8. Eredità Extra Cromosomica/ Eredità per via materna

Eredità Mendeliana

- **Modelli di eredità Mendeliana**
  - Coinvolgono geni che influenzano direttamente i caratteri
  - Rispettano le leggi di Mendel
    - Principio della segregazione
    - Principio dell'assortimento indipendente
  - Includono
    - Relazioni dominante/recessivo
    - Interazioni geniche
    - Ruolo del sesso e ambiente nell'influenzare il fenotipo
  - La maggior parte dei geni degli eucarioti seguono un modello di eredità di tipo Mendeliano

Modello mendeliano come regola generale

- **Il comportamento di un gene non dipende dal genitore che lo trasmette**





UGUALI
PROBABILITA' DI
TRASMISSIONE

Ereditarietà non-mendeliana

- Molti geni non seguono un modello di ereditarietà mendeliana, ad esempio, i geni strettamente associati non seguono la legge di assortimento indipendente di Mendel

- Questa lezione discuterà modelli di ereditarietà non mendeliani aggiuntivi e più singolari

- **Effetto materno**
- **Eredità extranucleare**
- Ereditarietà epigenetica

EREDITÀ MATERNA vs EFFETTO MATERNO

- **Eredità materna:** dovuta **all'esistenza di geni localizzati su genomi citoplasmatici**, che sono generalmente ereditati dalla madre

- **Effetto materno:** **non dovuto a geni extranucleari, ma a geni nucleari**
  - in questo caso il fenotipo si esprime prima della fecondazione della cellula uovo (e quindi è controllato dalla madre)

1. Effetto materno

- **Effetto materno**
  - Modello di ereditarietà per certi geni nucleari
  - **Il genotipo della madre determina direttamente il fenotipo della progenie**
    - Genotipo del padre e progenie sono irrilevanti
    - Spiegabile da **accumulazione di prodotti genici forniti dalla madre** durante lo sviluppo dell'uovo.

----> Effetto materno o influenza materna **implica che il fenotipo della progenie**, per un determinato carattere, **sia sotto il controllo di prodotti genici nucleari presenti nella cellula uovo**, contrariamente all'eredità biparentale, dove entrambi i genitori determinano l'informazione genetica.

Effetto Materno

- **Geni ad effetto materno**
  - Codificano RNA and proteine che giocano ruoli importanti nelle prime fasi dell'embriogenesi

    e.g., Divisione cellulare, modello di divisione, orientamento dell'asse del corpo
  - Alleli difettivi tendono a mostare effetti fenotipici drammatici

Maternal Effect

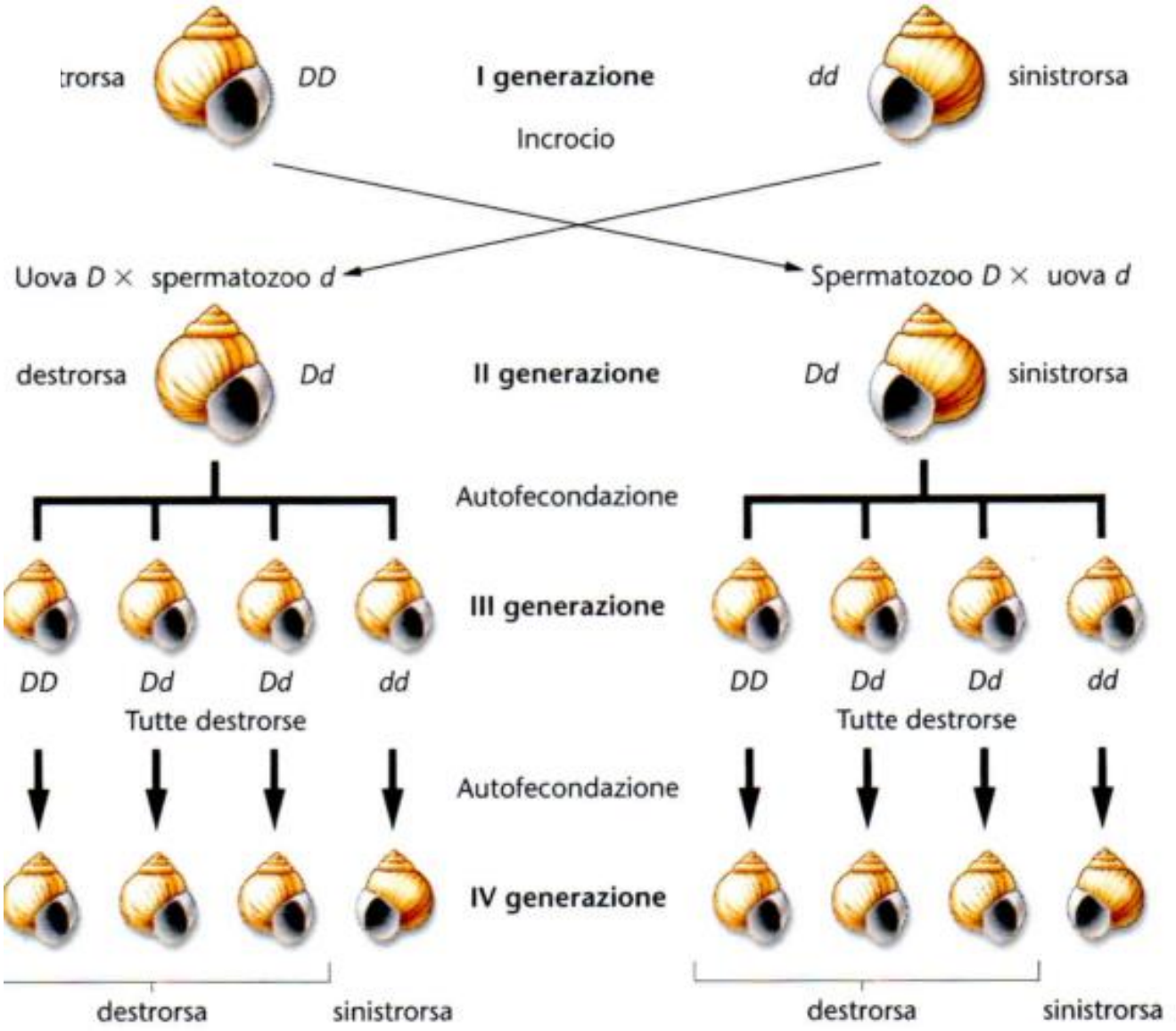
A. E. Boycott (1920s)

- First to study an example of maternal effect
- Involved morphological features of water snail
  - ***Limnea peregra***
    - Shell and internal organs can be either right- or left-handed
      - Dextral or sinistral, respectively
      - determined by **cleavage pattern of egg after fertilization**
    - Dextral orientation is **more common and dominant**



Effetto MATERNO

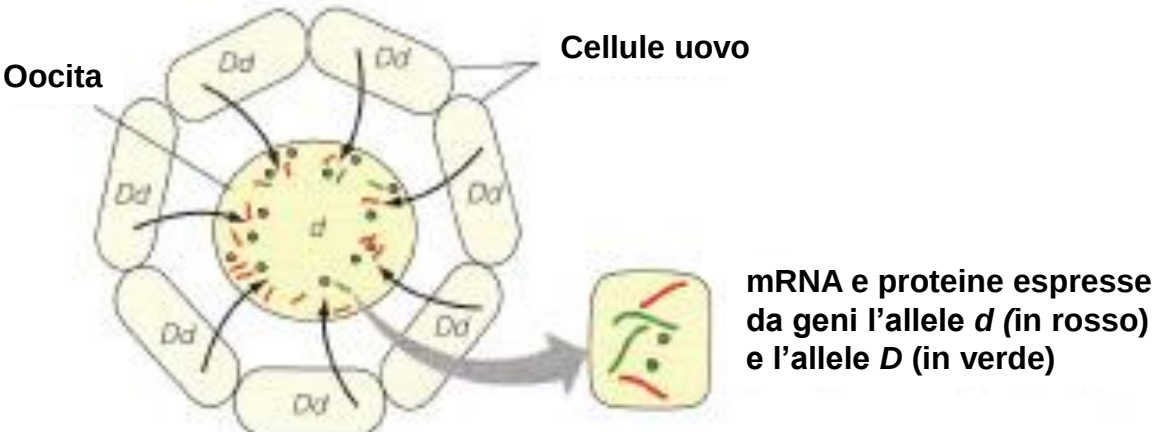
Nell'effetto materno, il genotipo materno ha un'influenza dominante sullo sviluppo embrionale



- Eredità della spiralizzazione della chiocciola *Limnea peregra.*
- La spiralizzazione può essere destrorsa o sinistrorsa.
- L'effetto materno è evidente nelle generazioni II e III, dove il **genotipo del genitore femminile**, e non il genotipo della progenie stessa, **controlla il fenotipo della prole.**

Spiegazione dell'effetto materno

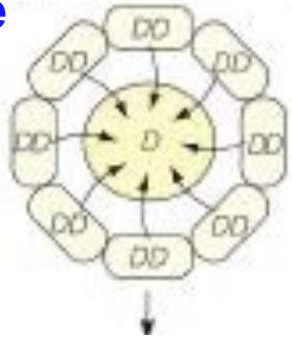
- I geni con effetto materno **codificano per mRNA e proteine** importanti per **l'embriogenesi**
  - l'effetto materno **dipende da prodotti genici** presenti nel citoplasma degli oociti o ricevuti da cellule nutrici



a) **Trasferimento dei prodotti genici dalle cellule nutrici all'ovocita**

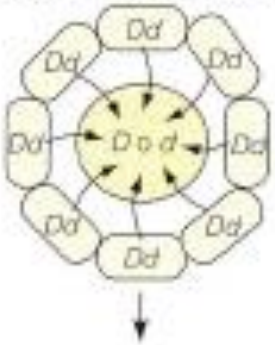
- il fenotipo dipende
  - dalla **presenza dell'allele dominante** nel genotipo femminile
  - è quindi **indipendente dal genotipo maschile**

La madre è *DD*
L'uovo è *D*



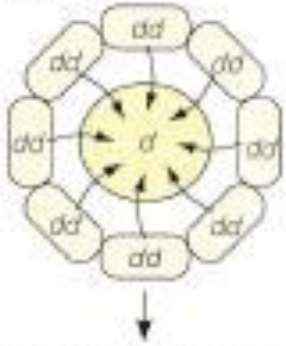
Tutti i discendenti sono destrorsi poiché l'uovo ha ricevuto i prodotti genici dell'allele *D*

La madre è *Dd*
L'uovo può essere *D o d*



Tutti i discendenti sono destrorsi poiché l'uovo ha ricevuto i prodotti genici dell'allele dominante *D*

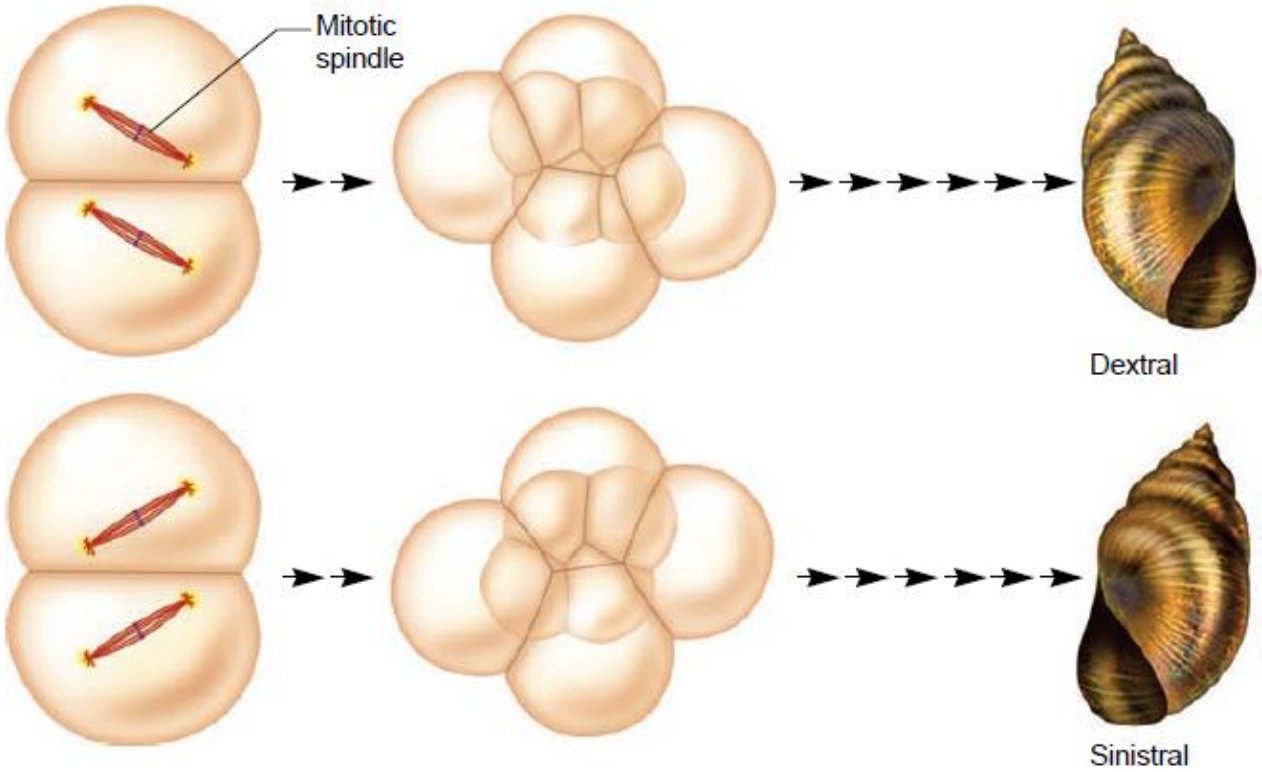
La madre è *dd*
L'uovo è *d*



Tutti i discendenti sono sinistrorsi poiché l'uovo ha ricevuto i prodotti genici dell'allele *d*

b) **Effetto materno nella torsione della chiocciola**

An explanation of coiling direction at the cellular level



The direction of snail coiling is determined by differences in the cleavage planes during early embryonic development.

Genes → Traits

- If the nurse cells are *DD* or *Dd,* **they will transfer the *D* gene product to the egg and thereby cause the resulting offspring to be dextral.**
- If the nurse cells are *dd*, only the *d* gene product will be transferred to the egg, so the resulting offspring will be sinistral

2. EXTRANUCLEAR INHERITANCE

- Most genes are found in the cell's nucleus
- **Some genes are found outside of the nucleus**
  - Some organelles possess genetic material
  - Resulting phenotypes display non-Mendelian inheritance patterns

    - "**Extranuclear inheritance**"
    - "**Cytoplasmic inheritance**"



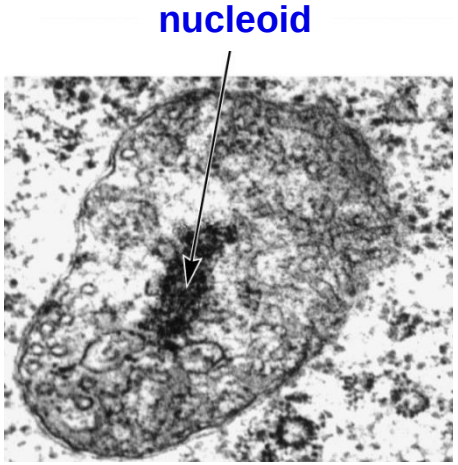
I genomi degli organuli

Caratteristiche comuni

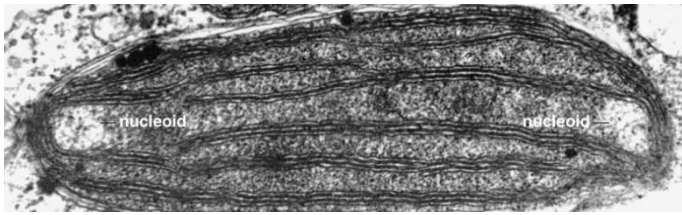
- Piccoli, ma essenziali
- Elevato numero di copie
- Organizzati in complessi nucleoproteici (nucleoidi)
- Eredità non mendeliana
- Informazione genetica necessaria ma non sufficiente per la completa funzionalità dell'organulo
- Interazione tra i sistemi genetici degli organuli e del nucleo

Extranuclear Inheritance

- **Mitochondria and chloroplasts possess DNA**
  - Circular chromosomes resemble smaller versions of bacterial chromosomes
  - Located in the **nucleoid** region of the organelles
    - Multiple nucleoids often present
    - Each can contain multiple copies of the chromosome



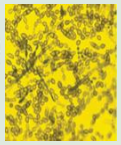
Species	Organelle	Nucleoids per Organelle	Total Number of Chromosomes per Organelle
Tetrahymena	Mitochondrion	1	6–8
Mouse	Mitochondrion	1–3	5–6
Chlamydomonas	Chloroplast	5–6	~80
Euglena	Chloroplast	20–34	100–300
Higher plants	Chloroplast	12–25	~60



Extranuclear Inheritance

- The **inheritance pattern** of mitochondria and plastids **varies among different species**

Transmission of Organelles Among Different Species

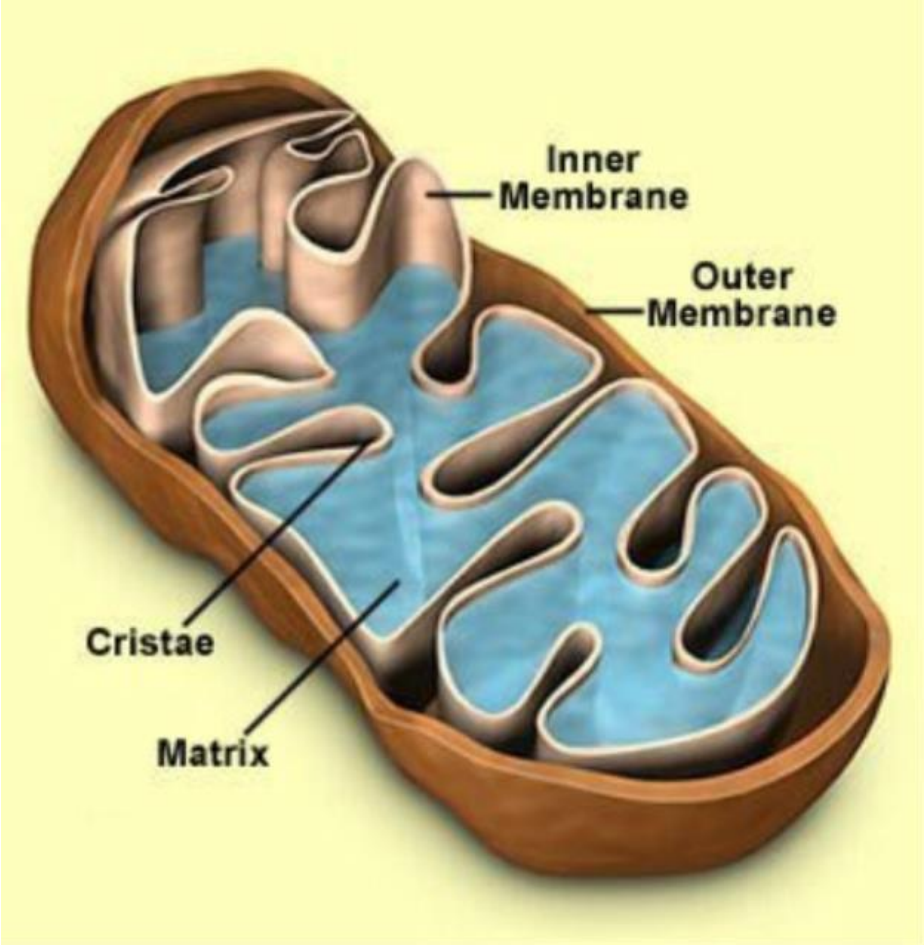
Species	Organelle	Transmission
Mammals	Mitochondria	Maternal inheritance
Mussels 	Mitochondria	Biparental
S. cerevisiae	Mitochondria	Biparental
Molds 	Mitochondria	Usually maternal inheritance; paternal inheritance has been found in the genus *Allomyces*
Chlamydomonas	Mitochondria	Inherited from the parent with the mt^- mating type
Chlamydomonas	Chloroplasts	Inherited from the parent with the mt^+ mating type
Plants		
Angiosperms	Mitochondria and plastids	Often times maternally inherited although biparental inheritance is not uncommon among many species
Gymnosperms	Mitochondria and plastids	Usually paternal inheritance

A)The mitochondrial genome

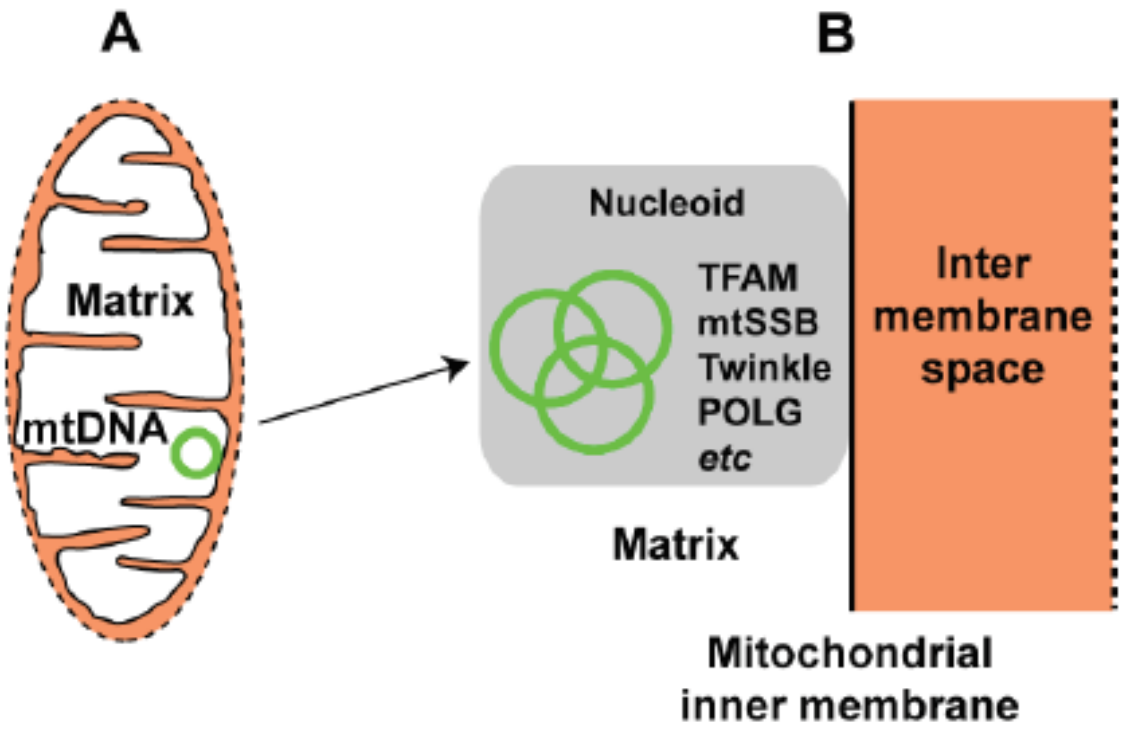


- 1-10 μm small
- Mitochondria are **present in the cytoplasm** of

- **all eukaryote cells** of animals and higher plants
- and also in some microorganisms (algae, fungi, protozoa).



Structural organization of mitochondrial DNA, mtDNA



(A) Mitochondria **have own genetic materials** within the matrix.

(B) mtDNA nucleoid structure.

Several **mtDNA molecules** within a nucleoid **associate to mitochondrial inner membrane** through the complex formed with nucleoid proteins such as TFAM, mtSSB, Twinkle, POLG and so on

Mitochondrion plays a role in:

- **Energy production** → **Oxidative phosphorilation (OXPHOS)**
- **Maintaining the intracellular homeostasis**
- **Protecting** the rest of the cell from reactive oxygen species (ROS)
- **Apoptosis** → important development and disease

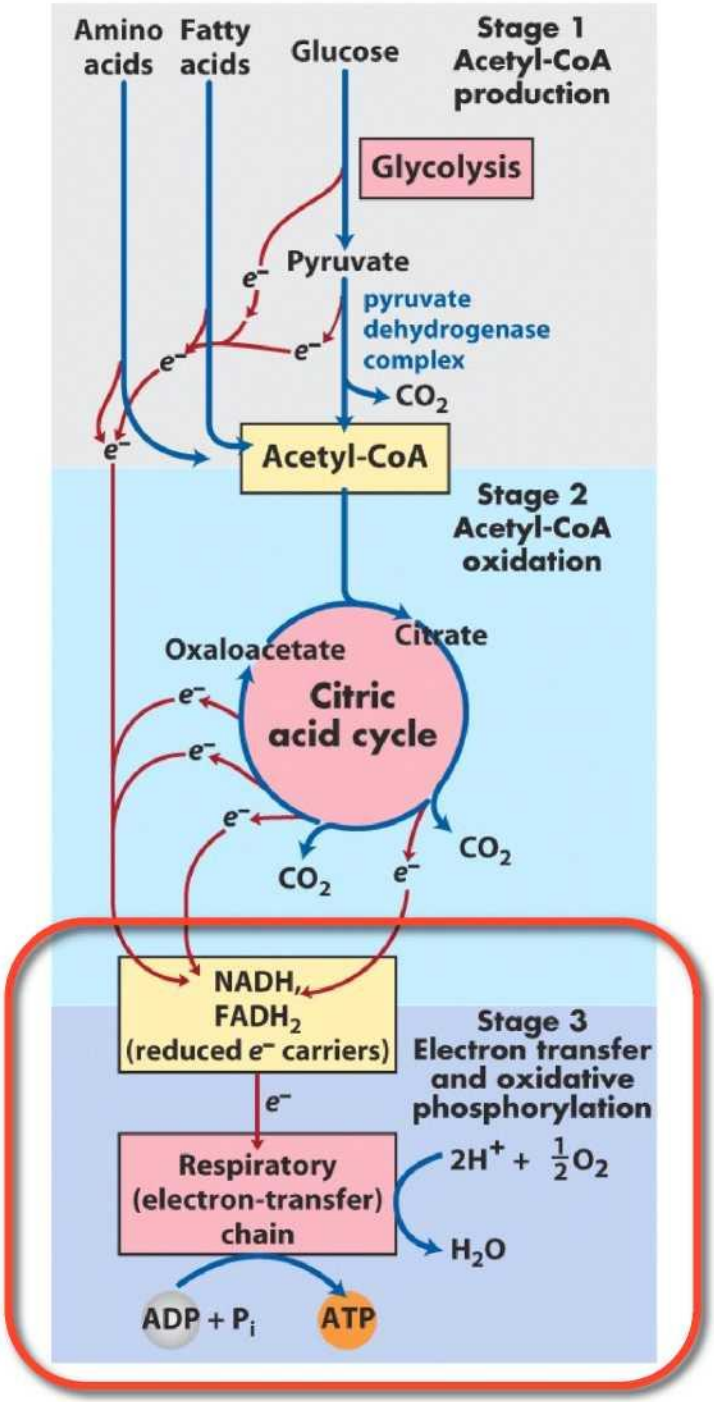
- **La fosforilazione ossidativa rappresenta il culmine del metabolismo energetico negli organismi aerobi**
- **E' costituita da due fenomeni strettamente accoppiati :**

1. **I coenzimi ridotti prodotti durante le reazioni ossidative vengono ossidati nella catena di trasporto degli elettroni, lungo la quale gli elettroni vengono trasferiti all'ossigeno molecolare**

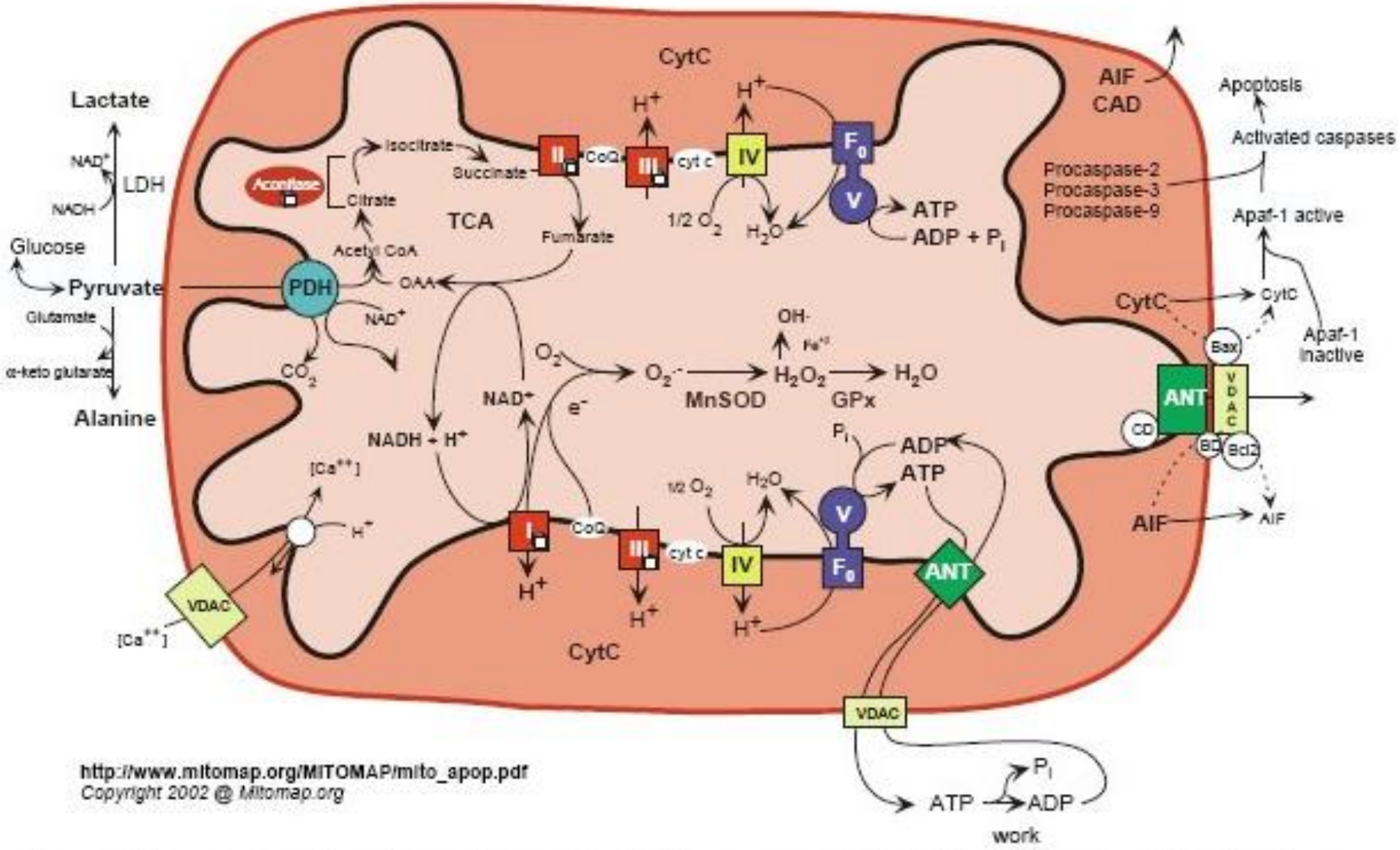
La catena respiratoria di trasporto degli elettroni è un sistema di complessi proteici localizzati nella membrana mitocondriale interna (membrana plasmatica delle cellule procariotiche)

Tre di questi complessi sono ossidoreduttasi che pompano protoni, cioè generano un gradiente di protoni attraverso la membrana mitocondriale interna

2. **Il gradiente di protoni è una fonte di energia libera che viene dissipata quando i protoni passano nuovamente attraverso la membrana mitocondriale interna tramite il complesso ATP sintasi In questo processo si forma ATP**



Mitochondrial Energetics

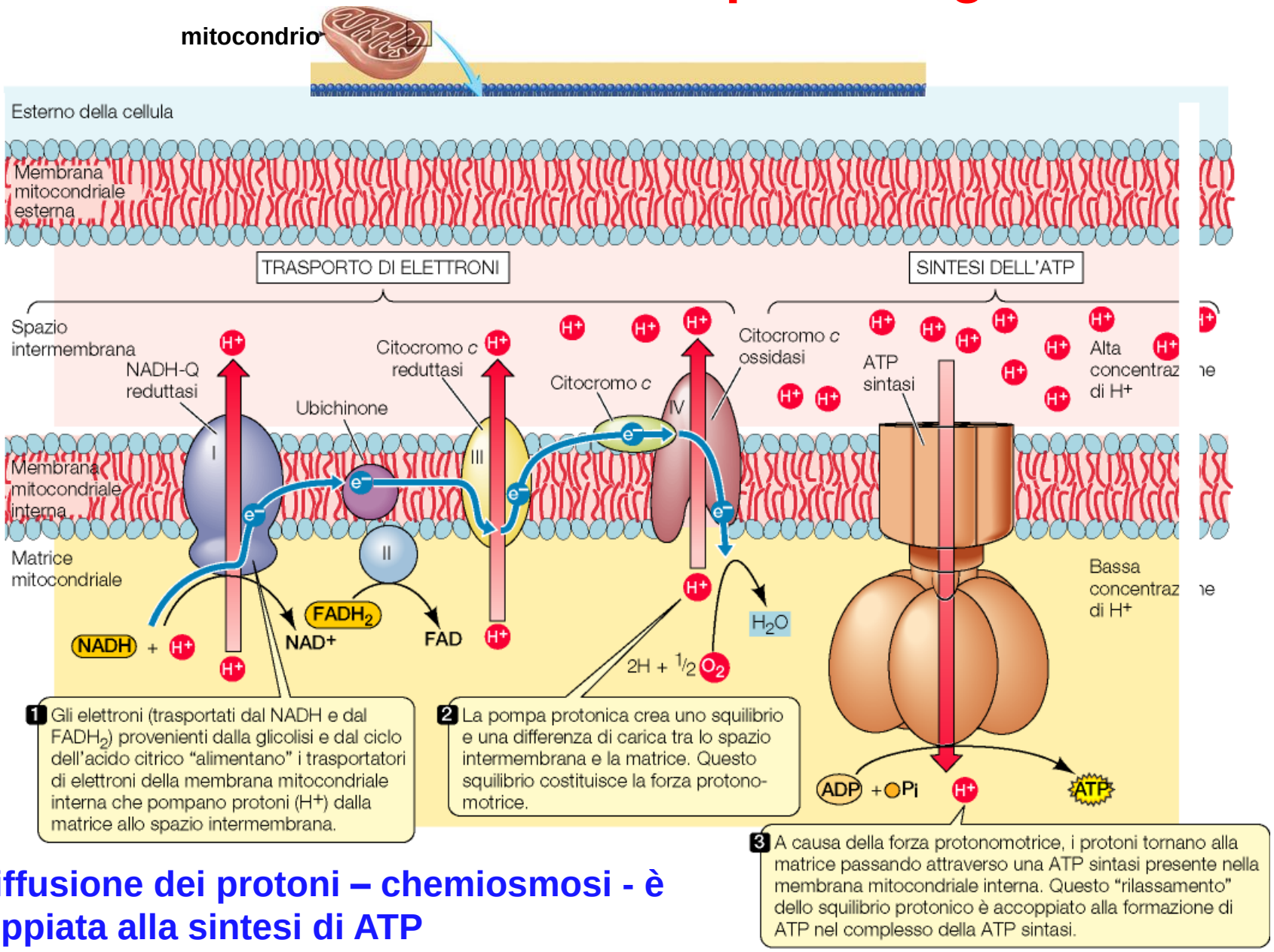


http://www.mitomap.org/MITOMAP/mito_apop.pdf

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Diagram of the mammalian mitochondrion showing the relationship between energy production, ROS generation, and regulation of apoptosis.

Chemiosmosi e catena di trasporto degli elettroni



La diffusione dei protoni – chemiosmosi - è accoppiata alla sintesi di ATP

EXTRANUCLEAR INHERITANCE

- **Mitochondrial genome**
  - size varies greatly among different species
    - **400-fold variation in mitochondrial chromosome size**
      - Mitochondrial genomes of animals tend to be fairly small
      - Mitochondrial genomes of fungi, algae, and protists tend to be intermediate in size
      - Mitochondrial genomes of plants tend to be fairly large

Diversità dei genomi mitocondriali

	Animals	Fungi	Plants
Size	14kb - 42kb	17kb - 180kb	184kb - 2,400kb
Non-coding DNA	Very Low	Variable	Very High
Mutation rate	High	Low	Very Low
Recombination	×	✓/×	✓
Introns	×	×	✓
Universal genetic code	×	Mostly	✓

Contenuto genico

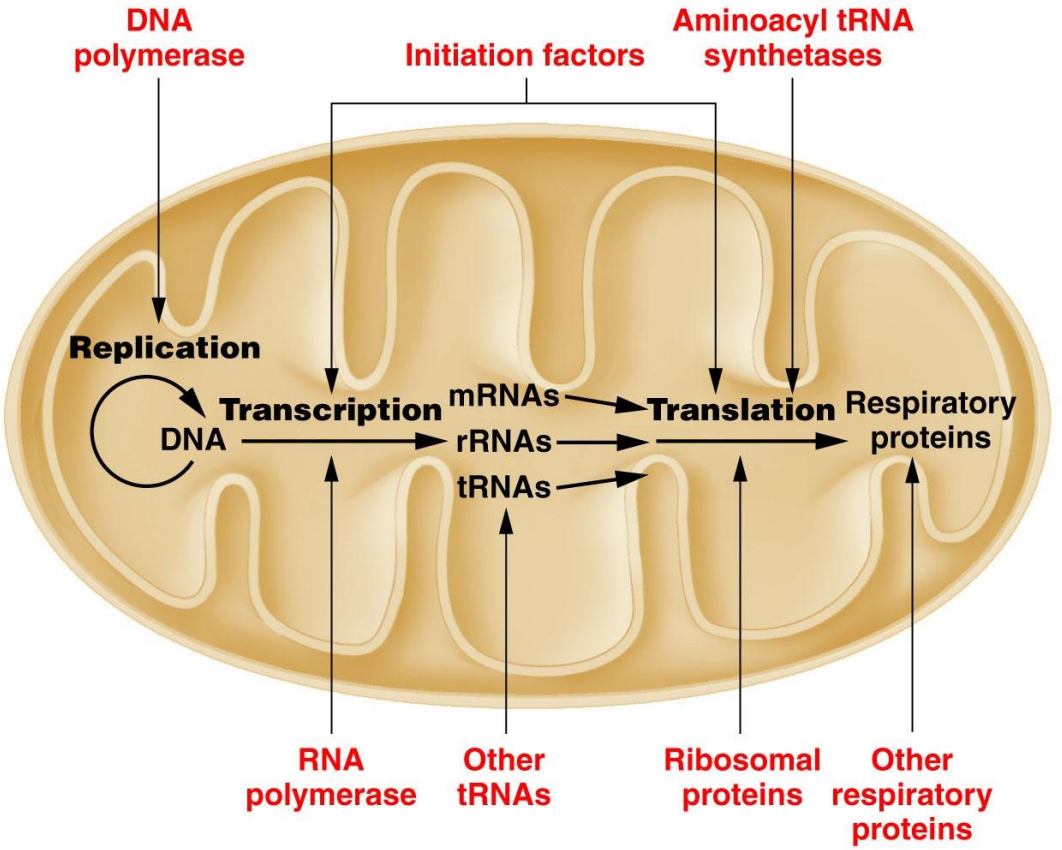
- Il contenuto in geni dei genomi mitocondriali è sostanzialmente conservato
  - comprende **essenzialmente geni coinvolti nella respirazione e nella traduzione.**
- Meno frequentemente vi sono geni per l'import e la maturazione delle proteine e il processing dell'RNA.
- Questi **geni derivano dal progenitore "-proteo-batterico"**.
- Il numero di **geni codificanti proteine** varia da **circa 100** nel protozoo *Reclinomonas americana* a 5 nel *Plasmodium falciparum.*
- Si osserva invariabilmente la presenza di 2 o 3 rRNA, mentre il numero di tRNA è estremamente variabile, da nessuno a >30.
- Una compilazione aggiornata delle caratteristiche dei genomi mitocondriali è disponibile nella banca dati GoBase:

http://gobase.bcm.umontreal.ca/searches/gene.php

Extranuclear Inheritance

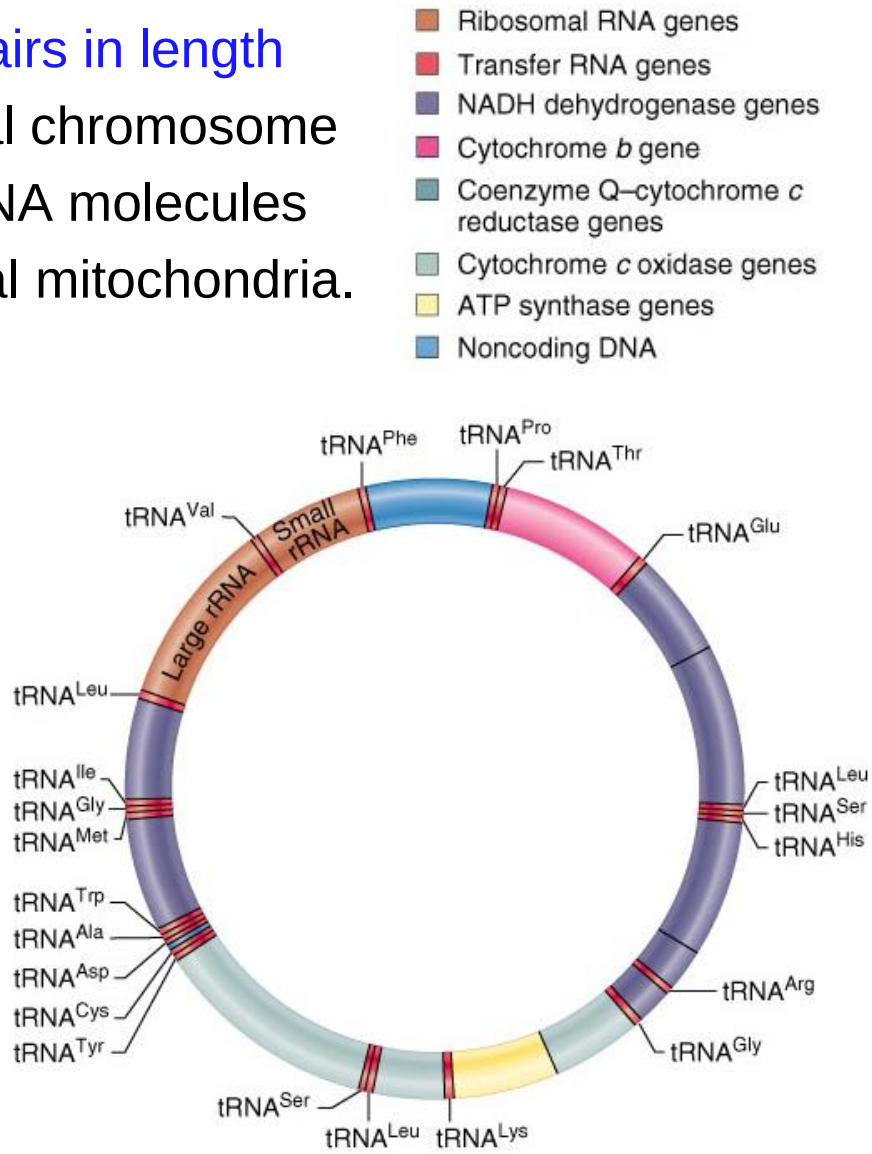
- **Most mitochondrial proteins are encoded by genes in the cell's nucleus**
  - Proteins are synthesized in the cytosol and transported into the mitochondria

Those shown entering the organelle are derived from the cytoplasm and encoded by nuclear genes.

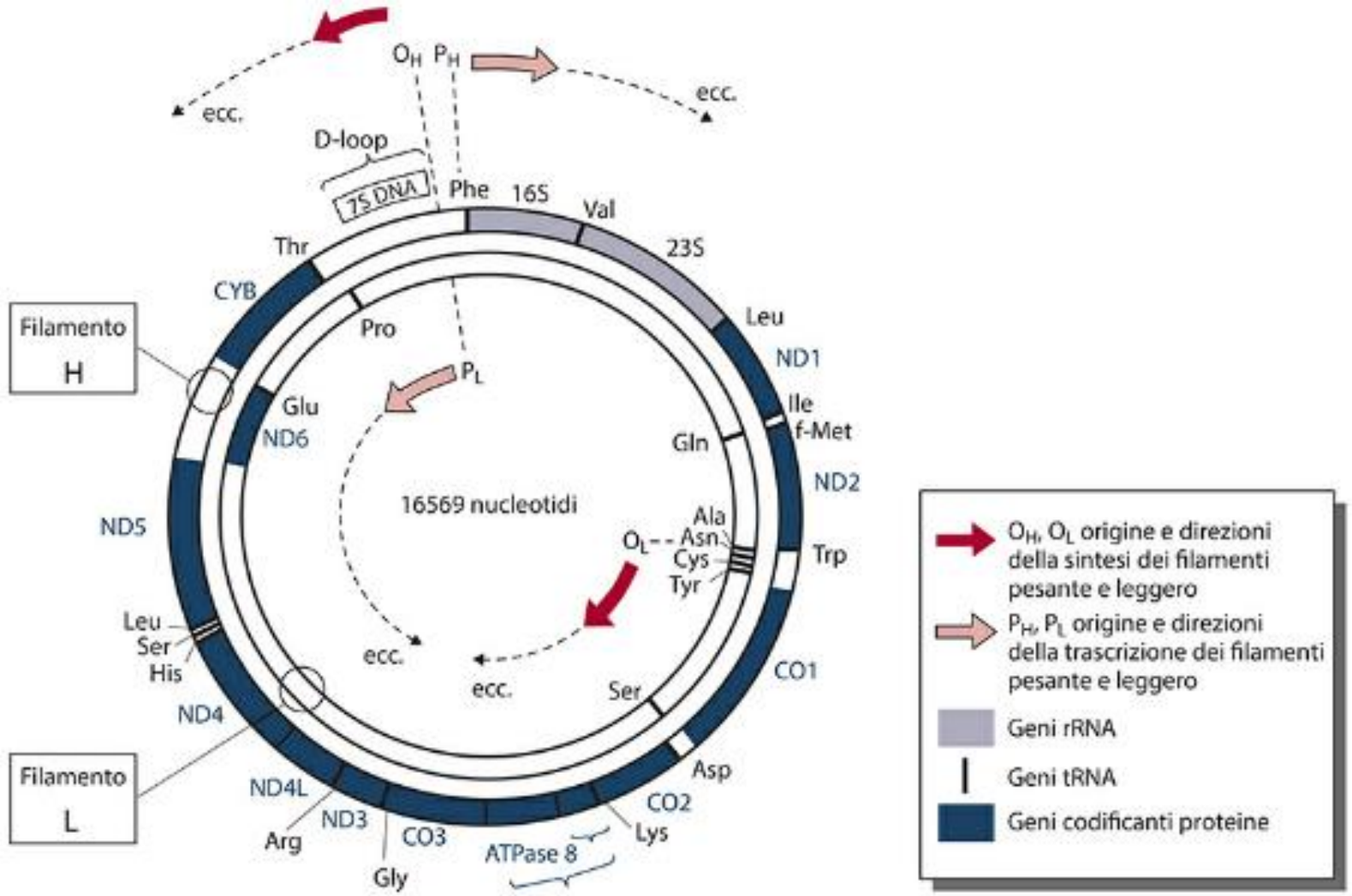


Extranuclear Inheritance

- **Human mitochondrial DNA is called mtDNA**
  - Circular chromosome 17,000 base pairs in length
    - Less than 1% of a typical bacterial chromosome
  - Most cells contain at least 1000 mtDNA molecules distributed among hundreds of individual mitochondria.
  - Not all the RNA and protein synthesized in a cell are encoded in the DNA of the nucleus.



DNA mitocondriale nell'uomo



Il genoma mitocondriale è una molecola circolare a doppio filamento (H e L), costituita da 16569 paia di basi contenenti 37 geni, di cui 13 producono mRNA, 2 rRNA e 22 tRNA. (Modificata da: Strachan T, Read AP. Human molecular genetics. 2nd ed. New York-London: Garland Science; 1999.)

Geni del DNA mitocondriale umano

- **Il genoma mitocondriale contiene 37 geni:**

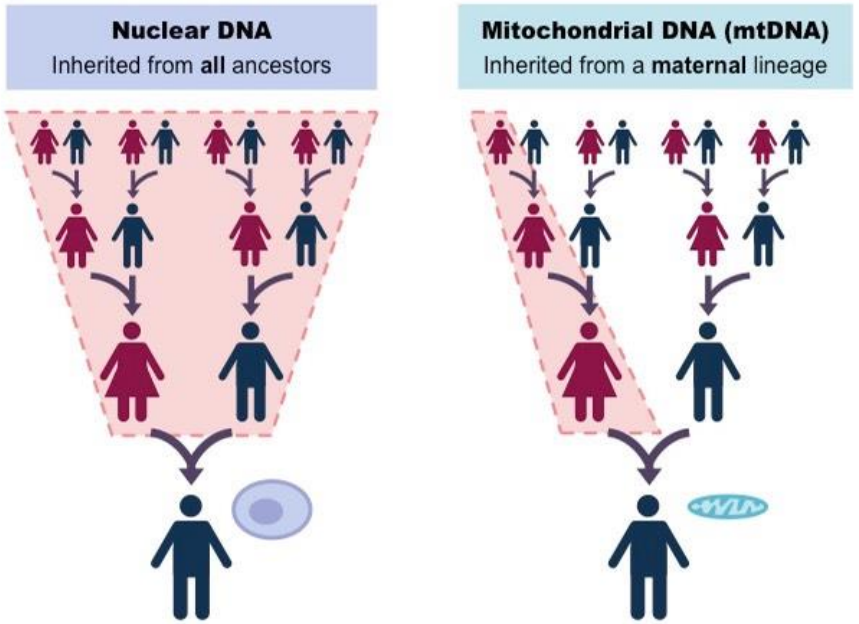
• 28 sul filamento H • 9 sul filamento L 13 geni codificano subunità proteiche dei complessi della catena respiratoria o del sistema della fosforilazione ossidativa:
• 7 subunità del complesso I (NADH-CoQ) ossidoreduttasi
• 3 subunità del complesso IV (citocromo c ossidasi)
• 1 subunità del complesso citocromo b (CoQH2-citocromo c reduttasi)
• 2 subunità del complesso V (ATP sintetasi)

- I complessi della catena respiratoria contengono 75 subunità codificate dal DNA nucleare, importate dal citoplasma nel mitocondrio

24 geni, che codificano molecole indispensabili alla sintesi di tali subunità:
- 2 RNA ribosomiali (rRNA)
- 22 RNA transfer (tRNA)

CHARACTERISTICS

- It is **inherited exclusively from the mother**!
- mtDNA is a c**ircular shape single** chromosome
- It is only 16 kb in length - contains 16,600 bp
- Codes for 37 genes
- Contains 22 tRNA and 2 rRNA coding genes



- Encodes **13 proteins that are subunits** of **oxidative phosphorilation**
- Contains only exons, no introns
- Has no reparation system – high mutation rate especially in D-loop!
- **No crossing over**
- **Replicative segregation, homoplasmy & heteroplasmy**

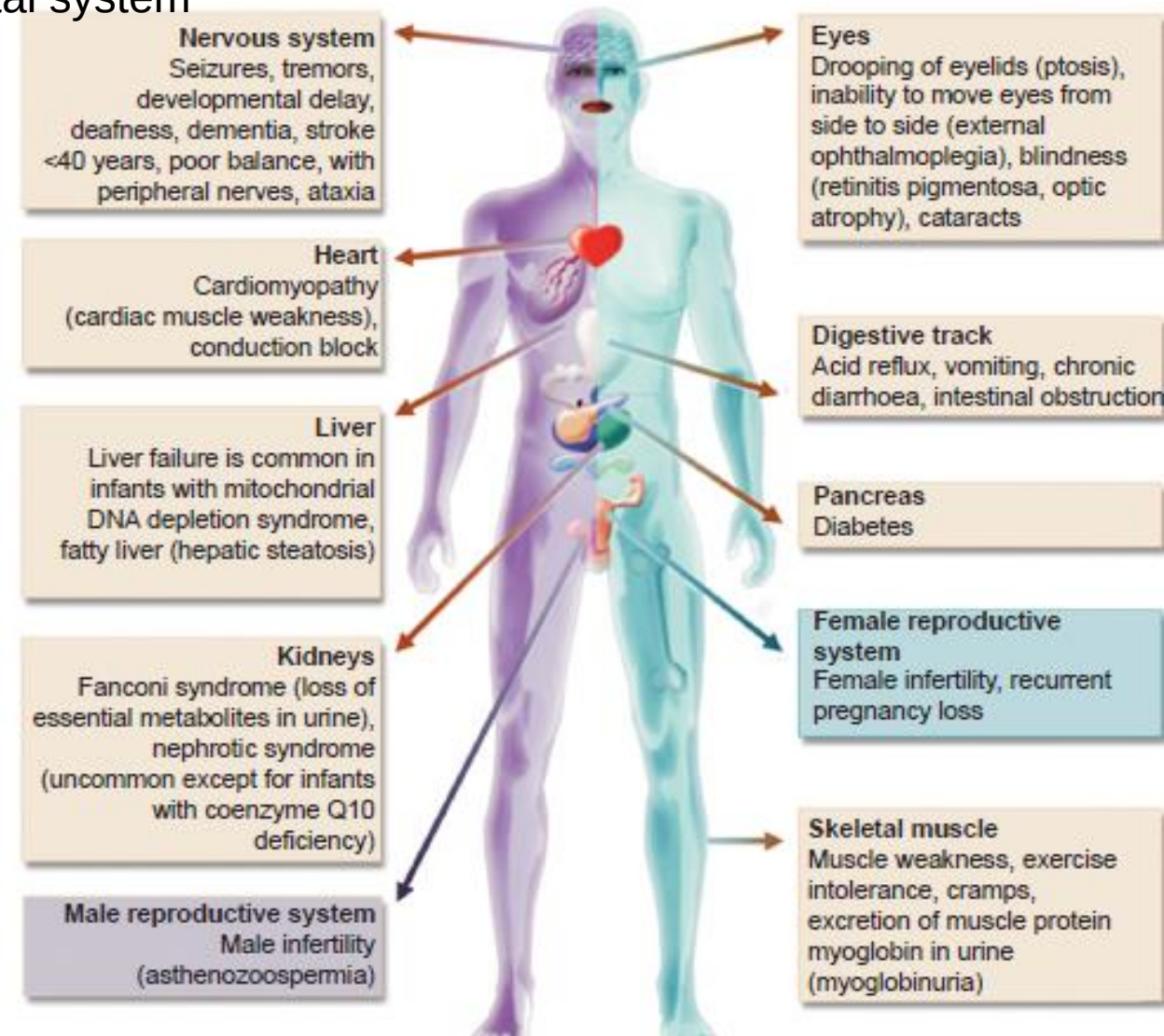
Mitochondrial disorders

- More than **100 different rearrangements and 100 different point mutations** have been identified in mtDNA that can cause human disease, often involving the CNS and musculoskeletal system

- **can be caused by mutations**: in mitochondrial DNA (mtDNA) or in nuclear genes that code for mitochondrial components.

- Can be **acquired mitochondrial dysfunction** due to adverse effects of drugs, infections, or other environmental causes

- **Most sensitive cells are:**
  - **Neurons**
  - **Muscle cells**



Clinical presentation of mitochondrial syndromes

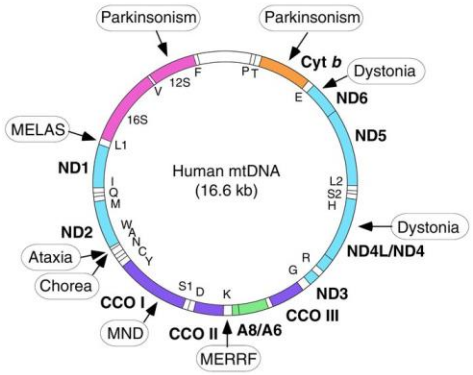


Table. Clinical presentations of mitochondrial syndromes

Syndrome	Common genotype	Age at onset	Clinical features
Mitochondrial encephalopathy with lactic acidosis and stoke-like episodes (MELAS)	Point mutations mtDNA (>80% *MTTL1* gene) (m.3243A>G)	Late childhood or adulthood	Stroke-like episode with seizures. Other features include intermittent episodes of encephalopathy, vomiting, migraine, diabetes mellitus, cardiomyopathy, SNHI, pigmentary retinopathy, cerebellar ataxia.
Myoclonic epilepsy with ragged red fibers (MERRF)	Point mutation mtDNA (*MTTK*) (m.8344A>G, m.8356C>T and m.8363 G>A)	Childhood or early adulthood	Myoclonus, epilepsy, cerebellar ataxia and myopathy; other features include dementia, optic atrophy, bilateral SNHI, pyramidal signs, spasticity, multiple lipomas.
Neuropathy, ataxia and retinitis pigmentosa (NARP)	Point mutation mtDNA (*MTATP6 / MTATP8*) (m.8993T>G)	Late child or adult	Peripheral neuropathy, ataxia, pigmentary retinopathy.
Leber hereditary optic neuropathy (LHON)	Point mutation mtDNA (*MTND1 / MTND4/ MTND6*)	Adult	Optic neuropathy with sub-acute bilateral deterioration in vision; 95% of patients harbouring one of three common point mutations: m.11778G >A, m.3460G > A or m.14484T >C.
Chronic progressive external ophthalmoplegia (CPEO)	Large-scale single or multiple deletions. Point mutation mtDNA (m.3243A>G and m.12316G>A)	Early adult	External ophthalmoplegia and bilateral ptosis. Other features may include proximal myopathy, cardiac conduction defects, and dysphagia, *etc*.
Maternally inherited Leigh syndrome (MILS)	Point mutation mtDNA(m.8993T>C,m.10158T>C, m.10191T>C)	Infancy and early childhood	Myoclonus, seizures, developmental delay, muscle weakness, problems with movement, heart disease, kidney problems and difficulty in breathing.
Pearson syndrome	Large-scale single mtDNA deletion	Early childhood	Sideroblastic anaemia, pancytopenia, exocrine pancreatic failure, renal tubular defects. Survivors develop Kearns–Sayre syndrome.
Kearns–Sayre syndrome (KSS)	Single/large-scale deletions	Later childhood	Progressive external ophthalmoplegia, pigmentary retinopathy, raised cerebrospinal fluid protein, cerebellar ataxia, cognitive impairment, deafness, cardiomyopathy, complete heart block, short stature, dysphagia and endocrinopathies.

Source: Refs 10, 14, 26, 30, 40-45

http://www.icmr.nic.in/ijmr/2015/janaury/0103.pdf

Genetic characteristics

1. Replicative Segregation
2. Homoplasmy and Heteroplasmy
3. Maternal Inheritance

1. Replicative segregation

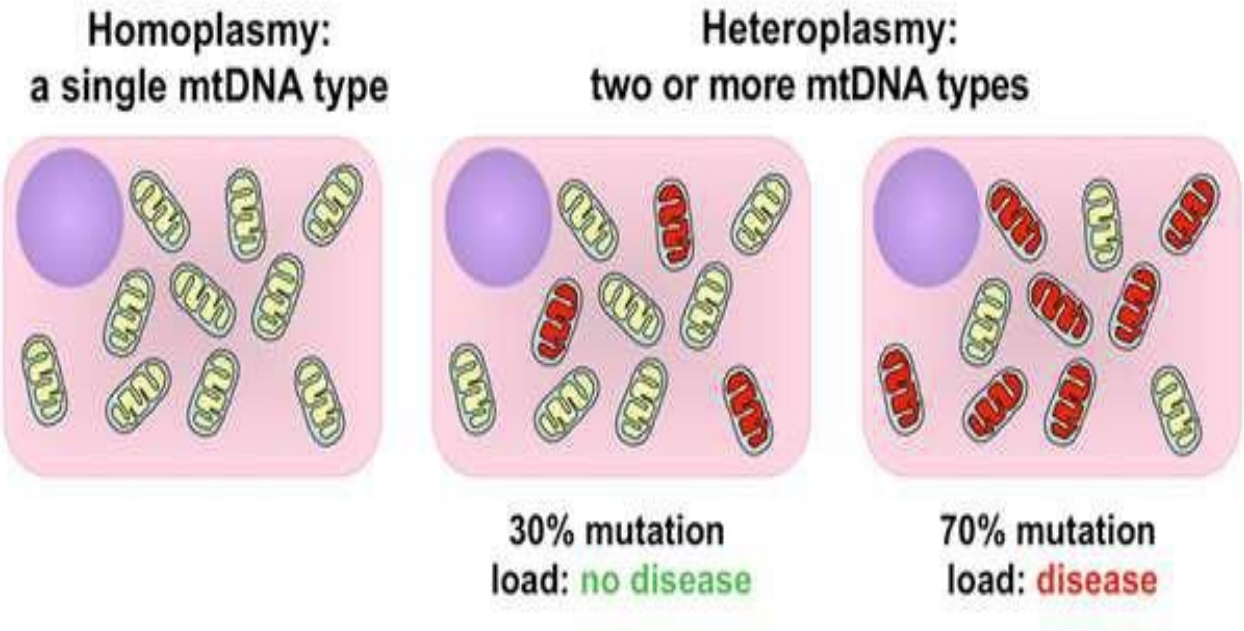
- At cell division, **multiple copies of mtDNA in each of the mitochondria** in a cell
  - **replicate and**
  - **sort randomly**

  among newly synthesized mitochondria.
- The mitochondria in turn **are distributed randomly between the 2 daughter cells.**
  - This is called as **replicative segregation.**
- The first unique feature of mitochondria is the absence of tightly controlled segregation seen during mitosis and meiosis of the 46 nuclear chromosomes.

Homoplasmy and Heterolasmy

- One **daughter cell may by chance receive mitochondria** that contain only **a pure population of normal mtDNA** or a **pure population of mutant** mtDNA **(Homoplasmy)**
- The **daughter cell may receive a mixture** of mitochondria some with and some without mutation **(Heteroplasmy)**

In normal conditions mtDNA is HOMOPLASMIC
Many pathogenic mutations are HETEROPLASMIC



Extranuclear Inheritance:

B) Chloroplast Genome

- organelle found in plant cells and eukaryotic algae
- Photosynthesis

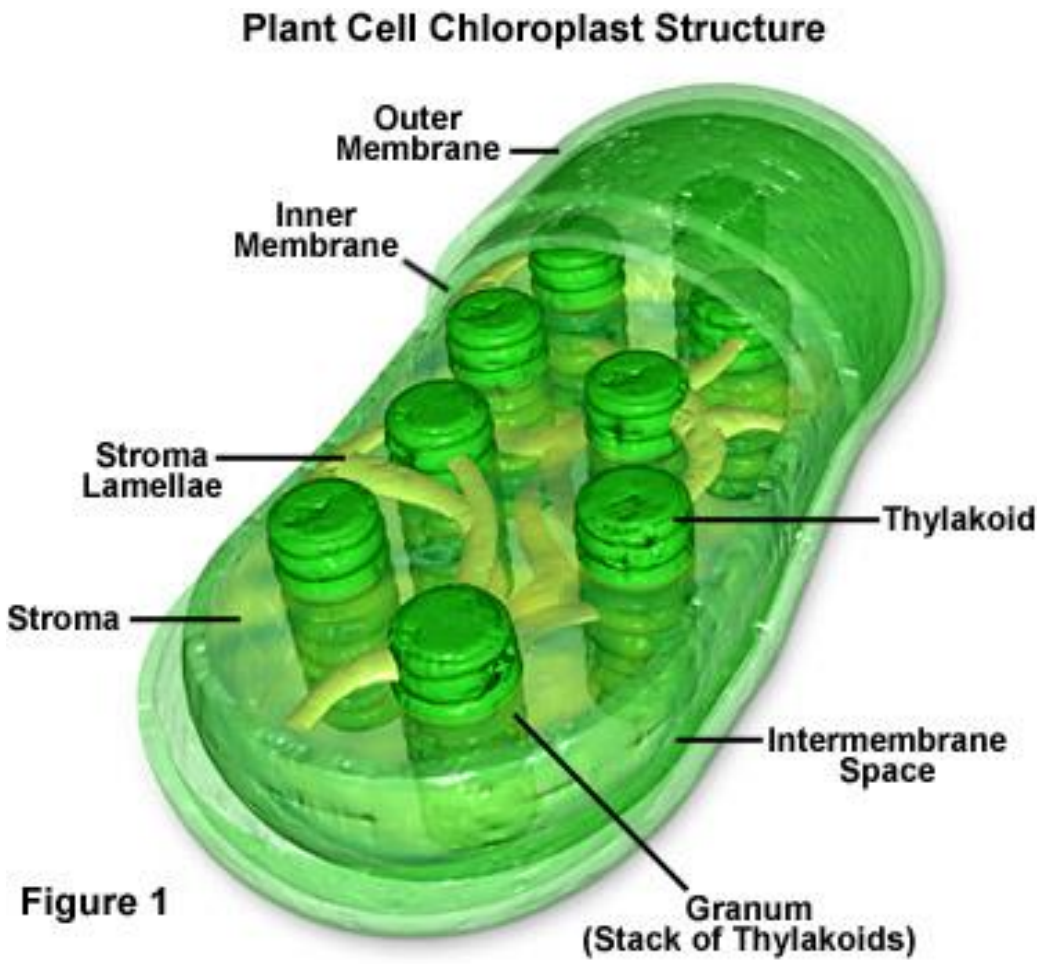


Figure 1

Chloroplast genome: characreristics

- Chloroplast DNA (cpDNA) is also known as plastid DNA (cpDNA).
- **Circular double stranded DNA molecule**
- Tend to be larger than mitochondrial genomes
  - Ten times larger than the mitochondrial genome of animal cells
- Chloroplast genome size ranges **120-217kb** with majority of plants fall into **120-160kb.** (*Pelargonium* has a chloroplast genome size 217kb)
- contain about **100 genes to synthesize proteins**
- cpDNA regions includes Large Single-Copy (LSC) & Small Single-Copy (SSC) regions, and Inverted Repeats (IR_A & IR_B).
- Conifers and a group of legumes lack Inverted Repeats.

Sizes of (plastid) cpDNA

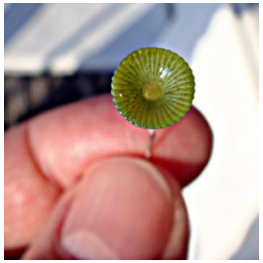
- Range is **70,000 bp (**70 kb) to ~**2,000,000 bp** (2,000 kb), but most are **less than 250,000 bp (250 kb**)

- Land plants typically **120 – 170 kb**

(70 kb – *Epifagus;* ~2,000 kb – *Acetabularia*







Chloroplast genome

- **Complete chloroplast DNA sequences** of four land plants (*Nicotiana tabacum*, *Marchantia polymorpha, Oryza sativa* and *Epifagus virginiana*) were available for comparative study on structure and gene content of chloroplast genomes in 1980s.
- At present, the number of **complete chloroplast genome sequences is 122** (from 114 different organisms).
- eg. *Arabidopsis thaliana, Coffea arabica, Eucalyptus globulus, Glycine max, Gossypium hirsutum, Helianthus annuus, Lycopersicon esculentum, Nymphaea alba, Phaseolus vulgaris, Pinus koraiensis, Piper cenocladum, Solanum tuberosum, Triticum aestivum, Vitis vinifera, Zea mays* etc.

Taxa	Genome size (in kb)	Inverse duplication (in kb)
Angiospermae		
Nicotiana tabacum	156	25
Spinacia oleracea	150	24
Pelargonium hortorum	217	76
Pisum sativum	120	Not present
Epifragus virginiana	70	22
Oryza sativa	134	21
Gymnospermae		
Pinus	120	Not present
Ginkgo biloba	158	17
Pteridophyta		
Osmunda cinnamomea	144	10
Bryophyta		
Marchantia polymorpha	121	10
Chlorophyta		
Codium fragile	85	Not present
Chlamydomonas reinhardtii	195	22
Chlamydomonas moewusii	292	41
Rhodophyta		
Cyanophora paradoxa	127	10
Chromophyta		
Dictyota dichotoma	123	5

Organization of typical (angiosperm) chloroplast chromosome

- inverted repeats (IRa and IRb) separate circle into large and small single-copy regions (LSC and SSC, respectively)
- IRs always contain the rRNA (*rrn*) genes, but also contain other genes
- **~125 genes are found**, encoded on both strands, without much overlap

cpDNA Gene Content

- Most cp genes fall into 2 functional groups:

1. **genes involved in the genetic apparatus** (replication, transcription, translation)
2. **genes involved in photosynthesis**

Also genes for protein degradation, **fatty acid synthesis**, and respiration (chlororespiration?).

Gene nomenclature

- Based on **bacterial naming syste**m, which uses lower case letters, and a descriptive prefix, based on the probable function.
- If the gene product is part of a multi-subunit complex, a letter of the alphabet is used to denote different subunits.

Examples:

***psa* for genes of photosystem** I (*psaA*, *psaB*, etc.)

***psb* for genes of photosystem II** (*psbA*, *psbB*, etc.)

A non-conforming example:

rbc for genes encoding ribulose-1,5-bisphosphate carboxylase (RuBPCase)

- RuBPCase has two subunits, large and small
- the genes are *rbcL* and *rbcS*;
- ***rbcL* is in cpDNA**,
- *rbcS* is encoded in the nucleus

CP GENES (land plants)

prefix	complex/enzyme	no. that are cp-encoded
Photosynthesis:		
psa	photosystem I	5 of >10
psb	" II	15 of >22
pet	cytochrome f/b_6	6 of 8
atp	ATP synthetase	6 of 9
rbc	RuBPCase	1 of 2
cem	inorganic carbon uptake	1
ndh	NADH dehydrogenase	11 of 11(?)
Genetic (transcription, translation, protein assembly/turnover) functions:		
RNAs		
rrn	ribosomal RNAs	3 of 3
trn	transfer RNAs	30-32 (all)
Proteins		
rpo	RNA polymerase	4 of 5
rpl	proteins of ribosome LS	9 of ~30
rps	" " " SS	12 of ~30
inf	transl. initiation factor	1 of 3
mat	maturase (splicing)	1
clp	protease	1 of 2
ycf3, ycf4	photosystem I assembly	
ccs	cytochrome c assembly	1 of ?
Others:		
acc	Acetyl-CoA carboxylase (fatty acid synthesis)	1 of 4

accD →

Still some conserved open-reading frames (ORF) or ycfs of unknown function, e.g. ycf1 and ycf2, which are essential and encode very large proteins.

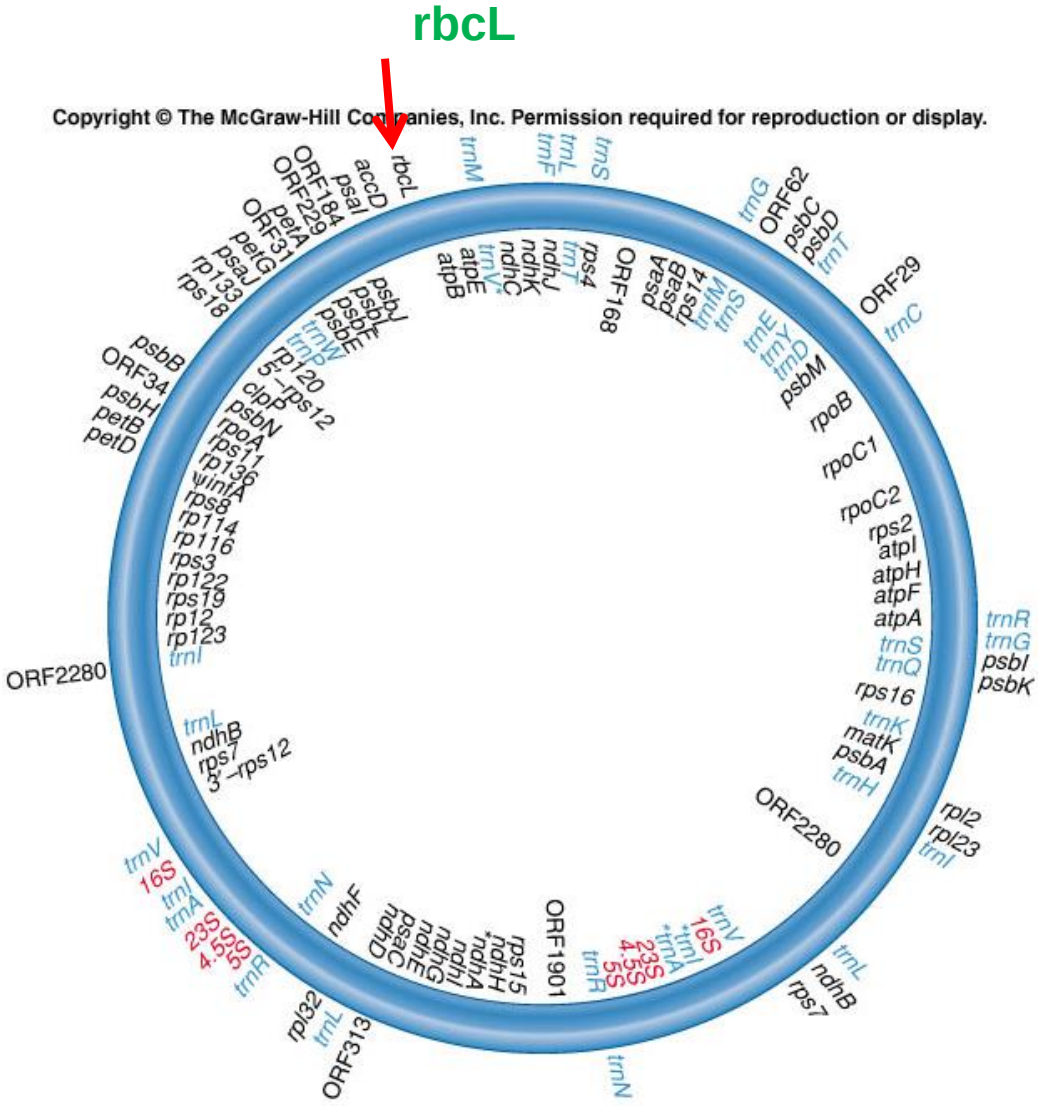
Characteristics of Chloroplast Genome

- cpDNA is a relatively abundant component of plant total DNA, thus facilitating extraction and analysis.
- Conservative rate of nucleotide substitution enables to resolve plant phylogenetic relationships at deep levels of evolution.
- *e.g.* familial level; mono- & dicotyledonous;
- **Chloroplast protein-coding genes evolve at a rate** that is on average

  five-fold slower than plant nuclear genes.

EXTRANUCLEAR INHERITANCE

- **Chloroplast DNA (cpDNA) of the tobacco plant**
  - 156,000 bp circular DNA molecule
  - 110 – 120 different genes
    - rRNAs, tRNAs, and many proteins required for photosynthesis
    - Many chloroplast proteins are encoded in the nucleus



Characteristics of Chloroplast Genome

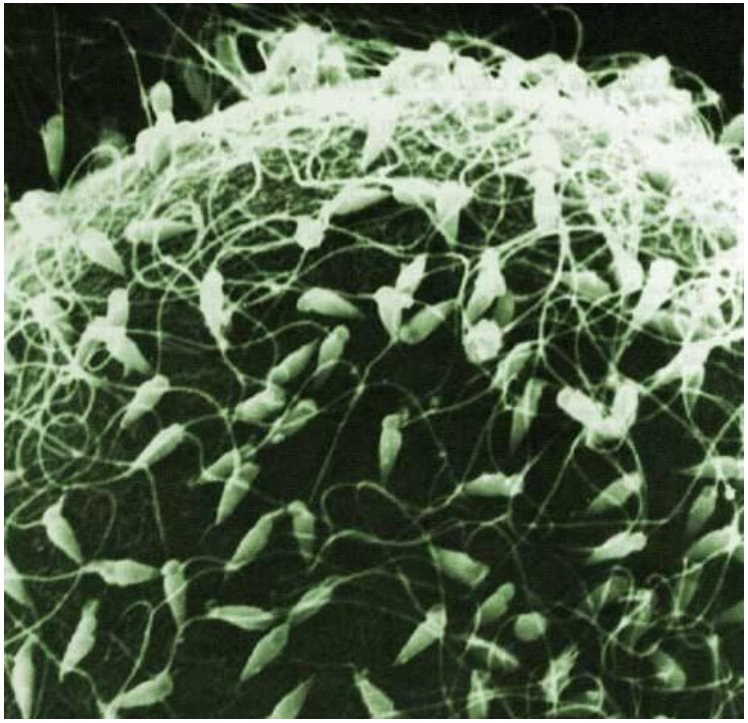
- **Strictly maternally inherited** in most angiosperms while in conifers, inheritance is paternal.
- Chloroplast DNA is passed on from one generation to the next with only an occasional mutation altering the molecule;
- **sexual recombination does not occur.**

Extranuclear Inheritance

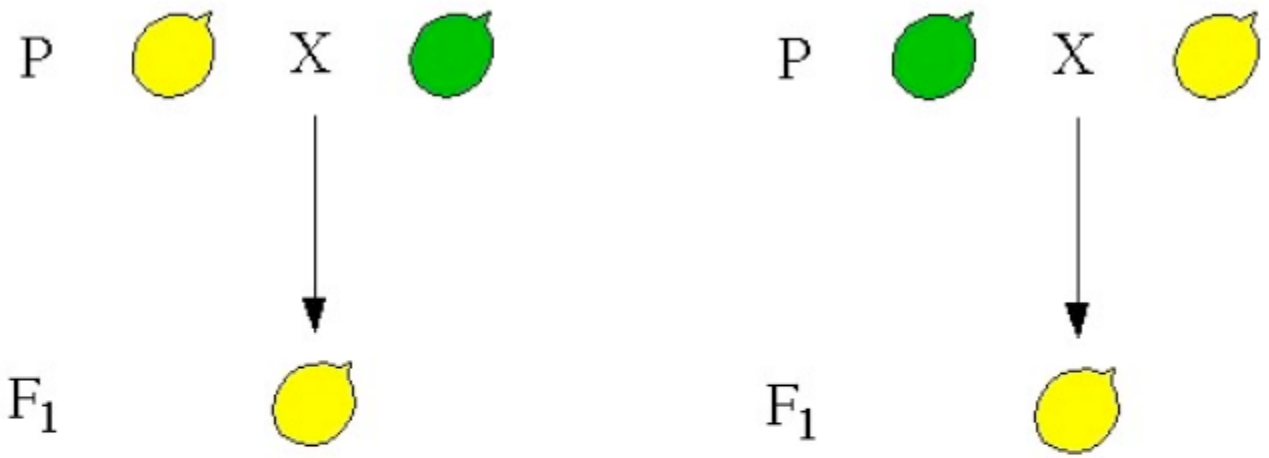
- Most nuclear genes in diploid eukaryotes display Mendelian inheritance patterns
  - Homologous chromosomes segregate during gamete production
  - Offspring inherit one copy of each gene from each parent
- **The inheritance pattern of extranuclear genetic material displays non-Mendelian inheritance**
- Mitochondria and plastids do not segregate into gametes as do nuclear chromosomes

Extranuclear Inheritance

- Many organisms are **heterogametic**
  - Two kinds of gametes are made
    - Female gamete tends to be large and provides most of the cytoplasm to the zygote
    - Male gamete is small and often provides little more than a nucleus
  - **Mitochondria and plastids are most often inherited from the maternal parent**

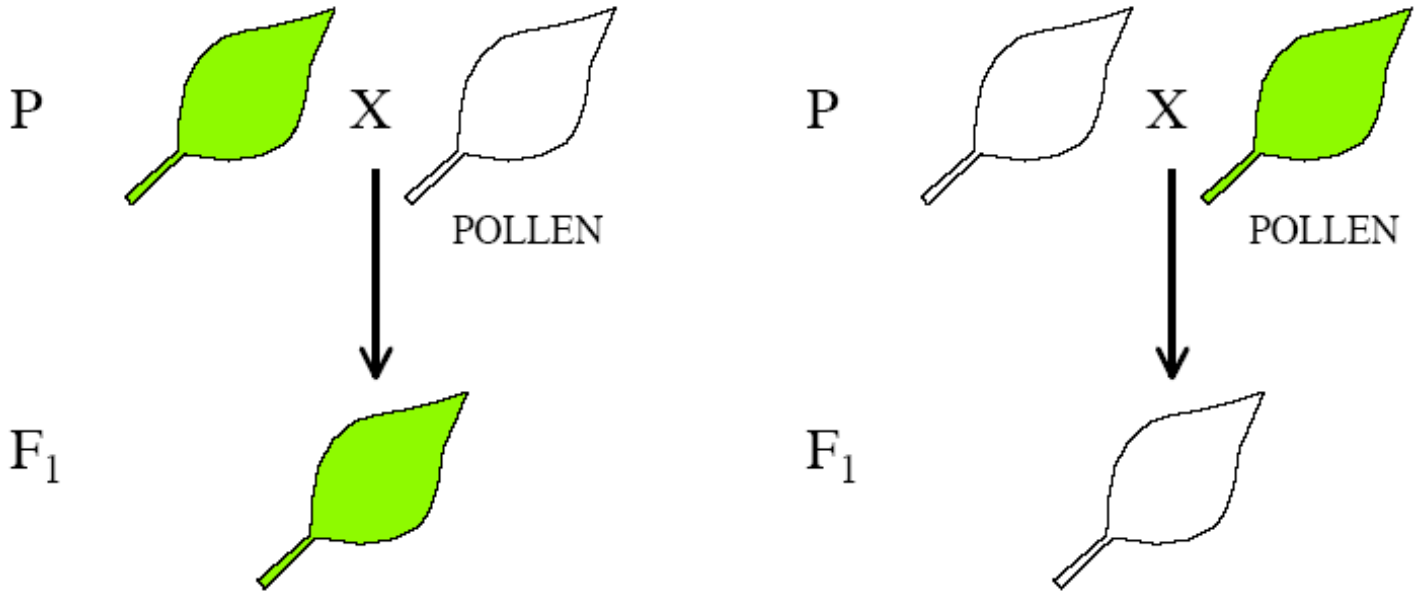


Nell'eredità nucleare incroci reciproci producono identiche F_1



Mendel used ***reciprocal crosses***, i.e. where the traits inherited maternally and paternally were reversed, to show that the trait observed in the F_1 was the same regardless of the source

Nell'eredità materna incroci reciproci danno luogo ad F1 diverse



- Il fenotipo del genitore materno (ovulo) è solo responsabile
- per determinare il fenotipo della progenie

Questo è eredità strettamente materna

Extranuclear Inheritance

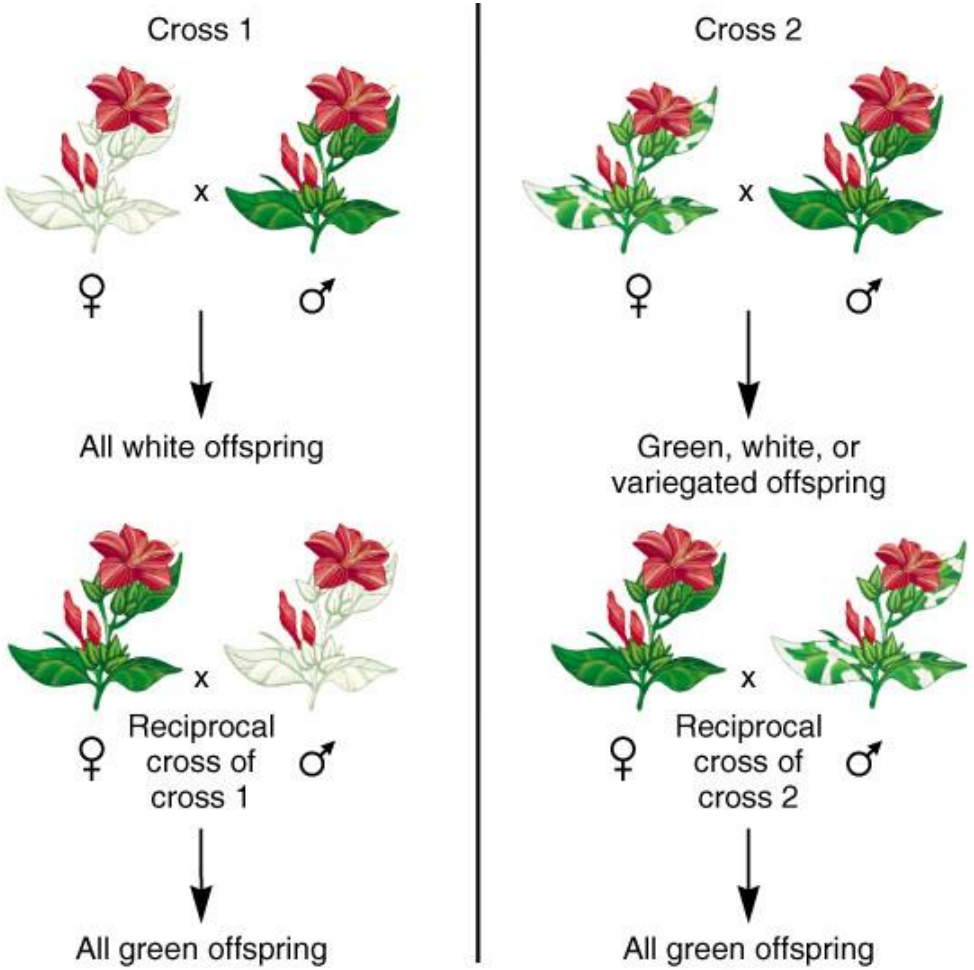
- **Pigmentation in *Mirabilis jalapa***
  - The four-o'clock plant
  - Pigmentation is determined by chloroplast genes
    - Green phenotype is the wild-type condition
      - Green pigment is formed
    - White phenotype is due to a mutation in a chloroplast gene
      - Synthesis of green pigment is diminished
    - Cells containing both types of chloroplasts display green coloration
      - Normal chloroplasts produce pigment
      - "Heterotroplasmy"

EXTRANUCLEAR INHERITANCE

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  - Cells containing both types of chloroplasts display green coloration
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    - "Heterotroplasmy"

- Pigmentation in the offspring depends solely on the maternal parent
  - "Maternal inheritance"
  - Chloroplasts are inherited only through the cytoplasm of the egg

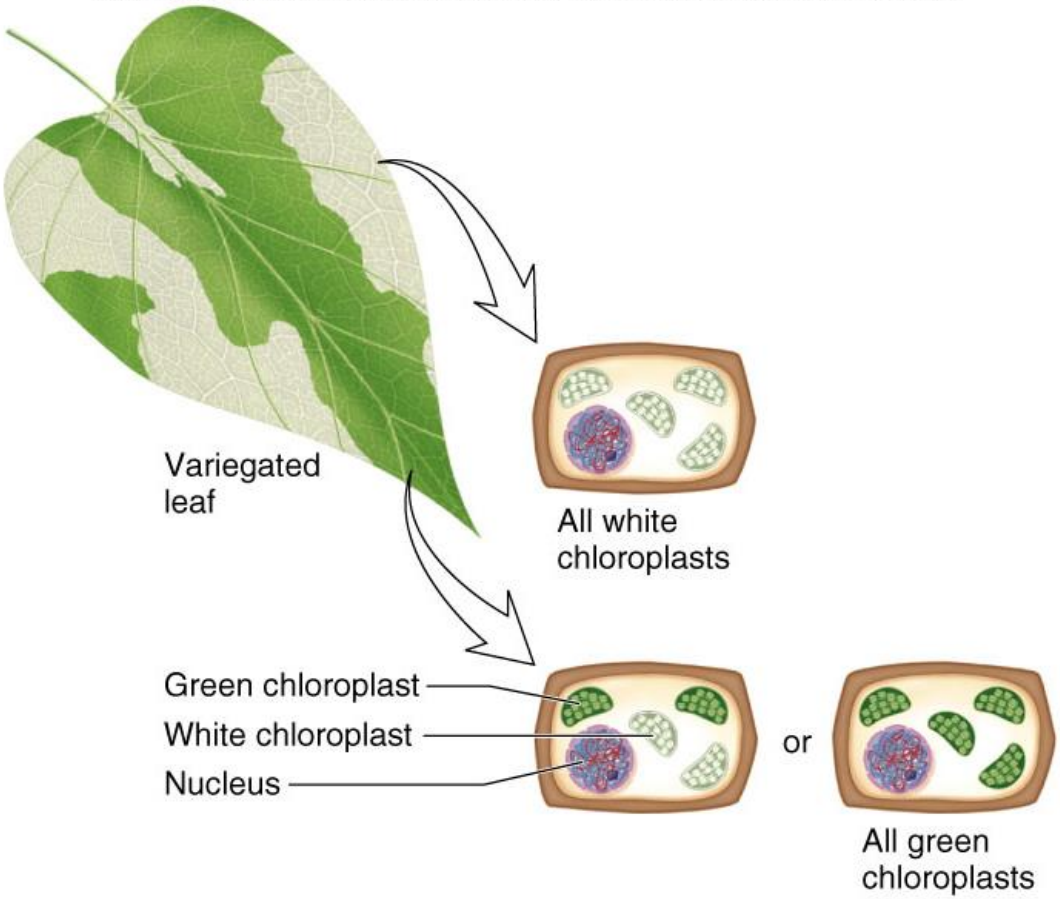


Extranuclear Inheritance

- A cellular explanation of the **variegated phenotype** in *Mirabilis jalapa.*
  - Cells can contain both types of chloroplasts
    - Coloration is green because pigment is produced
  - Chloroplasts are irregularly distributed to daughter cells during cell division

    (Cytoplasmatic segregation)
    - Some cells may receive only chloroplasts defective in pigment synthesis
      - The sector of the plant arising from such a cell will be white
    - Variegated phenotype

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Extranuclear Inheritance

- Studies in yeast and unicellular algae provided genetic evidence for extranuclear inheritance of mitochondria and chloroplasts
  - e.g., *Saccharomyces cerevisiae*
  - e.g., *Chlamydomonas reinhardtii*

Ciclo vitale e sessuale del lievito *S. Cerevisiae*

- **Organismo unicellulare**
  - nucleo aploide
  - si riproduce di norma per gemmazione

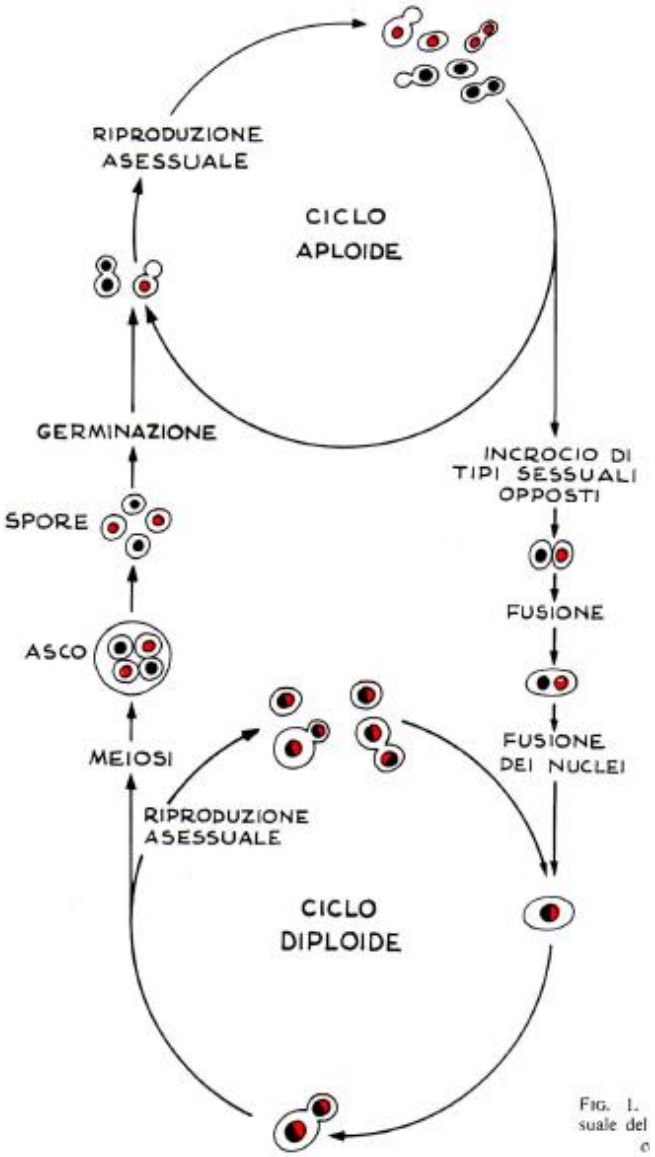


FIG. 1. suale del

1. Yeast can grow either

- anaerobically by fermentation (slow growth) or

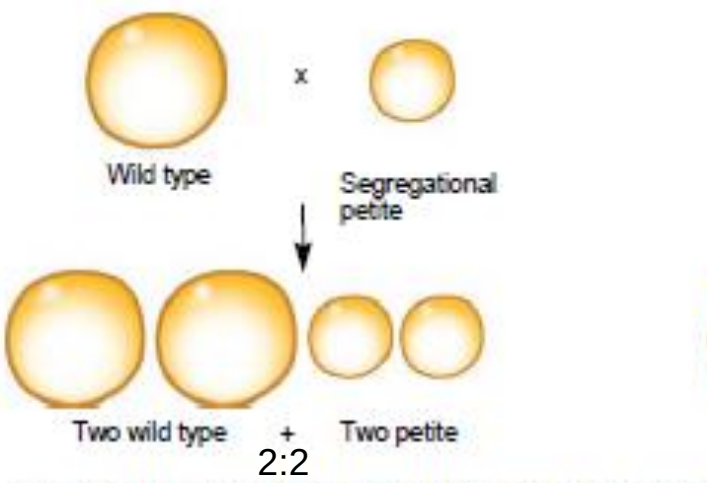
- **aerobically** using mitochondria (fast growth), forming colonies from single cells on solid media.

2. **Yeast petite colonies are much** smaller than those formed by wild-type cells,

- this is due to **cytochrome deficiencies that prevent aerobic respiration.**

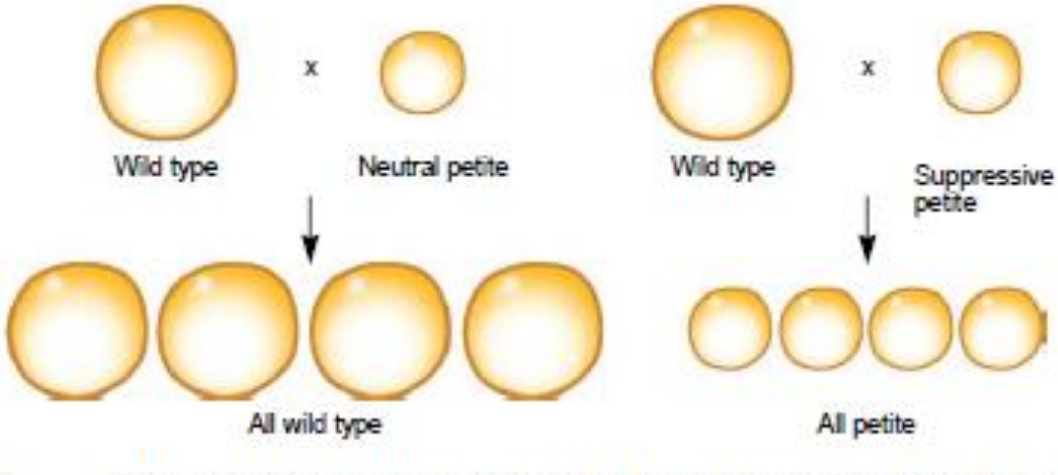
Transmission of the petite trait in *S. cerevisiae*

a) Petite nucleari o segreganti



(a) A cross between a wild type and a segregational petite

b) Petite citoplasmatici o neutri



(b) A cross between a wild type and a neutral or suppressive vegetative petite

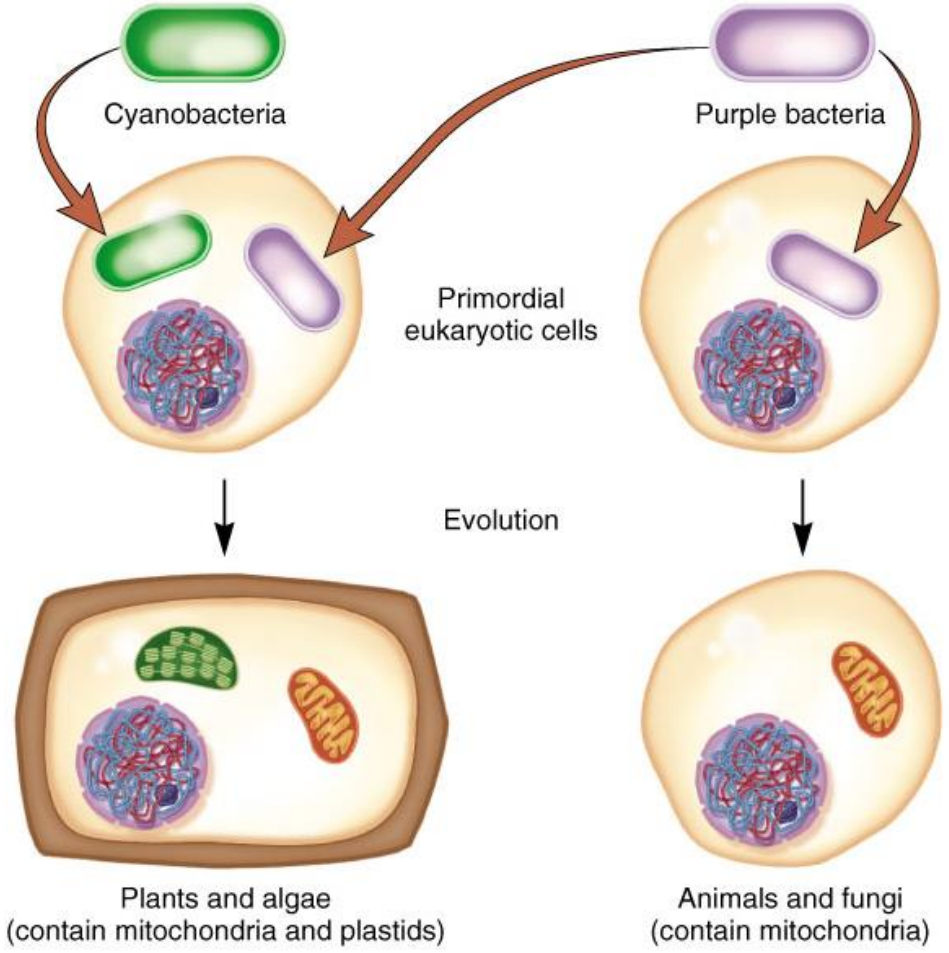
(a) A wild-type strain crossed to a segregational petite.

b) A wild-type strain crossed to a neutral vegetative petite and to a suppressive vegetative petite

- **Colonie *petite* :**
  - **cellule difettive nella respirazione;**
  - **formano colonie più piccole → producono quantità limitata di ATP**

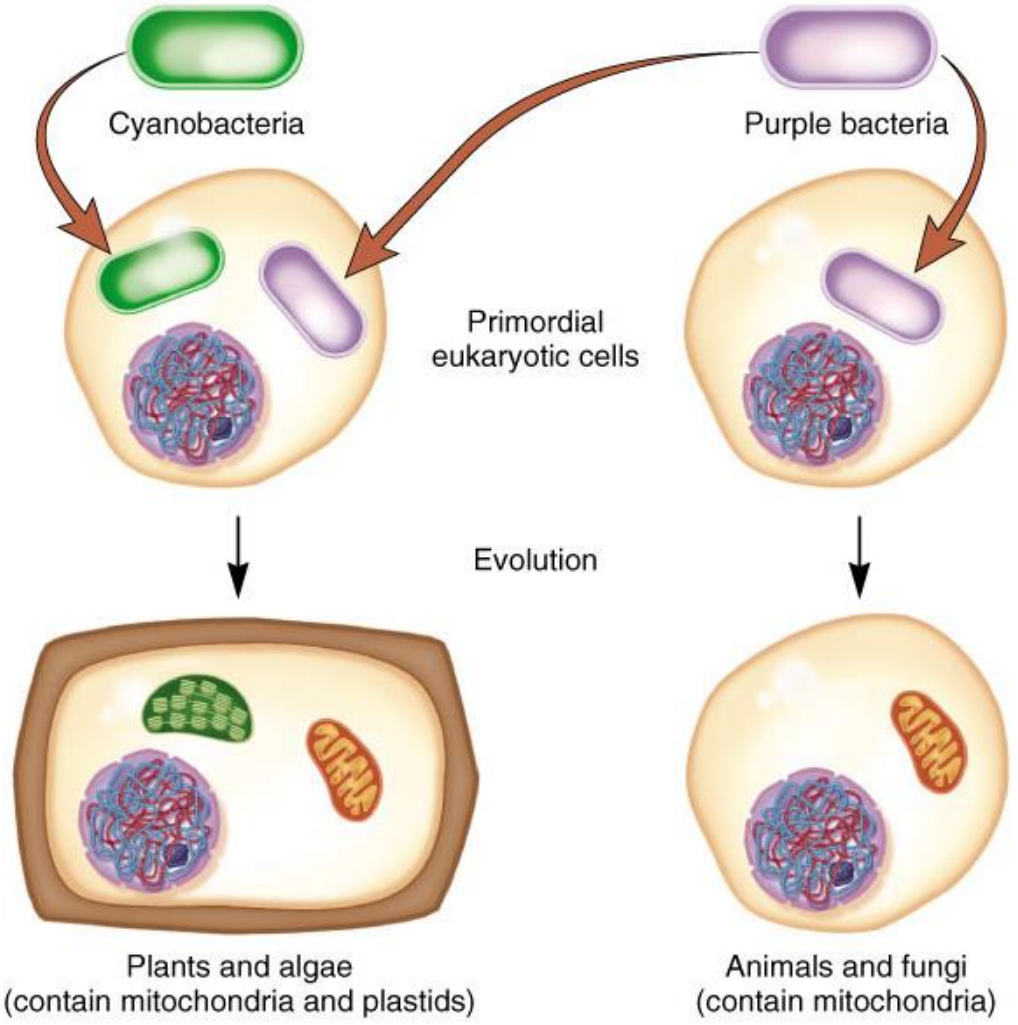
Extranuclear Inheritance

- Mitochondria and chloroplasts **were once free-living bacteria**
  - Engulfed and retained by early eukaryotes
  - Endosymbiosis



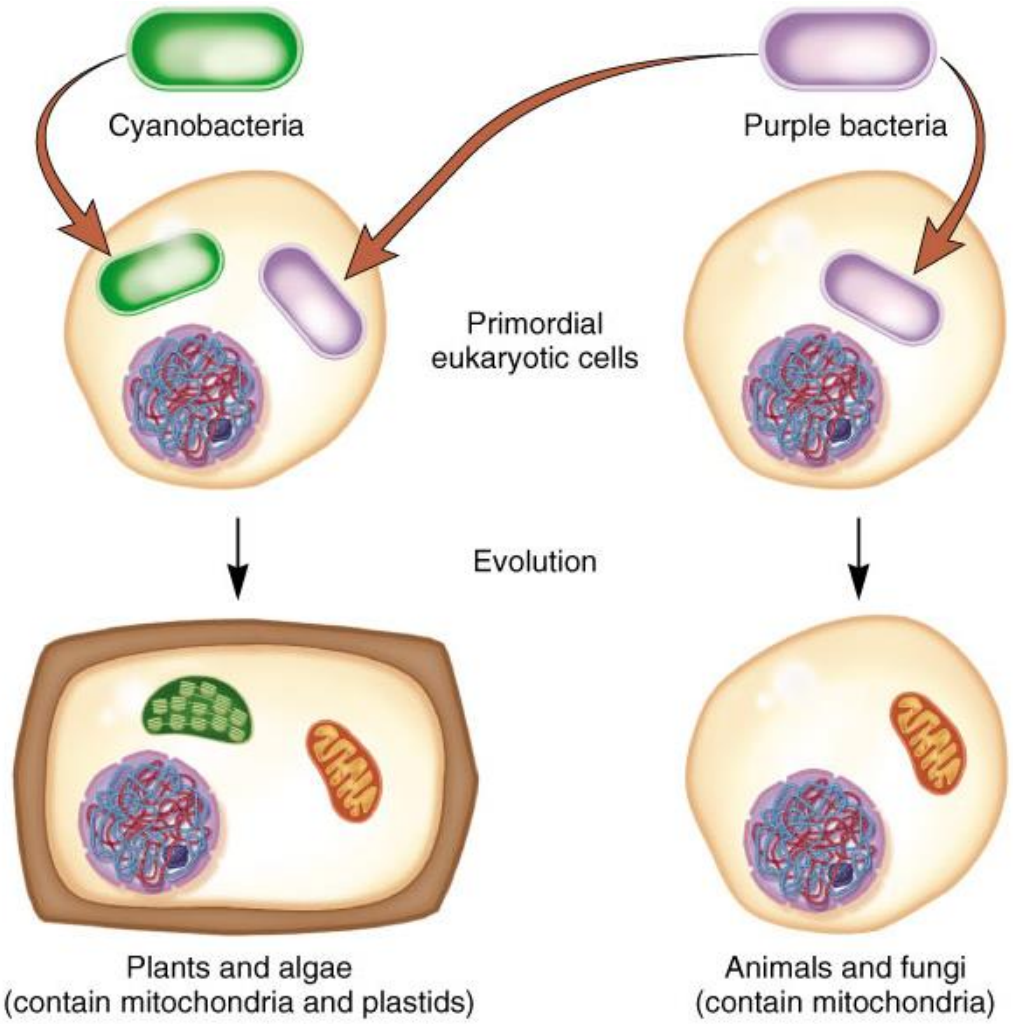
EXTRANUCLEAR INHERITANCE

- **Plastids were derived from cyanobacteria**
  - Photosynthetic bacteria
  - Relationship allows **plants and algae to obtain energy from the sun**
  - Benefit to the bacterium is less clear



Extranuclear Inheritance

- **Mitochondria were likely derived from gram-negative nonsulfur purple bacteria**
  - Relationship enabled eukaryotes to produce **larger amounts of ATP**



Extranuclear Inheritance

- **Endosymbiosis**
  - **Most genes originally found in these bacterial *genomes*** have been lost or transferred to the nucleus
    - The DNA sequence of some nuclear genes indicates horizontal gene transfer from bacteria
  - Biological benefits are unclear

EXTRANUCLEAR INHERITANCE

- **Endosymbiosis**
  - Transfer of **mitochondrial genes t**o the nucleus has apparently ceased in animals
  - Gene transfer from mitochondria and chloroplasts continues in plants at a low rate
  - Transfer from the nucleus to the organelles has apparently almost never occurred
    - One example in plants of transfer to the mitochondrion is known

Extranuclear Inheritance

- Eukaryotic cells occasionally contain **symbiotic infective particles**
  - Some individuals of the protozoan *Paramecia aurelia* possess the "killer" trait
    - Secrete the toxin *paramecin*
    - Many strains of paramecia are killed
- Killer strains contain cytoplasmic particles
  - "Kappa particles"
  - 0.4 μm long
  - Contain their own DNA
    - Gene encodes **paramecin** toxin
    - Genes encode resistance to this toxin
  - Kappa particles are infectious
    - Particles in extract from killer strains can infect nonkiller strains
    - Converted to killer strains

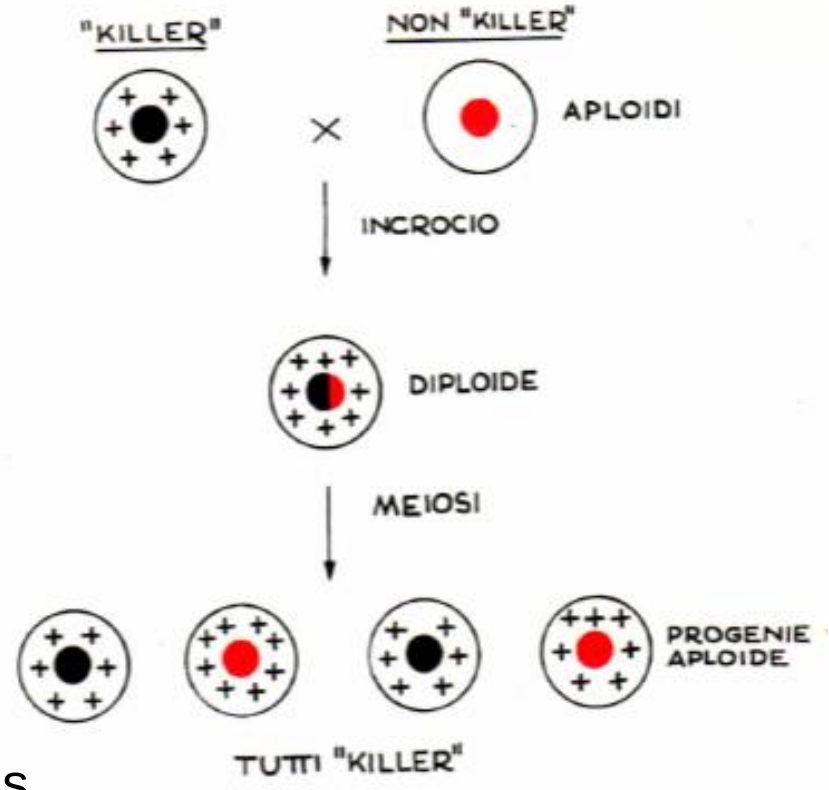
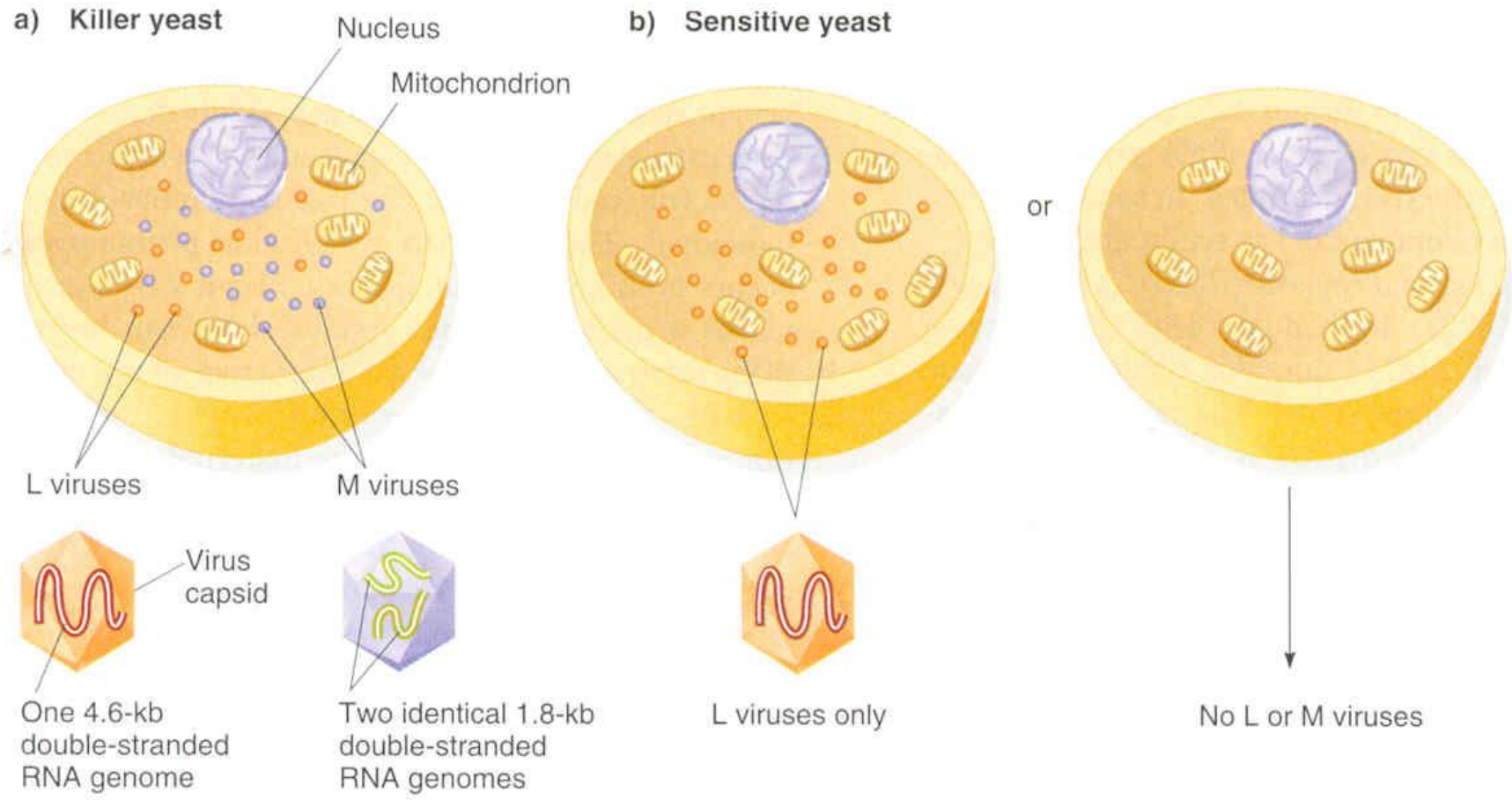


Figure 15.12

The killer phenomenon in yeast. **(a)** Killer yeast contain two virus types, L and M, each of which contains a double-stranded RNA genome. L-dsRNA encodes both virus particles and the enzyme required for L and M virus replication. M-dsRNA encodes the killer toxin. **(b)** Sensitive yeast, which can be killed by killer toxin, either have L viruses but no M viruses or have neither virus type.



Riassunto

L'ereditarietà extranucleare

- L'ereditarietà extranucleare comporta l'ereditarietà di geni che si trovano nei **mitocondri o nei cloroplasti.**
- Mitocondri e cloroplasti portano cromosomi circolari in una regione nucleoidica.
- Questi cromosomi circolari contengono relativamente pochi geni rispetto al numero nel nucleo cellulare.

• Ereditarietà materna si verifica quando organelli, come i mitocondri o cloroplasti, vengono trasmessi attraverso l'uovo.
• L'eteroplasmia dei cloroplasti può determinare un fenotipo variegato
• I petites neutri e soppressivi nel lievito sono dovuti a difetti nel DNA mitocondriale e mostrano un modello di ereditarietà non mendeliana.
• I pattern di trasmissione dei mitocondri e dei cloroplasti variano tra le diverse specie.
• Mitocondri e cloroplasti derivano da un'antica relazione endosimbiotica.
• In rare occasioni, le cellule eucariotiche possono contenere particelle infettive