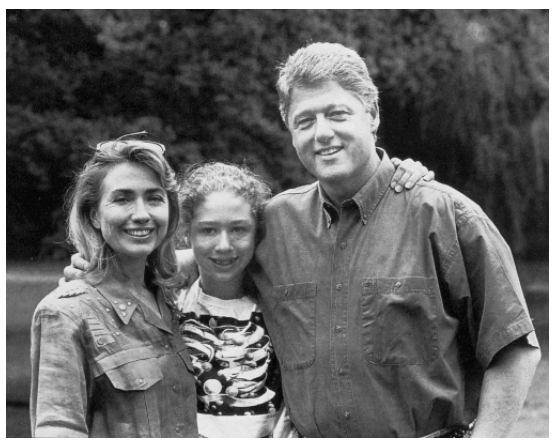
Genetics

- Blending Theory
- Mendel's Experiments
- Mendel Expanded
- Chromosomal Theory of Genetics
- Human Genetics

Blending Theory – offspring have a blend of traits of their parents



- Problems with blending theory

Problems with Blending Theory

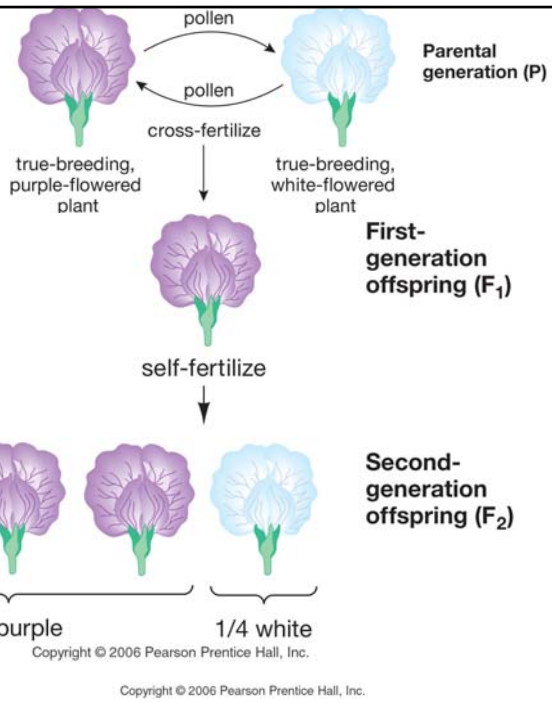
Mohan Begum

Orange Orange X Orange

Orange and White

1. Often offspring's trait reflect just one of the parents.
2. Often traits seem to skip generations

Mendel



Mendel's Explanation

1. Genes controlled Traits
   - Example :
     - Flower color gene controls flower color

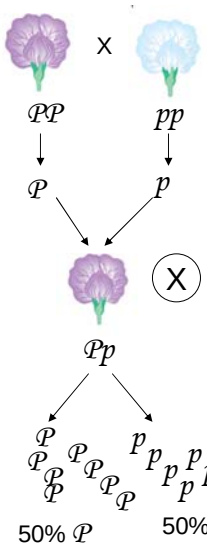
2. Genes exist in different forms called Alleles
   - Example
     - White flower color allele
     - Purple flower color allele

3. Individuals possess two alleles for every gene.
   - Example
     - Two white alleles – homozygous white – white flowers
     - Two purple allels – homozygous purple – purple flowers
     - One white and one purple – heterozygous – purple flowers
     - (One allele is dominant to recessive allele)

4. Law of Segregation – The two alleles separate during gamete formation and during fertilization the paired condition is restored when the egg and sperm fuse.

Follow the Alleles

Purple allele – P
White allele - p



P1 generation
(True breeding stocks)

Gametes

F1 generation
(Heterozygous)

Gametes

F1 generation
(Heterozygous)

Pp

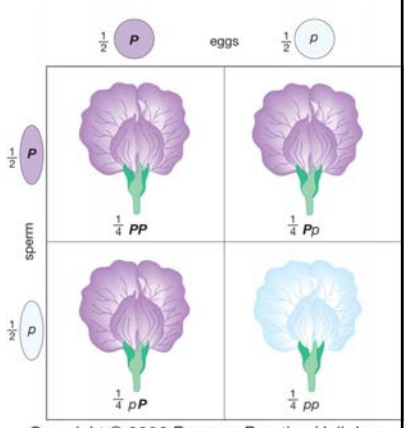
Gametes

P P P P p p p p

50% p 50% P

Random Fertilization

¾ Purple ¼ White



Phenotype – Observable trait of an organism
White Flower vs Purple Flower

Genotype – List of alleles
White Flower – Homozygous pp

Purple Flower – Homozygous PP
Heterozygous Pp

Mohan Revisted

Mohan Begum

Orange Orange X Orange

Orange and White

What are Mohan and Begum phenotypes? What are their genotypes?

What are their childrens genotypes

Why does the white trait skip a generation?

What are the genotypes of the offspring?

How would you determine the genotype of one of Mohan's orange grandchildren?

Testcross

- Goal – determine the genotype of an unknown individual (e.g. Mohans orange grandson)
- Experiment - Mate an individual of unknown genotype with a homozygous recessive and examine the offspring.
- Consider results

Mendel Expanded

- Allelic Interactions
  - Dominant/Recessive
  - Incomplete Dominance
  - Codominance
  - Multiple Alleles
- Polygenic Traits
- Pleiotropy

Human Example Dominant/Recessive

Free Earlobes
(Dominant)

Attached Earlobes
(Recessive)

Bent Pinkies
(Dominant)

•What is Dr. Wadsworths phenotype?
•What is Dr. Wadsworth's genotype?

•Suppose that you know Dr. Wadsworth's mother had attached earlobes, what could you conclude about his genotype?

Huntington's Disease

Dominant/Recessive Trait

Symptoms:

•First exhibited in mature adults

•Physical, early- *jerky, random, movements*
- loss of motor control

•Loss of Cognitive skills - *planning; abstract thinking, initiating appropriate actions, and inhibiting inappropriate actions*

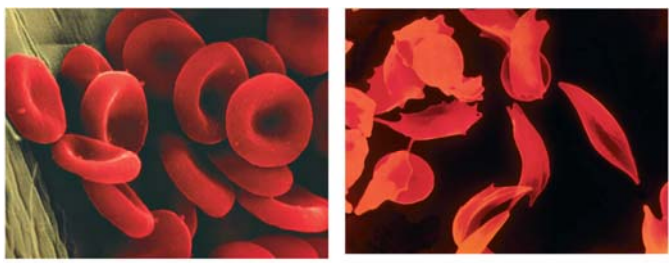
•Retain their memories of who and what they were before.

Dominant Genetic Trait (Hh)

Ann Bishop

If two heterozygotes have children what is the likelihood that their first child will develop Huntingtons?

Sickle Cell Anemia
Dominant/Recessive Trait

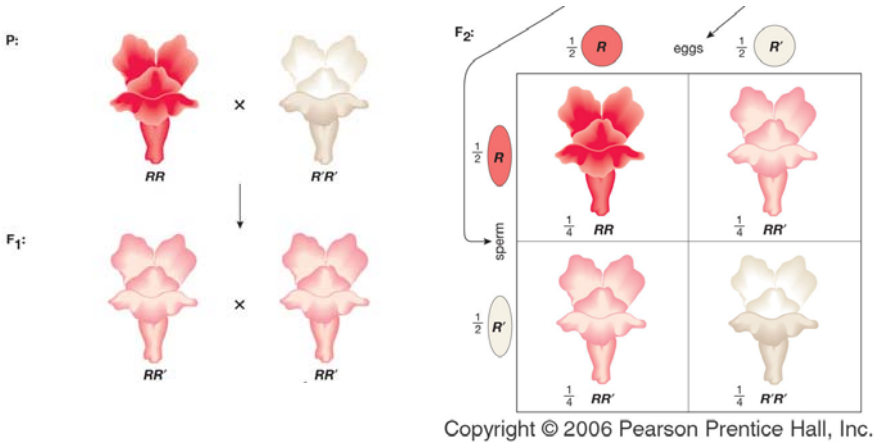


(a) (b)

S – healthy allele
s – recessive disease causing allele

SS – healthy
ss – sickle cell anemia
Ss – healthy carrier (resistance to malaria

Incomplete Dominance
Snapdragon flower color



Familial **Hypercholesterolemia**
Incomplete Dominance

- Two alleles –
  - FH, healthy allele for normal cholesterol controlled by environmental factors
  - fh, Disease allele causing supper high cholesterol
  - Homozygous FH,FH – normal levels cholestrol
  - Heterozygous FH,fh 300-600mg/ml LDL– heart disease young adult
  - Homozgyous fh,fh – 1200 mg/ml LDL – heart disease as young child.
  - Heterozygotes treated with statins
  - Homozygotes treated – liver transplant.

Codominance –both alleles expressed

- Blood type
  - I^A allele for A type blood
  - I^B allele for B type blood

  $I^A I^A$ – A type blood
  $I^B I^B$ – B type blood
  $I^A I^B$ – AB type blood

Multiple Allels

- Blood type
  - I^A allele for A type blood
  - I^B allele for B type blood
  - I^O allele for O type blood – always recessive

  Possible Genotypes
  $I^A I^A$ – A type blood, $I^A I^O$ – A type blood,
  $I^B I^B$ – B type blood, $I^B I^O$ – B type blood,
  $I^A I^B$ – AB type blood, $I^O I^O$ – O type blood,

Bob
O type

X

Sally
A type

Tom
O type

What is the probability that the next child will have O type blood?

Polygenic Traits

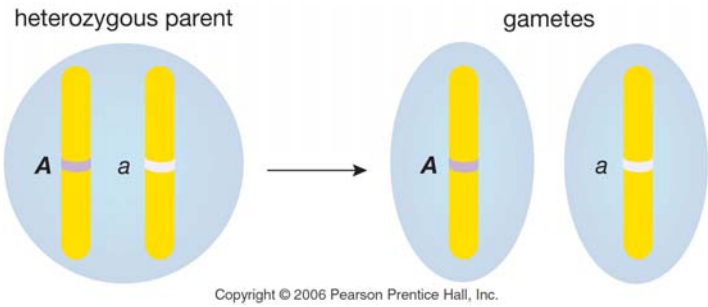
- One trait controlled by many genes
- Example skin pigmentation in humans – three genes each with two alleles
  - AABBCC – Highest level of melanin
  - AABBCc (or AaBBCC etc) – High levels
  - AaBbCC (or aaBBCC etc) - Still high level of pigment
  - AaBbCc (or AABbcc etc) - Intermediate levels
  - AAbbcc - lower levels
  - Aabbcc - low levels
  - Aabbcc - lowest levels
- The more pigmentation alleles the darker the skin

Pleiotropy
One gene many Traits

- Cystic Fibrosis
  - Recessive Trait
- Homozygote Symptoms
  - Mucus buildup on lungs
  - Salty sweat
  - Digestive Problems
  - Kidney Failure
  - Clubbing
- Cf gene encodes chloride membrane pump

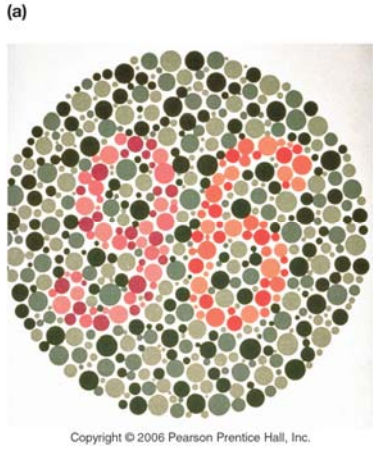
Chromosomal Theory of Inheritance

- Every gene is at a specific location on a specific chromosome
  - Example cystic fibrosis gene is on chromosome 7
- Meiosis accounts for segregation of alleles



Sex Linked Genes
Genes on X chromosome

- Example Red-Green Color Blindness
- Two Alleles
  - CB Normal color vision
  - cb Colorblind (recessive)
- Females have two X chromosomes and therefore 2 alleles
- Males have one X chromosome and only 1 allele



Example

Jayne
Normal Vision

X

James
Normal Vision

↓

Greg
Colorblind

What is Jayne's, James' and Greg's genotype?

Aneuploidy

Missing or having extra copies of a chromosome.

- Down Syndrome
  - Trisomy 21

(a) (b)

Symptoms – Distinctive eyelid, varying mental retardation, heart malformations

Sex Chromosome Aneuploids

- Turners Syndrome (XO)

- Trisomy X (XXX)

- Klinefelters XXY
- Jacob Syndrome XYY