Traits determined by a single gene

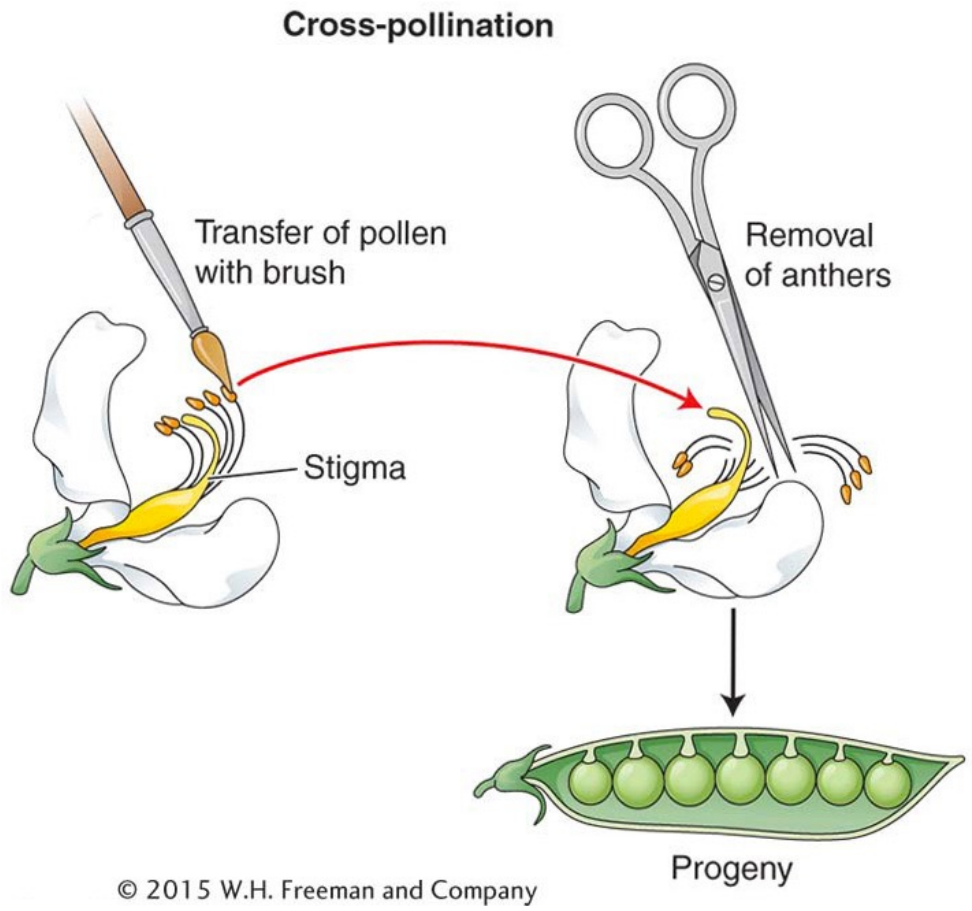
Ch1.1, Ch2.1-2.3

Blending theory (hypothesis) : traits of parents get mixed like fluid in the offspring, resulting into new traits that resembles parents.

Prediction: If blended like fluid, parent's traits are lost in the offspring and cannot be recovered!

Mendel's method

Purple petals

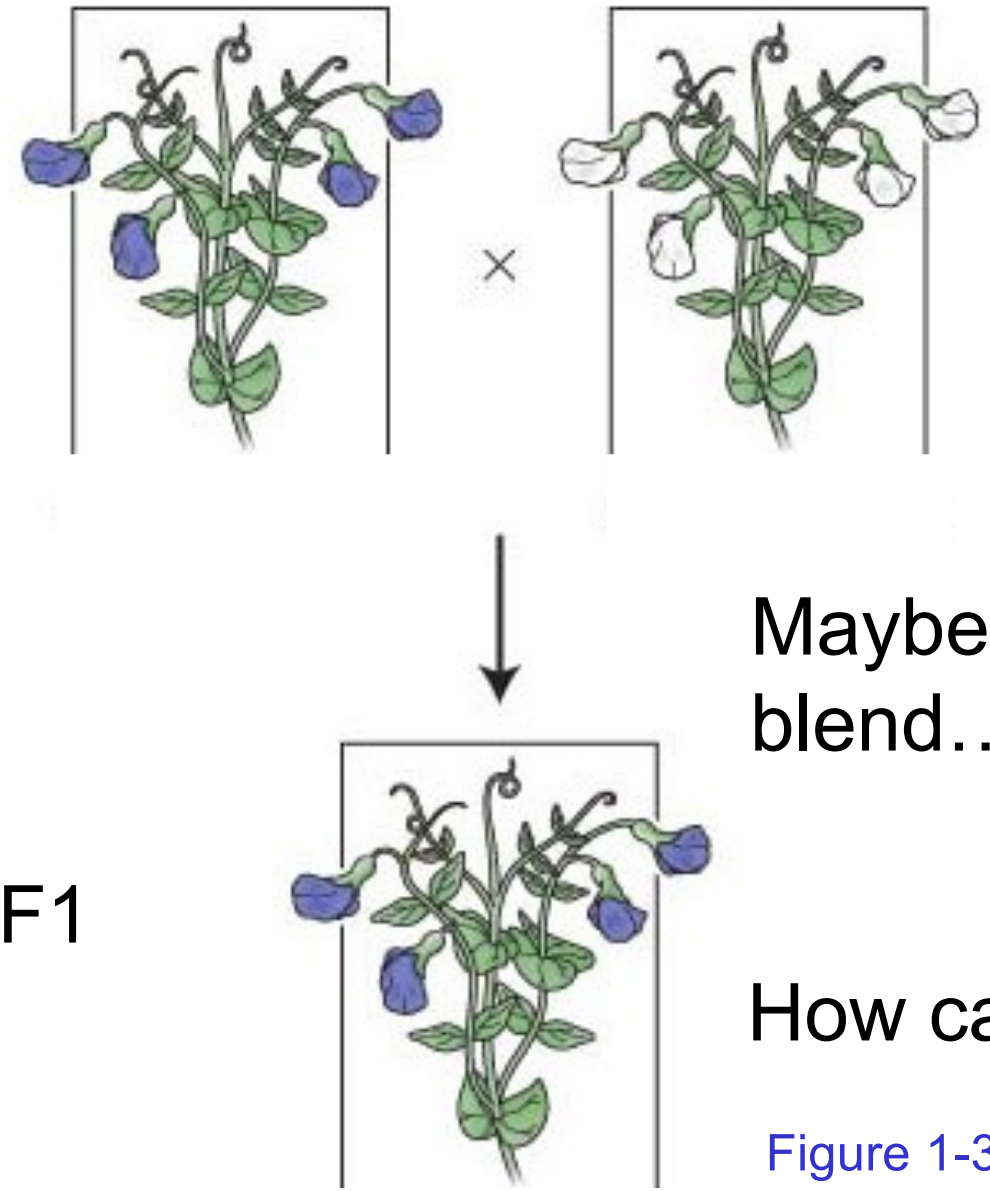


White petals

Figure 2-3

Mendel was able to <u>control</u> the crosses between different <u>purebred</u> pea plants.

Mendel's result #1



100% of F1

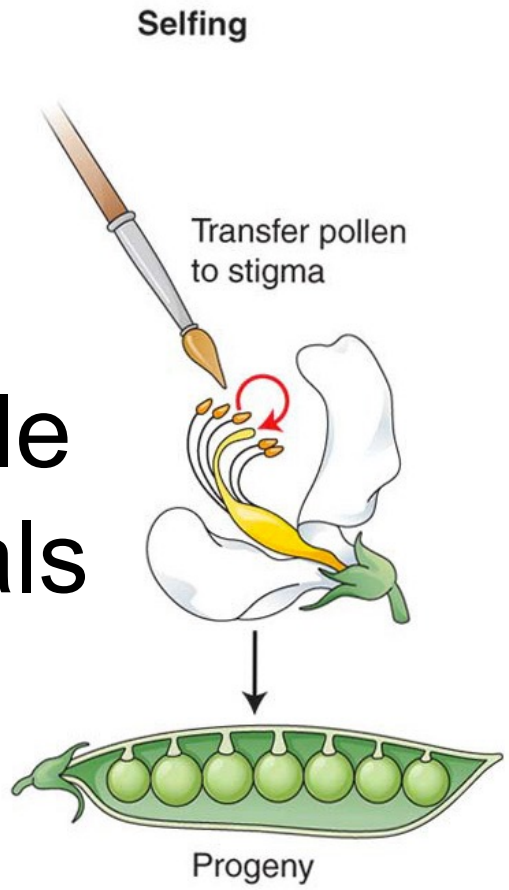
Maybe they don't blend…..

How can you test this?

Figure 1-3

Mendel's experiment #2

F1: Purple Petals



What is your prediction if "Blending theory" is correct?

Figure 2-3

Self-cross of F1 (monohybrid cross)

Mendel's result #2

705 224

Figure 1-3

The result argues against the "blending theory"

Presence of white flowers means recovery of the parental traits, indicating that they don't get lost.

This also suggests that the "element" responsible for the flower color (trait/phenotype) works **like particles, not like fluid**, not only maintained from one generation to the next but can be separated

Mendel did similar experiments with seven different traits

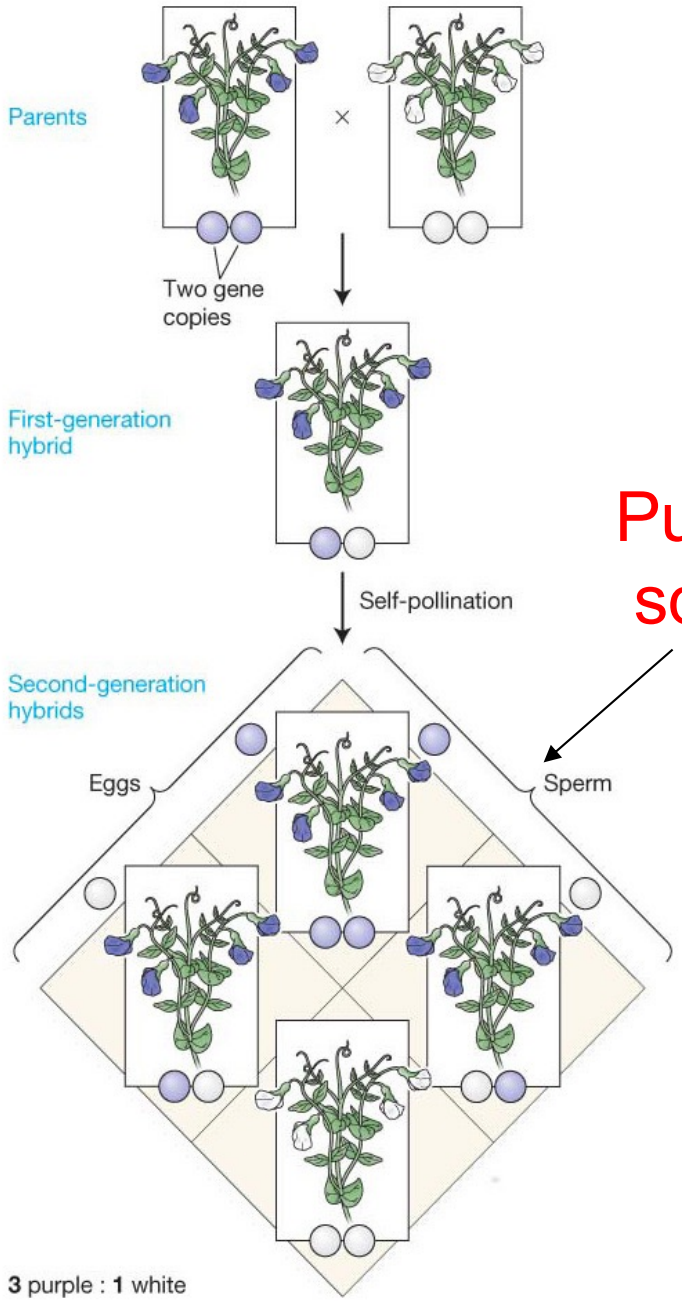
TABLE 2-1 Results of All Mendel's Crosses in Which Parents Differed in One Character

Parental phenotypes	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. inflated × 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

1. There is a **dominant** trait that hides the **recessive** trait

2. The "element" producing traits are transmitted to the next generation in a predictable pattern

Mendel's model (hypothesis) 1866



Punnett square

There are two copies of the "**element**" (associated with a trait/phenotype) that are inherited from the parents, but only one of the two is visible (**dominant/recessive**).

Only **one** of the two is transmitted to the next generation.

In the monohybrid cross, each of the two parental elements has an equal chance of transmitted to the next generation. However, because 3 out of the 4 possible element combinations in the offspring contain at least one **dominant trait**, the cross results in the 3:1 ratio of dominant and recessive traits.

Figure 1-3

The reason why Mendel's experiments worked

Traits affected by only one gene. Many traits (people's height) are affected by multiple genes

Pure genetic background and ability to cross- or self- pollinate (ability to control cross/mating).

Ability to obtain a large number of progeny

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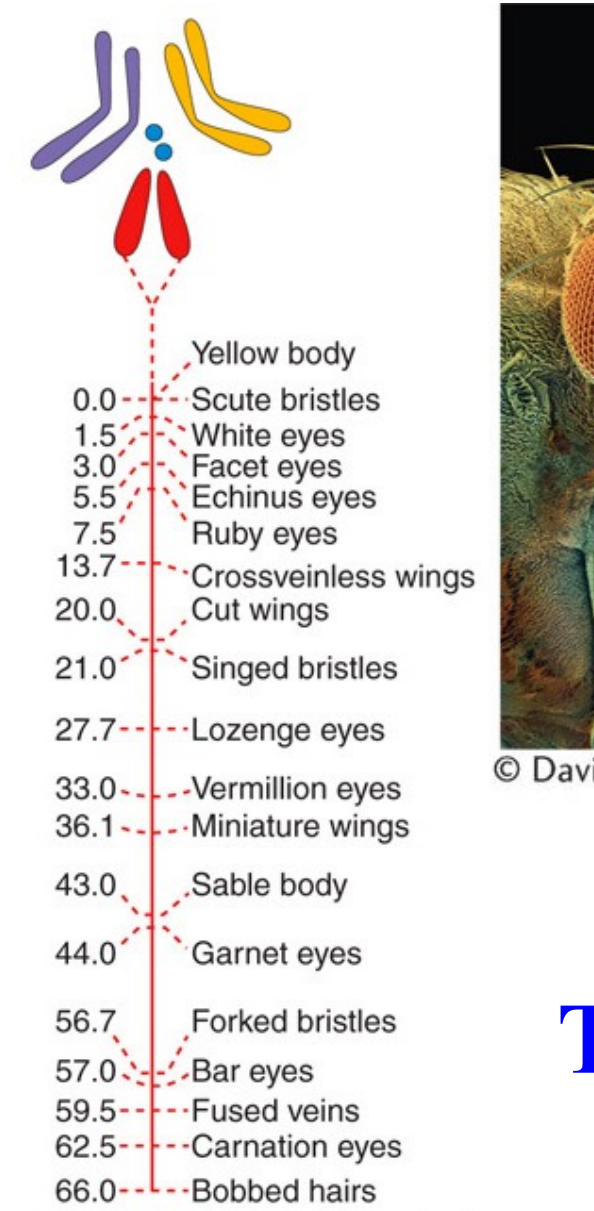
Why is the ability to obtain a large number of progeny important?

slido

Why is the ability to obtain a large number of progeny important?

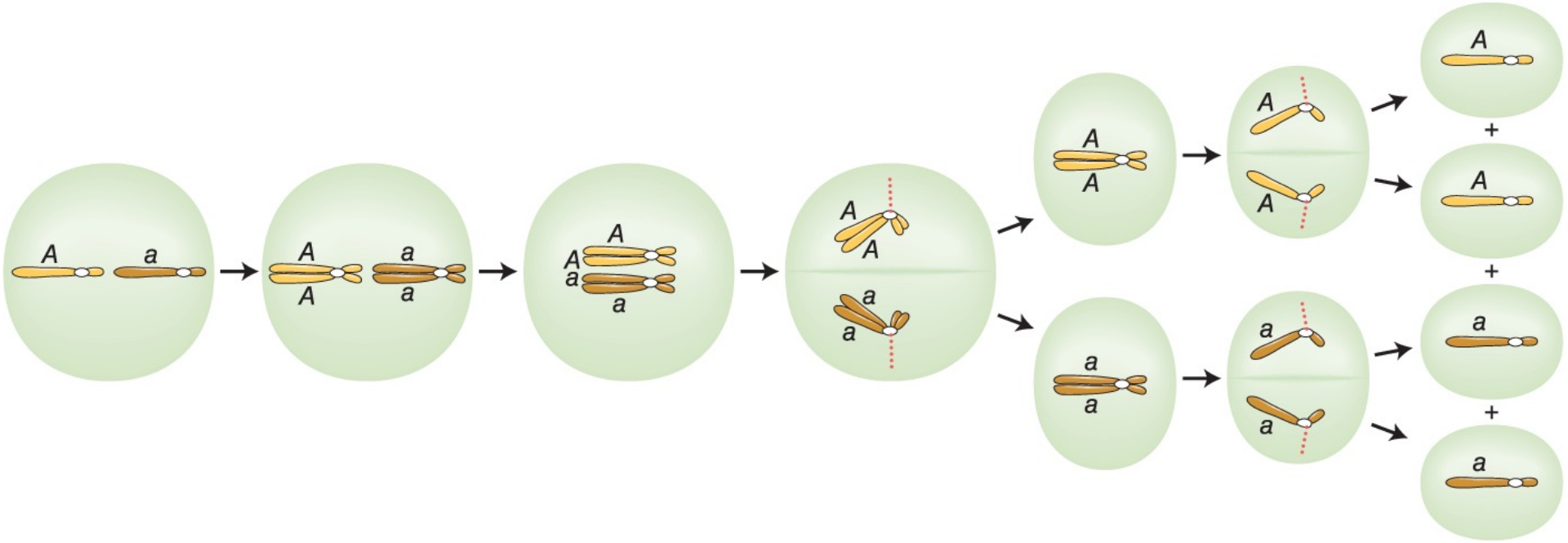
ⓘ Start presenting to display the poll results on this slide.

The “element” is the gene



Traits determined by a single gene

Meiosis in the GERM CELL is the cellular/molecular base of Mendel's law

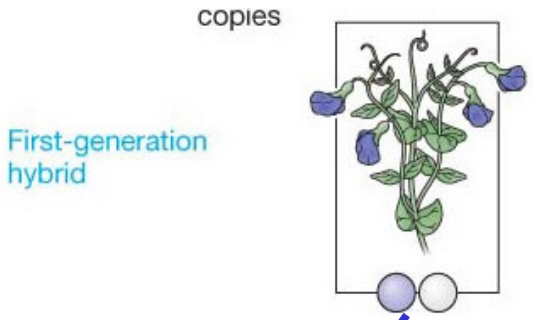


Griffiths et al., *Introduction to Genetic Analysis*, 12e, © 2020 W. H. Freeman and Company

During meiosis, a A/a cell produces 4 gametes, 2 X A & 2 X a. Therefore, meiosis produces the same ratio of A to a, giving an equal chance of transmitting between A or a.

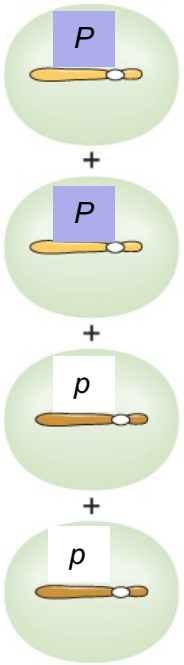
Figure 2-9

Monohybrid cross

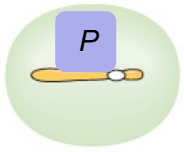


Monohybrid:
Heterozygote of a gene

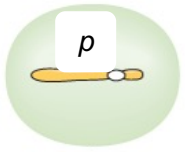
Meiosis



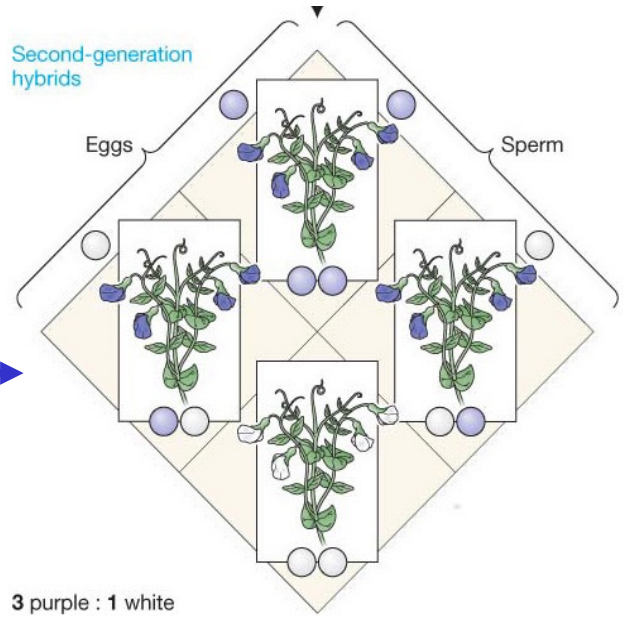
During self pollination, only 1 of the 4 gametes is transmitted to the next generation



or



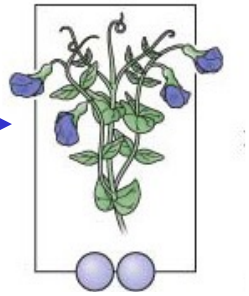
Phenotypically
3 purple
1 white



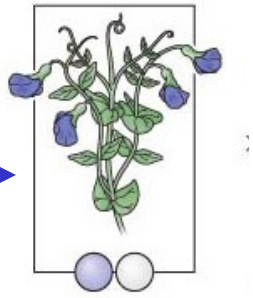
Testcross: the cross of an individual to a fully recessive individual

Testcross is very useful for determining the genotype a testee.

#1. Purple →



#2. Purple →



Cross with #1 will give 100% purple
Cross with #2 will give 50% purple & 50% white

Ch2.1 & 2.3: definition and terminology

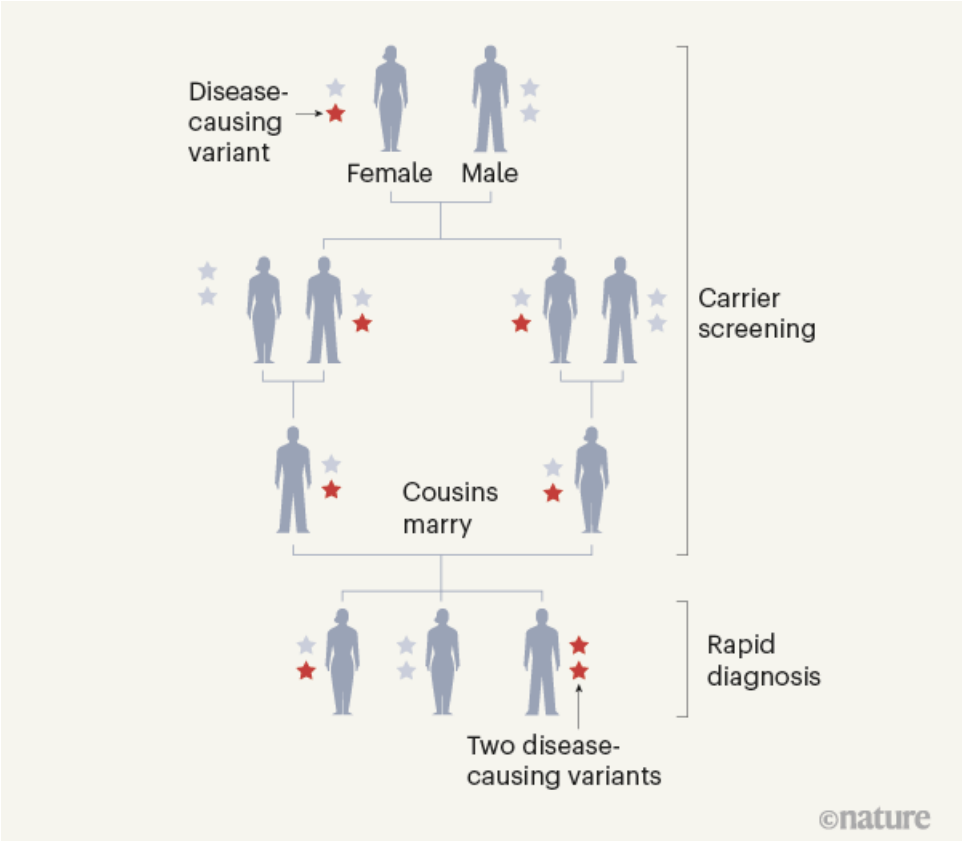
Loss of function vs. Recessive

Gene vs. allele

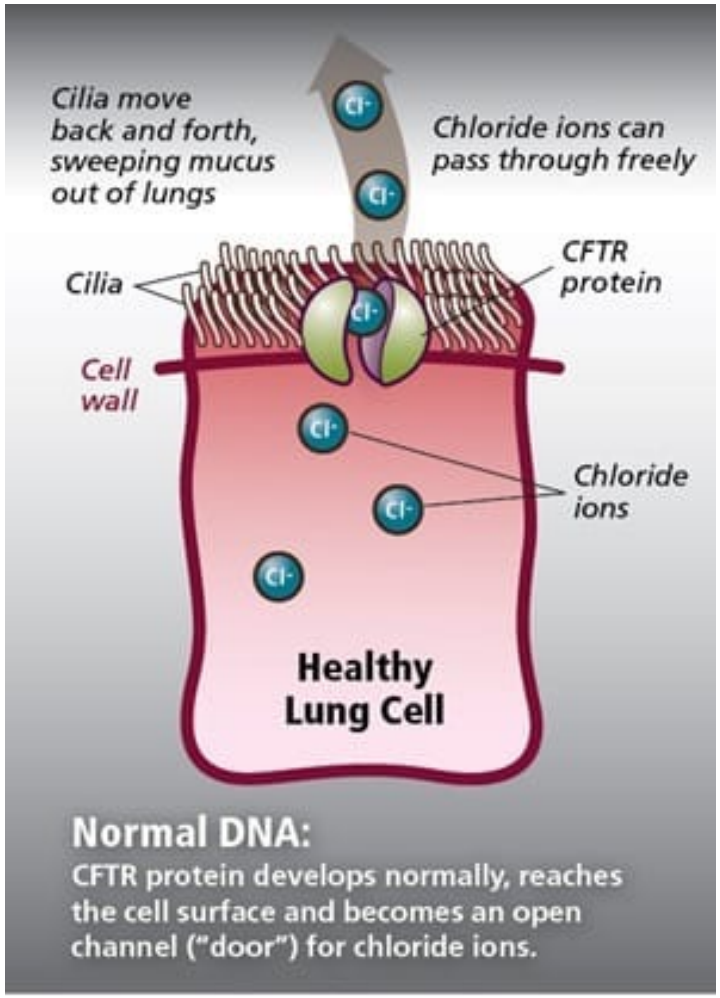
-dominant allele
-recessive allele

Loss of function mutations (molecular term) are often recessive (phenotype) because a single wild-type copy of the gene can often provide necessary functions for the organism. Most genes (wild-type) are haplosufficient

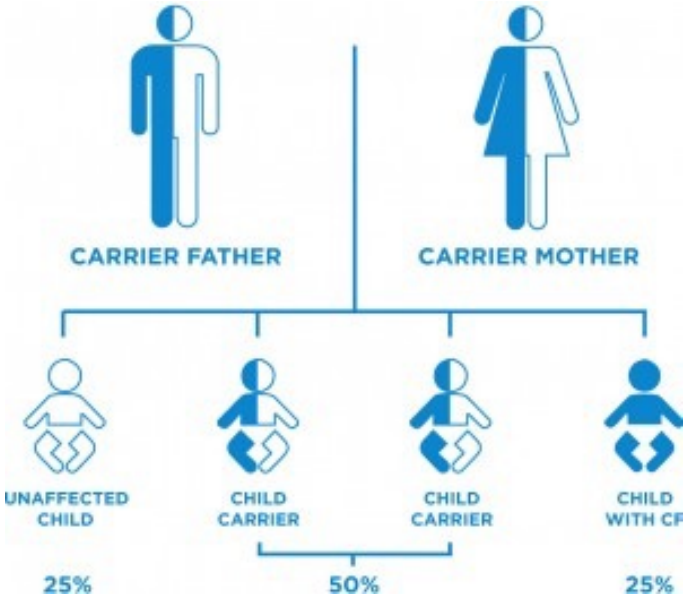
Genetic disorders and cousin marriage



Cystic Fibrosis as a Mendelian Disorder



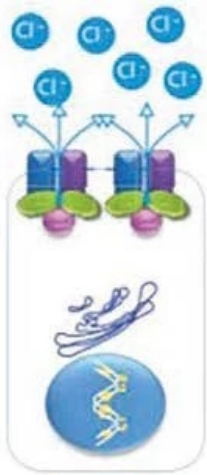
- Cystic fibrosis affects the cells that produce mucus, sweat and digestive juices such as the lung.
- Prevalence in Canada is around 1/3600, around 1/30 is a carrier.
- Caused by mutation in the Cystic Fibrosis Transmembrane Receptor (CFTR gene)
- Gene identified by researchers at Hospital for Sick Children - Toronto



Two carrier parents have 1 in 4 chances of having a child with CF because CF is a haplosufficient gene.

Alleles of a single gene

FIGURE 1 ***CFTR* MUTATIONS**

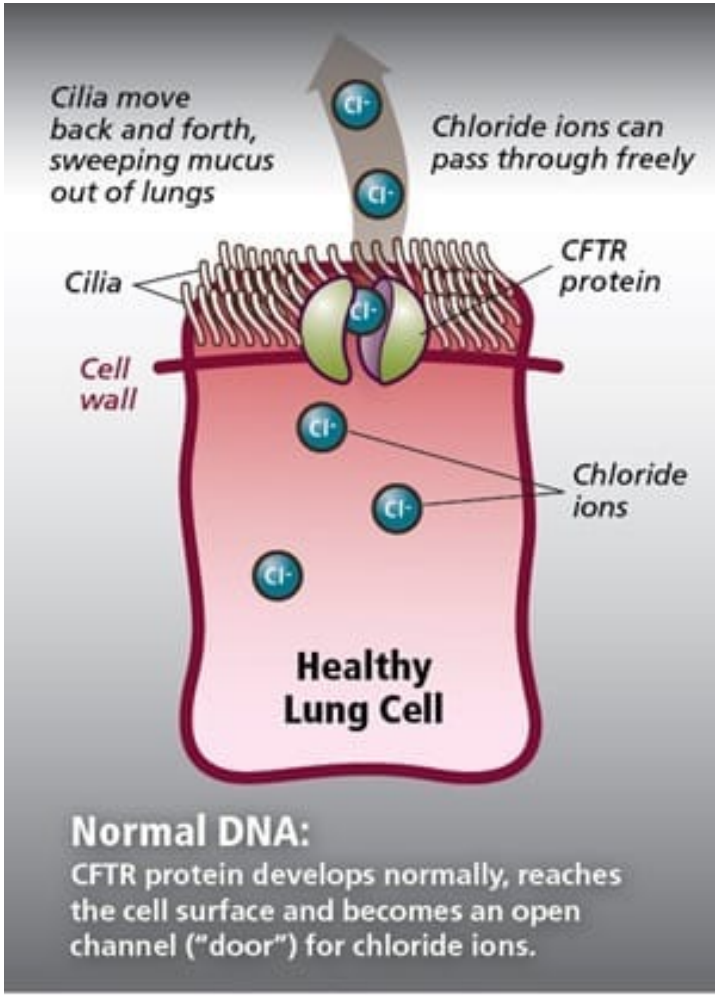





	Normal	Class I	Class II	Class III	Class IV	Class V
DESCRIPTION	CFTR is created, reaches cell surface and functions properly, allowing transfer of chloride and water.	No functional CFTR created.	CFTR protein is created, but misfolded, keeping it from reaching the cell surface.	CFTR protein is created and reaches cell surface, but does not function properly.	The opening in the CFTR protein ion channel is faulty.	CFTR is created in insufficient quantities.
EXAMPLES		G542X W1282X R553X	F508del N1303K I507del	G551D S549N V520F	R117H D1152H R347P	3849+10kbC->T 2789+5G->A A455E

SOURCE OF DATA: Cystic fibrosis patients under care at CF Foundation-accredited care centers in the United States, who consented to have their data entered.
Adapted from: CFTR Modulator Therapies; http://www.cff.org/Life-With-CF/Treatments-and-Therapies/Medications/CFTR-Modulator-Therapies/#section2.
Abbreviations: CF, cystic fibrosis; CFTR, cystic fibrosis transmembrane conductance regulator.

Why is the prevalence of CF in Canada around 1/3600 when1/30 people is a carrier?



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