HUMAN HEREDITY

Review

How many chromosomes do humans have?
46
How many autosomes do humans have?
23
How many sex chromosomes do humans have?
2
How many chromosomes do gametes have?
23
- autosomes?
22
- sex chromosomes?
1
Genes that are carried on sex chromosomes are called?
Sex linked

Inheritance of Human Traits

- Genotype determines phenotype
- Phenotype can be affected by environmental factors (complex characters)

Ex: humans: height affected by nutrition

hydrangeas: color determined by acidity

fox coloration: determed by temperature (enzymes affected)

Methods of studying Human Genetics

1. Population genetics: large groups that represent whole population are studied

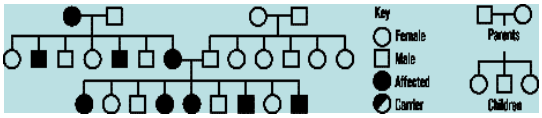
   ex: 65% population PTC tasters (phenylthiocarbomide)
   35% population non tasters

2. Twin studies: use identical twins to determine effect of environment on specific traits

3. Pedigree studies: family record (tree) that shows how a trait is inherited over generations

Pedigree

Chart of family tree
Used to study passage of trait through generations



Categories of Inherited Traits and Diseases

1. Multiple allele: three or more alleles for same gene code for a single trait

   ex: blood type

2. Autosomal dominant: affects body cells, one copy of gene needed to express trait

   ex: Huntingtons disease

3. Autosomal recessive: affects body cells, two copies of recessive gene needed to express trait

   ex: sickle cell anemia

Categories of Inherited Traits and Diseases

4. **Polygenic:** two or more genes control a specific trait

   ex: skin color, eye color, hair color

5. **Non-disjunction:** improper segregation of chromosomes during cell division

   ex: sex chromosomes: Kleinfelter, Turner syndromes
   autosomes: Trisomy 21 (Down syndrome)

6. **Sex linked:** gene carried on X or Y chromosome

   Hemizygous condition needed to express in males
   Homozygous recessive needed to express in females

   ex: hemophelia, color blindness

Multiple Allele Inheritance

A. Blood Types

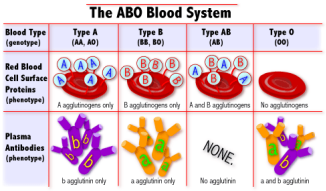
- "A,B,O" system
- 3 alleles for blood type gene
- determined by presence of absence of agglutinogen on RBC (type of antigen)

  antigen: protein substance on cell surface recognized by immune system that causes production of antibodies

Blood Types, cont.

A agglutinogen
B agglutinogen
(both have different enzyme activity)

O lack of agglutinogen
(no enzyme activity)



Blood Types, cont.

4 phenotypes, 6 genotypes

Genotype	Phenotype
IA IA	A
IA i	A
IB IB	B
IB i	B
IA IB	AB
i i	O

(capitals show co-dominance)

Genotypes and Blood Types

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
A	A	AA	A
A	B	AB	AB
A	O	AO	A
B	A	AB	AB
B	B	BB	B
B	O	BO	B
O	O	OO	O

Blood Type Inheritance:

Parents' Blood Types	Children's Possible Blood Types			
AB + AB	AB	A	B	
AB + A	AB	A	B	
AB + B	AB	A	B	
AB + O		A	B	
A + A		A		O
A + B	AB	A	B	O
B + B			B	O
O + A		A		O
O + B			B	O
O + O				O

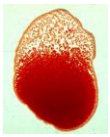
Blood Type Compatibility

If your blood type is...	You can receive....			
AB+	Any blood type			
AB-	O-	A-	B-	AB-
A+	O-	O+	A-	A+
A-	O-	A-		
B+	O-	O+	B-	B+
B-	O-	B-		
O+	O-	O+		
O-	O-			

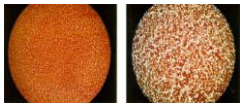
Why is blood typing important?

Mixing wrong types will cause agglutination (clumping) of RBC and destroy all blood cells causing death.

Agglutinins:
Antibodies in plasma that would destroy foreign RBC, ensure right type of blood circulates in your body



agglutination animation



B. Rh types (Rhesus monkey)

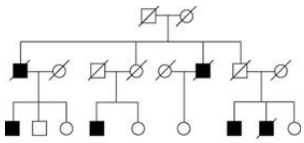
- humans either have antigen or lack it

Rh+ simply dominant (Rh+, Rh+ or Rh+, Rh-)
Rh- simply recessive (Rh-, Rh-)

Rh incompatability very important in pregnancy and transfusions.

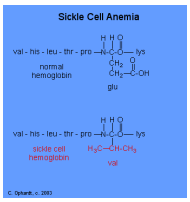
Autosomal Dominant Inheritance

Huntington's Disease
- progressive destruction of nervous system starting in 30- 40's
- only single copy of gene needed

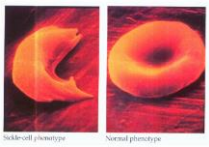


Autosomal Recessive Inheritance

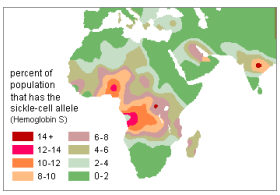
Sickle Cell Anemia



- discovered in 1904, afflicts individuals of African ancestry
- caused by point mutation (one codon) in hgb. molecule
- different protein produced , can't carry oxygen
- when O_2 deprevation occurs, RBC become sickle shaped and clog b.v. starving tissues of oxygen
- hgb. comes out of RBC and forms crystals , causes sickle shape



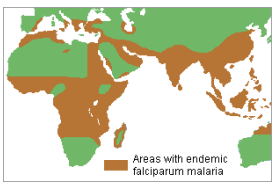
Sickle Cell Anemia, cont.



Occurrence
~ 10% in U.S.
~ 40% in Africa

Interesting reason for this pattern:

evolutionary relationship



Sickle Cell Anemia, cont.

- Genetic basis:

H^S normal RBC co-dominant
H^s sickle cell

- Genotypes

$H^S H^S$ normal

$H^S H^s$ carrier, but basically normal (1/2 cells normal)

$H^s H^s$ homozygous sickle cell – express disease

Practice problems

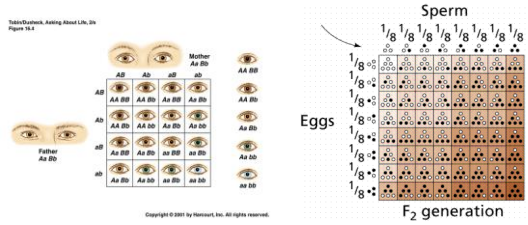
Determine the phenotypes and genotypes of offspring of the following crosses:

1. **sickle cell x carrier**
2. **sickle cell x sickle cell**
3. **carrier x normal**

Polygenic Inheritance

- **Two or more genes control one trait**

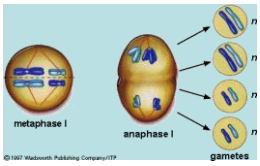
Eye color, skin color, hair color

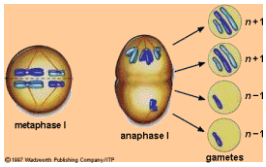


Non Disjunction Disorders

Chromosomes DO NOT PROPERLY separate during anaphase

- this is abnormal
- can occur in gametes or autosomes
- results:
  - too many chromosomes
    - ex: 47 chromosomes
  - missing chromosomes
    - ex: 45 chromosomes



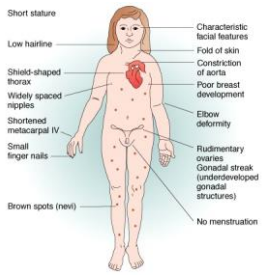


A. Sex chromosome non disjunction disorders

1. **Turners syndrome (Monosomy X)**
   - **female missing an X chromosome (45 XO)**

X + __ → X__

- **short, low mental capacity, sterile, bulging webbed neck, possible underlying cardiac, muscle problems**



How monosomy occurs

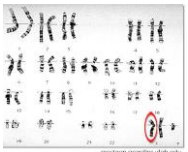
2. **Klinefelter's syndrome**

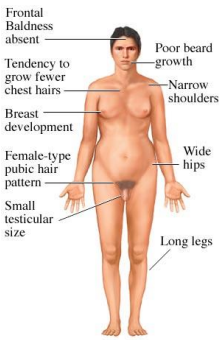
- **male with extra X chromosome (47 XXY)**

XY + X → XXY

- **tall, low mental capacity, sterile, wide hips, breast development**

"still male due to Y chromosome"





How trisomy occurs

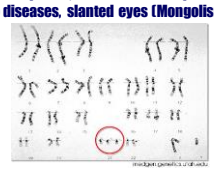
B. Autosomal non disjunction disorders

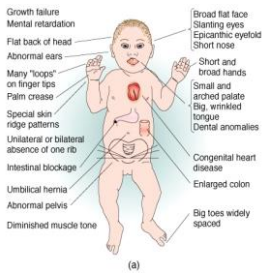
Down Syndrome (Trisomy 21)

- **not on sex chromosomes**

-extra copy of chromosome 21 (due to translocation)

- **mental retardation, poor muscle development, increased susceptibility to diseases, slanted eyes (Mongolism)**



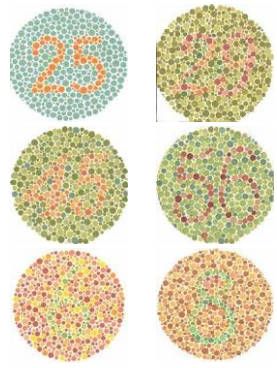


Sex Linked Inheritance

A. Color blindness

- recessive gene on X chromosome
- person can't distinguish between certain colors (red/green)
- occurrence: males 8% females 1%
- genetic basis
  - X^c normal vision
  - X^c color blind
- genotypes
  - X^cX^c normal female
  - X^cX^c carrier female
  - X^cX^c color blind female
  - X^cY normal male
  - X^cY color blind male

Ishahara Color blindness Test



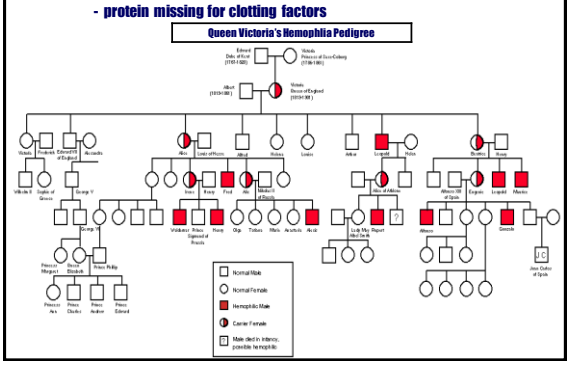
Practice Problems

Determine the phenotypes and genotypes of offspring of the following crosses:

1. female carrier x color blind male
2. normal female x color blind male
3. color blind female x normal male

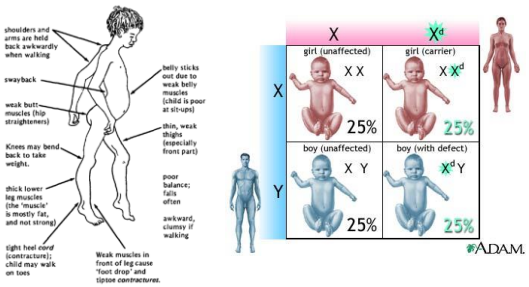
B. hemophelia "bleeders disease"

- recessive gene on X chromosome
- protein missing for clotting factors



C. muscular dystrophy

- most common form: defective gene on X chromosome
- codes for dystrophin (abnormal muscle protein)
- gradual progressive muscle loss



Sex Limited Traits

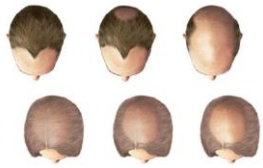
- genes located on both sex chromosomes
- only expresses in one sex (usually males) due to hormones
  - Ex: beard growth
    - Roosters – wattles and combs
    - Peacocks – fans
    - Birds, fish – brighter colors in males

Sex Influenced Traits

- genes found on both autosomes but different expression in each sex (due to hormones)
- dominant in one sex/ recesive in other sex
- if male has one recessive allele, he will show trait
- two recessive alleles needed for female to show same trait.

Ex: baldness

male pattern baldness



female pattern baldness

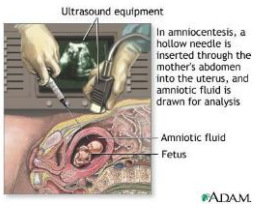
Lethal Genes

- a gene that leads to the death of an individual
- can be either dominant or recessive in nature
  - dominant: kills heterozygote
  - recessive: kills homozygote
- ex: NPHP2
  rare kidney disease that causes kideny failure in babies, children, and young adults
- ex: pyruvate dehydrogenase complex (PDC): defective enzyme prevents conversion of pyruvate to acetyl-CoA

Prenatal Diagnosis Techniques

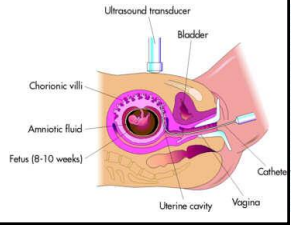
AMNIOCENTESIS

Small amount of amniotic fluid removed from amniotic sac around fetus
- done between weeks 15 – 18
- cells grown in lab and karyotype studied for chromosomal abnormalities



CHORIONIC VILLUS SAMPLING

Sample of embryonic cells taken directly from placenta surrounding fetus
- done between weeks 10 – 12
- more rapid results
- more chance of fetal harm than amnio



Ethical concerns

Euthenics: improve condition of human beings through improving environment

Eugenics: improving human species by either encouraging the breeding of persons with desired traits or discouraging breeding of persons with negative traits (Hitler)

Cloning: growing new organisms from cell lines of existing organism

Study for the test!!