

# 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: determined by temperature (enzymes affected)



## Methods of studying Human Genetics

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

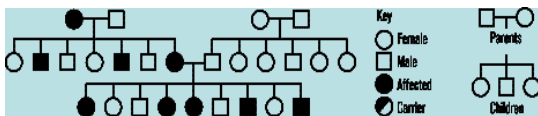
ex: 65% population PTC tasters (phenylthiocarbamide)  
35% population non tasters

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: Klinefelter, 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: hemophilia, color blindness

### Multiple Allele Inheritance

#### A. Blood Types

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

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

### Blood Types, cont.

**A** agglutinin  
**B** agglutinin  
 (both have different enzyme activity)

**O** lack of agglutinin  
 (no enzyme activity)

#### The ABO Blood System

Blood Type (genotype)	Type A (AA, AO)	Type B (BB, BO)	Type AB (AB)	Type O (OO)
Red Blood Cell Surface Proteins (phenotype)				
Plasma Antibodies (phenotype)				
	A agglutinins only	B agglutinins only	A and B agglutinins	No agglutinins
	b agglutins only	a agglutins only	NONE	a and b agglutins

### Blood Types, cont.

4 phenotypes, 6 genotypes

#### Genotypes and Blood Types

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

(capitals show co-dominance)

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

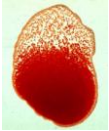
If your blood type is...	You can receive....			
AB+	Any blood type			
AB-	O-	A-	B-	AB-
A+	O-	O+	A-	A+
A-	O-	A-		
B+	O-	O+	B-	B+
B-	O-	B-		
O+	O-	O+		
O-	O-			

### Why is blood typing important?

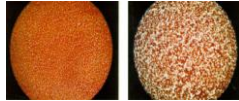
Mixing wrong types will cause agglutination (clumping) of RBC and destroy all blood cells causing death.

#### 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

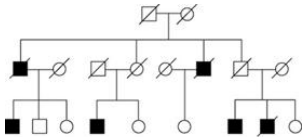
Rh+ simply dominant (Rh+, Rh+ or Rh+, Rh-)  
 Rh- simply recessive (Rh-, Rh-)

Rh incompatibility very important in pregnancy and transfusions.

### Autosomal Dominant Inheritance

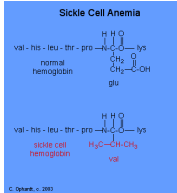
#### Huntington's Disease

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



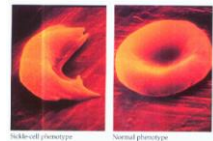
### Autosomal Recessive Inheritance

#### Sickle Cell Anemia

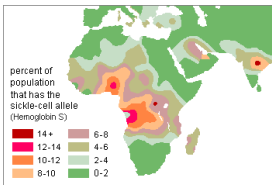


- discovered in 1904, afflicts individuals of African ancestry
- caused by point mutation (one codon) in hgb. molecule
- different protein produced , can't carry oxygen

- when O<sub>2</sub> deprivation occurs, RBC become sickle shaped and clog b.v. starving tissues of oxygen
- hgb. comes out of RBC and forms crystals, causes sickle shape

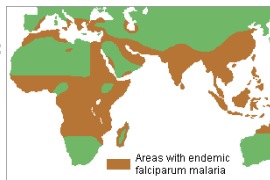


### Sickle Cell Anemia, cont.



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

Interesting reason for this pattern:  
 evolutionary relationship



### Sickle Cell Anemia, cont.

- Genetic basis:

H<sup>S</sup> normal RBC  
 H<sup>S</sup> sickle cell  
 co-dominant

- Genotypes

- H<sup>S</sup> H<sup>S</sup> normal
- H<sup>S</sup> H<sup>s</sup> carrier, but basically normal (1/2 cells normal)
- H<sup>s</sup> H<sup>s</sup> homozygous sickle cell – express disease

### Practice problems

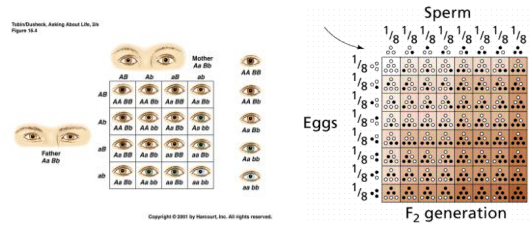
Determine the phenotypes and genotypes of offspring of the following crosses:

- sickle cell x carrier
- sickle cell x sickle cell
- carrier x normal

### Polygenic Inheritance

- Two or more genes control one trait

Eye color, skin color, hair color

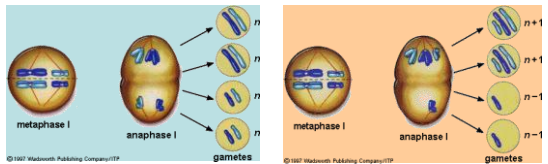


### Non Disjunction Disorders

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

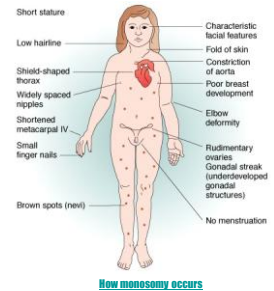
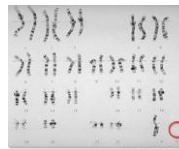


### A. Sex chromosome non disjunction disorders

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

$$X + \_ \rightarrow X\_$$

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



How monosomy occurs

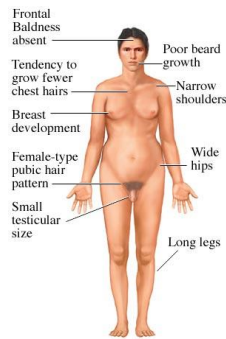
### 2. Klinefelter's syndrome

- male with extra X chromosome (47XXY)

$$XY + X \rightarrow XXY$$

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

"still male due to Y chromosome"



How trisomy occurs

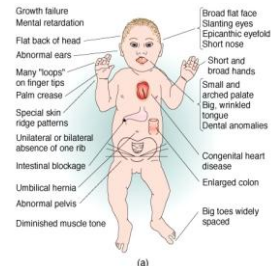
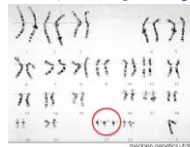
### B. Autosomal non disjunction disorders

#### 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)



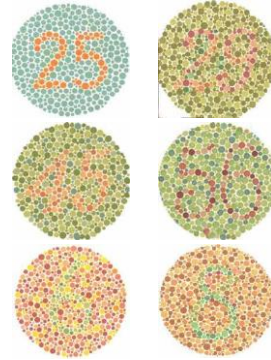
(a)

## 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
  - $X^C X^C$  normal female
  - $X^C X^c$  carrier female
  - $X^c X^c$  color blind female
  - $X^C Y$  normal male
  - $X^c Y$  color blind male

## Ishihara Color blindness Test



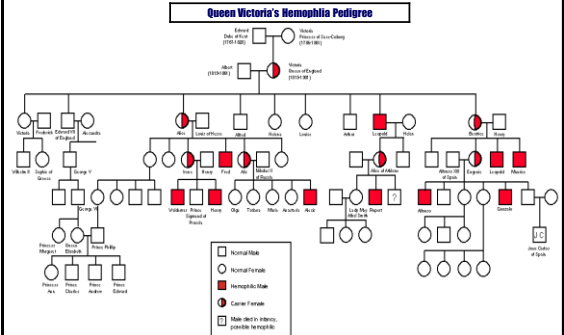
## Practice Problems

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. hemophilia "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

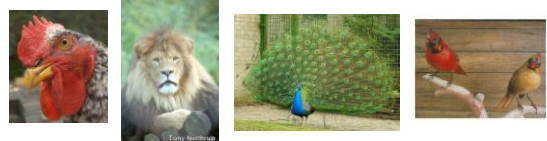
	X	$X^d$
X	girl (unaffected) $XX$ 25%	girl (carrier) $XX^d$ 25%
Y	boy (unaffected) $XY$ 25%	boy (with defect) $X^dY$ 25%

#ADAM

## 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/ recessive 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



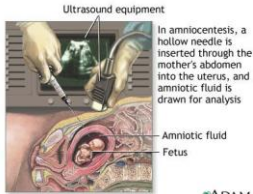
### Lethal Genes

- 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 kidney failure in babies, children, and young adults
- ex: pyruvate dehydrogenase complex (PDC): defective enzyme prevents conversion of pyruvate to acetyl-CoA

### Prenatal Diagnosis Techniques

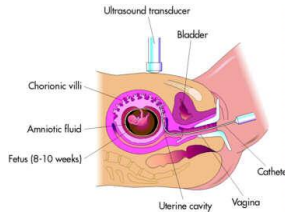
#### AMNIOCENTESIS

- 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!!

