


Chapter 14

Mendel and the Gene Idea



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Gregor Mendel

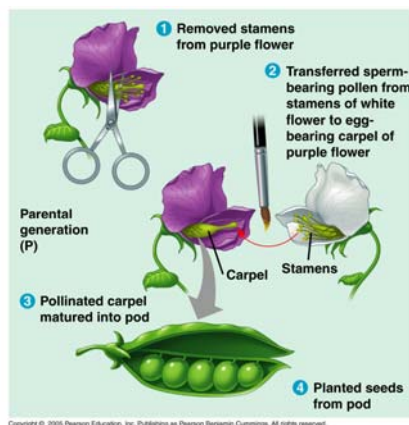
- Born and raised on a farm in the Czech Republic (Austria)
- 1840's entered a monastic lifestyle and studied science
 - During this time many scientists were monks
- In 1857, Mendel began to perform experiments with garden peas to gain an understanding of inheritance



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Why peas???

- Breeding could be strictly controlled
 - No random breeding
 - And no self-pollination, the male portions of the flowers were removed prior to maturity
 - Mendel performed cross-pollination
- Peas were removed from pods and Mendel could track the offspring from individual parents
- Lots of varieties of peas




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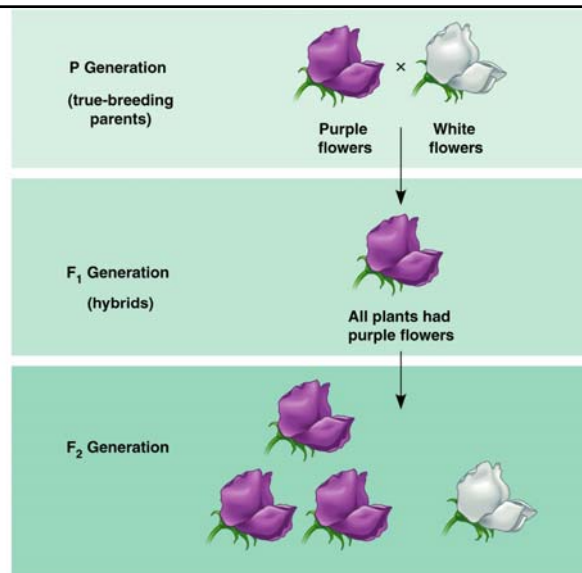
Mendel kept records of:

- Heritable features; **characters**
 - Flower color is a character
- Variants for a character; **traits**
 - Purple flowers *or* white flowers are traits
- Mendel also made sure his projects began with pea varieties that were **true-breeding**
 - self-pollination only produces offspring that are the same variety

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- In a typical breeding experiment, Mendel would cross-pollinate two contrasting, true-breeding pea varieties.
 - Hybridization
 - Crossing of two true-breeding parents
 - P generation
 - Hybrid offspring represent the first filial generation
 - F₁ generation
 - Mendel carried out most of his experiments at least to the F₂ generation
 - Allowing the F₁ generation to self pollinate

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- It was this second filial generation where Mendel noticed the fundamental principles of heredity

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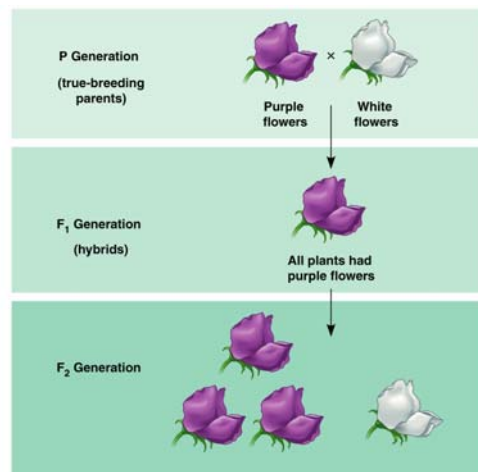
From these simple experiments:

- Law of segregation
- Law of independent assortment
 - Now keep in mind, all of this occurred well before the technology to understand how meiosis worked had been developed!!!
 - Some say Mendel got lucky!!!
 - Some say Mendel cheated and fudged the data??
 - Who knows??!!!

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Mendel's model


- Let's look at one of Mendel's experiments
- What happened to the white flowers in F_1 ?
- If it were totally lost, then how could white flowers be present in the F_2 ?
- Mendel collected data on the number of offspring
 - F_2
 - 705 purple
 - 224 white
 - ~3:1
- Mendel described traits as
 - dominant
 - recessive



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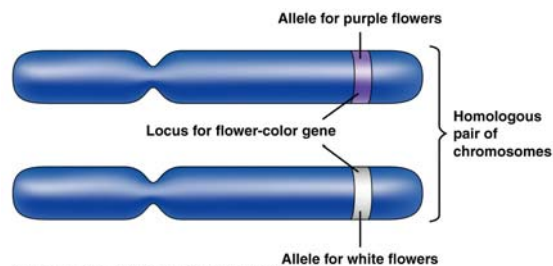
Mendel developed a hypothesis to explain the 3:1 inheritance ratios

- Four concepts to Mendel's model
 1. Alternative versions of genes account for variations in inherited characters
 - These are called alleles; correspond to loci on chromosomes
 2. For each character, an organism inherits two alleles, one from each parent
 3. If the two alleles at a locus differ, the dominant allele determines the physical appearance; the recessive allele has no noticeable effect on appearance
 4. These alleles for an inherited character separate during gamete formation – **Law of Segregation**

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Keep in mind..

- Mendel had no idea about
 - homologous chromosomes
 - where alleles were actually located at loci



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P Generation
 Appearance: Purple flowers × White flowers
 Genetic makeup: PP × pp
 Gametes: P × p

F₁ Generation
 Appearance: Purple flowers
 Genetic makeup: Pp
 Gametes: $\frac{1}{2} P$ × $\frac{1}{2} p$

F₂ Generation

	P	p
P	PP	Pp
p	Pp	pp

3 Purple : 1 White

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- Punnett Squares are useful to illustrate likely combinations of alleles
- Also give insight to ratios and probabilities of offspring with a given
 - **Genotype** – the genetic makeup
 - **Phenotype** – physical appearance

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- Phenotype
 - Ratio 3:1
- Genotype
 - **Homozygous**
 - Identical pair of alleles for a gene
 - Homozygous dominant or homozygous recessive
 - **Heterozygous**
 - Two unlike alleles for a gene
 - Ratio
 - 1:2:1

Phenotype

Purple
Purple
Purple
White

Ratio 3:1

Genotype

PP (homozygous)
Pp (heterozygous)
Pp (heterozygous)
pp (homozygous)


Ratio 1:2:1

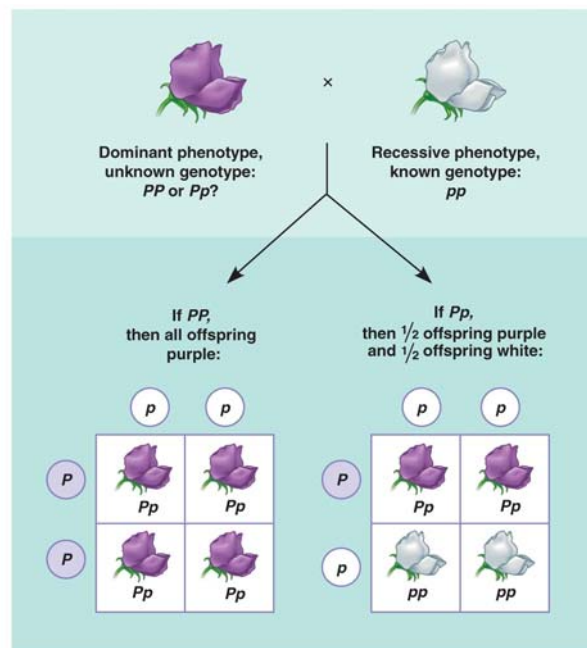
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This same principle can also be used to determine if an organism which exhibits a dominant trait (phenotype) is homozygous or heterozygous (genotype) for a given trait

- **Testcross** is used to make this determination
- example
 - We have a pea plant that has purple flowers
 - phenotype = purple
 - Genotype = PP *or* Pp
 - A testcross will be performed using a white flowered mate (white phenotype; must be homozygous recessive genotype, pp)

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- In these experiments where Mendel tracked a single character all of the F₁ offspring produced were hybrids of true-breeding parents for a single character
 - Monohybrids
- In his experiments Mendel tracked seven different characters

Table 14.1 The Results of Mendel's F₁ Crosses for Seven Characters in Pea Plants

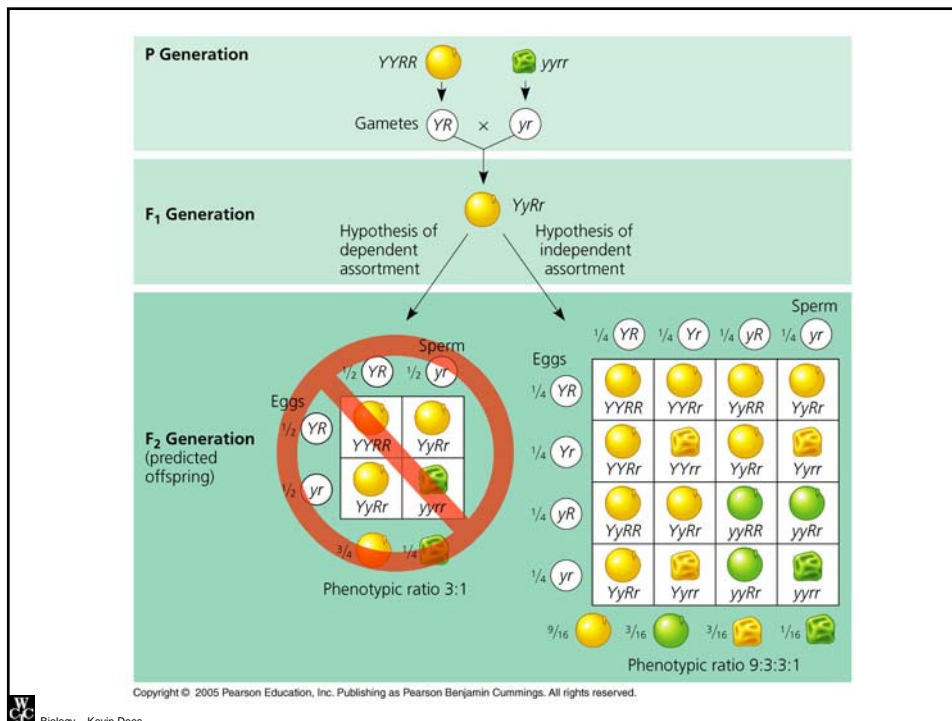
Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6022:2001	3.01:1
Seed shape	Round 	×	Wrinkled 	5474:1850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

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Law of Independent Assortment


- To derive his second law, Mendel had to track two characters at one time
- Used two true-breeding pea varieties
 - yellow round seeds (YYRR) x green wrinkled seeds (yyrr)
- The F₁ offspring are known as **dihybrids**
 - YyRr genotype; yellow round seeds phenotypes
- When the dihybrid cross is performed, the phenotypical ratio of 3:1 is not seen
- This means that the alleles are segregated or separated at some point
- This illustrates the **law of independent assortment**
 - Each pair of alleles segregates independently of other pairs of alleles during gamete formation

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Mendel was lucky!!

- He chose – characters with alleles located on different chromosomes – not homologous chromosomes
 - Genes located near each other on the same chromosome tend to be inherited together and have more complex patterns of inheritance
- He chose – characters which exhibited **complete dominance** of one allele over another
- He chose characters controlled by only two alleles; no **multiple alleles**

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Spectrum of dominance

- Complete dominance
 - The dominant allele determines phenotype over recessive allele
 - Ex: purple vs. white pea flowers

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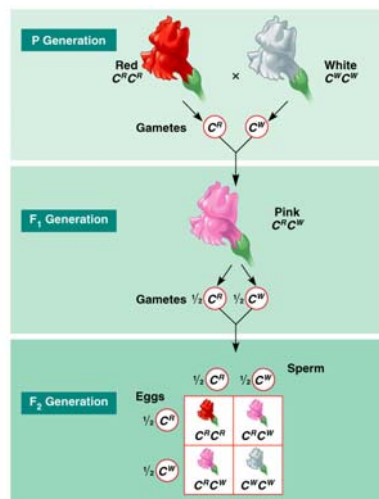
Spectrum of dominance

- Codominance
 - Both alleles affect the phenotype in separate distinguishable ways
 - EX: MN blood groups
 - Not blood type!!!!!!
 - Codominant alleles for the synthesis of two specific glycoproteins
 - Individuals that are MM have RBC with M glycoproteins
 - Individuals that are NN have RBC with N glycoproteins
 - Individuals that are MN have RBC with both M and N glycoproteins

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Spectrum of dominance

- Incomplete dominance
 - Alleles for some characters fall in the middle of the spectrum of dominance; phenotype represents a 'blending' of the two parental varieties
 - EX: snapdragon flower color
 - F² ratio 1:2:1







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Multiple alleles

- More than two alleles control the phenotype
- EX: Human blood type
 - There are four possible phenotypes
 - A, B, AB, O
 - There are three alleles for the enzyme (I) that attaches the A or B carbohydrate to the RBC
 - I^A , I^B or i (neither)
 - Matching blood type is essential!
 - If a person with type A blood receives blood from type B or AB their immune system attacks the cells with the B and can cause clumps/clots
 - AB – universal recipient
 - O – universal donor

Table 14.2 Determination of ABO Blood Group by Multiple Alleles

Genotype	Phenotype (Blood Group)	Red Blood Cells
$I^A I^A$ or $I^A i$	A	
$I^B I^B$ or $I^B i$	B	
$I^A I^B$	AB	
ii	O	

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So far we have treated inheritance as though each gene effects one character...

- This is not the norm.
- Most genes have multiple phenotypic effects
 - Pleiotropy
 - Pleiotropic alleles are responsible for multiple symptoms associated with some hereditary diseases
 - Cystic fibrosis
 - Sickle-cell disease (sickle-celled anemia)

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Cystic fibrosis

- The most common lethal genetic disorder in the USA
- Most common in European lineages
- It is a recessively inherited disorder
 - Must be homozygous recessive)
- It is estimated that 1 in 25 Americans of European descent are **carriers** (heterozygotes) and have normal phenotypes
 - Normal allele codes for membrane protein that functions in chloride transport across mucous membranes
 - Homozygous recessive phenotypes exhibit multiple (pleiotropic) effects
 - Poor absorption of nutrients
 - Chronic bronchitis
 - Untreated is usually lethal before age 5 or 6
 - Aggressive treatment with antibiotics can allow for survival into early adult hood

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Sickle-cell disease

- Most common in African lineages
 - Estimated that it affects 1 in 400 African Americans
- Recessively inherited disorder
- Homozygotes have malformed RBC due to slight change in hemoglobin protein
 - Poor oxygen transport
 - Irregular clotting/clumping of sickle shaped cells
 - Pleiotropic effects
 - Also exhibits incomplete dominance; heterozygotes (about 1 in 10 African Americans) may suffer some reduced symptoms
 - Why so common???
 - Possible link to malaria
 - Malarial parasite not able to infect sickle-shaped cells

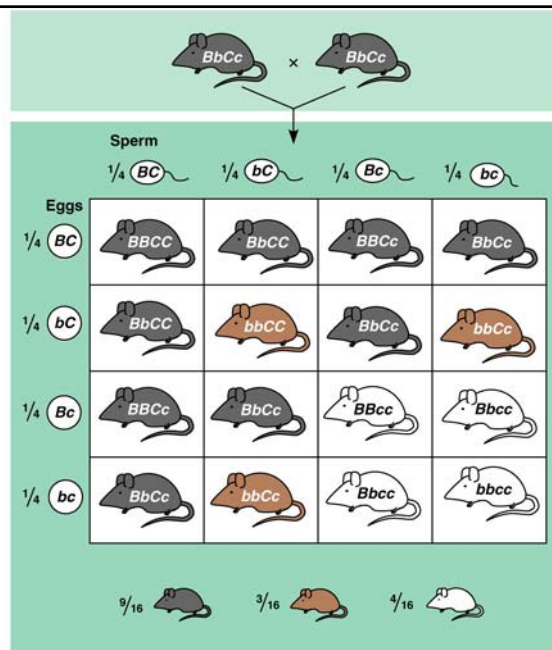
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Epistasis

- A gene at one locus alters the phenotypic expression of a gene at another locus
 - “Stop gene”
 - Example
 - Hair color in many mammals
 - Black (B) is dominant to brown (b)
 - » So to have brown fur → bb
 - A second gene determines if pigment will be deposited in the hair; dominant (C) is to have pigment deposition
 - If the mammal is homozygous recessive at the locus for the second gene (cc) then the coat is white regardless of what the first gene’s alleles say

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- Note genotypes still 9:3:3:1
- But phenotypes are now 9:3:4!!!

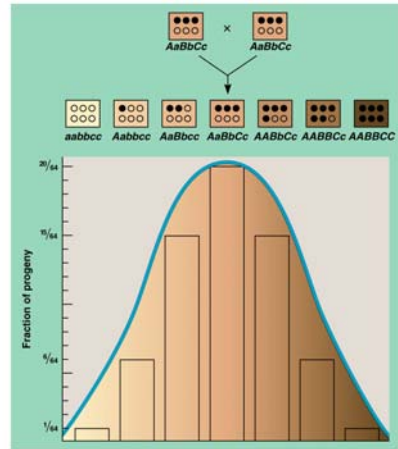


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Polygenic inheritance

- Effect of two or more genes on a single phenotype
 - Opposite of pleiotropy
- Example
 - Skin pigmentation in humans
 - At least three separate genes
 - From gradations in phenotypic expression



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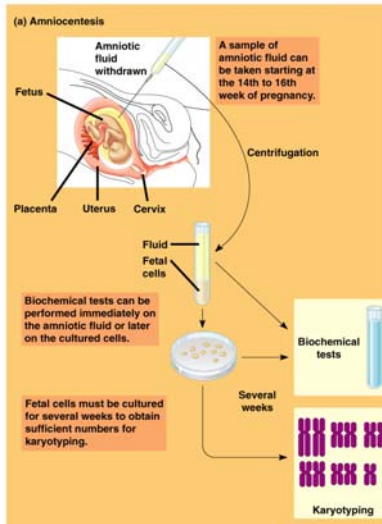
Not all human genetic disorders are recessive

- Achondroplasia – form of dwarfism
 - 99.99% of population is homozygous recessive
 - About 1: 250,000 exhibit phenotype
 - (AA or Aa)
- Huntington's disease
 - Degenerative disease of nervous system
 - Usually fatal by age 40
 - Approx 1 in 10,000 in USA

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These and other genetic disorders have led to many advances in genetic testing and counseling

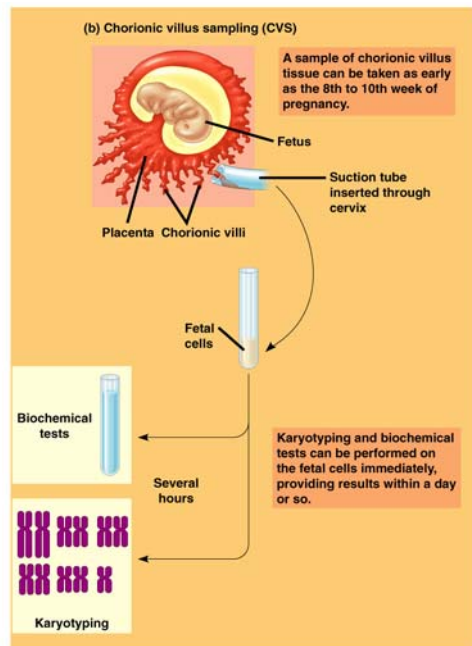
- Amniocentesis - testing of amniotic fluid
 - Can test fetal cells or presence of chemicals in fluid



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- Chorionic villus sampling (CVS)
 - Tests a sample of tissue from placenta
 - Faster than amniocentesis



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It is important to ponder

- Are we who we are because of our genes?
 - nature
- Are we who we are because of our environment?
 - nurture
- Nature vs. Nurture
 - Even identical twins differ slightly (or markedly)
 - Generally many factors (both genetic and environmental) affect phenotype – **multifactorial**