / cell (haploid)

2 copy / cell (diploid)

Organelle chromosome : x00~x,000 copy/ cell

* regulation of copy number is relatively loose

ex.) Chloroplast : Leaf cells of the garden beet

~ 40 chloroplast / cell

4 ~ 8 nucleoids / chloroplast

(nucleoid : Specific heavily DNA area in chloroplast)

4 ~ 18 cpDNA molecules / nucleoid

MAX : 40 x 8 x 18 = 5760 copy of cpDNA / cell

Chlamydomonas

1 chloroplast / cell ⟶ 500 ~ 1500 cpDNA molecules

# How many copies?

ex.) Mitochondria : haploid yeast

1 ~ 45 mitochondria / cell

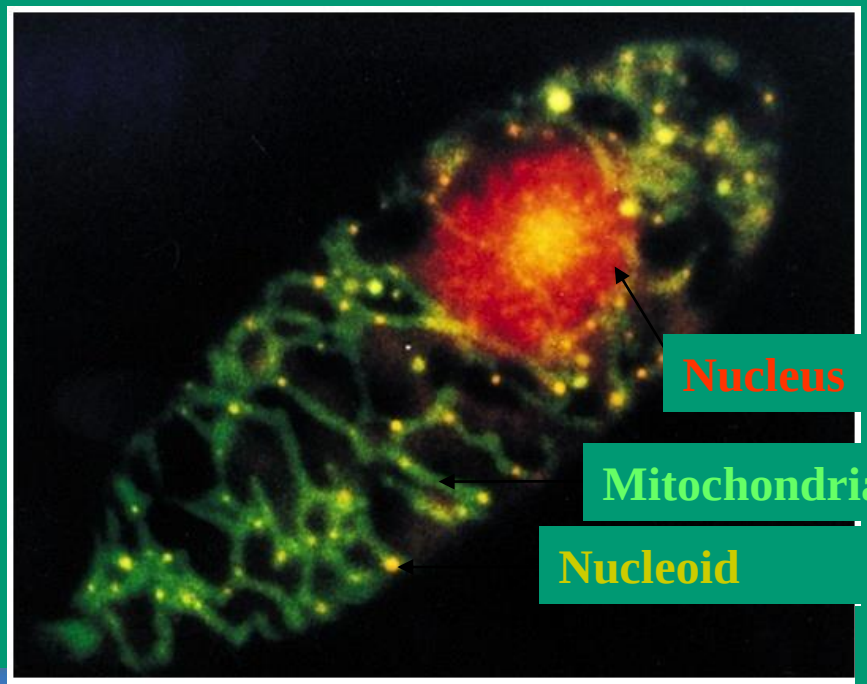
10 ~ 30 nucleoids / mitochondria

4 ~ 5 mtDNA molecules / nucleoid

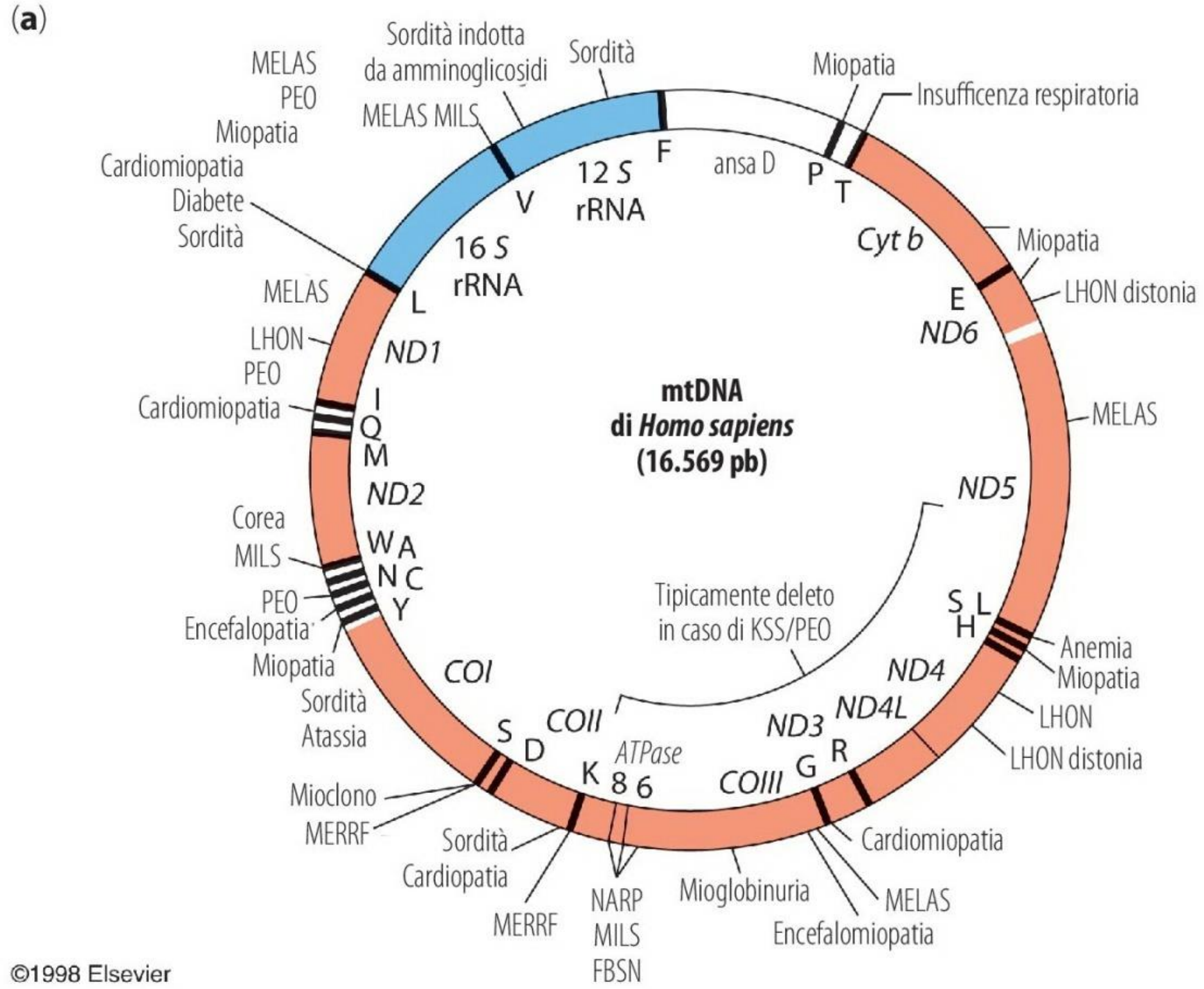
MAX : 45 x 30 x 5 = 6750

Human

2 ~ 10 mtDNA mol. / mitochondria




**Fluorescent staining of a cell of *Euglena gracilis.***


(a)
Sordità indotta da amminoglicosidi
Sordità
Miopatia
Insufficenza respiratoria
MELAS
PEO
MELAS MILS
Miopatia
Cardiomiopatia
Diabete
Sordità
12 S rRNA
ansa D
F
P
T
V
16 S rRNA
Cyt b
Miopatia
LHON distonia
MELAS
L
E
ND6
LHON
PEO
Cardiomiopatia
ND1
I
Q
M
mtDNA
di Homo sapiens
(16.569 pb)
MELAS
ND2
ND5
Corea
MILS
PEO
Encefalopatia
Miopatia
W
A
N
C
Y
Tipicamente deleto
in caso di KSS/PEO
S
L
H
Anemia
Miopatia
COI
ND4
LHON
Sordità
Atassia
COII
ND4L
ND3
LHON distonia
S
D
ATPase
K
8
6
R
G
COIII
Mioclono
MERRF
Sordità
Cardiopatia
Cardiomiopatia
MERRF
NARP
MILS
FBSN
Mioglobinuria
MELAS
Encefalomiopatia
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# Mitochondrial genomes

## Function

**Some of the proteins:** ~ oxidative phosphorylation

**tRNA, rRNAs, some proteins:** ~ mitochondrial protein synthesis

(some genes are encoded in nucleus)

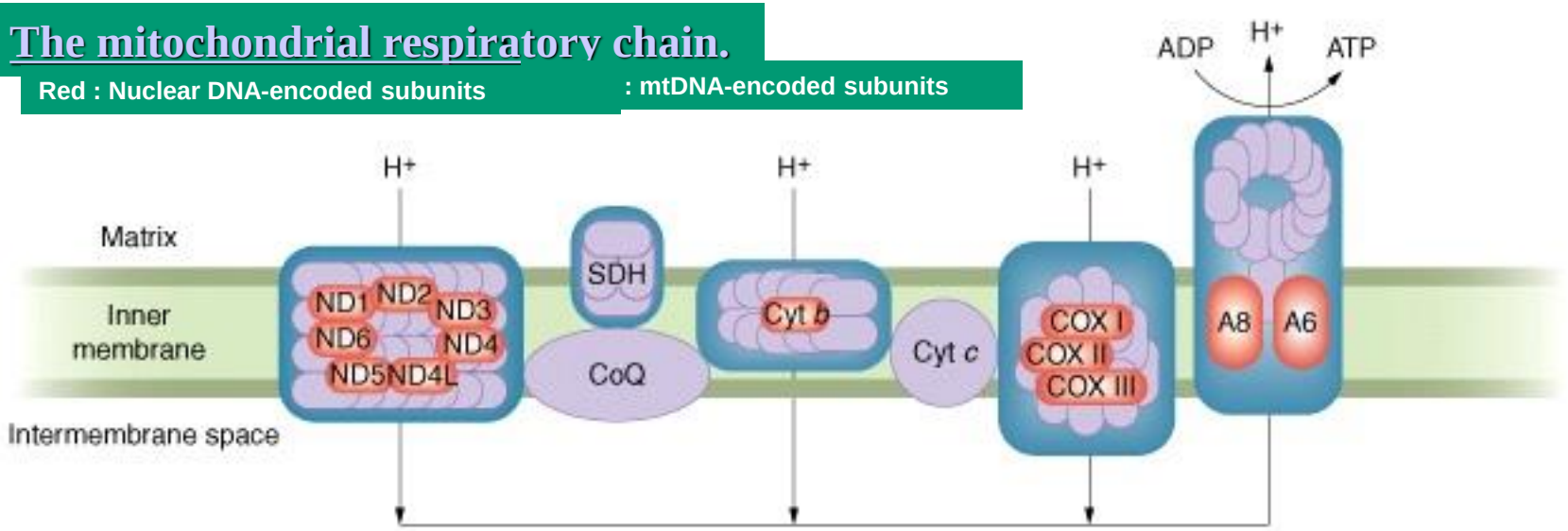
⇨ mRNA is translated outside the mitochondria on cytosolic ribosomes

Synthesized proteins are transported into the mitochondria

Complete system is assemble in mitochondrial inner membrane

## The mitochondrial respiratory chain.

Red : Nuclear DNA-encoded subunits : mtDNA-encoded subunits




| Subunits | Complex I | Complex II | Complex III | Complex IV | Complex V |
|---|---|---|---|---|---|
| nDNA-encoded | 35 | 4 | 10 | 10 | 12 |
| mtDNA-encoded | 7 | 0 | 1 | 3 | 2 |

# Mitochondrial genomes

| First letter | Second letter: U | Second letter: C | Second letter: A | Second letter: G | Third letter |
|---|---|---|---|---|---|
| U | Phe | Ser | Tyr | Cys | U |
| | Phe | Ser | Tyr | Cys | C |
| | Leu | Ser | *Stop* | (*Stop*) Trp | A |
| | Leu | Ser | *Stop* | Trp | G |
| C | Leu | Pro | His | Arg | U |
| | Leu | Pro | His | Arg | C |
| | Leu | Pro | Gin | Arg | A |
| | Leu | Pro | Gin | Arg | G |
| A | Ile (Met) | Thr | Asn | Ser | U |
| | Ile | Thr | Asn | Ser | C |
| | (Ile) Met | Thr | Lys | (Arg) *Stop* | A |
| | Ile | Thr | Lys | (Arg) *Stop* | G |
| G | Val | Ala | Asp | Gly | U |
| | Val | Ala | Asp | Gly | C |
| | Val | Ala | Glu | Gly | A |
| | Val | Ala | Glu | Gly | G |

Differ with nuclear code

**The genetic code of the human mitochondria.**

**22 tRNA types by the 22 boxes that do not contain stop codons**

# Mitochondrial genomes

## Yeast

**The intron in several mitochondrial gene**

Ex.) Subunit I of cytochrome oxidase - 9 introns

nuclear gene - rare intron

**The existence of unassigned reading frames (URFs) within the yeast intron**

* URF - sequences that have correct initiation codons & are uninterrupted by stop codons

* some URF - important in the splicing out of the introns themselves at the RNA level ∴ Specifying proteins

## Human

**Much smaller & more compact than yeast mtDNA**

>> much less spacer DNA

# Chloroplast genomes

**cpDNA : 120 ~ 200Kb in different species**

**ex.) Liverwort *Marchantia :* 136 genes**

**4 kinds of rRNA**

**31 kinds of tRNA**

**90 proteins (20: photosynthesis & electron-transport functions)**

**half the chloroplast genome: relate with translational function**

**Large inverted repeats of virtually all species of plants**

**(the sequences of the repeats are same!)**

**Like mtDNA, cpDNA cooperates with nuclear DNA to provide subunits for functional proteins**

# In Maternal Effect, the Maternal Genotype Has a Strong Influence during Early Development

• Maternal effect implies that an offspring's phenotype is under the control of nuclear gene products present in the egg. These factors influence patterns established early in development. For maternal effect genes, the genotype of the female parent and not that of the embryo determines the phenotype of the offspring.

# La spiralizzazione in Limnea

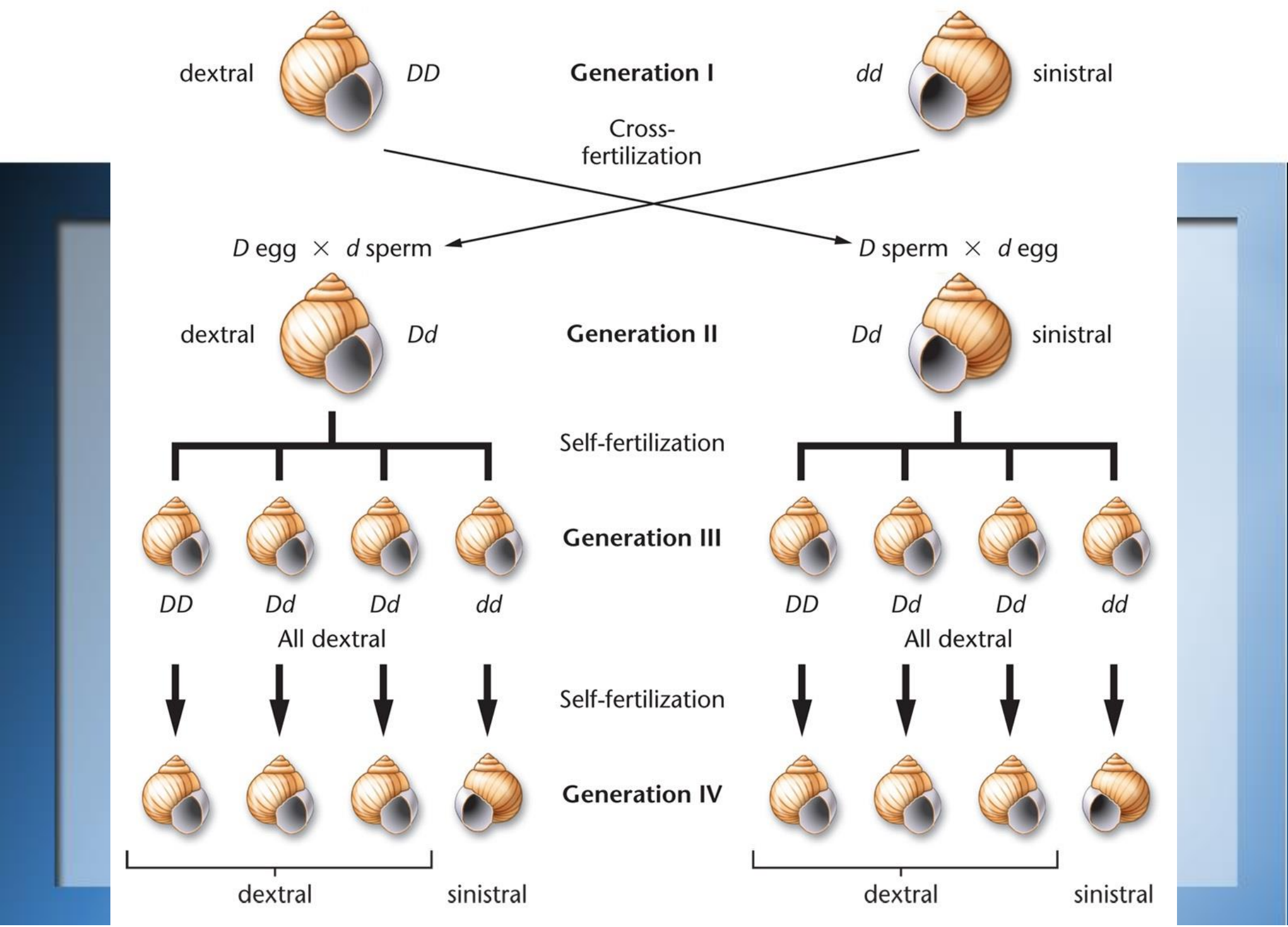
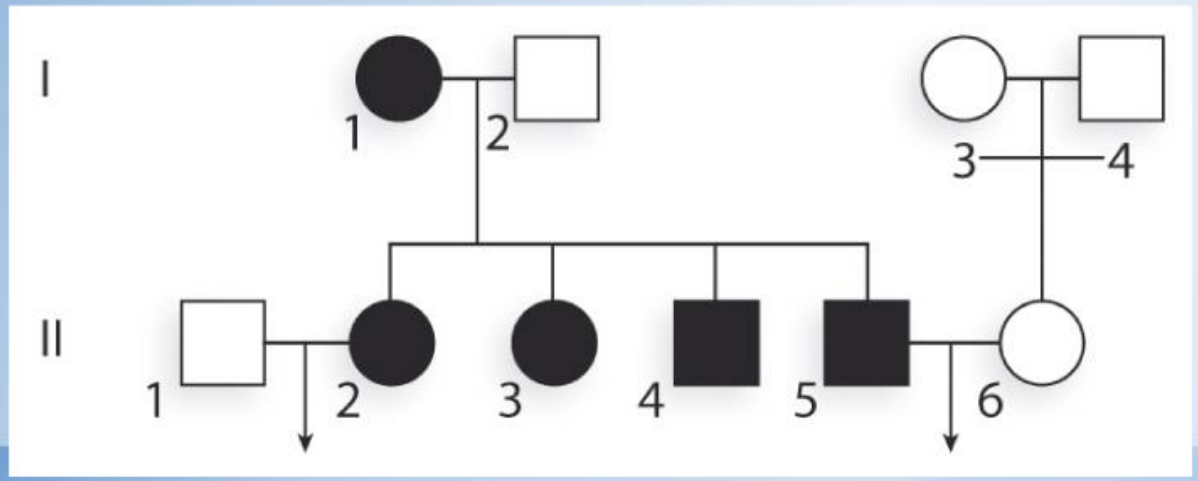
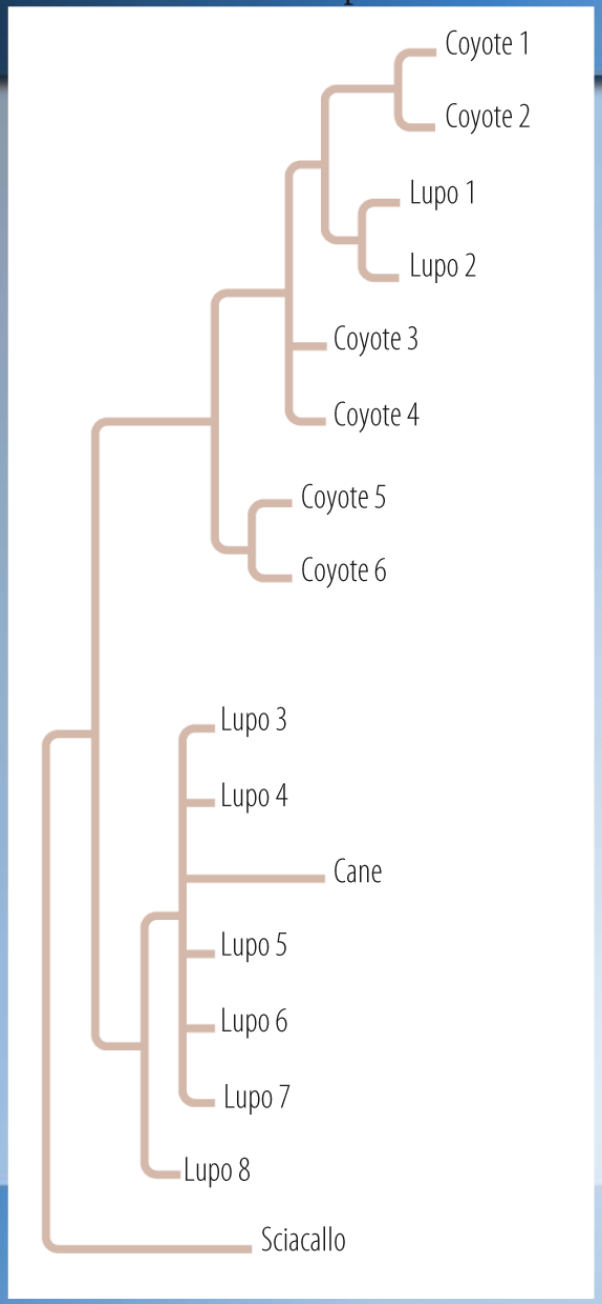



**Figure 9-13-02** 

Il seguente albero genealogico raffigura una famiglia in cui diversi individui presentano i sintomi della malattia mitocondriale detta MERRF. Un fratello e una sorella (II-5 e II-2) si rivolgono a voi per sapere se anche i loro figli saranno affetti da MERRF. Cosa rispondete?






Sulla base di questo albero filogenetico (analizzando il DNA mitocondriale), cosa concludete relativamente alla potenziale ibridazione interspecifica tra lupi e coyote?