11.1 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

Mendel 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 è**F_2 generation**

Fig 11.3 How Mendel conducted his experiments

11.2 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**

11.2 What Mendel Observed

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

Table 11.1 Seven Characters Mendel Studied in his Experiments

11.2 What Mendel Observed

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

<u>11.2 What Mendel Observed</u>

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

Fig 11.5 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

Fig 11.6 Alternative alleles of genes are located on homologous chromosomes

Table 11.2 Some Dominant and Recessive Traits in Humans

11.3 Mendel Proposes a Theory

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**

Fig 11.7 Punnett square analysis

Figure 11.8 How Mendel analyzed flower color

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

Figure 11.9 How 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

11.4 Mendel's Laws

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**

Figure 11.10 Analysis of a dihybrid cross

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

Mendel's 2nd Law: INDEPENDENT ASSORTMENT

genes located on different chromosomes are inherited independently of one another

Figure 11.11 The journey from DNA to phenotype

Some Traits Don't Show Mendelian Inheritance

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**

Figure 11.12 Height is a continuously varying character

Some Traits Don't Show Mendelian Inheritance

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

Figure 11.13 Pleiotropic effects of the cystic fibrosis gene

11.6 Why Some Traits Don't Show Mendelian Inheritance

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)

Figure 11.14 Incomplete dominance

11.6Environmental effects

Expression of some alleles depends on environment

Ie: some alleles are heat-sensitive

arctic foxes only produce fur pigment when temperatures are warm

Fig 11.15 Environmental effects on an allele

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 Fig 11.22 Linkage

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

11.8 Human Chromosomes

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

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

Fig 11.23 Nondisjunction in anaphase I (Down Syndrome)

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

Figure 11.26 Nondisjunction of the X chromosome

- 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**

Table 11.3 Some Genetic Disorders

11.9 The Role of Mutations in Human Heredity
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

Figure 11.27 A general pedigree
Fig 11.28 Royal hemophilia pedigree

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

Sickle-cell Anemia

- **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

Figure 11.33 Huntington's disease is a dominant genetic disorder

11.10 Genetic Counseling and Therapy

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

11.10 Genetic Counseling and Therapy

identify high-risk pregnancie & 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

Figure 11.34 Amniocentesis

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

chromosomal karyotype: analysis can reveal aneuploidy or gross chromosomal alterations

enzyme activity: in some cases, it is possible to test directly for the proper functioning of enzymes associated with genetic disorders

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**

Figure 11.36 Preimplantation genetic diagnosis

The **End** of Chapter 11

On to chapter 12: DNA, Protein Synthesis