Genetics

Mendel Genetics

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A good Mendel Organism

- Good known genetic past
- Short Generation Interval
- Large litter size (lots of progeny)
- Easily controlled; small size
- Some individuals have mutations
  - Good genetic variation, which is required for everything.

Mendel Genetic Variations

- **Continuous – Many levels with small differences**
- **Discrete – Couple of different levels**
- **Mendel picked the second**

Gregor Johann Mendel

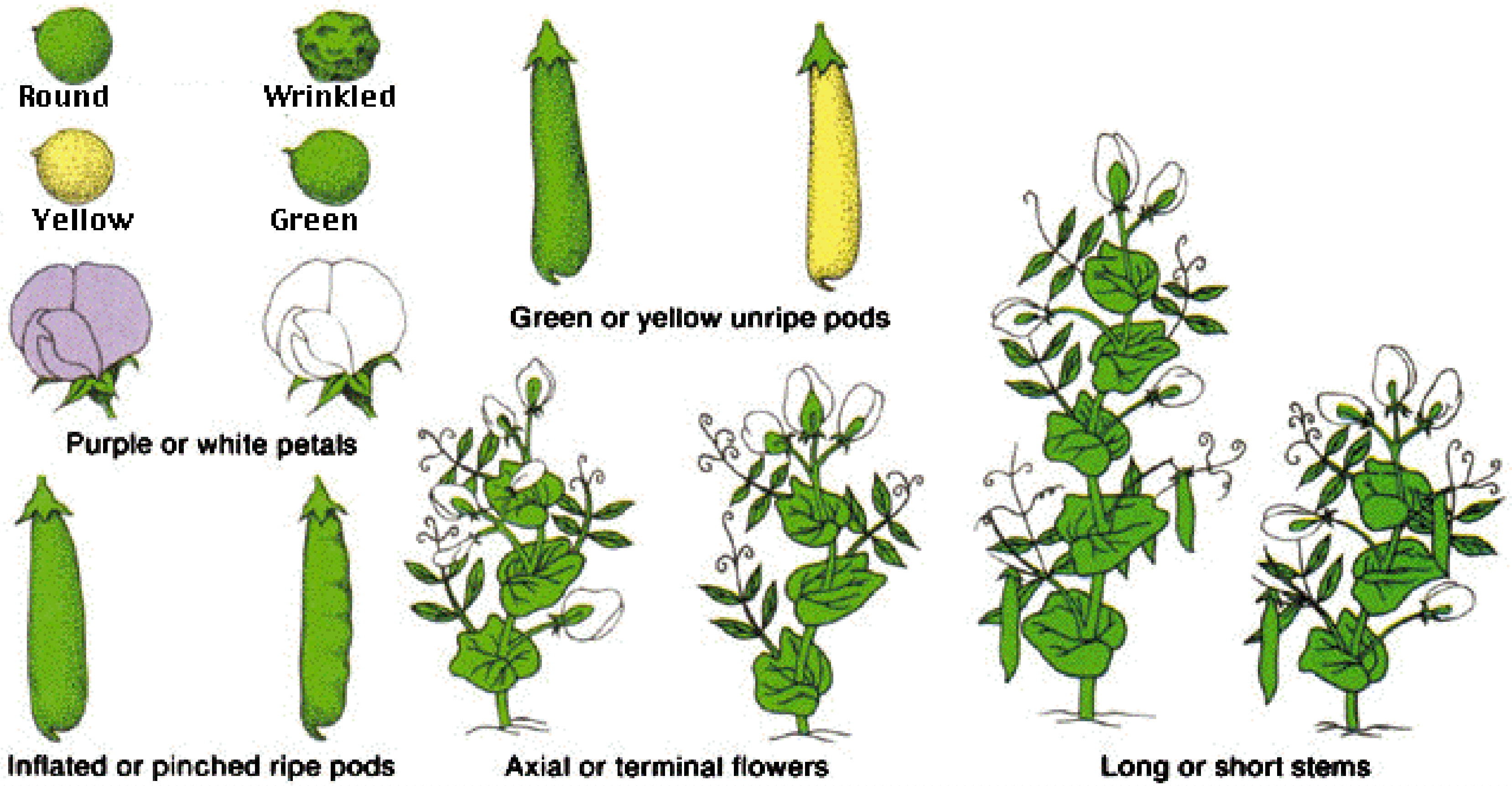
- Austrian Monk, born in what is now Czech Republic in 1822
- Son of peasant farmer, studied Theology and was ordained priest Order St. Augustine.

Gregor Mendel

- Went to the university of Vienna, where he studied botany and learned the Scientific Method
- Worked with pure lines of peas for eight years
- Prior to Mendel, heredity was regarded as a “blending” process and the offspring were essentially a “dilution” of the different parental characteristics.

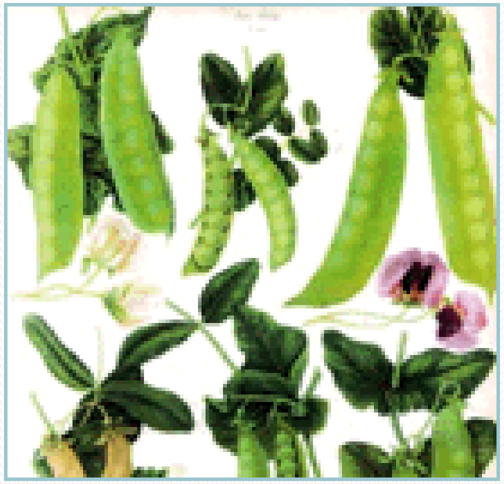
Mendel's peas

- Mendel looked at seven traits or characteristics of pea plants:



- In 1865, he presented his theory to his peers.
- In 1866 he published *Experiments in Plant Hybridization*, (*Versuche über Pflanzen-Hybriden*) in which he established his three Principles of Inheritance
- Work was largely ignored for 34 years, until 1900, when 3 independent Botanists rediscovered Mendel's work.

- Mendel was the first biologist to use Mathematics – to explain his results quantitatively.
- Mendel predicted
  - The concept of genes
  - That genes occur in pairs
  - That one gene of each pair is present in the gametes



7 Characteristics in Peas

Trait	Stem length	Pod shape	Seed shape	Seed color	Flower position	Flower color	Pod color
Characteristics	Tall	Inflated	Smooth	Yellow	Lateral	Purple	Green
	Dwarf	Constricted	Wrinkled	Green	Terminal	White	Yellow

Qualitative traits?

- Fur color
- Conception rate
- Having horns or being polled
- Tall or short?
- Level of coloration?

Monohybrid cross

- Parents differ by a single trait.
- Crossing two pea plants that differ in stem size, one tall one short

T = allele for Tall

t = allele for dwarf

TT = homozygous tall plant

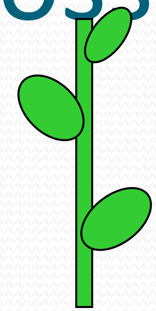
t t = homozygous dwarf plant

$$TT \times tt$$

Long or short stems

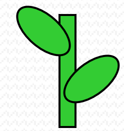
Monohybrid cross for stem length:

P = parentals
true breeding,
homozygous plants:



$\mathrm{T}\,\mathrm{T} \times t\,t$

(tall) (dwarf)

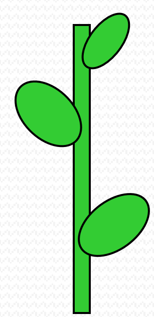


↓

F$_1$ generation
is heterozygous:

$\mathrm{T}\,t$

(all tall plants)

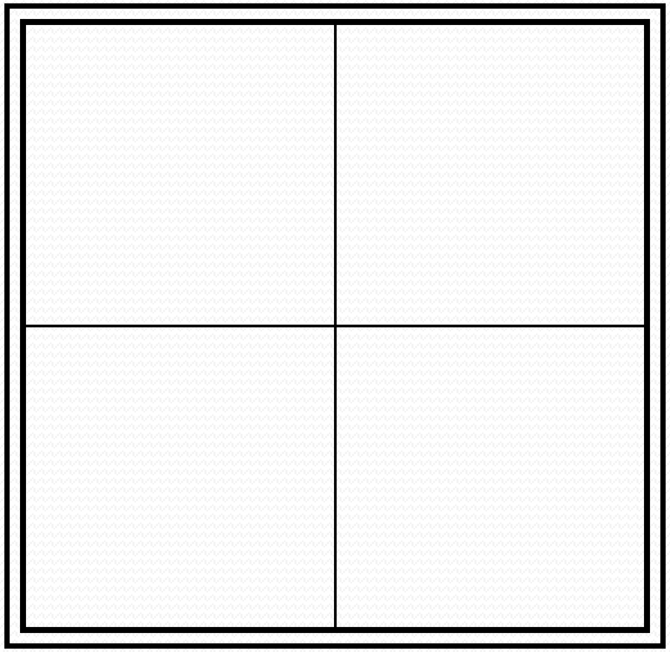



HETEROZYGOATS
Just allele uneven.

Punnett square

- A useful tool to do genetic crosses
- For a monohybrid cross, you need a square divided by four….
- Looks like a window pane…

  We use the Punnett square to predict the genotypes and phenotypes of the offspring.



Using a Punnett Square

STEPS:

1. determine the genotypes of the parent organisms
2. write down your "cross" (mating)
3. draw a p-square

Parent genotypes:

TT and *t t*

Cross

T T $\times$ ***t t***

Punnett square

4. **"split" the letters of the genotype for each parent & put them "outside" the p-square**
5. determine the possible genotypes of the offspring by filling in the p-square
6. summarize results (genotypes & phenotypes of offspring)

T T × *t t*

	T	**T**
t	**T *t***	**T *t***
t	**T *t***	**T *t***

Genotypes:
100% T t

Phenotypes:
100% Tall plants

Monohybrid cross: F_2 generation

- If you let the F1 generation self-fertilize, the next monohybrid cross would be:

T *t* × **T *t***
(tall) (tall)

	T	***t***
T	**T T**	**T *t***
t	**T *t***	***t t***

<u>Genotypes:</u>
1 TT= Tall
2 T*t* = Tall
1 *tt* = dwarf
Genotypic ratio= 1:2:1

<u>Phenotype:</u>
3 Tall
1 dwarf
Phenotypic ratio= 3:1

Secret of the Punnett Square

- Key to the Punnett Square:
- Determine the <u>gametes</u> of each parent…
- How? By "splitting" the genotypes of each parent:

If this is your cross

T T × ***t t***

The gametes are:

T **T** ***t*** ***t***

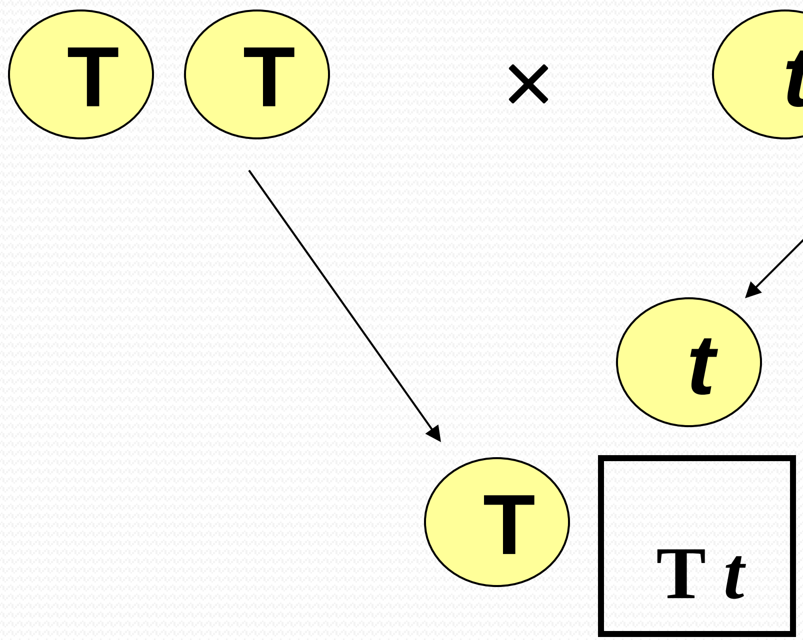
Once you have the gametes…

T T × *t* *t*

	t	*t*
T	T*t*	T*t*
T	T*t*	T*t*

Shortcut for Punnett Square…

- If either parent is HOMOZYGOUS



Genotypes:
100% T t

Phenotypes:
100% Tall plants

- **You only need <u>one</u> box**

Understanding the shortcut…

	t	*t*
T	T*t*	T*t*
T	T*t*	T*t*

=

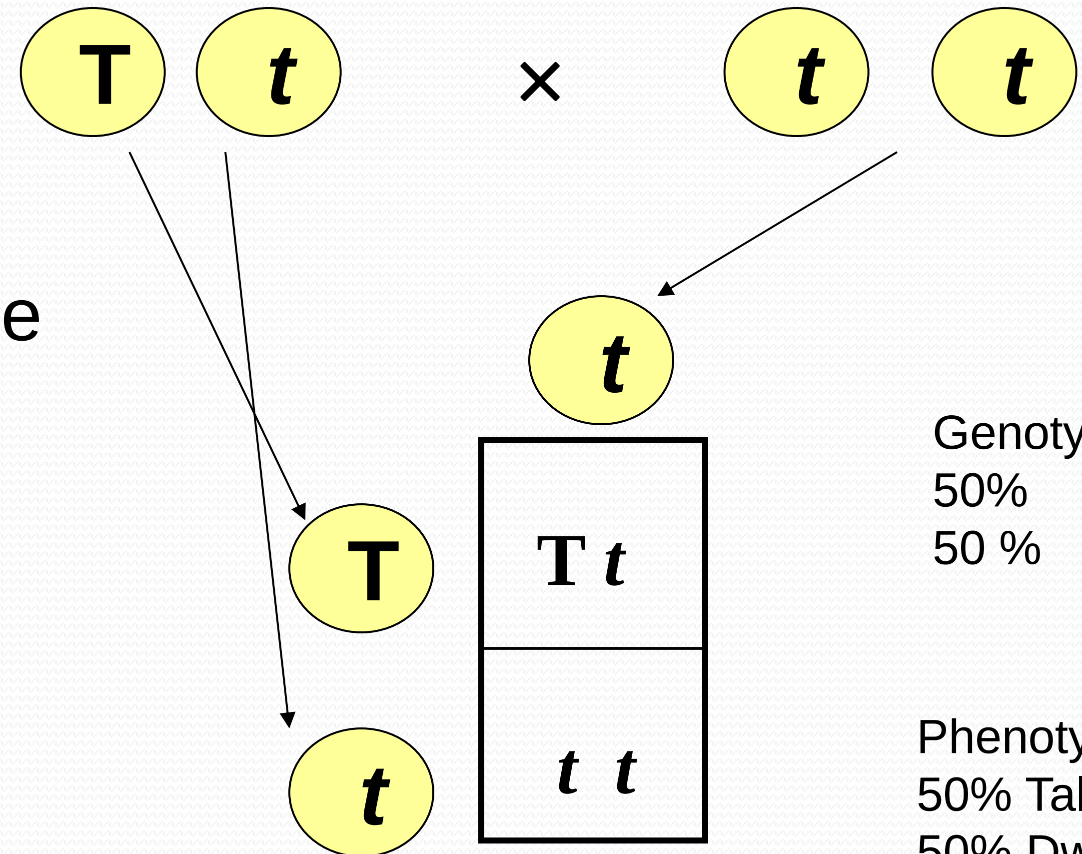
	t
T	T*t*

Genotypes:
100% T t

Phenotypes:
100% Tall plants

If you have another cross…

- A heterozygous with a homozygous



You can still use the shortcut

Genotypes:
50% T t
50 % *t t*

Phenotypes:
50% Tall plants
50% Dwarf plants

Another example: Flower color

For example, flower color:

P = purple (dominant)

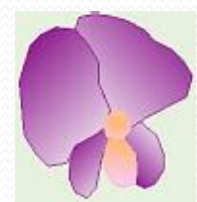
p = white (recessive)

If you cross a homozygous Purple (PP) with a homozygous white (*pp*):

P P × *p p*

↓

P *p*



ALL PURPLE (P*p*)

Cross the F1 generation:

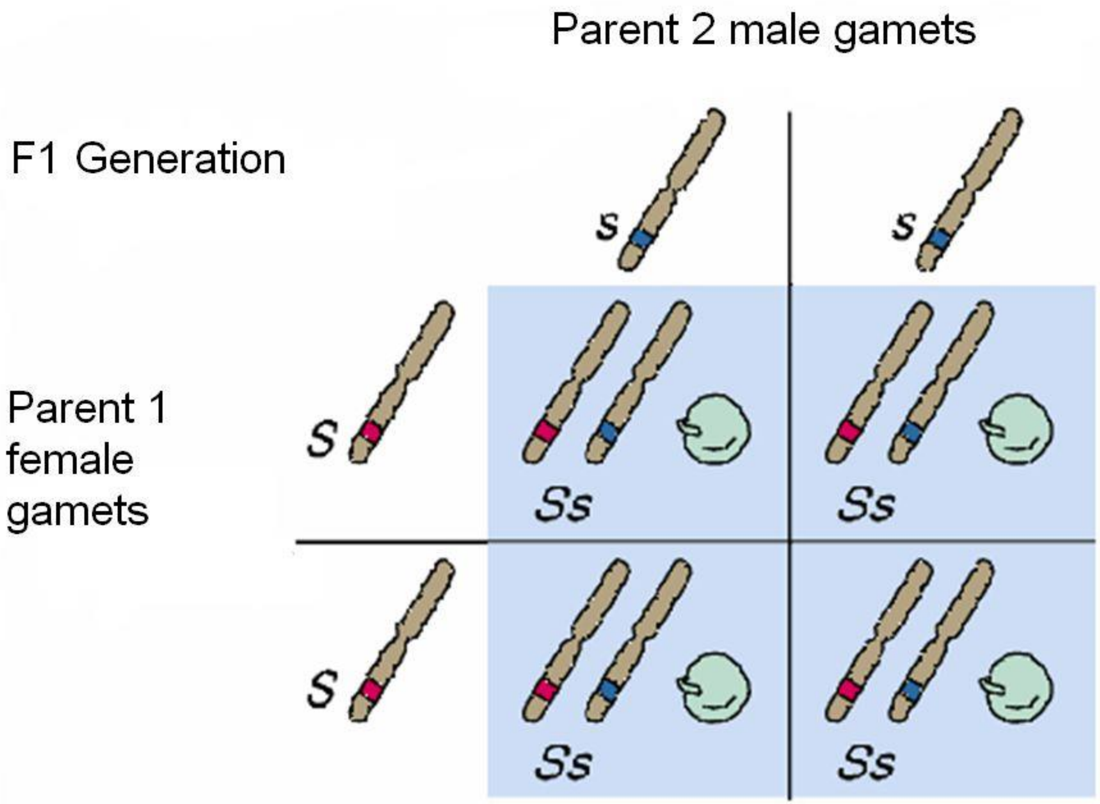
P *p* × P *p*

	P	*p*
P	**P P**	**P *p***
p	**P *p***	***p p***

Genotypes:
1 PP
2 Pp
1 pp

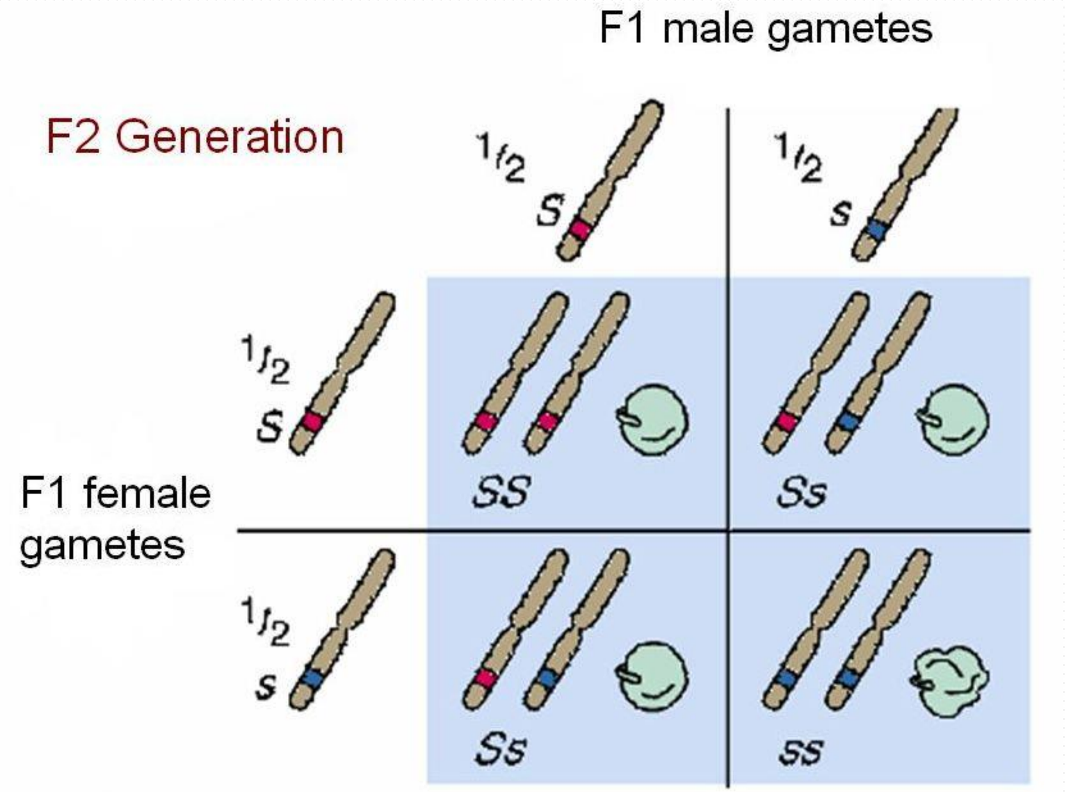
Phenotypes:
3 Purple
1 White

Mendel’s Monohybrid Crosses



F1 genotypes: all Ss
F1 phenotypes: all smooth
Prof. Dr. Akin Pala

Mendel's Monohybrid Crosses F2



- F2 genotypes: 1/4SS, ½ Ss, ¼ ss
- F2 phenotypes: 3/4 smooth, ¼ wrinkled EXPECTED

Mendel's Principles

- **1. Principle of Dominance**:

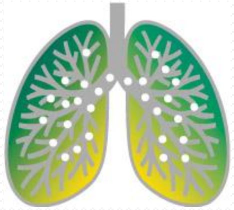
  One allele masked another, one allele was dominant over the other in the F_1 generation.

- **2. Principle of Segregation**:

  When gametes are formed, the pairs of hereditary factors (genes) become separated, so that each sex cell (egg/sperm) receives only one kind of gene.

Human case: CF

- Mendel's Principles of Heredity apply universally to all organisms.
- Cystic Fibrosis: a lethal genetic disease affecting Caucasians.
- Caused by mutant recessive gene carried by 1 in 20 people of European descent
- One in 400 Caucasian couples will be both carriers of CF – 1 in 4 children will have it.
- CF disease affects transport in tissues – mucus is accumulated in lungs, causing infections.



Inheritance pattern of CF

IF two parents carry the recessive gene of Cystic Fibrosis (*c*), that is, they are heterozygous (C *c*), one in four of their children is expected to be homozygous for *cf* and have the disease:

	C	*c*
C	C C	C *c*
c	C *c*	*c c*

C C = normal
C *c* = carrier, no symptoms
c c = has cystic fibrosis

Probabilities…

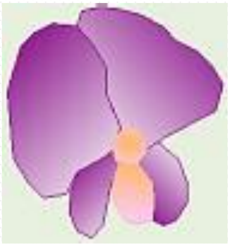
- Of course, the 1 in 4 probability of getting the disease is just an **expectation**, and in reality, any two carriers may have normal children, or all 4 may get the disease.
- However, the greatest probability is for 1 in 4 children to be affected.
- Important factor when prospective parents are concerned about their chances of having affected children.
- 1 in 29 Americans is a symptomless carrier (Cf *cf*) of the gene.

Polyhybrid: Dihybrid crosses

- Matings that involve parents that differ in **<u>two</u>** genes (two independent traits)

For example, flower color:

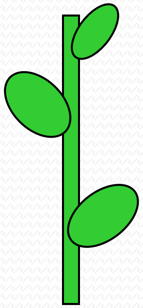
P = purple (dominant)



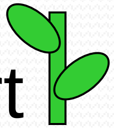
p = white (recessive)

and stem length:

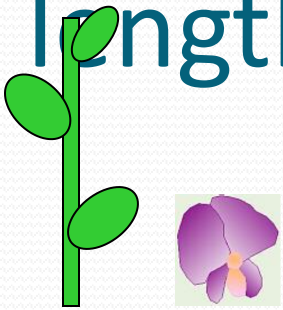
T = tall



t = short

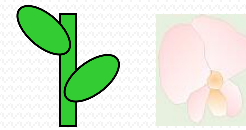


Dihybrid cross: flower color and stem length



TT PP × *tt pp*

(tall, purple) (short, white)



Possible Gametes for parents

T P and *t p*

	tp	*tp*	*tp*	*tp*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*
TP	T*t*P*p*	T*t*P*p*	T*t*P*p*	T*t*P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross: flower color and stem length (shortcut)

TT PP × *tt pp*

(tall, purple) (short, white)

Possible Gametes for parents

T P *t p*

	T P
t *p*	T*t* P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

$$\mathrm{T}t\ \mathrm{P}p \quad \times \quad \mathrm{T}t\ \mathrm{P}p$$

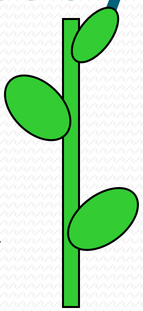
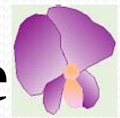
(tall, purple) (tall, purple)

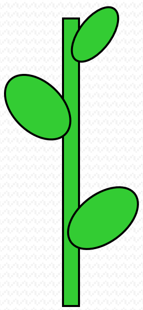
Possible gametes:
TP T*p* *t*P *tp*

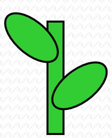
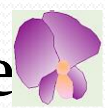
	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

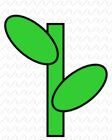
Four phenotypes observed
Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Dihybrid cross

9 Tall  purple 

3 Tall  white 

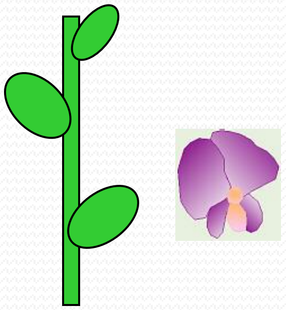
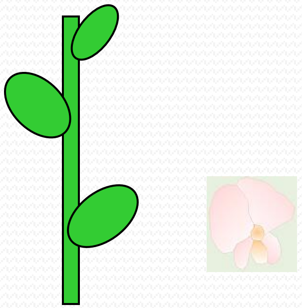
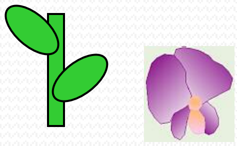
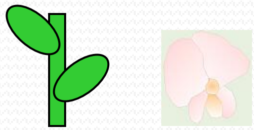
3 Short  purple 

1 Short  white 

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Phenotype Ratio = 9:3:3:1

Dihybrid cross: 9 genotypes

Genotype ratios (9):		Four Phenotypes:	
1	TTPP	Tall, purple (9)	
2	TTP*p*		
2	T*t*PP		
4	T*t*P*p*		
1	TT*pp*	Tall, white (3)	
2	T*tpp*		
1	*tt*PP	Short, purple (3)	
2	*tt*P*p*		
1	*ttpp*	Short, white (1)	

Principle of Independent Assortment

- Based on these results, Mendel postulated the
  3. Principle of Independent Assortment:

  "Members of one gene pair segregate independently from other gene pairs during gamete formation"

Genes get shuffled – these different combinations are one of the advantages of sexual reproduction

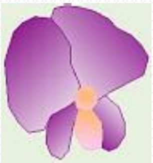
Test cross

When you have an individual with an unknown genotype, you do a **test cross**.

Test cross: Cross with a homozygous recessive individual.

For example, a plant with **purple** flowers can either be **PP** or **P*p***… therefore, you cross the plant with a *pp* (white flowers, homozygous recessive)

P ? × *pp*



Test cross

- If you get all 100% purple flowers, then the unknown parent was PP…
- Could be due to chance, so increasing the # of obs. is important.

In probability theory, the **central limit theorem** (CLT) states that, given certain conditions, the arithmetic mean of a sufficiently large number of iterates of independent random variables, each with a well-defined expected value and well-defined variance, will be approximately normally distributed, regardless of the underlying distribution.

	P	**P**
p	**P *p***	**P *p***
p	**P *p***	**P *p***

- If you get 50% white, 50% purple flowers, or even some white flowers, then the unknown parent was Pp

	P	***p***
p	**P *p***	***p p***
p	**P *p***	***p p***

Homework Q1 (use turnitin)

- Black coat (B) is dominant over red coat (b) in cattle
- Polled (P) is dominant over having horns (p)
- If you had a black polled animal, how would you test its genotype?

Beyond Mendelian Genetics: Incomplete Dominance

- Mendel was lucky.
- Traits he chose in the pea plant showed up very clearly.
- One allele was dominant over another, so phenotypes were easy to recognize.
- But sometimes phenotypes are not very obvious…

Incomplete Dominance

Snapdragon flowers come in many colors.

If you cross a red snapdragon (RR) with a white snapdragon (rr)

You get PINK flowers (Rr)

R R × *r r*

↓

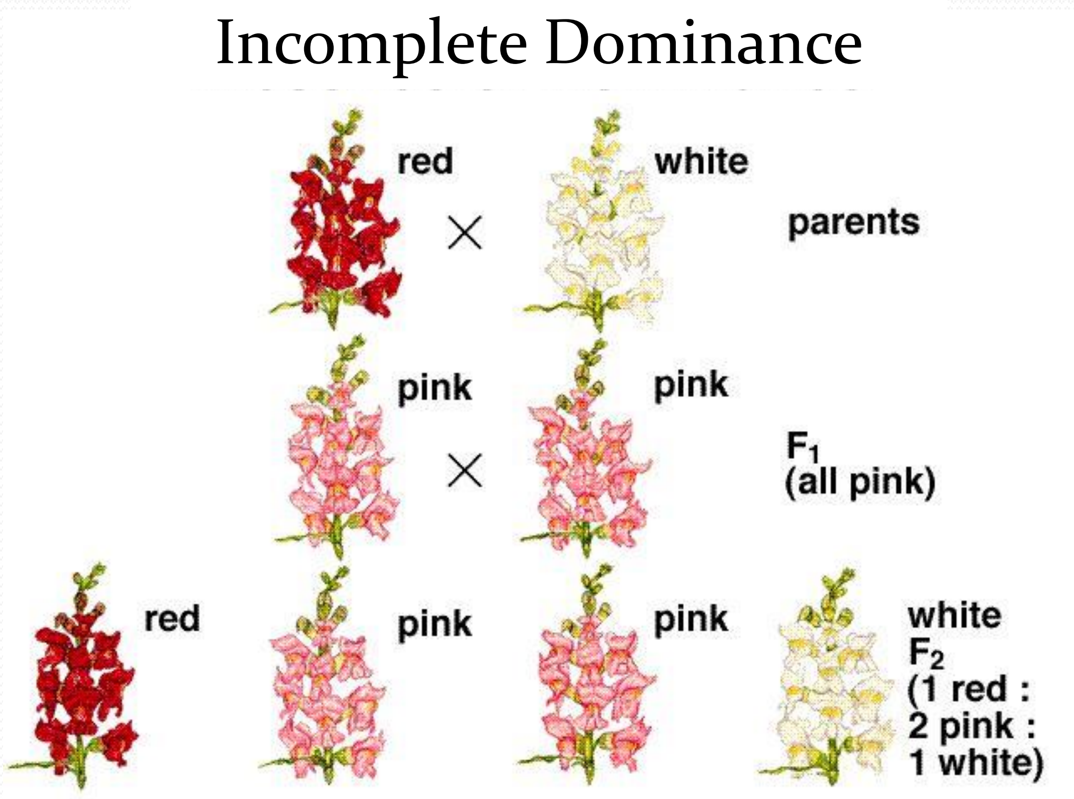
R *r*

Genes show incomplete dominance when the heterozygous phenotype is intermediate.

Incomplete dominance

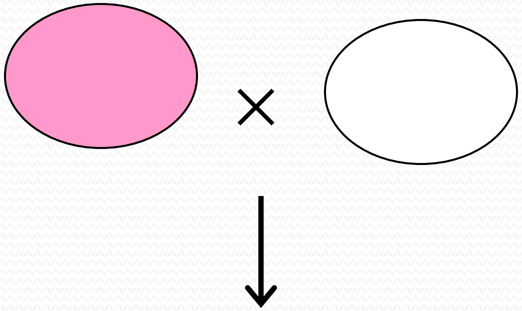
When F1 generation (all pink flowers) is self pollinated, the F2 generation is 1:2:1 red, pink, white

	R	*r*
R	R R	R *r*
r	R *r*	*r r*

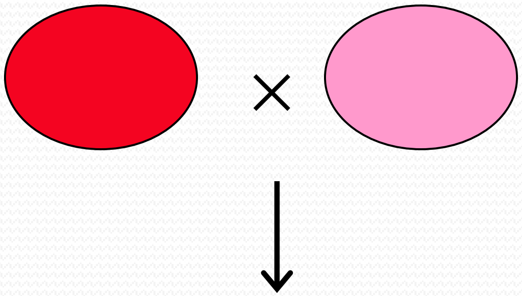


Incomplete dominance HOMEWORK Q 2

What happens if you cross a pink with a white?



A pink with a red?



Summary of Mendel Genetics

- Chromosomes carry hereditary info (genes)
- Chromosomes (and genes) occur in pairs
- New combinations of genes occur in sexual reproduction
- Monohybrid vs. Dihybrid crosses
- Mendel's Principles:
  - Dominance: one allele masks another
  - Segregation: genes become separated in gamete formation
  - Independent Assortment: Members of one gene pair segregate independently from other gene pairs during gamete formation

GENETİCS-BREEDING

- Animal breeding or plant breeding is genetics
  - just like what Mendel did.

What is classical breeding?

- Animals and plants are bred, artificially selected to increase their
  - Milk yield and quality
    - Milk fat, milk protein, somatic cell count
  - Meat production and quality
    - Weaning weight, yearling weight, marbling
  - Wheat production
- They can also be crossbred to increase their
  - Corn production
  - 40 days weight in poultry
  - Feed efficiency
  - The number of eggs.

What is classical breeding?

- Selection
- Crossbreeding

are the two main methods used.

- GWAS (Genome Wide Associated Selection) is used to help selection. Lactoferrin, IGF-1 etc, 50k SNP chips.
  - Shortens generation interval
  - Increases accuracy
  - But costs more for analysis.
  - Not everyone can do it.

What is classical breeding?

- USA: eat more, produce more (large land),
- Europe: eat less, produce more (smaller land)

What is classical breeding?

- Livestock is bred for economical reasons.
- Therefore, the purpose is to increase the ratio of economic, high performing genotypes in the next generation of the population.

What is classical breeding?

- Selection and crossbreeding are artificial tools.
- Normally, a cow has only milk sufficient enough for its progeny.
- Normally, a chicken reaches 2kg body weight when it is 5 months old
- These are all changed using normal, slow breeding processes.
- Molecular genetics simply speed these up.

Roadmap for Breeding

- Calculate the variation
- How many of the individuals are in the desired performance level, how much of their difference with the others is hereditary.
- What will be the selection method
- What will be the mating method
  - Superior males with superior females, or select only males and mate with all females
  - Keep purebreds or make crossbreds.
    - Crossbreeding may be used even when one of the parents is considered “inferior”

Roadmap for Breeding

- Keeping records/numbers and regulating, summarizing them
- Calculating genetic parameters
  - Genetic correlations, sire variance, heritability, accuracy
- Selection or selecting animals for crossbreeding
  - Crossbreeding is also a selection method, you select breeds or lines instead of individuals
- Mating the animals
- Evaluating the progeny crop.

Genotype or Environment

- Phenotype is made up of genetics and environmental factors.
  - P=G+E
- Individuals in a herd may be have different weights and may produce different milk yield even when they are the same breed, same age and same sex.
- Monozygotic ("identical") twins, those developing from one zygote that splitting into two embryos, may have phenotypic differences. Why?
  - Environmental effects

Environmental factors (Genotype is later)

I. Environmental factors with discrete variation
- Gender
- Year
- Season
- Location (city, herd, region born and raised)
- Age of dam
  - Factors that can be corrected statistically

II. Environmental factors with continuous variation
- Birth weight
- Milk suckled from the dam/mother
- Suckling time
- Rent of the house raised in (may affect IQ)
- Weaning weight
- Funding of the school graduated
- Dry period (in days/hours/seconds/miliseconds etc.)

Environmental factors

III. Error
- Light received by the plant
- Protein content of grazed feed on pasture
- How much being picked on by bullies in high school
- Others?

- Error (within group variation) is variation that cannot be simply eliminated or corrected statistically.
  - Error is not to be confused with “Mistake”

Genotype? Environment?

- In quantitative traits that are economically important, usually environmental factors > genotype
  - Heritability of different traits
- Improving the environment is more effective, and it is effective in the same generation
- Even when the environment is improved to the maximum, genotype limits performance.
  - Weight lifting by an XYY tall person
  - Turkish Black Cattle

Genotype? Environment? Turkey?

- Local livestock breeds of Turkey cannot raise above a certain level, no matter how much the environment is improved.
  - Local breeds should be crossed with higher performing breeds and then selected for Turkish conditions.

Genotype? Environment? Turkey?

- Why not import these animals and directly use them in Turkey?
  - Genotype*environment interaction
- Princess living in the woods?
  - First you have to make the environment ready for her.
- Son of a villager and the princess may live in the woods and show good performance, whatever it is.

Breeding for better Genotype

1. Determine and select individuals that show the best reaction to the present environmental conditions
2. Use the selected individuals as parents in the population,
3. Try to increase the ratio of such individuals in the population in the upcoming generations.

Genotype * Environment

- Genotype and environment should be improved at the same time
- However, some traits may be improved only using genetics.

Genotype * Environment

- Traits that may be improved using mainly genetics:
  - Qualitative traits such as presence of horns or color
  - Egg size in chickens
  - # of bones in spine/backbone (vertebral column)
  - Wool quality in sheep

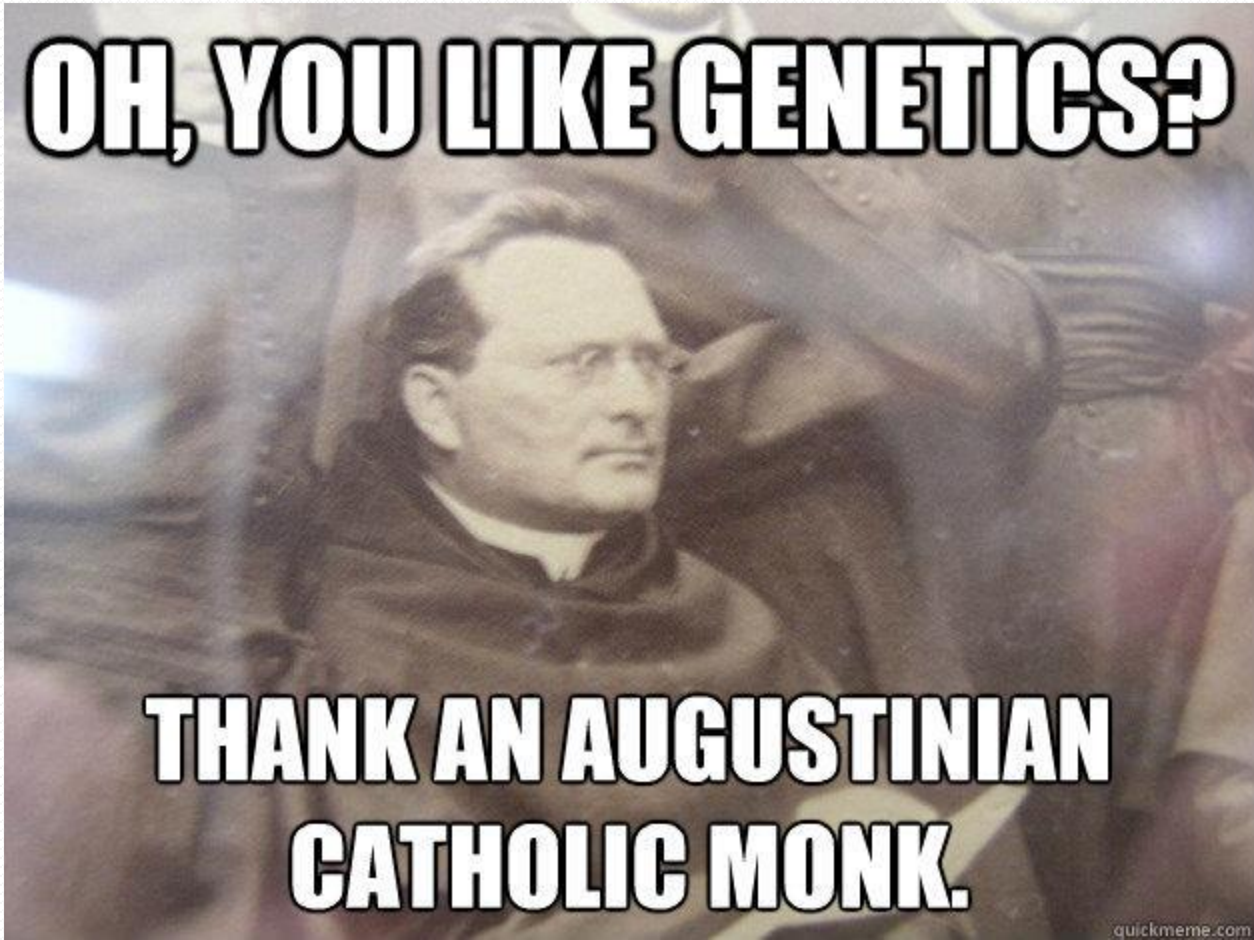
Breeding-Mendel Genetics

- Heredity of quantitative variance depend on multiple genes.
- Transmission and features of these genes are just like the transmission and features of the genes determining the qualitative variance.
- Quantitative Genetics is an extension of Mendel genetics and it uses the same principles.

Homework (Use Turnitin)

- Give an example to F1 and F2 generations, write down genotypes and phenotypes.


Gregor Mendel Gets His Genome Sequenced
Mind Blown


OH, YOU LIKE GENETICS?
THANK AN AUGUSTINIAN CATHOLIC MONK.
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Next Lesson

- **An Introduction to Population Genetics**