Human Heredity
Chapter 5
Human Genetics

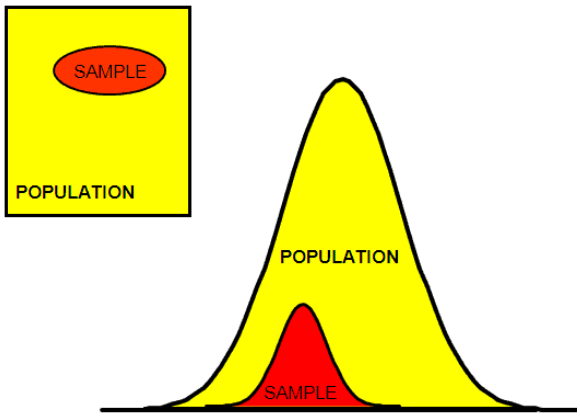
5:1 Studying Human Genetics

Humans are not good subjects for genetic research because:

1. Humans cannot ethically be crossed in desired combinations.
2. Time between human generations is too long.
3. Humans produce a small number of offspring.

The basic methods for studying human genetics are OBSERVATIONAL, not EXPERIMENTAL.

POPULATION SAMPLING: when researchers use carefully formulated statistical rules to select a small number of individuals that represent the whole population



A subset of the population.

5:2 Twin Studies

Because monozygotic twins have the same genetic inheritance they may be used to study environmental influences.

Dizygotic twins are no more alike than siblings.

Monozygotic twins occur from a random splitting of a single embryo.

RATE: 1 in 250 births
CAUSE: unknown

Dizygotic twins occur when a woman ovulates twice during the same menstrual cycle.

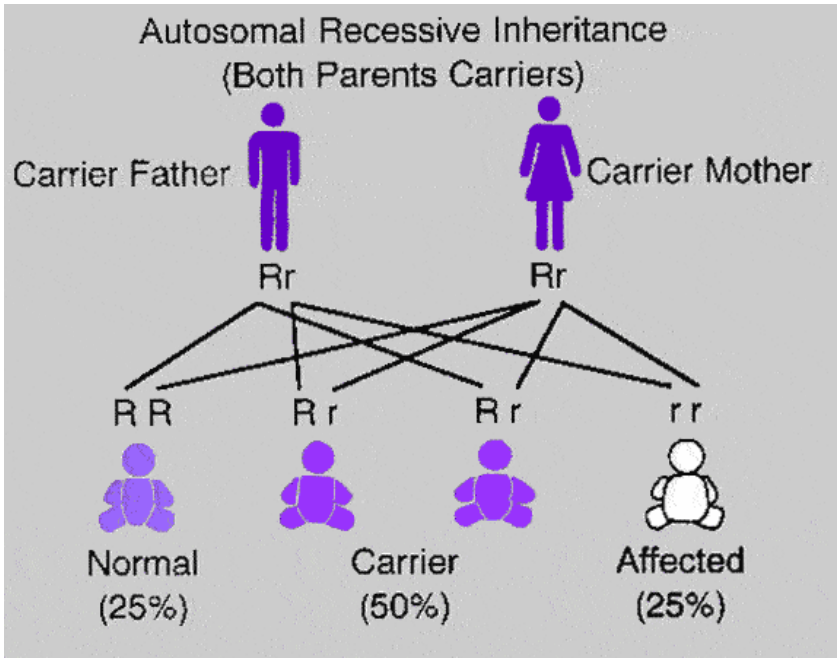
RATE: 1 in 80 births
CAUSE:Race – African high, Asian low
Heredity – family history
Age: 33 or older
Parity: having DZ twins already
Fertility Drugs:
Pergonal – 20% to 40%
Clomid – 5% - 7%

Triplets and more may occur from multiple embryo splitting, multiple ovulation, or a combination of both.

CONCORDANT: when both twins exhibit a trait
DISCORDANT: when only one twin has a trait

5:3 Autosomal Recessive Traits

Patterns of Autosomal Recessive Inheritance



1. Trait appears in offspring of unaffected parents.
2. Unless both parents are affected, affected individuals are usually members of alternate generations.
3. Two affected individuals cannot have an unaffected offspring.
4. Both sexes are equally affected.

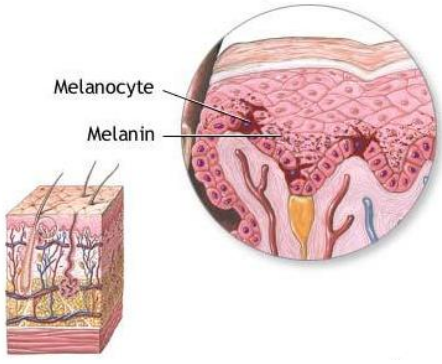
Autosomal Recessive Diseases

1. ALBINISM: genetic disorders associated with lack of pigmentation in skin and eyes.

GENEOTYPE: aa

PHENOTYPIC CHARACTERISTICS:

- Melanocytes (pigment containing cells in skin, eyes, and hair) contain no melanin
- Ocular albinism (OA) affects only the eye
- Ocular-cutaneous albinism (OCA) affects both eye and skin



- Reduced visual acuity and involuntary rapid eye movements

RATE OF OCCURRENCE

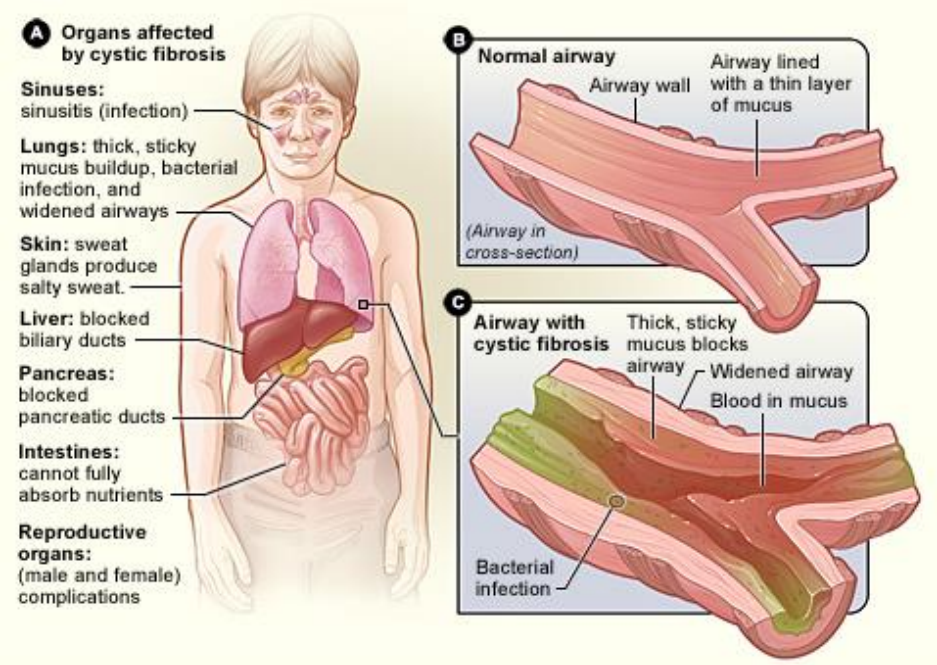
- US Caucasian population 1:37,000 OCA
- US African-American population 1:15,000 OCA
- San Blas Indians of Panama 1:132 OCA
- Hopi and Navajo in US 1:200 OCA

2. CYSTIC FIBROSIS: fatal recessive genetic disorder associated with abnormal secretions of the exocrine glands that produce mucus and sweat

GENOTYPE – cc

PHENOTYPIC CHARACTERISTICS

- Excessive salt in sweat (may give diagnosis)
- Thick mucus may clog ducts of pancreas causing both improper digestion-malnutrition and cysts in pancreas which then becomes fibrous
- Thick mucus causes blocked air passages and obstructive lung diseases
- Life expectancy increases as treatments improve
- Disease is passed on through heterozygous carriers



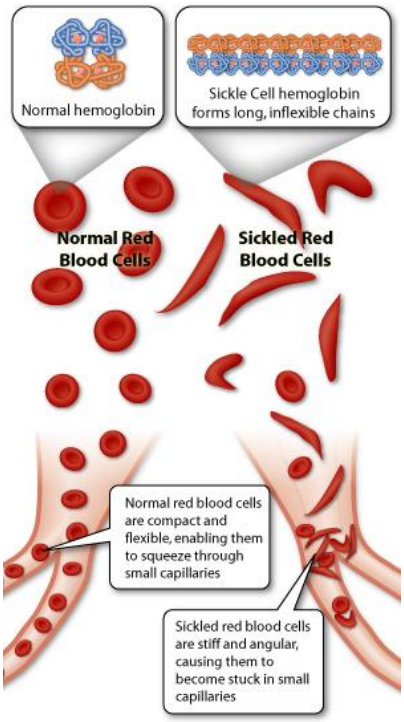
RATE OF OCCURRENCE

- US Caucasian population 1:2000, heterozygous carriers 1:20
- US African-American population 1:100,000-150,000

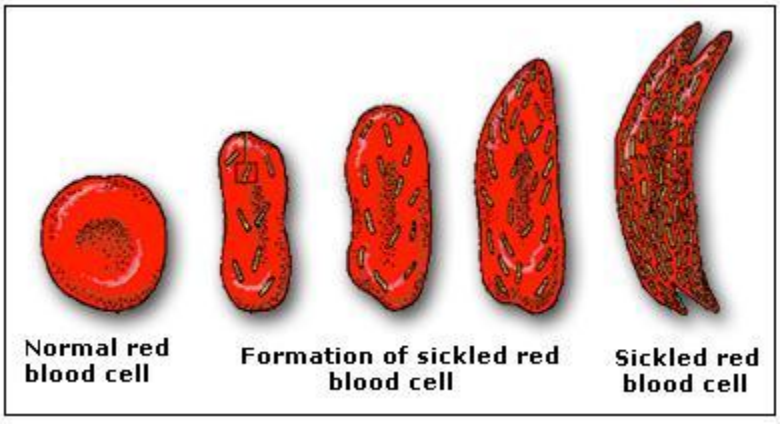
3. SICKLE CELL ANEMIA: fatal recessive genetic disorder associated with abnormal hemoglobin, the protein that transports oxygen in blood

GENOTYPE – hh

PHENOTYPIC CHARACTERISTICS



- When oxygen levels are low, abnormal hemoglobin causes RBC's to change shape and become sickle shaped and are easily broken
- Oxygen carrying capacity of blood is reduced, anemia results, heart may fail
- Sickle shaped cells may clog small blood vessels, further reducing oxygen transport, and producing more sickle cells
- Reduced circulation cause intense pain and skin ulcers and sores
- Individuals homozygous or heterozygous for sickle cell anemia are resistant to malaria
- Life expectancy can reach 50 years, women live longer than men



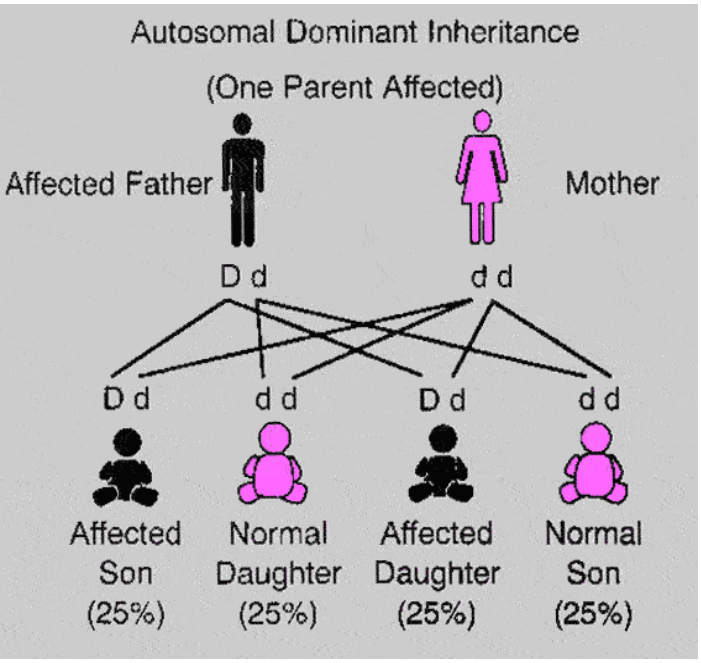
RATE OF OCCURRENCE:

- US African-American population 1:500
- US African-American heterozygous carriers 1:12

5:4 Autosomal Dominant

Traits

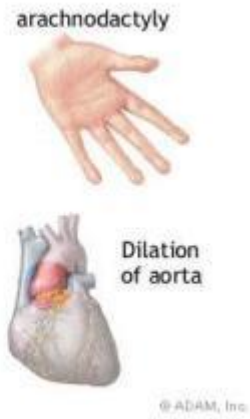
Patterns of Autosomal Dominant Inheritance



1. Trait passed directly from affected individual to their offspring.
2. Trait is present in all generations.
3. Approximately 50% of the offspring of an affected individual exhibit the trait.
4. Two affected parents MAY have unaffected children.
5. Both sexes are equally affected.

Autosomal Dominant Diseases

1. MARFAN SYNDROME: autosomal dominant disease affecting skeletal and cardiovascular systems and eye



GENOTYPE – MM, Mm

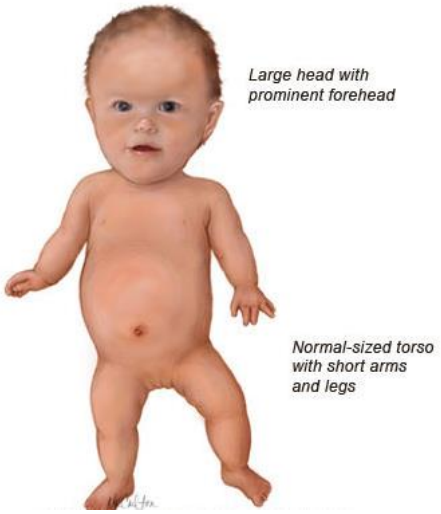
PHENOTYPIC CHARACTERISTICS

- Tall; thin; long arms, legs, and fingers
- Arm span greater than height
- Nearsighted and defects in the lens of the eye
- Weakness of the connective tissue around the aorta, causing it to enlarge and rupture
- Usually identified by autopsy or post mortem examination

RATE OF OCCURRENCE

- 1 in 10,000

2. ACHONDROPLASIA: autosomal dominant form of dwarfism



GENOTYPE – AA, Aa

PHENOTYPIC CHARACTERISTICS

- Arms and legs do not develop properly
- Skull is too large
- Body trunk is normal size
- Good muscular development

RATE OF OCCURANCE

- 1 in 10,000

3. NEUROFIBROMATOSIS: autosomal dominant disease causing abnormal pigmentation and tumors of the skin and nervous system

GENOTYPE – NN, Nn

PHENOTYPIC CHARACTERISTICS

- Café-au-lait spots (abnormal pigmentation)
- Large skin tumors
- Noncancerous tumors of nervous system
- Large tumors may compress vital nerves, causing blindness or paralysis
- Death may occur if tumors convert to cancerous form that quickly invades nervous system

RATE OF OCCURRENCE

- 1 in 3000 births, about ½ to unaffected parents because of the extremely high rate of mutation in the gene causing the disorder

All you have to do is ask.

If you don't already know him, meet Reggie Bibbs. He's kind of a celebrity around the Houston area.

Reggie is on a mission to raise awareness of neurofibromatosis.

Every weekend, Reggie wears his "Just Ask!" T-Shirt and attends Houston area events to help spread the word, have some fun and make new friends.

He wants you to feel as comfortable asking about his disorder as he is talking about it.

The next time you see Reggie, go on up and say hi. Who knows, you just might walk away with your very own "Just Ask!" T-Shirt.

And a new friend.

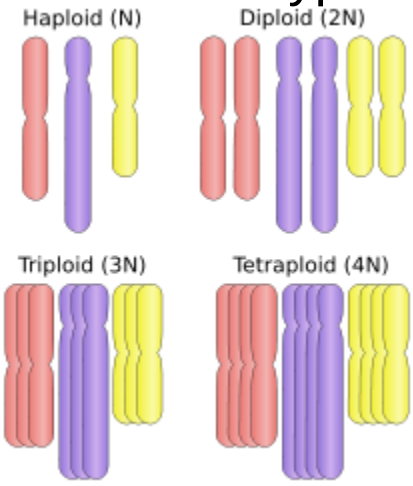
JUST ASK!

reggiebibbs.com

May is National Neurofibromatosis Month.

PHOTO: MARCUS MEDELLIN

5:5 Polyploidy



POLYPLOIDY: a chromosome number that is a multiple of the normal diploid number

Polyploidy is caused by errors in gamete formation, events at fertilization, or errors in mitotic cell division after fertilization.

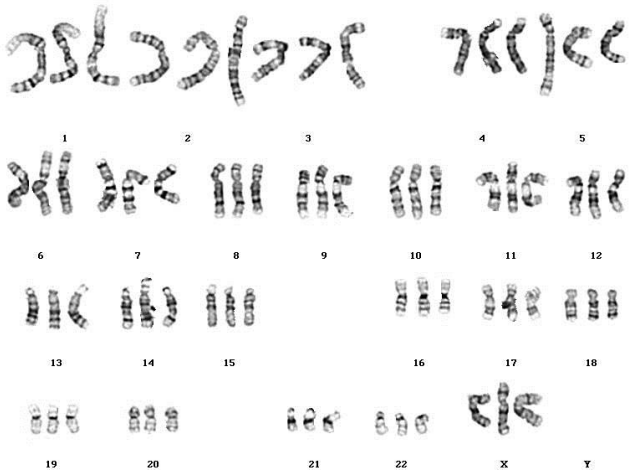
Types of Polyploidy

1. TRIPLOIDY: a chromosome number that is three times the haploid number, having 3 copies of autosomes and 3 sex chromosomes

GENOTYPE – 69 XXY, 69 XXX, 69 XYY

PHENOTYPIC CHARACTERISTICS

- Multiple abnormalities
- Enlarged head
- SYNDACTYLY: fusion of fingers and toes
- Malformation of mouth, eyes, genitals
- Most die within one month



CAUSE

- 75% of triploidy cases have 2 sets of paternal chromosomes, caused by 2 sperm fertilizing the egg.

RATE OF OCCURRENCE

- 1% of all conceptions, 99% die before birth
- 1:10,000 live births
- Observed in 15-20% of spontaneous abortions (miscarriages)

2. TETRAPLOIDY: a chromosome number that is four times the haploid number, have 4 copies of autosomes and 4 sex chromosomes
   GENOTYPE – 92 XXXX, 92 XXYY
   PHENOTYPIC CHARACTERISTICS
   - Lethal

   CAUSE
   - During the first mitotic division after fertilization, replication and separation of chromosomes is not followed by cytokinesis

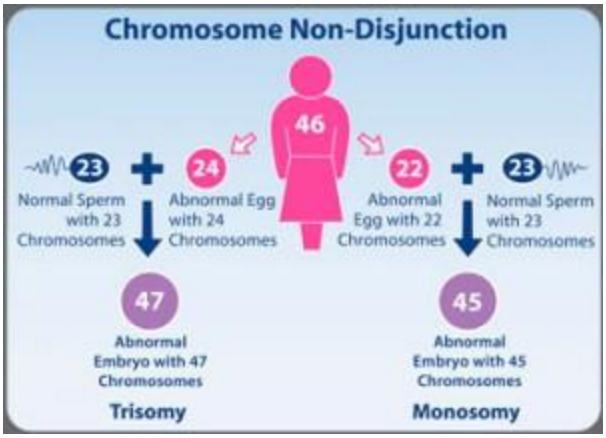
   RATE OF OCCURRENCE
   - 5% of all spontaneous abortions
   - Rare in live births

5:6 Aneuploidy

ANEUPLOIDY: a chromosome number that is not an exact multiple of the haploid, associated with the addition or deletion of an individual chromosome, caused by nondisjunction

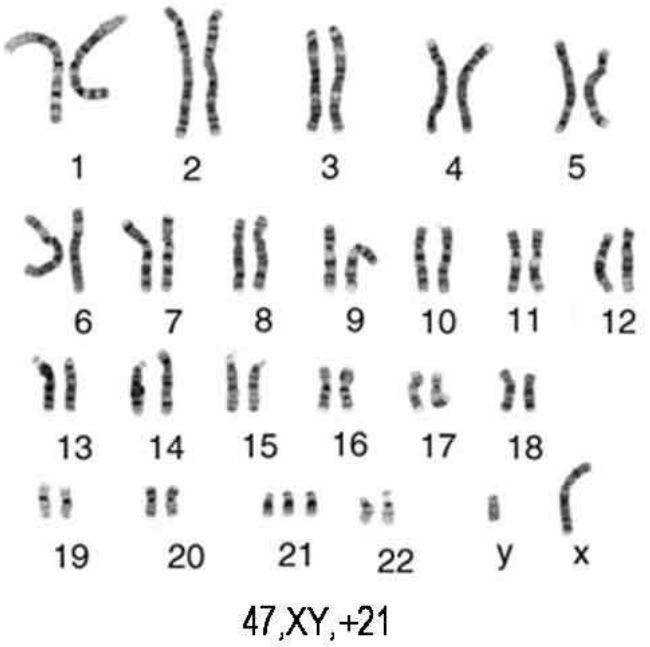
Types of Aneuploidy

1. MONOSOMY: when one chromosome of a pair is missing, causing one less than the diploid number (2n – 1)
2. TRISOMY: when one chromosome is present in 3 copies while all others are diploid, causing one more than the diploid number (2n + 1)



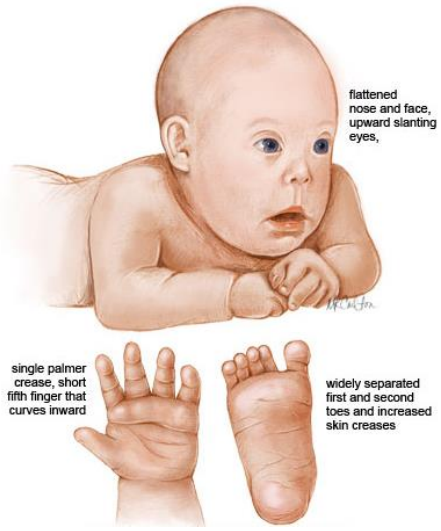
TRISOMY 21 (Down Syndrome)

GENOTYPE – 47, +21



PHENOTYPIC CHARACTERISTICS

- 40% have congenital heart defects
- Intelligence near normal to profound retardation
- Wide skull that is flatter than normal at the back
- Eyelids have epicanthic fold
- Tongue furrowed and protruding, mouth partially open
- Physical growth, behavior, mental development are retarded
- Prone to respiratory infections
- 15 times greater chance of leukemia
- Most survive to adulthood, life expectancy 60 years



CAUSE

- Maternal nondisjunction
- Increases with maternal age

RATE OF OCCURRENCE

- 1 in 700 live births
- 0.5% of all conceptions
- Most common chromosomal defect in humans