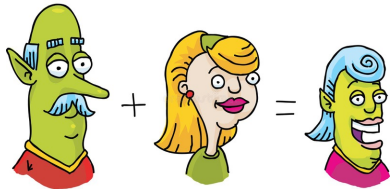


GENÉTICA E MELHORAMENTO DE PLANTAS

Licenciaturas em Eng^a Agronómica

Hereditariedade Mono-Híbrida

Manuela Gomes da Silva
manuelasilva@isa.ulisboa.pt



Bibliography:

An Introduction to Genetic Analysis
Molecular Biology of the Cell
The Cell: A Molecular Approach

Estudo da transmissão de caracteres:

Monohibridismo

Transmissão **ligada ao sexo**

Alelos **letais**

Dihibridismo

Interacções génicas

U LISBOA

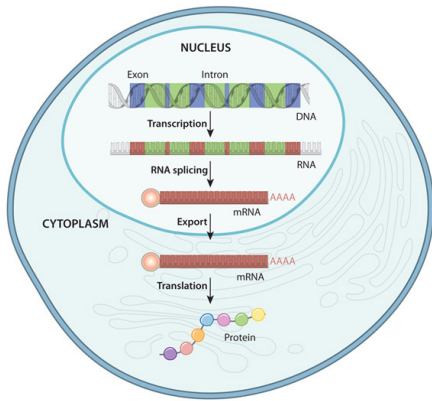
UNIVERSIDADE
DE LISBOA



INSTITUTO
SUPERIOR DE
AGRONOMIA

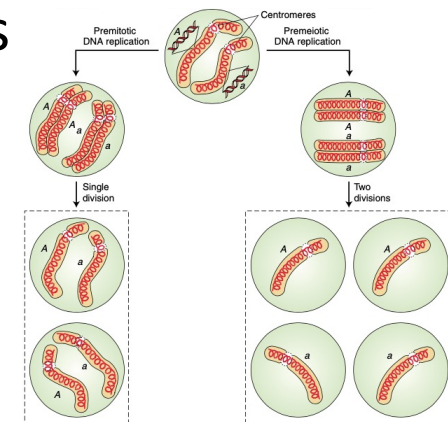
GENÉTICA

Estudo de **genes**



Como são expressos para conferir fenótipos
→ Genética da **expressão**

Como são passados entre gerações
→ Genética de **transmissão**



O que é um **GENE**?

- **Unidade física e funcional** básica da hereditariedade

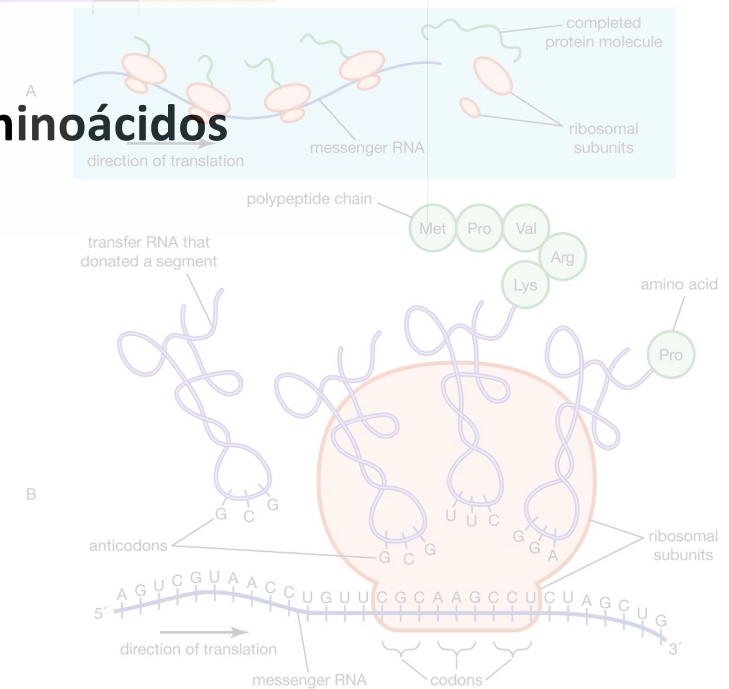
- Muitos genes codificam polipeptídeos

Transcrição → **mRNA** + Tradução → cadeia de **aminoácidos**

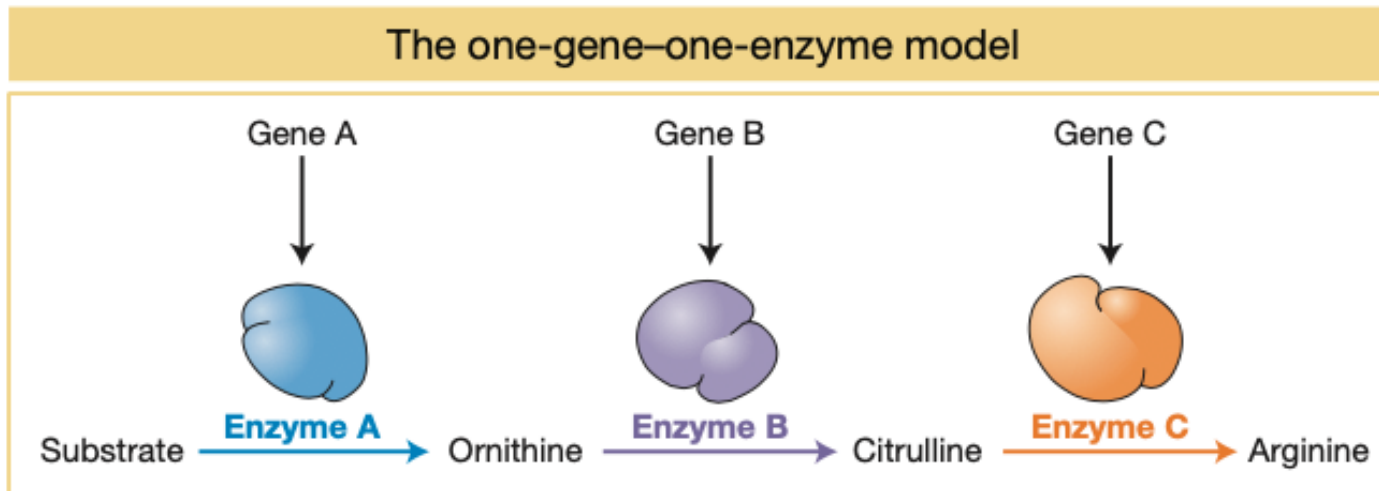
- Muitos genes não codificam para proteínas

→ RNAs funcionais

- Genes ribossomais (RNA ribossomal, **rRNA**)
- Genes de **tRNA**



One gene–one enzyme model



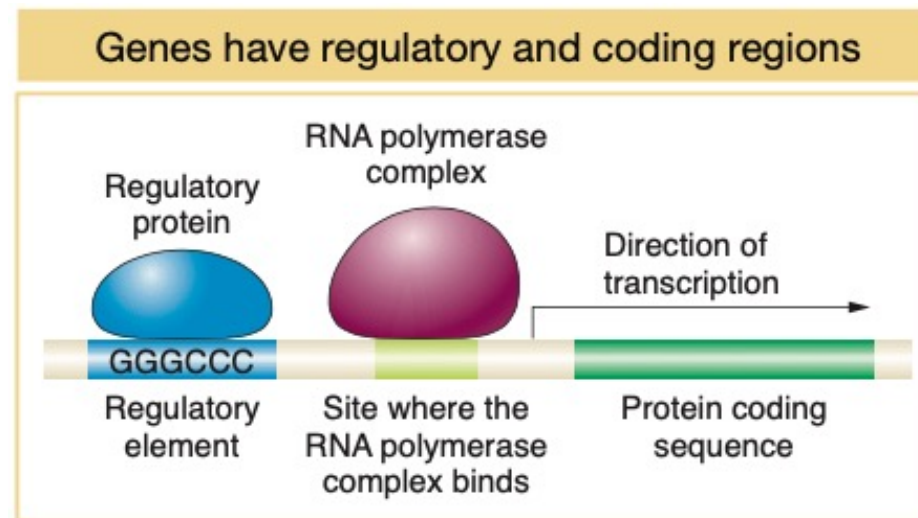
One-gene–one-enzyme model:

Genes encode enzymes responsible for biochemical cell functions
Ex. different genes coding for several enzymes involved in arginine biosynthesis
(1941, Tatum and Beadle)

Protein-coding gene

Structure of a protein-coding gene

- regulatory DNA element to which a regulatory protein binds
- promoter region where the RNA polymerase complex binds to initiate transcription
- protein-coding region



Transmissão e expressão da Informação genética

Para a transmissão da informação tem que ocorrer a **replicação** de **todo o genoma**

A informação genética contida nos **genes** é lida e executada num processo de 2 etapas:

1º **transcrição** - segmentos de uma sequência de DNA (uma das cadeias simples) são usados para guiar a síntese de moléculas de **RNA**

2º **tradução** - as moléculas de RNA são usadas para guiar a síntese de moléculas de **proteínas**

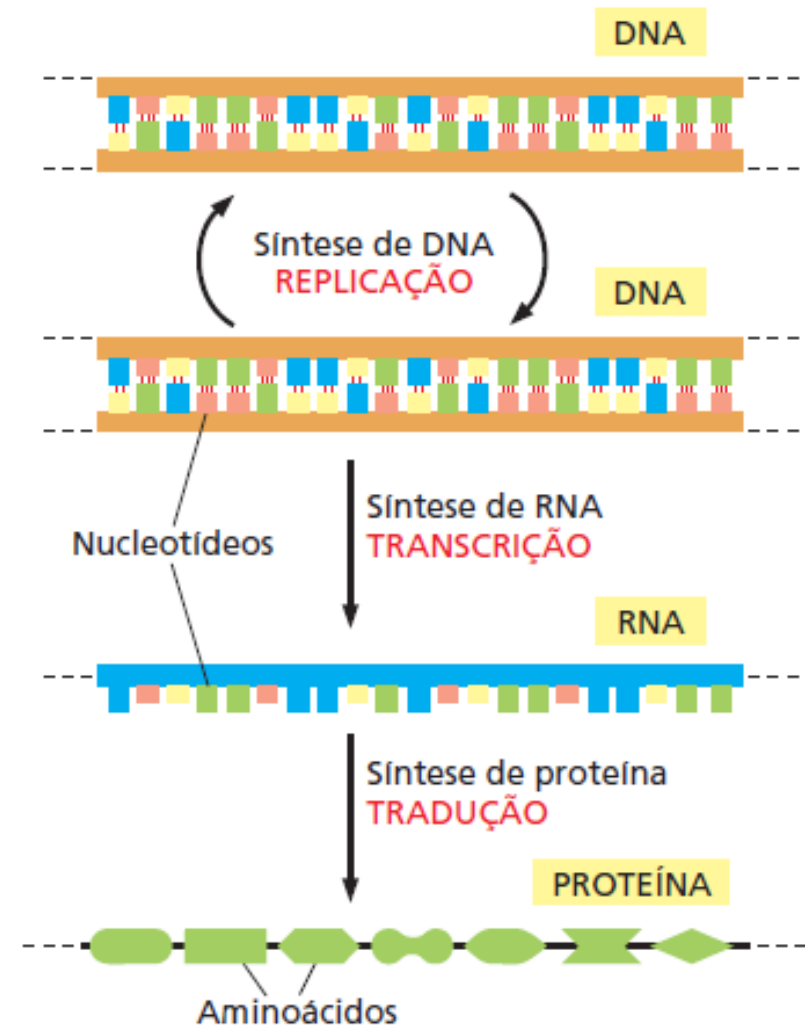
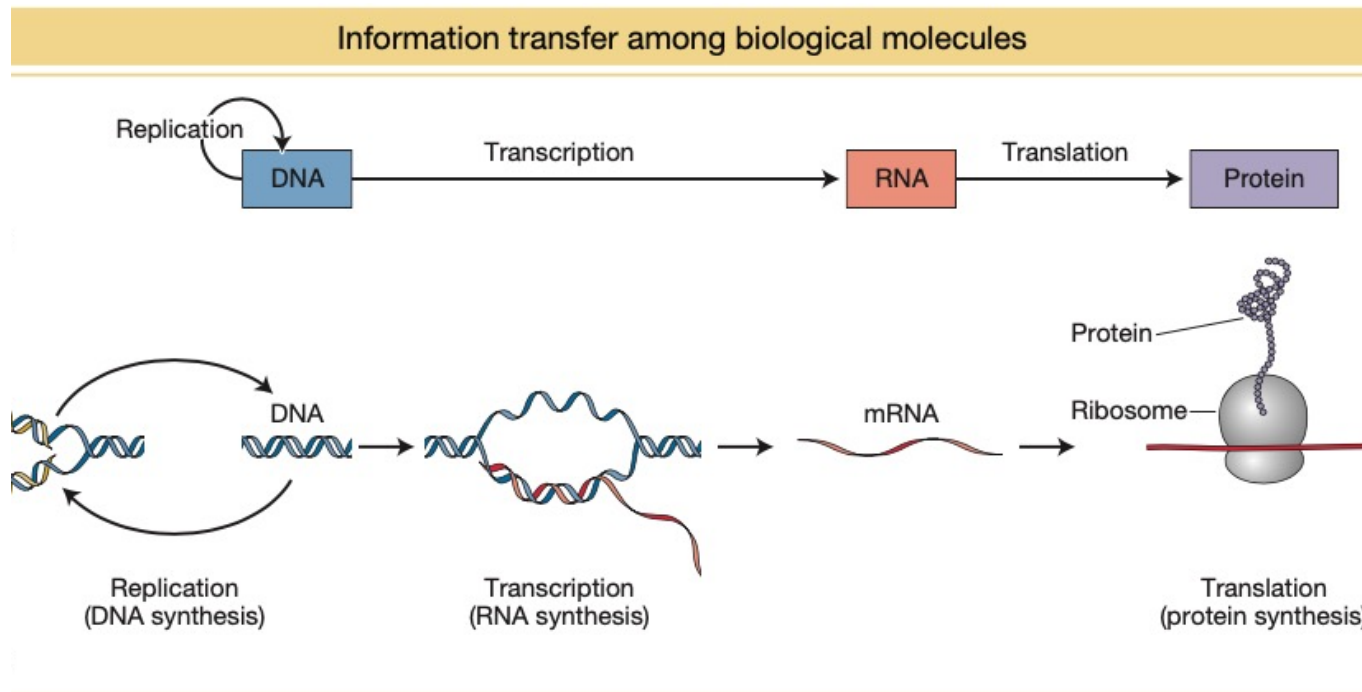


Figura 1-4 Do DNA à proteína. Alberts et al. (2017) *Biologia Molecular da Célula*. 6ªEd. Artmed.

Central dogma

Central dogma - information flow between biological molecules

Genética de **transmissão** → - **Replication** DNA → DNA
Genética de **expressão** → [- **Transcription** DNA → messenger RNA (mRNA)
- **Translation** mRNA → protein (ribosome)



CHROMOSOMES IN CELL DIVISION

Chromosomes – transmission vehicles of genetic information
sequences essential for correct **replication** and **segregation**

Telomeric sequences

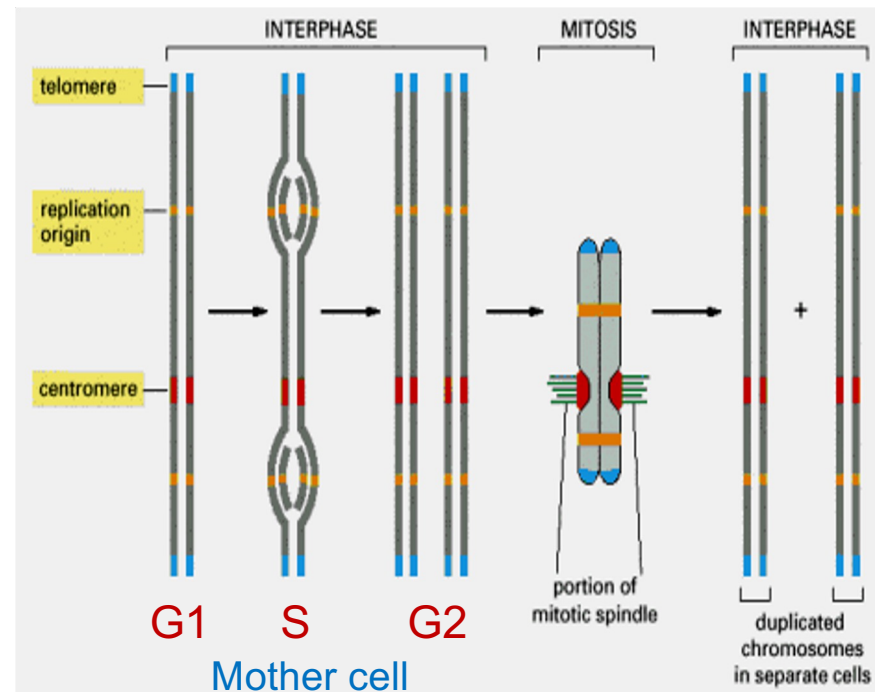
Replication origins

Centromeric sequences

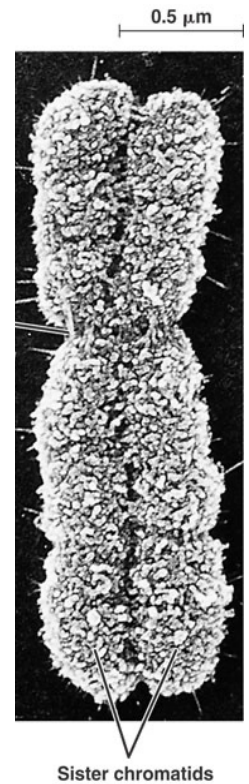
S phase - DNA **replication**

each chromosome = two DNA molecules
sister chromatids

Mitosis - sister chromatids separation

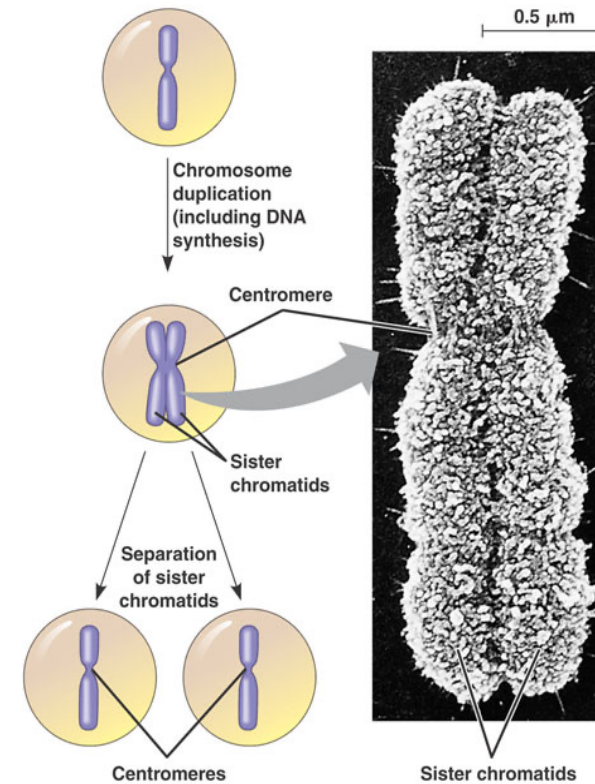
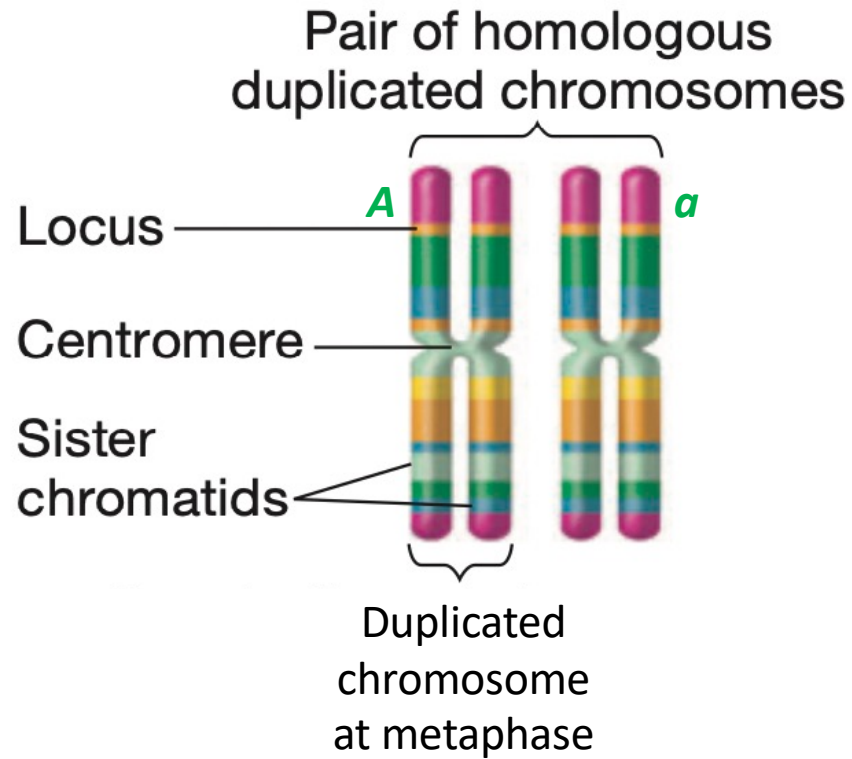


G1 G1
Daughter cells



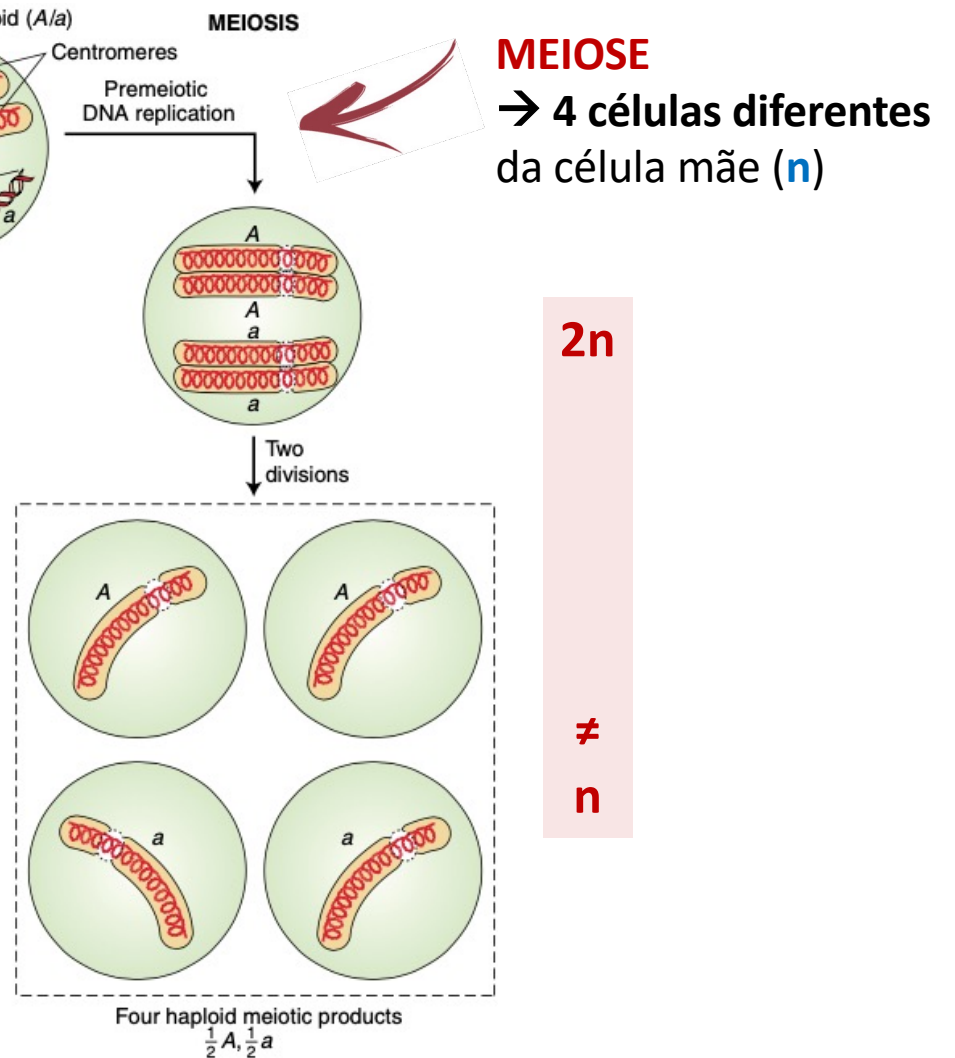
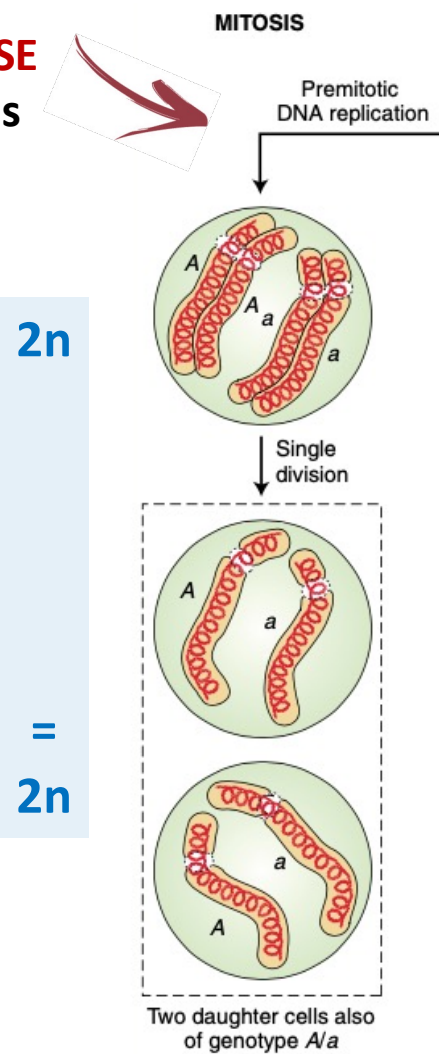
CHROMOSOMES VS ALLELES

Chromosomes vs alleles

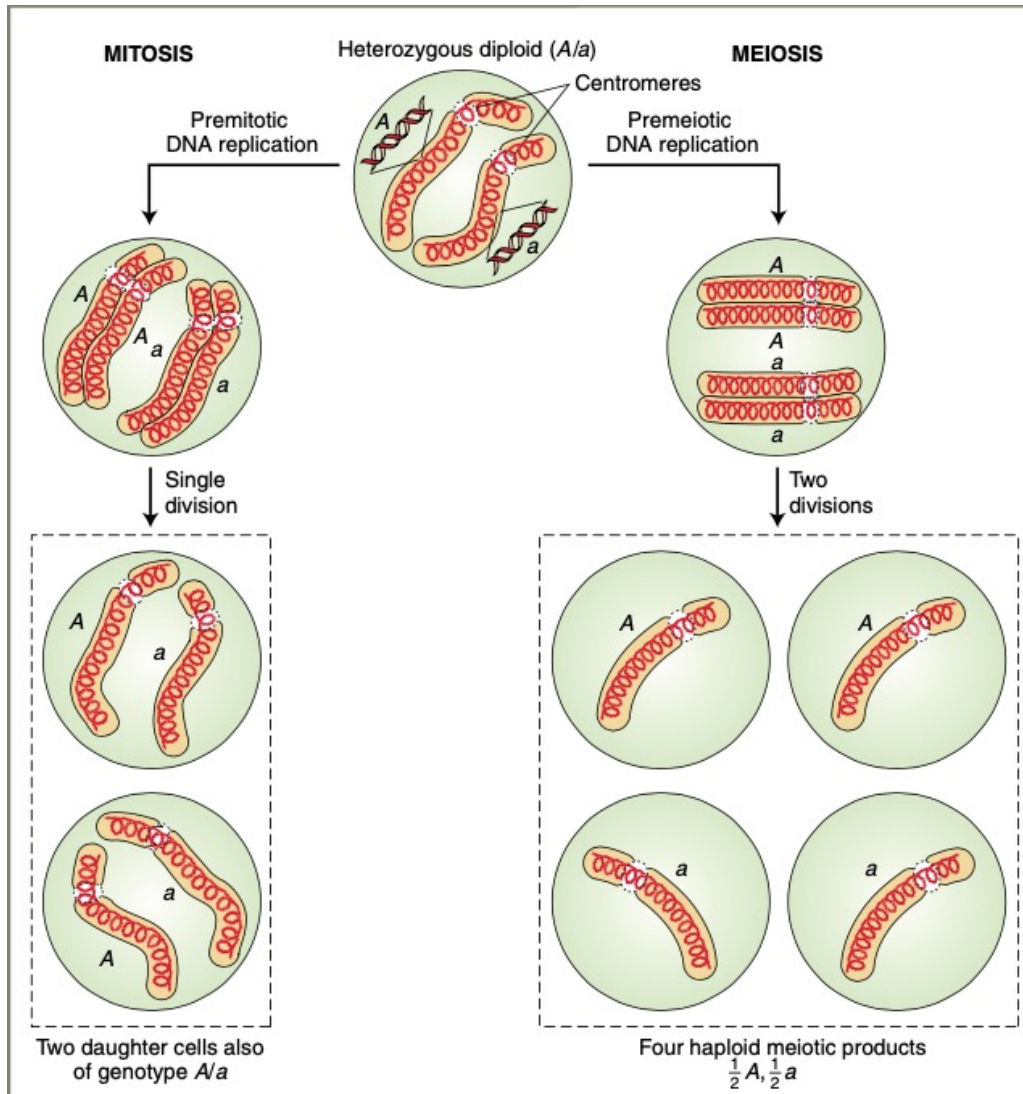


CHROMOSOMES — VEHICLES OF GENETIC INFORMATION

MITOSE
→ 2 células iguais
à célula mãe ($2n$)



THE CHROMOSOMAL BASIS OF (MONOHYBRID) INHERITANCE



Heterozygote somatic diploid cell (Aa)

MITOSIS

- A and a alleles replicate
- 1 division
- 2 daughter cells **genetically identical** (Aa)

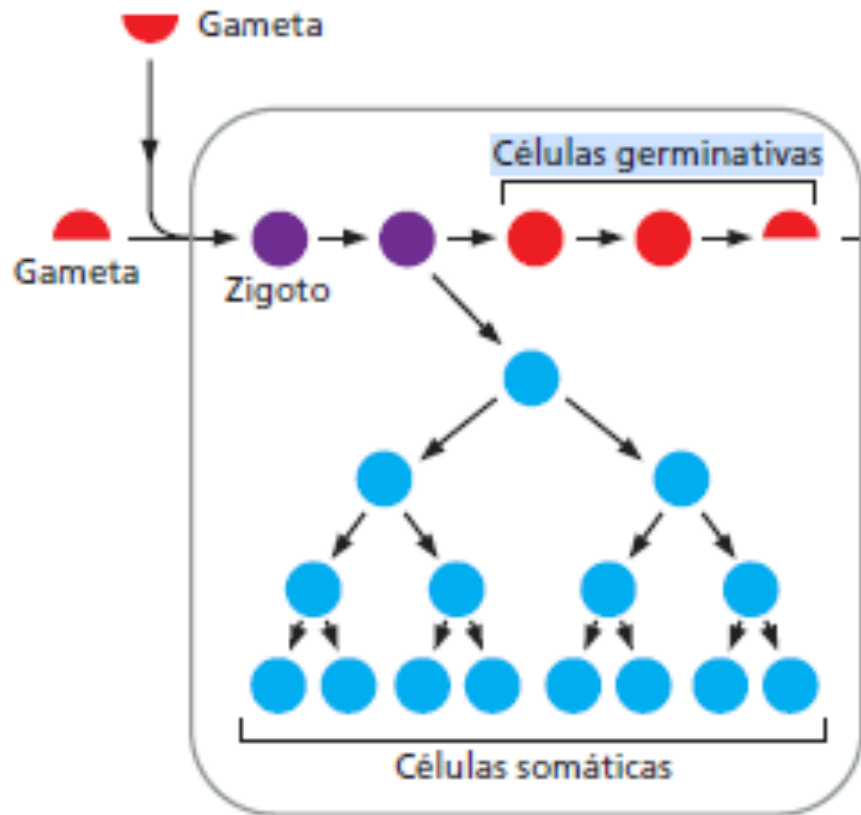
MEIOSIS

- A and a alleles replicate
- 2 consecutive divisions
- 4 **haploid** cells ($\frac{1}{2} A + \frac{1}{2} a$)

Separation of alleles into different haploid cells

→ **Mendel's first law** of **equal segregation**

CHROMOSOMES —VEHICLES OF GENETIC INFORMATION



Células germinais **haploides - n**

Em organismos que se reproduzem sexualmente, dão origem aos **gâmetas** e passam informação genética do organismo progenitor à **descendência**

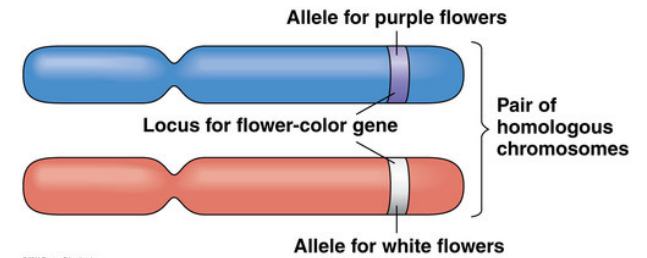
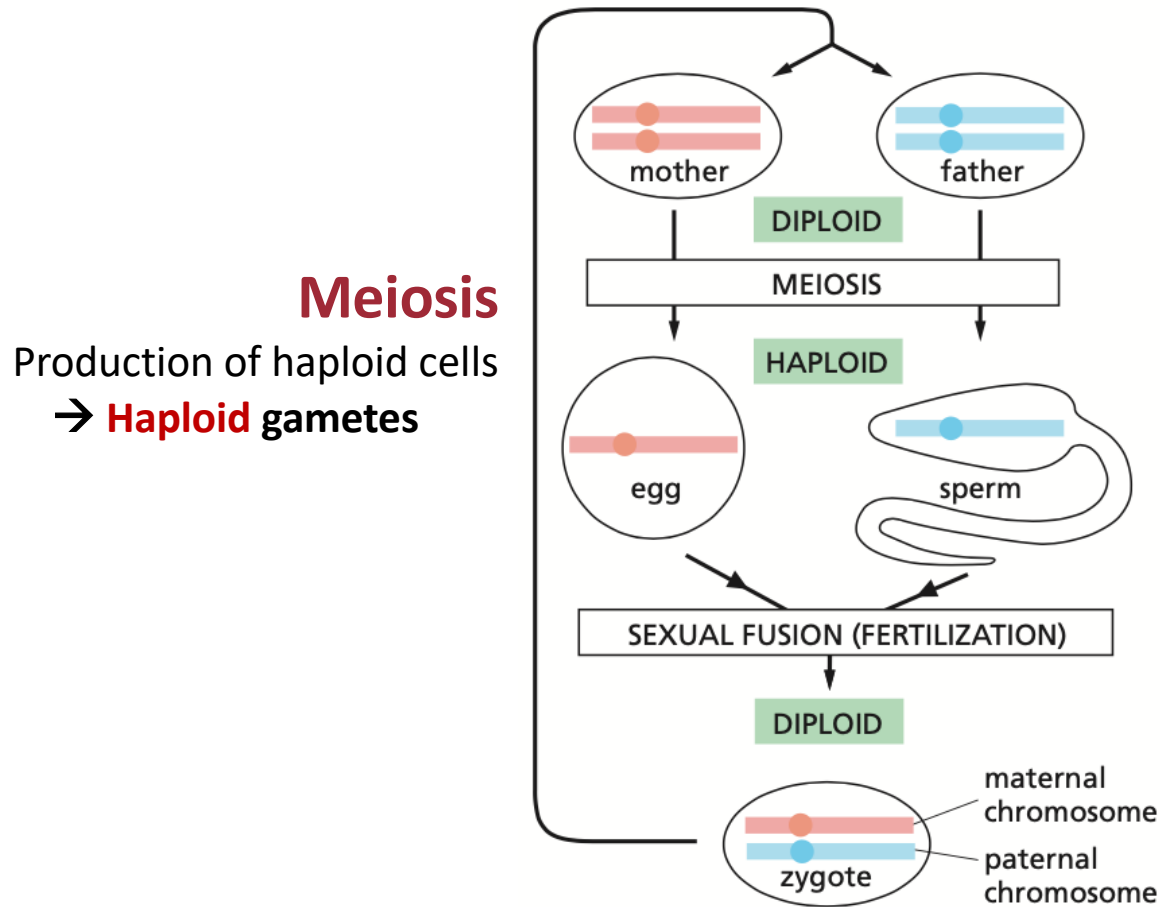
Células somáticas **diploides - 2n**

Formam o **corpo de um organismo** multicelular

SEXUAL REPRODUCTION

Meiosis - Alternation of **diploid** and **haploid** cell generations

THE HAPLOID-DIPLOID CYCLE OF SEXUAL REPRODUCTION



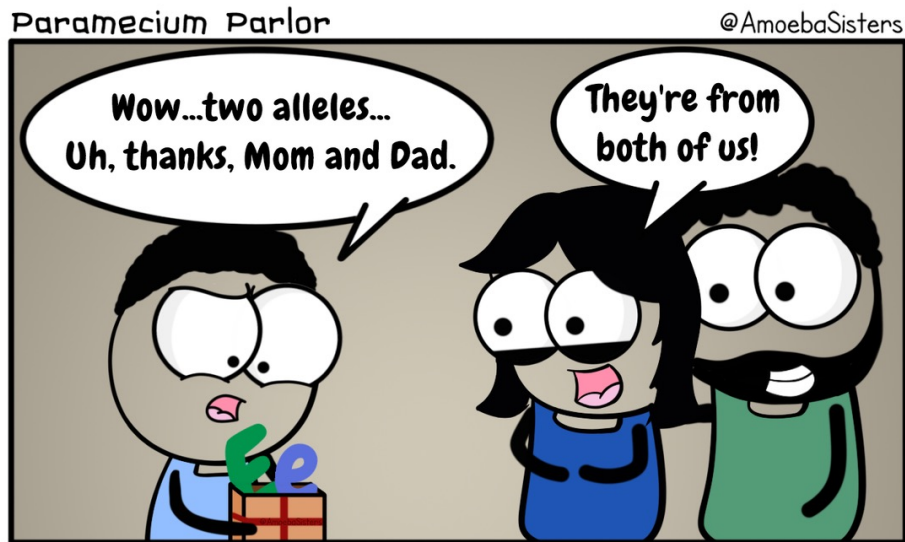
Fertilization

Fusion of haploid cells

→ **Diploid** zygote

GENES / ALELLES

Chromosome behavior and inheritance patterns in eukaryotes



Possible Parent Genotypes

Will Work:	Will Not Work:
Parent 1: Tt Parent 2: Tt Child: tt	Parent 1: TT Parent 2: TT Child: tt
Parent 1: tt Parent 2: tt Child: tt	Parent 1: Tt Parent 2: TT Child: tt

@AmoebaSisters

One allele from each parent!

Genes e ALELOS

- Qual a natureza molecular dos **alelos**?

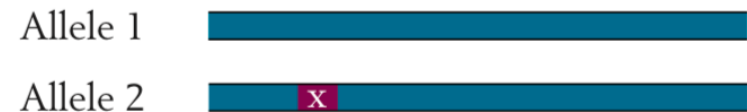
- **Alelos** são diferentes versões de um mesmo gene

- **Alelo standard** designa-se **Tipo Selvagem** (Wild type)

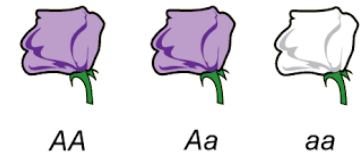
Ex: cor **púrpura** das pétalas da ervilha

Via metabólica que produz o pigmento responsável (antocianina)

mutação → alelo recessivo não produz pigmento → pétalas brancas



AA → active enzyme → purple pigment
Aa → active enzyme → purple pigment
aa → no active enzyme → white

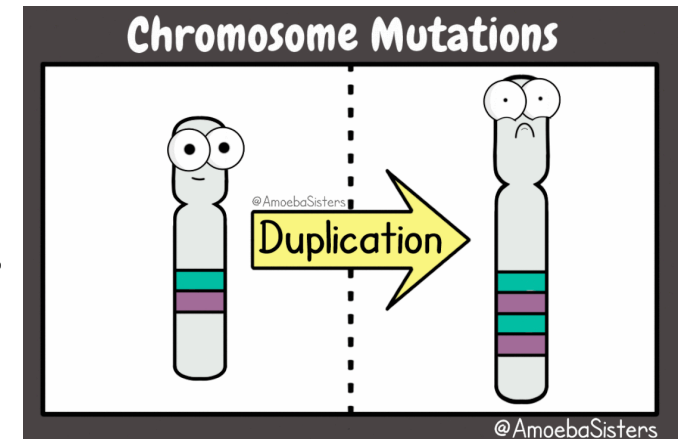


A dominante

→ **Heterozigótico** púrpura - produzida enzima suficiente pela produção de pigmento

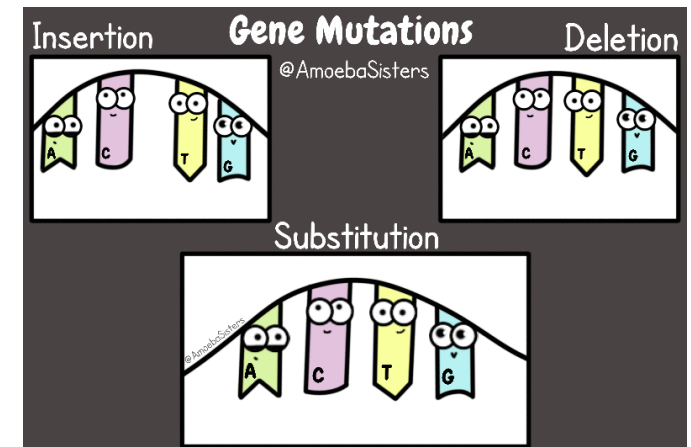
ORIGEM DA DIVERSIDADE - Tipos de mutações

- Mutação **Cromossômica**:
alteração do número / estrutura dos cromossomas

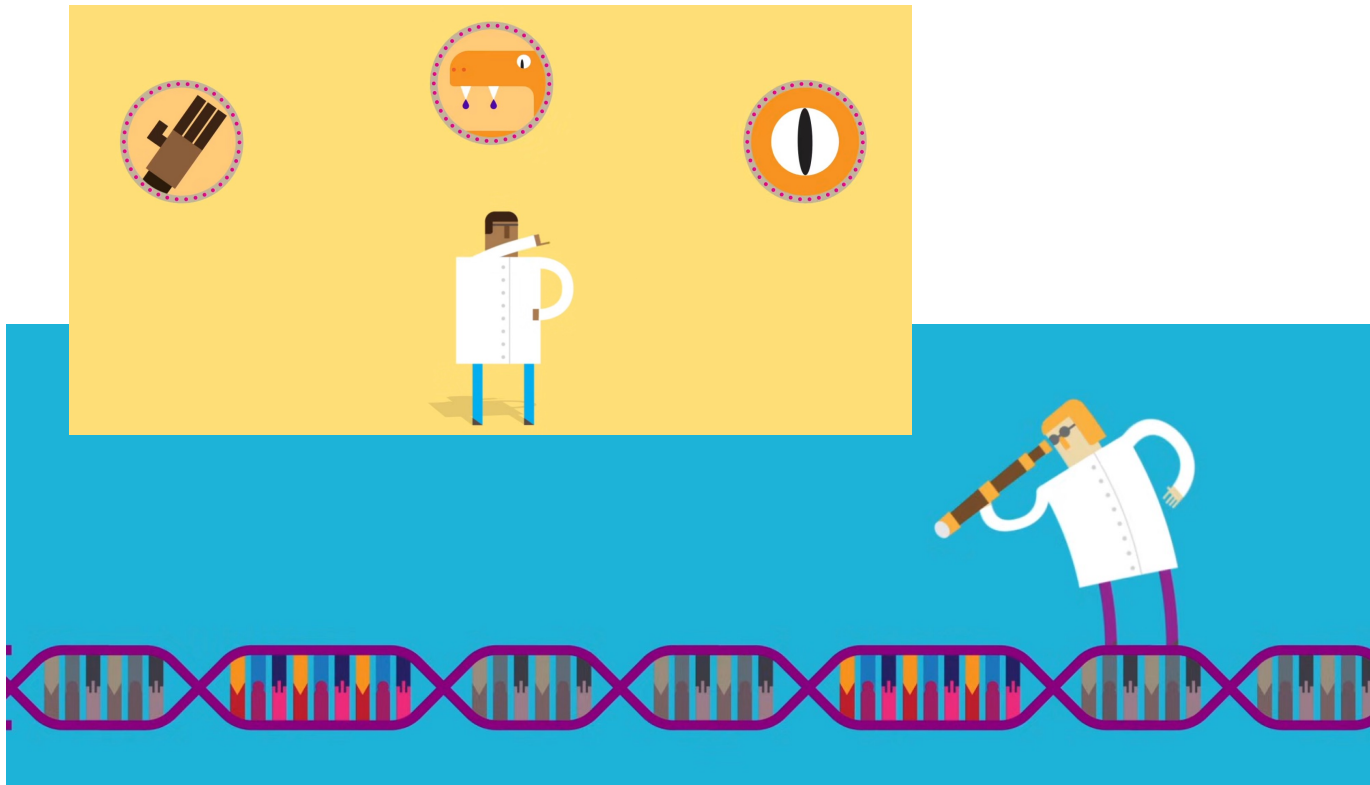


- Mutação pontuais / **Génica**:
alteração de genes individuais a nível molecular

Dois principais tipos de mutação pontual:
substituições ou **Indel** (inserção, deleções)



Where do genes come from? - Carl Zimmer



<https://www.youtube.com/watch?v=z9HIYjRRaDE&list=PLJicmE8fK0EiuxzlxoeC7R3-EFUrtaf9t&index=2>
4:23

Activity – explore the outcome of Mutation

Genetic Variation: The Outcome of Mutation

Mutation is a process that changes a DNA sequence. But how does a difference in a gene's DNA sequence (or genotype) lead to a difference in an observable trait (or phenotype)?

Learn more by exploring the examples below:

Shar Pei dogs



Double-muscled cattle



Extra-toed cats



Curly hair



Mendel's wrinkled peas



Disease resistance / Cystic Fibrosis



Red hair



Song learning in birds



05:00



The Outcome of Mutation

<https://tinyurl.com/MutationOutcome>

Activity – explore the outcome of Mutation

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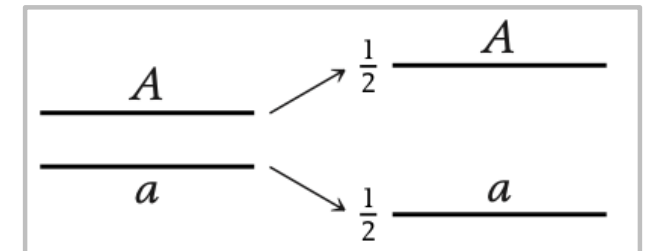
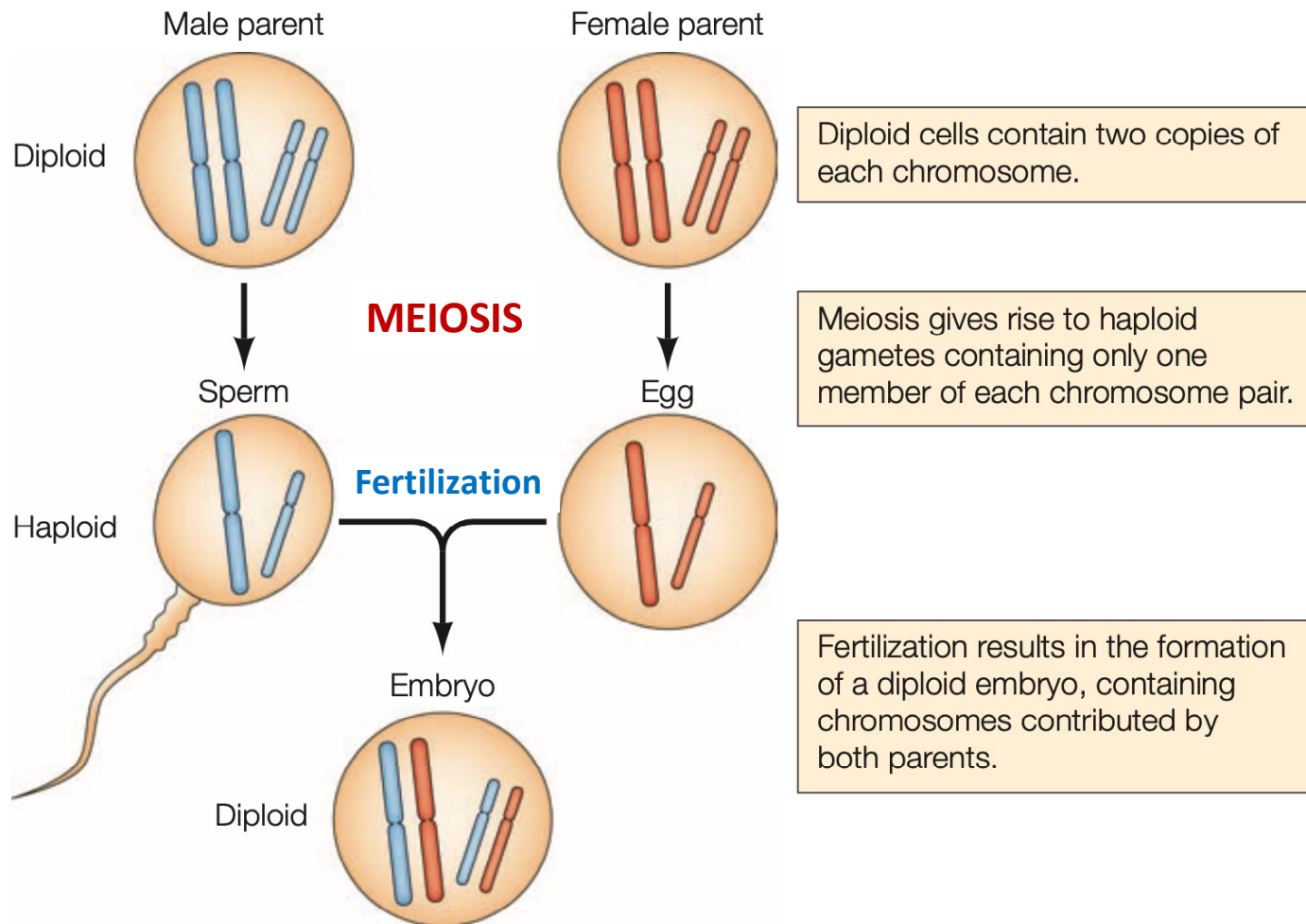
05:00



The Outcome of Mutation

<https://tinyurl.com/MutationOutcome>

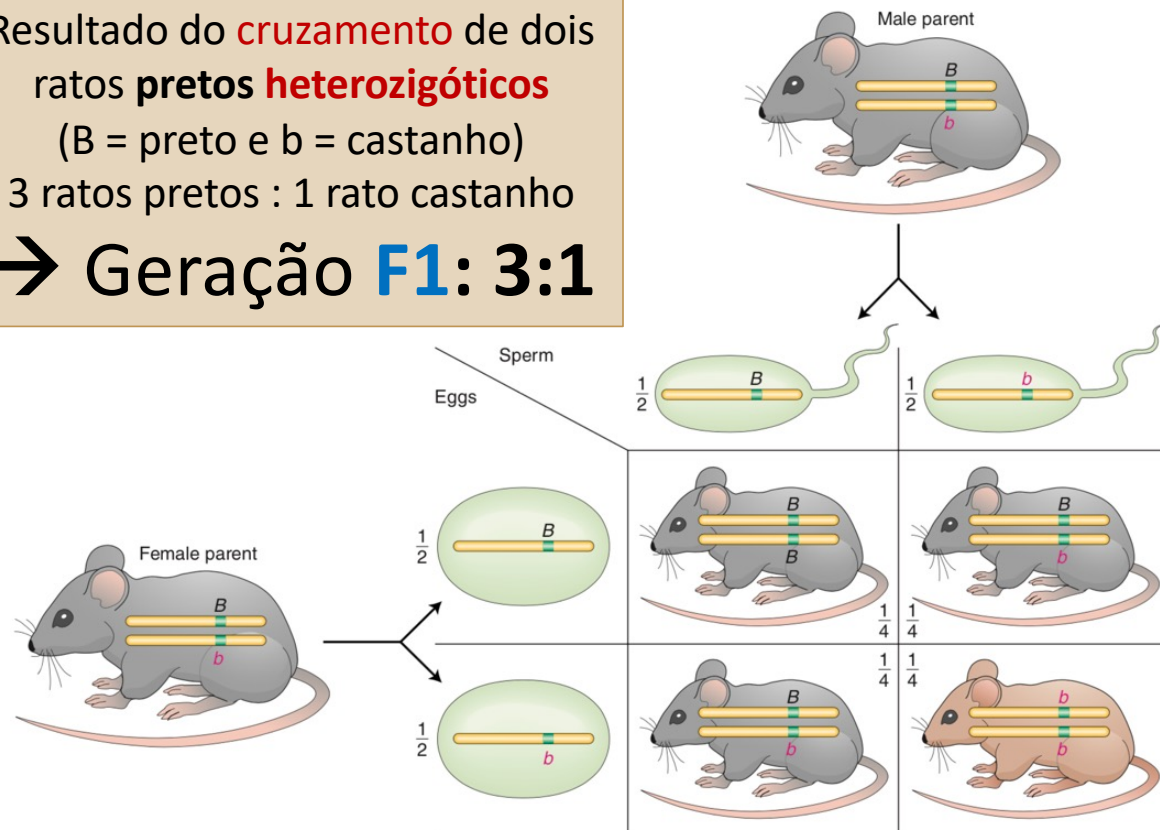
Chromosomes – Meiosis and Fertilization



Heterozigótico Aa - Meiose
→ $\frac{1}{2}$ gâmetas **A** + $\frac{1}{2}$ gâmetas **a**

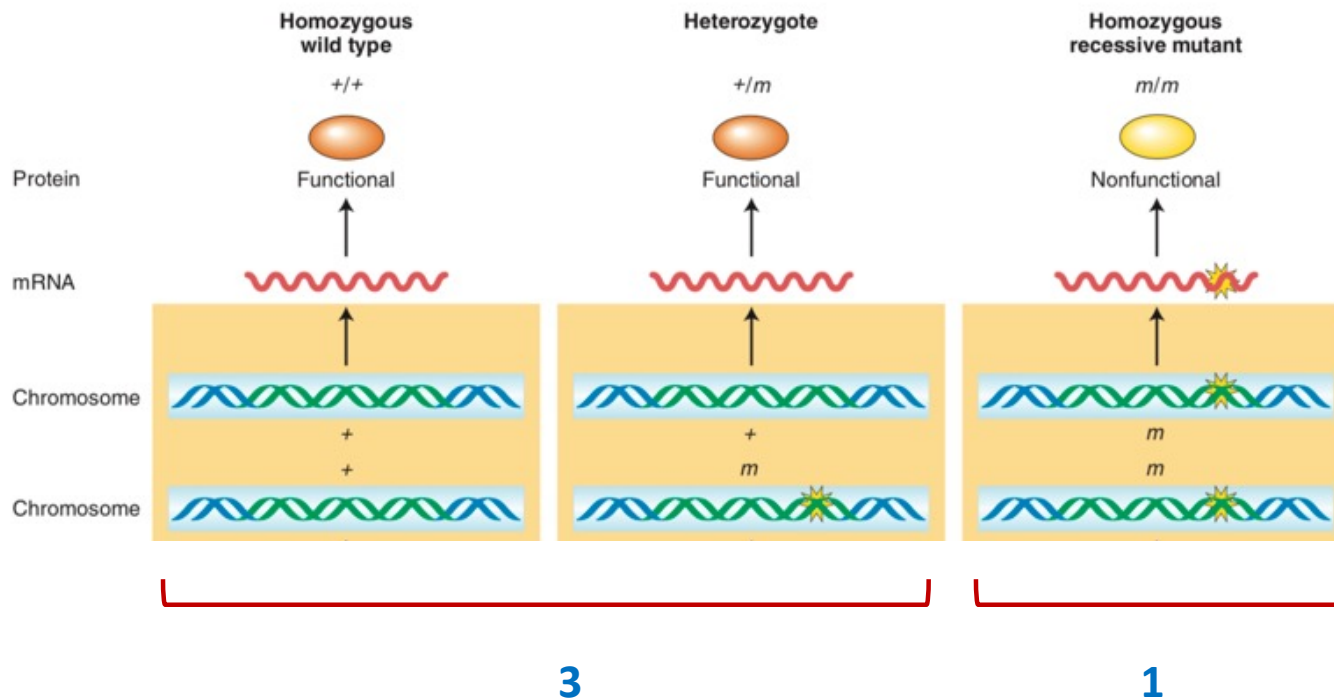
Padrão hereditário típico de um gene com dois alelos um dominante e outro recessivo

Resultado do **cruzamento** de dois
ratos **pretos heterozigóticos**
(B = preto e b = castanho)
3 ratos pretos : 1 rato castanho
→ Geração **F1**: **3:1**



4 possibilidades diferentes de **fecundação** → **2** **fenótipos** diferentes **3:1**

Interações entre os alelos de um gene: dominância e recessividade



- Mutações **recessivas**
Alelo selvagem é **haplosuficiente**

– uma dose é suficiente para produzir o fenótipo selvagem (dominante)

→ O heterozigótico terá o fenótipo do alelo dominante

Padrões hereditários



1822-1884

- Existência de **genes** – inferida por **Mendel** pela observação de **razões matemáticas precisas** nos descendentes de dois indivíduos parentais com fenótipos contrastantes
 - Esses **padrões** ainda são usados para
 - deduzir a existência de genes específicos que afetam determinados caracteres
- Os genótipos são avaliados **indiretamente** a partir dos fenótipos

Genética Classica

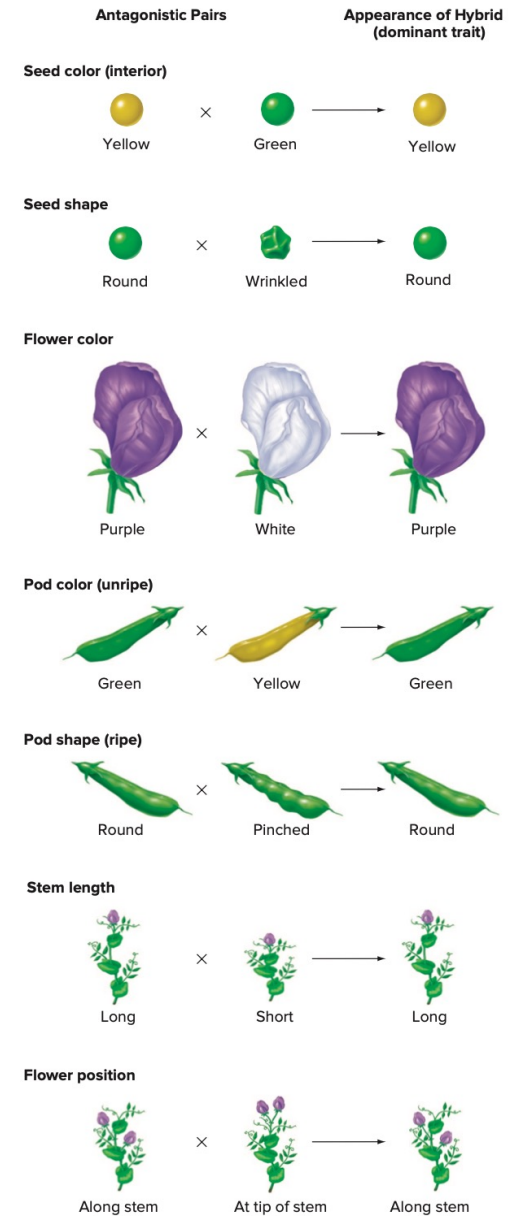
Quais são essas razões?

MENDEL'S EXPERIMENTAL DESIGN: pure lines and hybrids

- 1 - Establishment of **pure-breeding lines** for several sets of characteristics
- 2- Series of artificial **crosses** between individuals that differed in only one trait

Hybrids from parents with **antagonistic traits**

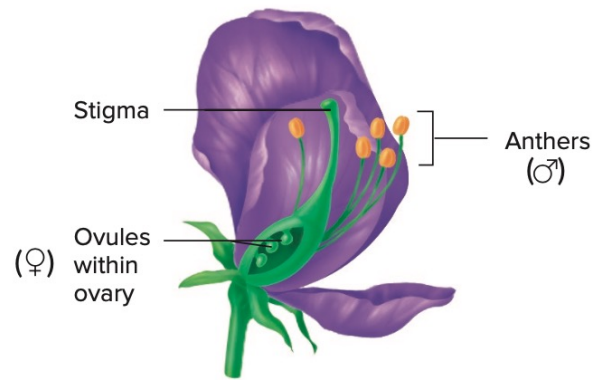
→ **F1** hybrids for 7 traits
= observation of only one of the parental traits
- **Dominant**



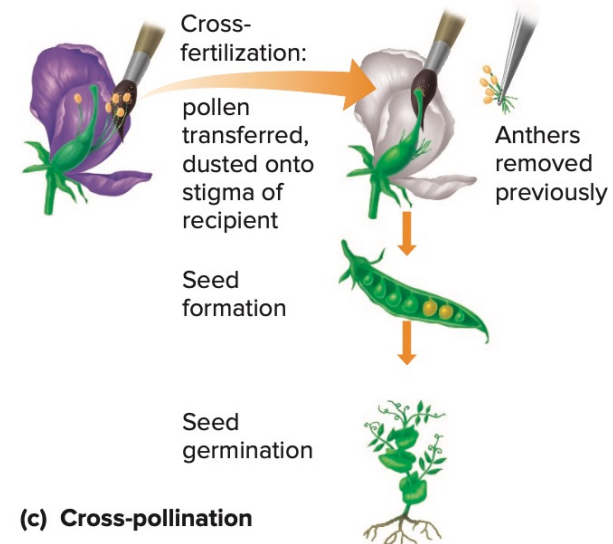
MENDEL'S EXPERIMENTAL ORGANISM: *Pisum sativum*



(a) *Pisum sativum*



(b) Pea flower anatomy



(c) Cross-pollination

Hybrid production:

- **Emasculation** - remove anthers of the female parents before pollen maturation
- **Pollen transfer** - from anthers of male parent to the stigma of female parent
→ **artificial cross-pollination**

Fertilized ovule → **pea** (mature seed)

Mendel artificial crosses and ratios

Flower color	Purple	×	White
			
Flower position	Axial	×	Terminal
			
Seed color	Yellow	×	Green
			
Seed shape	Round	×	Wrinkled
			

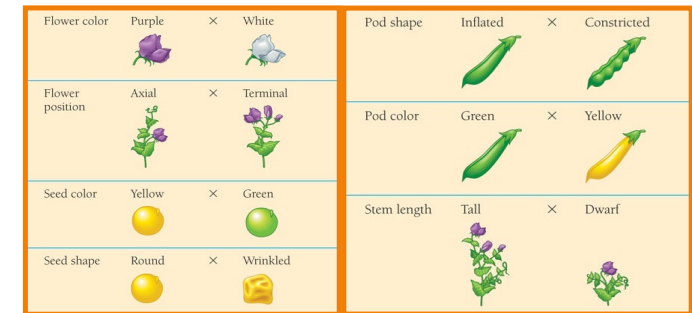
Pod shape	Inflated	×	Constricted
			
Pod color	Green	×	Yellow
			
Stem length	Tall	×	Dwarf
			

Parental phenotype	F ₁	F ₂	F ₂ ratio
1. Round × wrinkled seeds	All round	5474 round; 1850 wrinkled	2.96 : 1
2. Yellow × green seeds	All yellow	6022 yellow; 2001 green	3.01 : 1
3. Purple × white petals	All purple	705 purple; 224 white	3.15 : 1
4. Inflate × pinched pods	All inflated	882 inflated; 299 pinched	2.95 : 1
5. Green × yellow pods	All green	428 green; 152 yellow	2.82 : 1
6. Axial × terminal flowers	All axial	651 axial; 207 terminal	3.14 : 1
7. Long × short stems	All long	787 long; 277 short	2.84 : 1

For all **7 traits** studied, a **~3 : 1 ratio** in the F₁

3 : 1 Ratio

Mendel artificial crosses and ratios



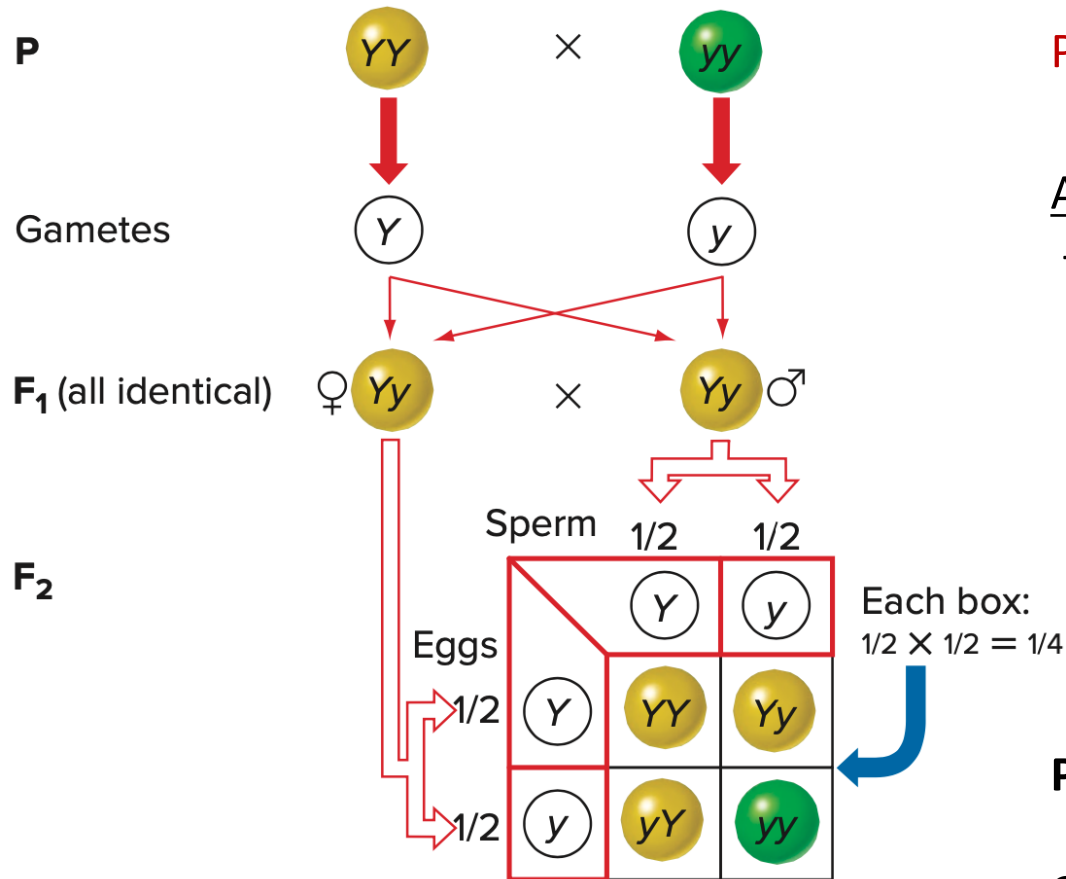
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For all **7 traits** studied, a **~3 : 1 ratio** in the F₁

3 : 1 Ratio

Why the proportion is **not exactly 3:1**?

MONOHYBRID CROSSES - hybrids for a single trait

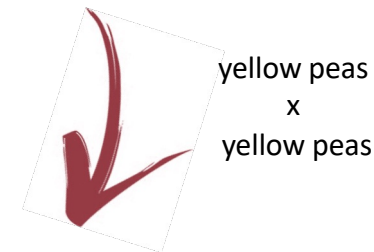


Pure-breeding parental (P) generation

Artificial crosses:

- female green-pea x male yellow-pea

→ F₁ generation (first filial)
all yellow peas



→ F₂ generation

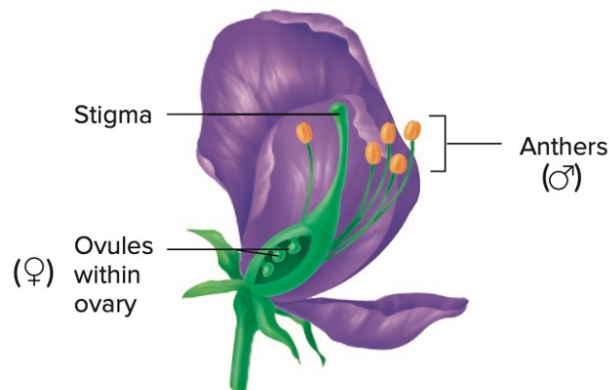
Phenotype: 6022 yellow, 2001 green → ratio 3:1

dominant recessive

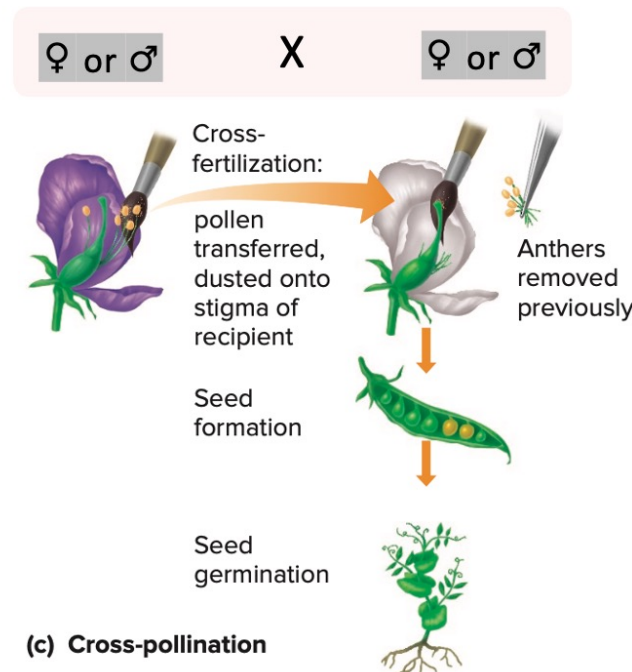
Genotype: yellow ($1/2 Yy + 1/4 YY$) + $1/4$ green yy

MONOHYBRID CROSSES - Reciprocal crosses

Gametic origin of an allele – no influence on the phenotype of the offspring



(b) Pea flower anatomy



Reciprocal crosses
sex of two parents is reversed

Ex. green pea ♀ x yellow pea ♂
reciprocal to
yellow pea ♀ x green pea ♂

→ F₂

Ratio **3 yellow** : **1 green**
in both reciprocal crosses

- **RECIPROCAL CROSSES** → **=** FENOTIPIC RATIOS

MENDEL FIRST AND SECOND LAWS

Mendel's Laws

1st LAW: Segregation of Alleles

A gene can exist in multiple forms called Alleles

(F₁)

Alleles or a gene that specifies height
This plant is heterozygous

Tt

2 types of gametes

T
 t

let's self this individual

sperms: T t

Eggs: T t

Tt tt

Tt tt

F₂

1 : 2 : 1

Genotypes TT Tt tt

Phenotypes tall Tall short

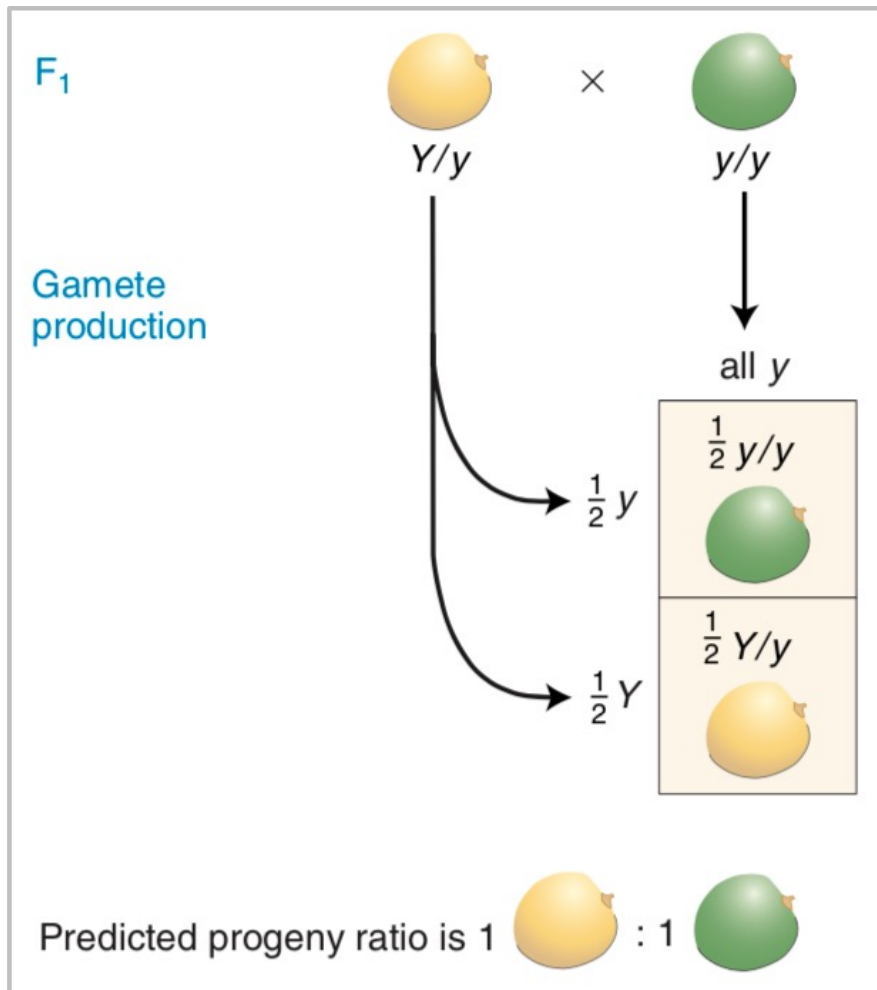
this is a Punnett Square

T = dominant
 t = recessive

https://www.youtube.com/watch?v=MDTs1ZgQ9E4&ab_channel=Genetics

Until 1.5 min

Cruzamento Teste



fenótipo **dominante**

(genótipo ??)

X

fenótipo **recessivo**

(homozigótico recessivo)

Permite saber o genótipo desconhecido

A descendência mostra a probabilidade dos gâmetas formados pelo **genótipo dominante ??**

Descendência: proporção fenotípica **1 : 1**

= proporção de **gâmetas** Y e y

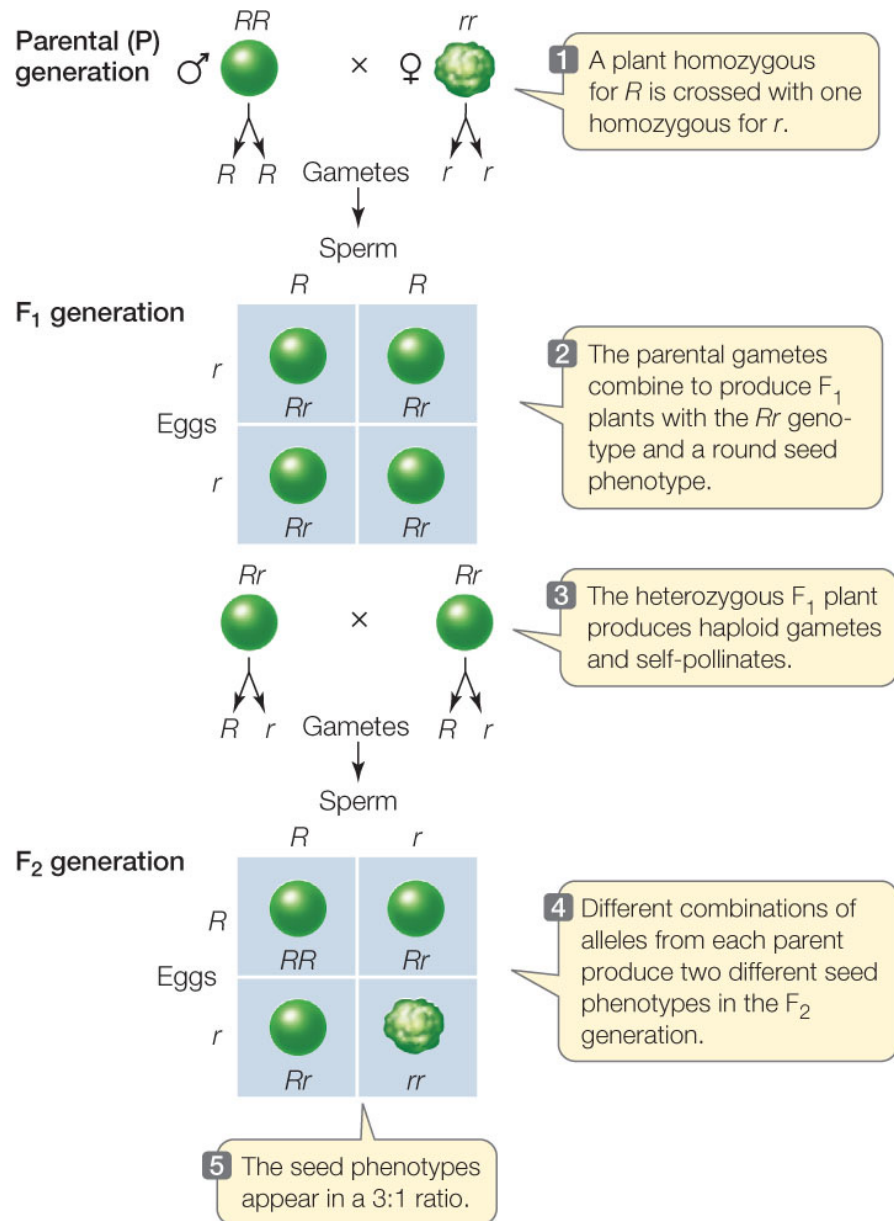
→ **heterozigótico**

Padrão hereditário típico

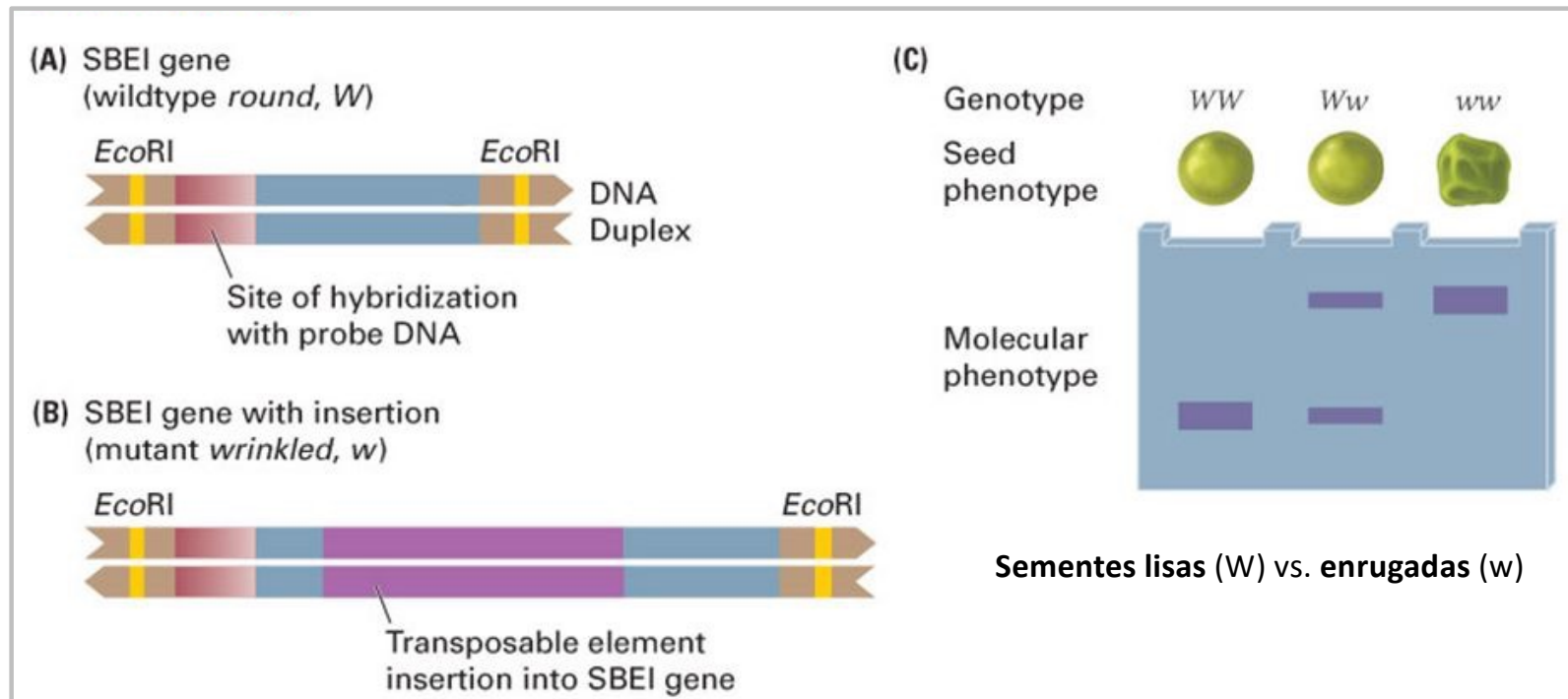
– Cruzamento monohíbrido

Forma das ervilhas

- lisas (**dominante**)
- enrugadas



Sementes enrugadas - Base molecular



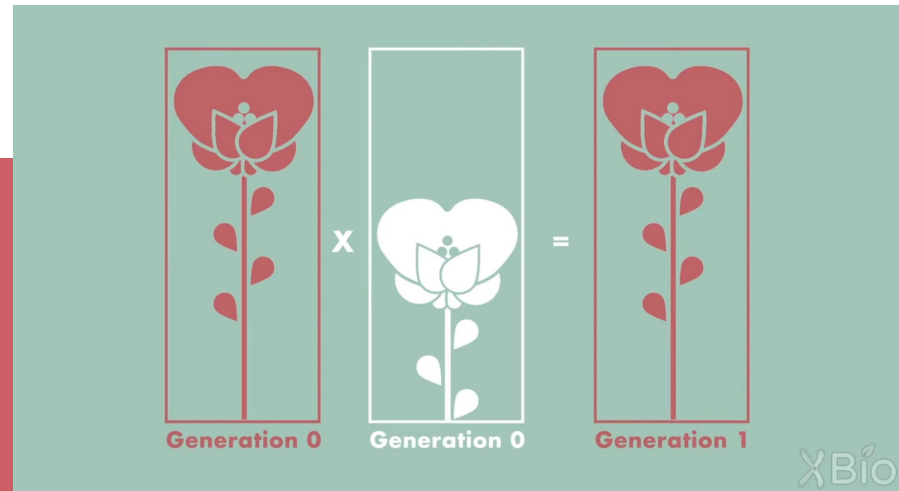
W alelo selvagem codifica starch branch enzyme I (SBEI))

Homozygous mutant allele → no enzyme activity → starch synthesis affected → wrinkled peas

Mutation = sequence interrupted by a **transposon**

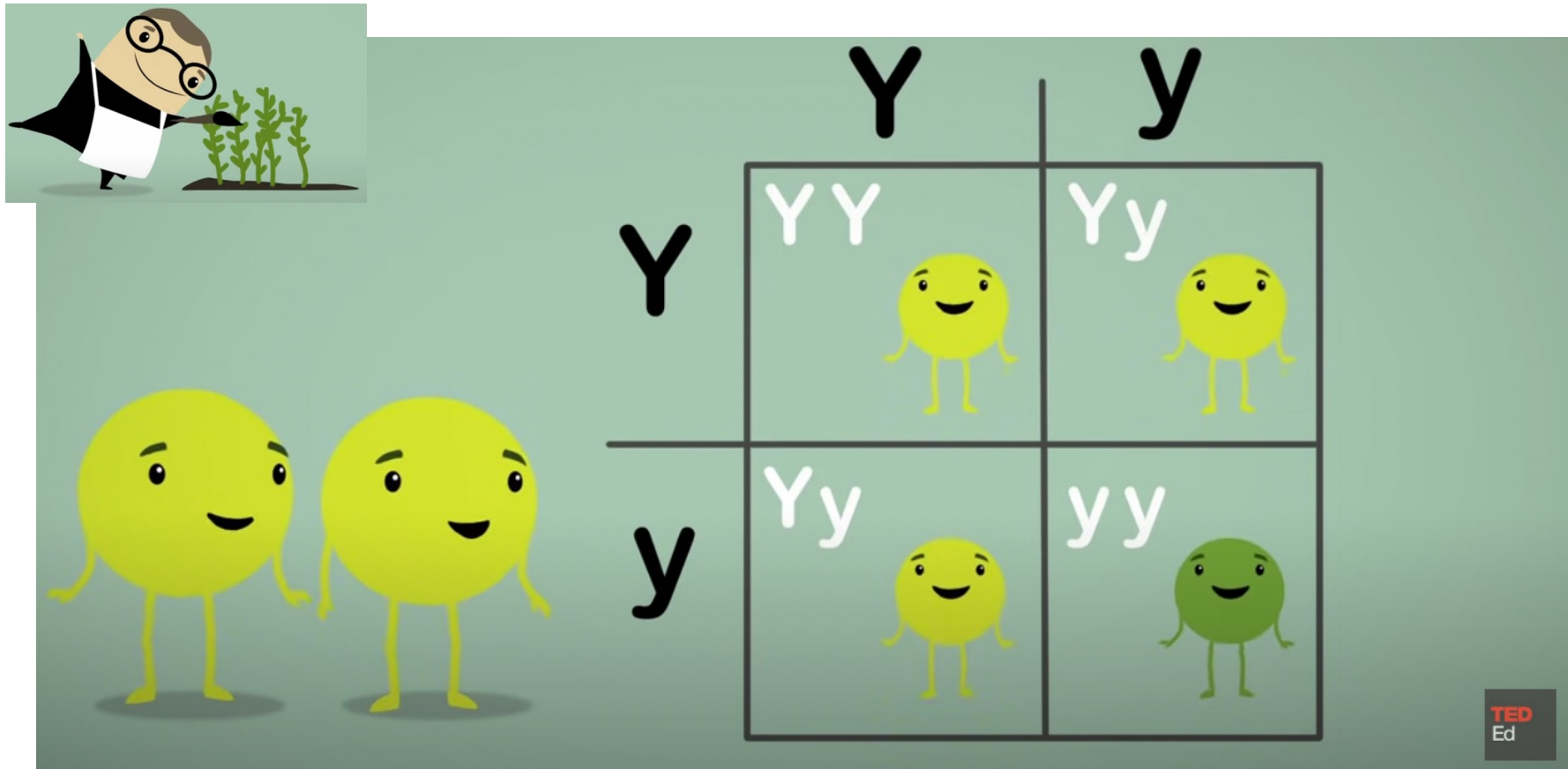
GREGOR MENDEL'S FAMOUS GENETICS EXPERIMENT

SHIRLEY TILGHMAN



<https://www.youtube.com/watch?v=XgXLVaBjQqw>

How Mendel's pea plants helped us understand genetics

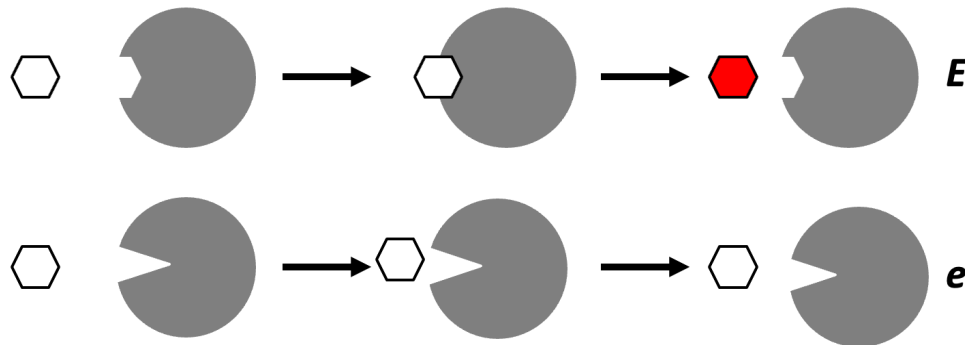


https://www.youtube.com/watch?v=Mehz7tCxjSE&ab_channel=TED-Ed

3:06

Ex: Flor branca recessiva em relação a **vermelha**
→ sugere que uma só dose do **alelo selvagem**
é **suficiente** para conferir a **pigmentação normal**

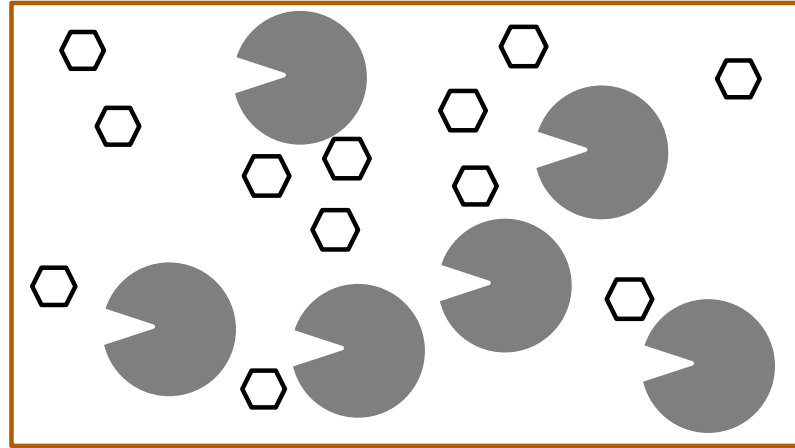
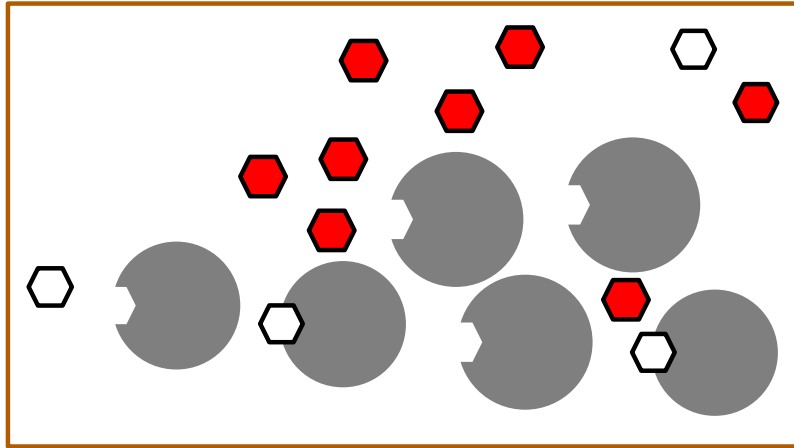
O alelo *e* pode conter uma mutação **que** impede a ligação da enzima ao substrato



EE

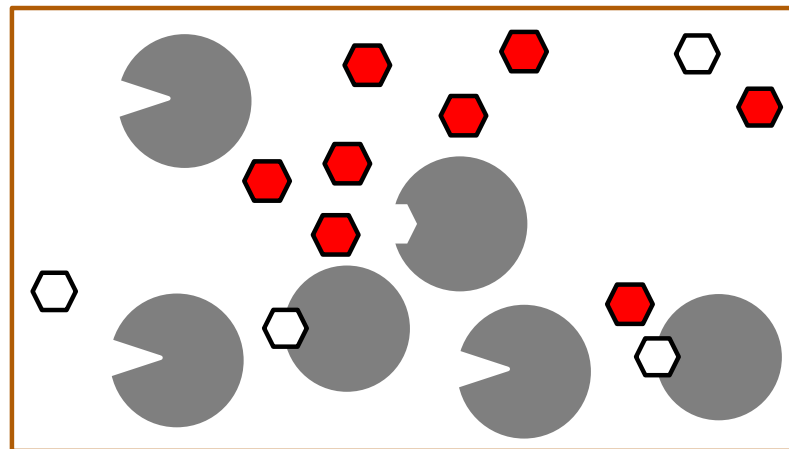
Homozigóticos

ee



Heterozigótico

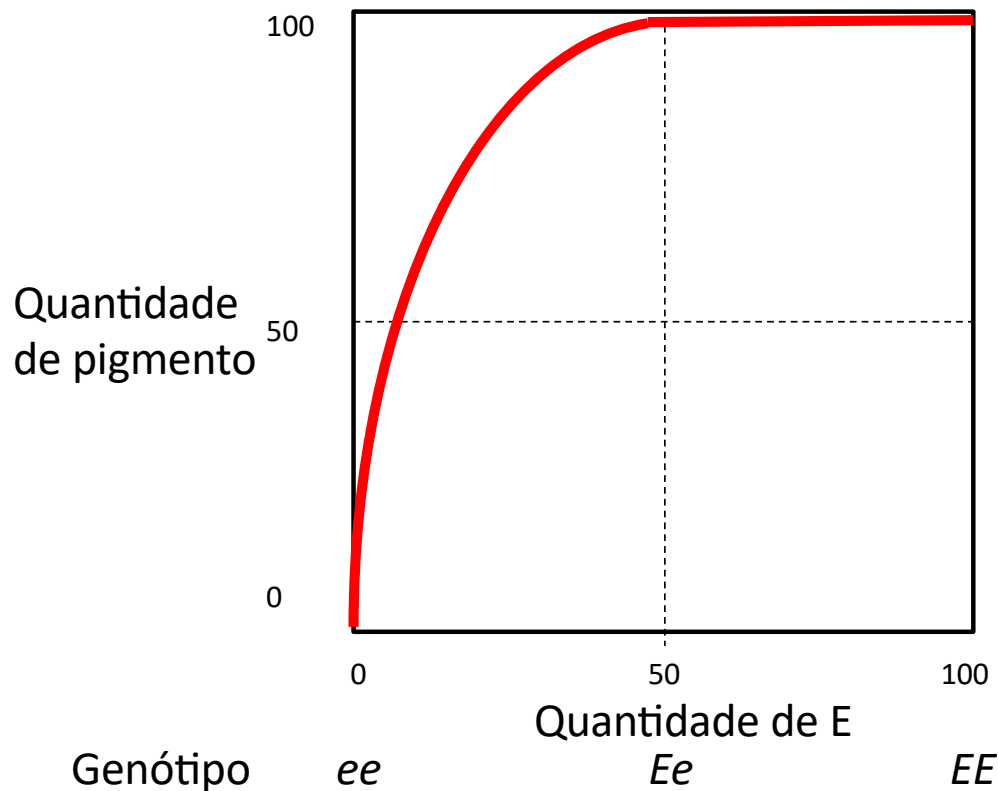
Ee



Heterozigótico Ee

produz apenas metade da quantidade de enzima do que EE

Se a curva de resposta à variação na quantidade de proteína é como na figura,
dose suficiente para produzir 100% de pigmento



Dominância

Alelo selvagem **Haplosuficiente**

→ Uma só cópia do alelo do tipo selvagem é **suficiente**

Alelo selvagem **haploinsuficiente**

→ **relação** entre os dois alelos é de **dominância incompleta**

