

Human Inheritance

Recessive Genetic Disorders

aa, bb, nn, rr

- Recall a recessive trait is expressed when an individual is homozygous recessive for a trait

ex. yy = green color tt = short

- If an individual is heterozygous for a trait, the dominant allele shows through, but the individual is known as a carrier

*AA, BB, NN, RR**Aa, Bb, Nn, Rr*

an individual who is heterozygous for a genetic disorder

Types of Recessive Genetic Disorders

Cystic Fibrosis

- excessive mucus production in the lungs
- can lead to digestive or respiratory failure

Albinism

- no color in skin eyes or hair
- skin susceptible to UV damage
- vision problems

Tay-Sachs Disease

- buildup of fatty deposits in the brain
- mental disabilities
- death by age of 5

Galactosemia

- mental disabilities
- enlarged liver
- kidney failure

There is no cure for any of these disorders

Dominant Genetic Disorders

- * diseases caused by having dominant alleles
- * if you do not have it, you are homozygous recessive for the trait

Huntington's disease

HH Hh hh

- affects the nervous system
- gradual loss of brain function, uncontrollable movement, emotional disturbances

Achondroplasia

- small body and limb size
- commonly called dwarfism

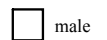
AA, Aa, aa

	A	a
A	AA	Aa
a	Aa	aa

Pedigrees

- A diagram that traces the inheritance of a particular trait through several generations

Symbols for reading a pedigree



female



male who is carrier for trait



female who is carrier for trait



male who expresses the trait

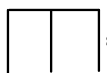


female who expresses the trait

Roman Numerals (I, II, III) = generation #

Regular Numbers (1,2,3) = # of individuals in a generation

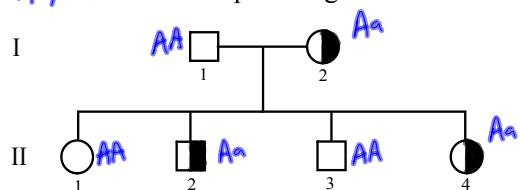
— parents



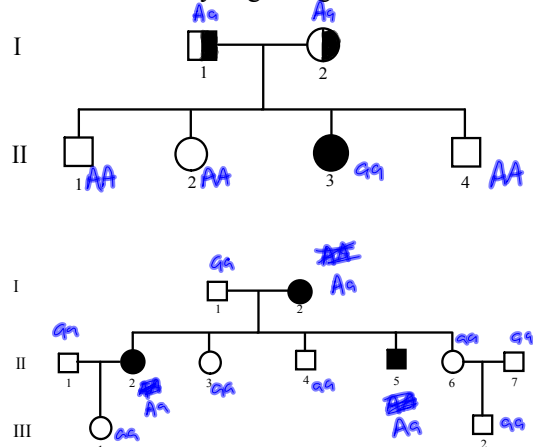
siblings

generation

Example Pedigree



Analyzing Pedigrees



- Pedigrees are used to infer genotypes from the observation of phenotypes
- genealogists can determine what genes an individual is most likely to have
- help genetic counselors determine whether inheritance patterns are dominant or recessive
- If good records are kept, within a families, disorders in future offspring can be predicted

	T	t
T	TT	Tt
t	Tt	tt

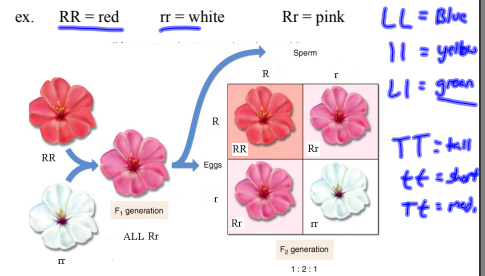
short = 75
 long = 25
 TT = 25
 Tt = 50
 tt = 25

	T	t
t	Tt	tt
t	Tt	tt

50 short
 50 long

Incomplete Dominance

- When the heterozygous phenotype is an intermediate phenotype between the 2 homozygous phenotypes



Codominance

- When both alleles are expressed in the heterozygous condition

ex. sickle cell disease

Normal	RR = red	rr = white	Rr = red
Inc. Dominance	RR = red	rr = white	Rr = pink
Codominant	RR = red	rr = white	Rr = Red & white



Multiple Alleles

Blood Types - ABO

- have 3 forms of alleles (not just 2 like other traits)

$I^A I^A$ or $I^A i$ → Blood Type A

$I^B I^B$ or $I^B i$ → Blood Type B

$I^A I^B$ → Blood Type AB I^A and I^B are codominant

$i i$ → Blood Type O has no AB markers

Epistasis

- * - the result of one allele hides the effect of another allele *

ex. Coat color in Labradors

- Labradors coat color is controlled by 2 sets of alleles

- The dominant allele (E) determines whether the dog will have dark pigment

- The dominant allele (B) determines how dark the pigment will be

** If a dog's genotype is EEbb or Eebb the dogs fur will be chocolate brown

** If a dogs genotype is eeBB, eeBb, and eebb will produce yellow coat because the (e) allele masks the effect of the dominant (B)

Sex Determination

- Each cell in the human body contains 46 chromosomes or 23 pairs of chromosomes

- the sex chromosomes determine the individuals gender 1 pair

2 types of sex chromosomes: X^* and Y

XX = female

XY = male

- the other 22 pairs of chromosomes are known as autosomes

	X	Y
X	XX	XY
X	XX	XY

50%
50%

- the X chromosome is larger than the Y chromosome

- the X and Y chromosomes contain genes that are vital to the development of an individual

- Since women do not need a double dose of X chromosome, one of the X chromosomes is inactivated

ex. calico cats fur

Sex-Linked Traits

XX = female

XY = male

Sex-Linked traits - traits controlled by genes located on the X chromosome

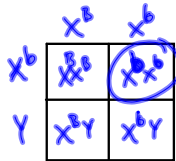
- also called X-linked traits

- Since males only have 1 X chromosome they are effected by recessive X-linked traits more than females (dominant X masks the recessive)

Red-Green color blindness

- the trait for red-green color blindness is a recessive x-linked trait

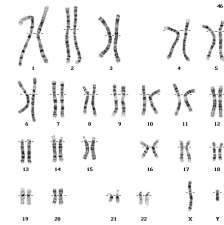
X^B = Normal
 X^b = Red-Green color blind
 Y = Y chromosome



- * what sex is the only child that could possibly be effected?
- * why are there fewer females who have red-green color blindness than males

Chromosomes and Human Heredity

Karyotype - pairs of homologous chromosomes arranged by decreasing size to produce a micrograph



22 pairs autosomes

1 pair sex chromosomes

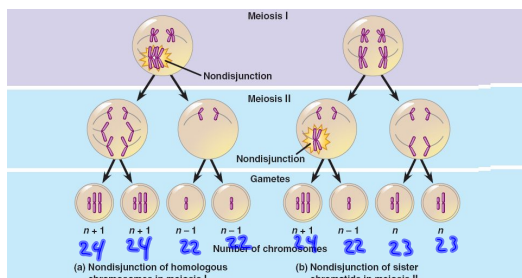
Telomeres

protective caps at the end of chromosomes that consist of DNA associated with proteins

- protects the structure of the chromosome
- * - might be involved with aging and cancer *

Nondisjunction

- cell division during which sister chromatids fail to separate properly
- if nondisjunction occurs in meiosis I or II the resulting gametes will not have the correct number of chromosomes
- when one of these gametes fertilizes another gamete the resulting offspring will not have the correct number of chromosomes
- can result in extra copies of a chromosome or only 1 copy of a chromosome



* Alters the # of chromosomes

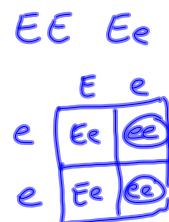
Down Syndrome

- also known as trisomy 21
- organism has 3 copies of chromosome 21



Nondisjunction and sex chromosomes

- XX = normal female
- XO = female with Turner's syndrome
- XXX = nearly normal female
- XY = normal male
- XXY = male with Klinefelter's syndrome
- XXY = nearly normal male
- OY = results in death



9:3:3:1

