GENETICS and Heredity

Mendel and the Garden Pea

- **Heredity:** the tendency for traits to be passed from parent to offspring

- heritable features: characters

- **traits** are alternative forms of a character
  - Genes
  - Alleles
  - loci

Gregor Mendel solved the puzzle of heredity

- performed experiments with garden peas

- Why study the garden pea?
  - many varieties/ easily distinguishable traits that can be quantified

  - small, easy to grow, and produce large numbers of offspring quickly

  - their reproductive organs easily manipulated so that pollination can be controlled

  - self-fertilize

Mendel had a specific experimental design

- 1st: establish **true-breeding** varieties
- Plants self-fertilize for several generations, (each variety had only 1 type of trait)
- pure lines: **P generation**
- Mating P generation è F_1
- F_1 generation self-fertilize
- **F_2 generation**

What Mendel Observed

- for each pair of contrasting varieties that he crossed, one of the traits disappeared in the F1 gen but reappeared in the F2 gen
- trait expressed in F1 gen: **dominant**
- trait not expressed in F1 gen: **recessive trait**
- Mendel counted # of each type of plant in the F2 generation
- 3/4of F2 individuals expressed the dominant trait while 1/4 expressed the recessive trait
- the dominant:recessive ratio among the F2 plants was always close to 3:1
- recessive trait hidden? in F_1 gen & not expressed
- He allowed the F_2 to self-fertilize and form the F_3 generation
- he found that one-fourth of the plants from the F_2 that were recessive were true-breeding in the F_3
- he found that of the three-fourths of the plants from the F_2
- only one-third were true breeding in the F_3
- the remaining half showed both traits
- He determined that the ratio of 3:1 ratio that he observed in the F_2 generation was in fact a disguised 1:2:1 ratio
    - 1: true breeding dominant
    - 2: not true breeding
    - 1: true breeding recessive
- The F_2 gen is a disguised 1:2:1 ratio

Mendel's 5 Hypothesis Theory

Hypothesis 1	*parents do not transmit traits directly to offspring* parents transmit information about the trait in the form of what Mendel called factors Now called **genes**
Hypothesis 2	*each parent contains 2 copies of factor governing each trait* the 2 copies of the factor may or may not be same **Homozygous:** two of the same copies **Heterozygous:** two different copies:
Hypothesis 3	*alternative forms of a factor lead to alt. traits* **Alleles:** alternative forms of a factor appearance is determined by the alleles a plant receives from its parents (**genotype)** expression of the alleles =appearance (**phenotype)**
Hypothesis 4:	2 *alleles do not affect each other*
Hypothesis 5	—*presence of allele does not ensure expression of trait* —in heterozygotes, only dominant allele is expressed

By convention, genetic traits are assigned a letter symbol referring to their more common form

—dominant traits: capitalized

—recessive trait: lower-case

—Ie: flower color in peas is represented as follows

—*P* signifies purple

—*p* signifies white

Punnet Square

—**Testcross: determine the genotype of unknown individuals in the F_2 gen**

- unknown individual is crossed with a homozygous recessive individual
- if the unknown is homozygous, then all of the offspring will express dominant traits
- if the unknown is heterozygous, then one-half of the offspring will express recessive traits
- Mendel used the testcross to detect heterozygotes

Mendel's 1st Law: Segregation

- *the two alleles of a trait separate from each other during the formation of gametes, so that half of the gametes will carry one copy and half will carry the other copy*
- Mendel also investigated the inheritance pattern for more than one factor
- when crossing individuals who are true-breeding for 2 different characters, the F1 individual that results is a *dihybrid*

  BBFf X BbFF

- after the dihybrid individuals self-fertilize, there are 16 possible genotypes of offspring

—*Conclusion: the inheritance of one trait does not influence the inheritance of the other trait*

Mendel's 2nd Law: <u>INDEPENDENT ASSORTMENT</u>

- *genes located on different chromosomes are inherited independently of one another*

The journey from DNA to phenotype

<u>Some Traits Don't Show Mendelian Inheritance</u>

Often the expression of phenotype is not straightforward

- **Continuous variation**
  - characters can show a range of small differences when multiple genes act jointly to influence a character
  - this type of inheritance is called **polygenic**
  - Height is a continuously varying character

- **Pleiotropic effects**
  - an allele that has more than one effect on a phenotype is considered **pleiotropic**
  - these effects are characteristic of many inherited disorders, such as cystic fibrosis and sickle-cell anemia

- **Incomplete dominance**
  - not all alternative alleles are dominant or recessive in heterozygotes
  - some alleles exhibit **incomplete dominance:** produce a heterozygous phenotype (intermediate between 2 parents)

- **Environmental effects**
  - Expression of some alleles depends on environment
    - ie: some alleles are heat-sensitive
    - arctic foxes only produce fur pigment when temperatures are warm

- **Codominance:**
  - often, in heterozygotes, there is not a dominant allele but, instead, both alleles are expressed
  - these alleles are said to be **codominant**

Chromosomal theory of inheritance was first proposed in 1902 by Walter Sutton

- supported by several pieces of evidence
- reproduction involves union of only eggs & sperm
- each gamete contains only 1 copy of the genetic information
- since sperm have little cytoplasm, the material contributed must reside in the nucleus
- chromosomes both segregate and assort independently during meiosis

Linkage: the tendency of close-together genes to segregate together

- ***the further*** two genes are from each other on the same chromosome, the more likely crossing over is to occur between them
- this would lead to independent segregation
- ***the closer*** that two genes are to each other on the same chromosome, the less likely that crossing over will occur between them
- these genes almost always segregate together and would, thus, be inherited together

- Each human somatic cell normally has 46 chromosomes, (23 pairs)
- #1-22 pairs are perfectly matched in both males and females and are called **autosomes**
- #23 pair are the **sex chromosomes**
- females are designated XX while males are designated XY
- the genes on the Y chromosome determine "maleness"

Sometimes errors occur during meiosis

1. Nondisjunction: failure of chromosome to separate during meiosis I or meiosis II

- leads to **aneuploidy:** abnl chromosome #
- most result in failure to develop/early death before adulthood
- extra copy of chromosome 21 or, more rarely, chromosome 22 can survive to adulthood
- delayed development and mental impairment
- **Down syndrome:** extra copy of # 21

Nondisjunction may also affect the sex chromosomes

- ***nondisjunction*** of the X chromosome creates three possible viable conditions

- **XXX female:** usually taller than average but other symptoms vary

- **XXY male (Klinefelter syndrome)**: sterile male with many female characteristics and diminished mental capacity

- **XO female (Turner syndrome)**: sterile female with webbed neck and diminished stature

Nondisjunction of the Y chromosome also occurs

- in such cases, YY gametes are formed, leading to XYY males

- these males are fertile and of normal appearance

- Accidental changes in genes are called **mutations**

- occur only rarely and almost always result in recessive alleles

- not eliminated from the population because they are not usually expressed in most individuals (heterozygotes)

- When mutant alleles produce harmful effects: **genetic disorders**

To study human heredity, scientists examine crosses that have already been made

- Family trees or **pedigree**

- often one can determine whether a trait is sex-linked or autosomal and whether the trait's phenotype is dominant or recessive

- for example, hemophilia is a sex-linked trait

Sickle-cell anemia:

- recessive hereditary disorder
- Affected: homozygous recessive
- carry a mutated gene that produces a defective version of hemoglobin
- hgb sticks together inappropriately and produces *a stiff red blood cell with a sickle-shape*
- the cells cannot move through the blood vessels easily and *tends to clot*
- this causes sufferers to *have intermittent illness and shortened life spans*

Heterozygous individuals have some of their TBC's sickled when O2 levels become low

- the sickle-cell allele more frequent among people in malarial regions
- the presence of the allele increases resistance to malaria infection

Tay-Sachs disease is another disease caused by a recessive allele

- it is an incurable disorder in which the brain deteriorates
- sufferers rarely live beyond five years of age

Huntington's disease is a genetic disorder caused by a dominant allele

- it causes progressive deterioration of brain cells
- every individual who carries the allele expresses the disorder but most persons do not know they are affected until they are more than 30 years old

Genetic counseling is the process of identifying parents at risk of producing children with genetic defects and of assessing the genetic state of early embryos

- ❖ identify high-risk pregnancies
- ❖ identify the chances of both parents being heterozygote carriers of an allele for a recessive genetic disorder
- ❖ high-risk also identified when the mothers are > 35 years old
- ❖ Genetic counselors also utilize genetic screening

Amniocentesis: amniotic fluid is sampled and isolated fetal cells are then grown in culture and analyzed

chorionic villus sampling: fetal cells from the chorion in the placenta are removed for analysis

Genetic counselors look at 3 things from the cell cultures obtained from either amniocentesis or chorionic villus sampling

1. **chromosomal karyotype**
   analysis can reveal aneuploidy or gross chromosomal alterations
2. **enzyme activity**
   in some cases, it is possible to test directly for the proper functioning of enzymes associated with genetic disorders
3. **genetic markers**
   test for the presence of mutations at the same place on chromosomes where disorder-causing mutations are found

DNA screening

- most recent form of genetic counseling
- screens DNA for the presence of key genes
- utilizing information from the Human Genome Project, the DNA of patients is assessed for copies of genes that lead to hereditary disorders,
- in addition, parents conceiving by in vitro fertilization (i.e., test-tube babies) can screen zygotes for potential genetic anomalies
- this procedure is called **preimplantation screening**