Mendelian Genetics

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Gregor Mendel

- 1865
- Particulate (discrete) bodies = genes

St Thomas Abbey, Brno

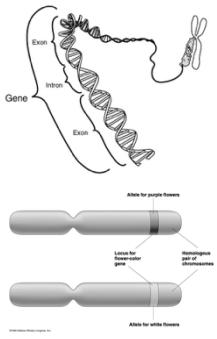
Hugo de Vries

Carl Correns

Erich von Tschermak-Seysenegg

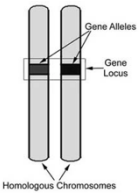
Mendelian Inheritance

- gene:basic unit of inheritance (segment of the DNA)
- Allele: alternative expression of a gene (e.g., A, B, O)



More Terms

- Locus:gene's specific position on chromosome
- Genotype: genetic constitution or allelic pair
- Phenotype: observable or testable characteristics



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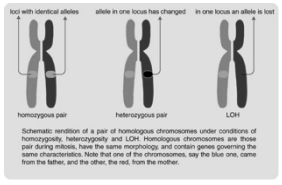
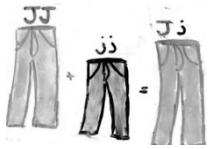
More Terms

- heterozygous – allelic pair differ (e.g., AO, BO, AB)
- homozygous – allelic pair identical (e.g., AA, BB, OO)





Dominant vs. Recessive

- dominance – allele expressed in homo- or heterozygous state (e.g., AO, AA, BO, BB)
- recessiveness – allele expressed only in homozygous state (e.g., OO)
- co-dominance – alleles equally expressed in phenotype (e.g., AB)

Phenotype (blood type)	Genotype	Antibodies in serum
A	A A or A O	Anti-B
B	B B or B O	Anti-A
AB	A B	None
O	OO	Anti-B and Anti-A

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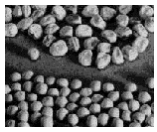
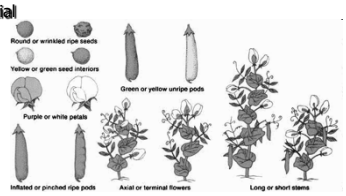
Mendel's Pea Plants

- color of endosperm (seed): yellow or green
- stature: short or tall
- shape of ripe seed: smooth or wrinkled
- pod color: green or yellow
- position of blossom: axial or terminal
- In all, 7 traits were investigated by Mendel





Mendel's Pea Plants Experiments

1. Observed each plant separately
2. Kept generations separate
3. Quantified results

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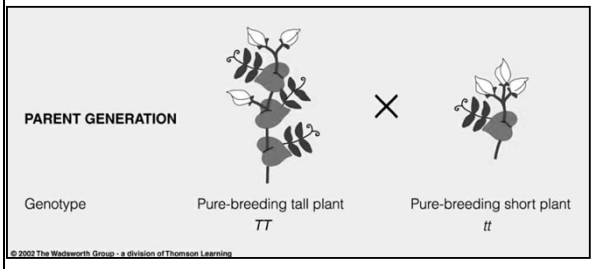
Typical Experiment: Tall x Short



First Generation

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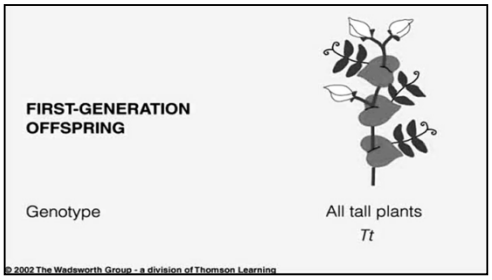

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Second Generation

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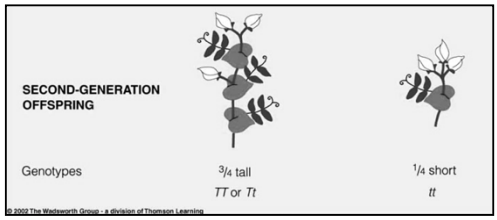


Important Points

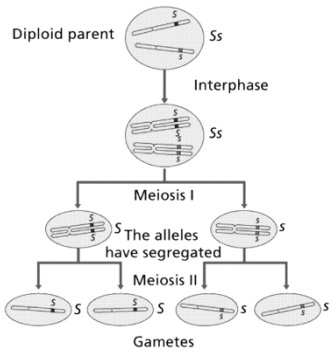
- Particulate inheritance
- Pair of alleles is responsible for proportions
- Pair of alleles in pure breeding parents is identical
- Pair of alleles in hybrids was different
- Some alleles dominant, others recessive

Mendel's Laws

Law of Segregation

- pair of genes (alleles) of a parent separate or segregate in the formation of gametes such that an offspring receives only one member of each allelic pair from each parent

Segregation



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Source of Variation

- segregation and random assortment of alleles during gamete formation is one way in which variability within a population (species) is maintained and transmitted across generations

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Dominant condition = Tall
Recessive condition = short
T = dominant allele for Tall plants
t = recessive allele for short plants

P_1	TT		tt	
F_1	Tt		Tt	
F_2	TT	Tt	Tt	tt

P matings to produce F_1 generation

- All Tall

Parent's alleles	t	t
T	Tt	Tt
T	Tt	Tt

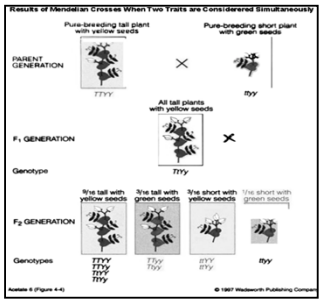
TT or Tt = tall
tt = short

Matings among F_1 offspring to produce F_2 generation

- 1 TT: 2 Tt: 1 tt
- or 3 tall, 1 short

T/ t	T	t
T	TT	Tt
t	Tt	tt

Tall Yellow x short green



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Matings to produce F_1 Generation

Parent's alleles	ty	ty
TY	TtYy	TtYy
TY	TtYy	TtYy

T = tall
t = short
Y = yellow
y = green

Phenotypes:
9 tall yellow
3 tall green
3 short yellow
1 short green

TtYy
(hybrid parents)

TY Ty tY ty
(gametes)

F_2 generation - TtYy x TtYy

Parent/ parent	TY	Ty	tY	ty
TY	TTYY	TTYy	TtYY	TtYy
Ty	TTYy	Ttyy	TtYy	Ttyy
tY	TtYY	TtYy	ttYY	ttYy
ty	TtYy	Ttyy	ttYy	ttyy

Mendel's 2nd Law

Law of Independent Assortment
- different pairs of alleles are passed to offspring independently so that new combinations of genes are possible

Mendel's laws are the foundation of modern genetics.

Review: Mendel's laws

0. *Genes* are particulate and come in different forms known as *alleles*.
- Organisms (peas or humans!) have two copies of each gene but transmit only one to each offspring. Which one is transmitted is chosen at random. i.e. if you are *heterozygous* for two different alleles, the alleles will *segregate* from each other in your offspring.
- Where alleles of more than one gene are segregating, segregation at each gene occurs independently of the others.

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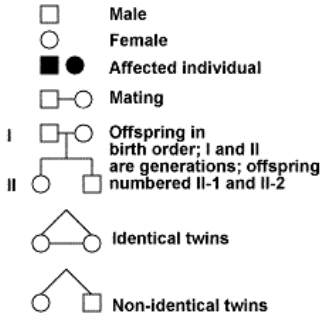
Human Inheritance Patterns

- whether genes are located on autosomal chromosomes or on sex chromosomes
- dominant, recessive allele
- co-dominant alleles
- pedigree analysis

Modes of Inheritance

1. autosomal dominance (e.g., brachydactyly)
   - allele is expressed in the phenotype if either in the homozygous or heterozygous state
2. autosomal recessive (e.g., albinism)
   - simple recessive traits appear only in the homozygous condition
3. sex-linked recessive
4. sex-linked dominant

Pedigree Analysis



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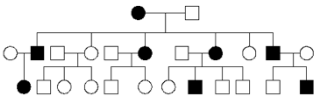
Autosomal dominant inheritance



- All affected individuals should have at least one affected parent
- Two unaffected parents can only have unaffected offspring
- Both sexes should be equally affected
- Roughly 50% of the offspring of an affected individual should also be affected
- Affected phenotype appears every generation
- Huntington's disease, Achondroplastic dysplasia, Neurofibromatosis, brachydactyly

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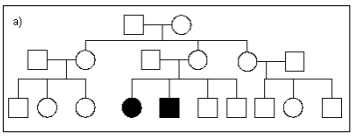
Autosomal Recessive Inheritance



- Usually there is no previous family history
- The most likely place to find a second affected child is a sibling of the first
- affected progeny are both male and female

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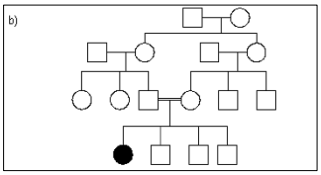
Autosomal recessive



- unaffected parents can have affected offspring
- Inbreeding increases the chance of observing an autosomal recessive condition
- E.g. Cystic fibrosis, sickle cell disease, Tay Sachs disease, albinism.

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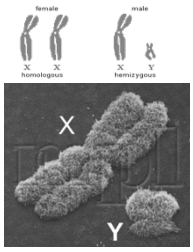
Sex-linked Inheritance

- Gene located on X or Y chromosome
- Males are hemizygous
- Females homozygous
- Genes on Y chromosome: holandric



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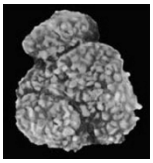
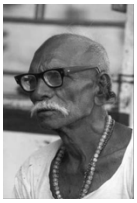
Y Chromosome



- TDF (for determining maleness)
- H-Y (sperm production)
- Hairy pinnae (hair on rims of ears)



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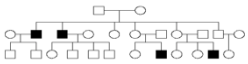
X-Linked Inheritance

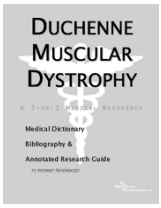
- X-linked recessive (e.g., hemophilia, G-6PD)
- X-linked dominant (e.g., Xg blood group system)

Czar Nicholas II of Russia and his family, photographed c. 1916, showing his wife Alexandra (who was a carrier of hemophilia), his four daughters, and (in the foreground) his son Alexis, perhaps the most famous European royal with hemophilia.

X-Linked Recessive Inheritance

- Clusters of affected males (each brother will have a 50% chance of being affected) connected through unaffected carrier females. There will be *no cases of direct male to male transmission WHY?*
  E.g.,
  Duchenne Muscular Dystrophy, Hemophilia)





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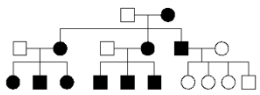
Sex linked dominant genes

- Sex linked dominant conditions are extremely rare, examples include incontinentia pigmenti (which is lethal in males) and congenital generalized hypertrichosis (wolf man syndrome).

Exceptions to clear cut Mendelian inheritance

- Lethal alleles
- Incomplete dominance
- Codominance
- Silent alleles
- Epistasis
- Pleiotropy
- Genetic heterogeneity
- Variable expressivity
- Incomplete penetrance
- Anticipation
- germline mosaicism
- Phenocopies
- Incomplete ascertainment



Mitochondrial inheritance

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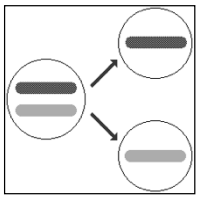
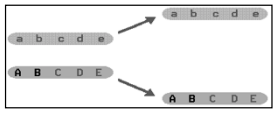
Causes of Variation

1. random assortment of chromosomes during gamete formation



Homologous chromosomes separating in the production of sex cells



Genetic linkage continues as homologous chromosomes separate in the formation of sex cells

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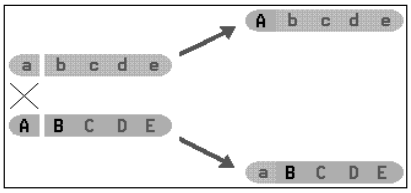
2. Crossing over - exchange of material on chromatids during meiotic prophase prior to the first division; results in a recombination



Crossing-over unlinks genes as homologous chromosomes separate in the formation of sex cells

3. Allelic differences
4. Mutations