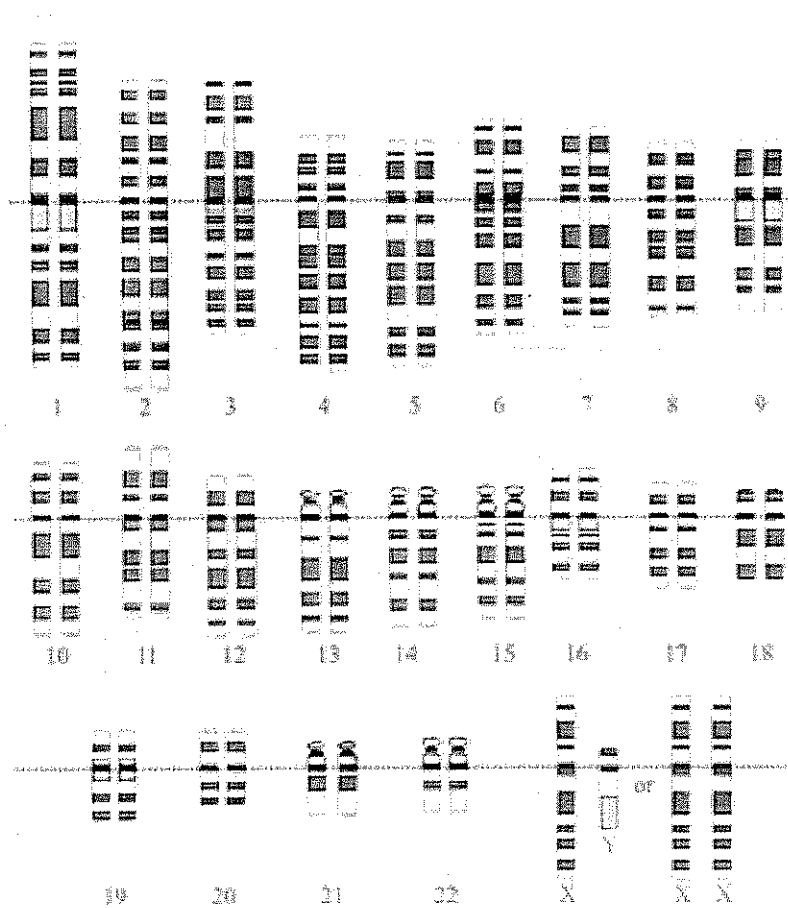


# GENETICS PART 1:

## *Mendelian & Human Genetics*



Name: \_\_\_\_\_

Hour: \_\_\_\_\_

## WORDS TO KNOW

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Genetics	Heredity	Mendel	Traits
Gamete	Fertilization	Homologous	Diploid
Haploid	Zygote	Autosome	Sex Chromosome
Allele	Gene	Genotype	Phenotype
Homozygous	Heterozygous	Dominant	Recessive
Segregation	Independent Assortment	Codominance	Punnett Square
Monohybrid Cross	Pedigree	Karyotype	Sex-linked
Polygenic Inheritance	Incomplete/Complete Dominance		Down's Syndrome
Turner's Syndrome			

## LEARNING GOALS

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After completing this unit you should be able to...

- Explain the significance of Gregor Mendel
- Understand the Laws of Segregation and Independent Assortment
- Describe the difference between homozygous and heterozygous allele combinations
- Compare dominant and recessive alleles
- Predict monohybrid crosses using Punnett Squares
- Predict genotypes and phenotypes based on given information about offspring and parents
- Recognize inheritance patterns and use them in a Punnett square
- Read and interpret karyotypes
- Compare normal chromosome patterns to Down's and Turner Syndrome patterns

# Mendelian Genetics

## History

- Gregor Mendel conducted the earliest genetic research and is known today as the "Father of Genetics" for his many discoveries of genes and inheritance
- He found that organisms closely resembled their parents with slight variations and their characteristics are influenced by heredity and the environment.

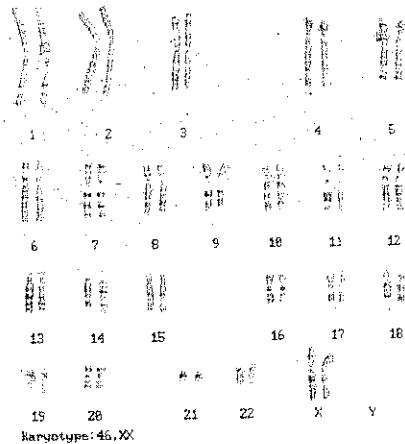
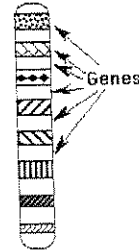
## II: Genetics

- Branch of biology that studies \_\_\_\_\_ or the passing on of characteristics from parent to offspring during sexual reproduction

## III: Conclusions made by Mendel:

- \_\_\_\_\_ or physical characteristic are determined by \_\_\_\_\_ on chromosomes.

- \_\_\_\_\_ : segment of DNA
  - 1 or more genes can determine a hereditary trait
- There are two types of chromosomes: all having different genes.
  - \_\_\_\_\_ chromosomes
  - \_\_\_\_\_ chromosomes: \_\_\_\_\_



- Genes have alternative forms called \_\_\_\_\_.

- Offspring will receive \_\_\_\_\_ allele from mom and dad
- \_\_\_\_\_ alleles determine how trait is expressed

### 3. Allele Combinations on each homologous pairs:

A. HOMOZYGOUS: \_\_\_\_\_ alleles that result in a dominant or recessive trait.

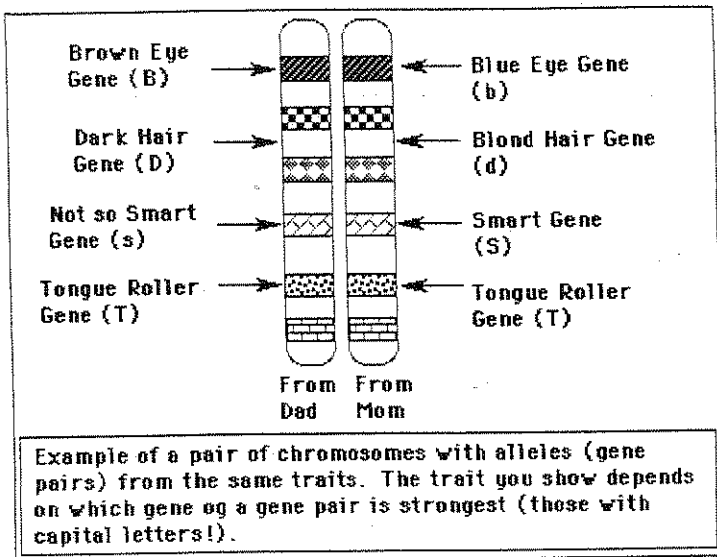
B. HETEROZYGOUS: \_\_\_\_\_ alleles that result in a dominant trait

- one allele dominant and one that is recessive

- \_\_\_\_\_ : observed or expressed trait
- \_\_\_\_\_ : weaker or hidden trait

**HOMOLOGOUS PAIRING**

**NEW GENE COMBINATIONS**



**IV. Prediction of Traits:**

1. \_\_\_\_\_ used to predict the **probability** or likelihood of a genetic cross

**Types of Genetic Crosses:**

a. \_\_\_\_\_ cross that involves \_\_\_\_\_ trait and predicts the **genotype** and **phenotype** of new gene combinations.


**A. GENOTYPE:** The allele combinations of an individual (genetic makeup)

Ex. Trait: Flower color

**B. PHENOTYPES:** physical appearance

Ex. Trait: Eye color

# Punnett Square Practice

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**Stop and Jot Space**



# Human Genetics

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## Patterns of Inheritance

- a. \_\_\_\_\_
- b. \_\_\_\_\_
- c. \_\_\_\_\_
- d. \_\_\_\_\_
- e. \_\_\_\_\_

### a. Complete Dominance:

- a. Traits inherited are either \_\_\_\_\_ or \_\_\_\_\_
- b. Ex. BB, Bb, bb

### b. Incomplete Dominance:

- Traits inherited shows a new phenotype that is a \_\_\_\_\_ or \_\_\_\_\_ of parental alleles.
    - a. \_\_\_\_\_ results in a "blending" of two alleles
    - b. Use both **capital letters** to represent (neither allele is dominant or recessive)
- Ex. 4 o'clock flowers, snapdragons, Tay Sach's Disease

### c. Codominance:

- i. Trait inherited shows a new phenotype that \_\_\_\_\_ parental alleles
  - c. \_\_\_\_\_ condition results in the expression of both alleles
  - d. Use both **capital letters** to represent
- Ex. Roan Horse (Red and White Hair), Dalmatian fur coat

d. **Sex-Linked Traits:** Human patterns

- traits are controlled by genes carried on the \_\_\_\_\_ or \_\_\_\_\_ chromosome
- a. \_\_\_\_\_-linked traits can be passed onto both male and female
- b. \_\_\_\_\_-linked traits can be passed on only to males

X-Linked Traits:

1. Affect the X chromosome
  2. \_\_\_\_\_ tend to be \_\_\_\_\_ and pass on traits to their sons
  3. Females can be affected but \_\_\_\_\_ chromosomes must have gene for trait
- Ex. Color blindness, Hemophilia

e. **Polygenic traits:** Human patterns

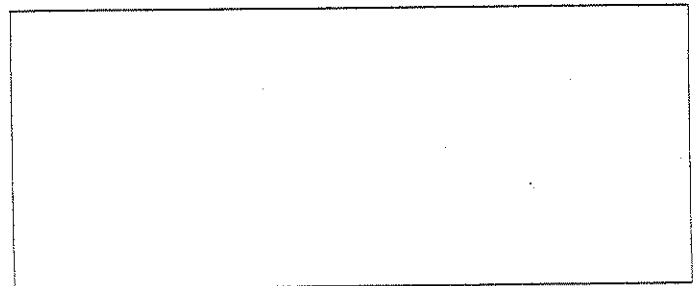
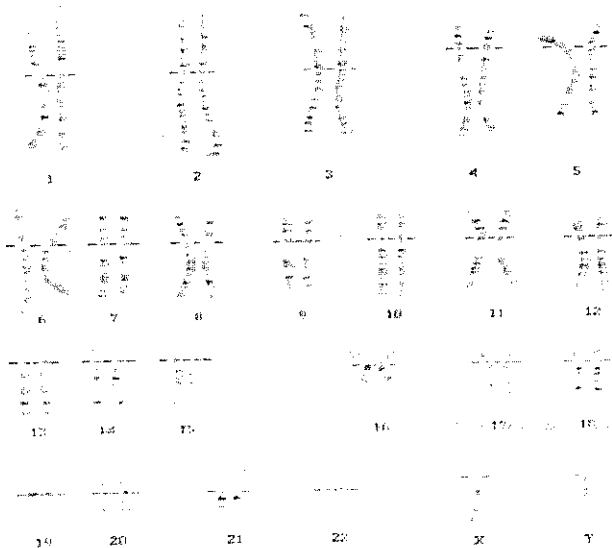
- Traits are controlled by \_\_\_\_\_ gene pairs
  - a. genes may be on **same** or **different** chromosomes
  - b. Expressed trait varies greatly
- Ex. hair color, eye and skin color, height, weight

II. How to study patterns of inheritance in human families.

Two Methods:

1. \_\_\_\_\_: Photograph of an individual's chromosomes arranged in **homologous pairs** and can be viewed underneath the microscope.
  - a. Used to identify problems with chromosomes
    - Abnormal numbers (non-sex and sex)
    - Damaged or broken
    - To detect genetic diseases

Typical Karyotype: Normal Individual



2. \_\_\_\_\_: diagram to show genetic inheritance and is used to map genetic traits.
  - a. shows \_\_\_\_\_ and \_\_\_\_\_ of family members
  - b. can predict patterns of inheritance
  - c. uses symbols



# Codominance & Incomplete Dominance Practice

1. Read each example and determine if it is an example of Codominance (C) or Incomplete Dominance (I)

- a. \_\_\_\_\_ is an example of 2 alleles being expressed
- b. \_\_\_\_\_ is an example of 2 alleles blended or mixed together

2. Above each phenotype listed in each set write the correct genotypes. Remember that the "intermediate" trait must always be heterozygous.

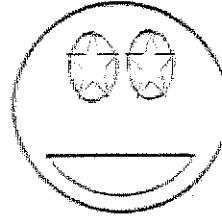
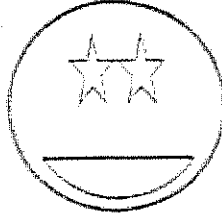
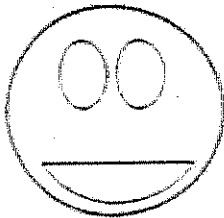
BB      YY      BY

A wocket in my pocket can be blue, yellow and green - \_\_\_\_\_

- a) \_\_\_\_\_ Birds can be blue, white, or white with blue-tipped feathers.
- b) \_\_\_\_\_ Flowers can be white, pink, or red.
- c) \_\_\_\_\_ A Hoo can have curly hair, spiked hair, or a mix of both curly and spiked.
- d) \_\_\_\_\_ A Sneech can be tall, medium, or short.
- e) \_\_\_\_\_ A Bleexo can be spotted, black, or white.

3. View the smiley pictures below – is this incomplete dominance or codominance? Circle

4. In smiley's, eye shape can be starred, circular, or a circle with a star. Write the genotypes for the pictured phenotypes



5. Show the cross between a star-eyed (SS) and a circle eyed (CC).

What is the phenotype of the offspring? \_\_\_\_\_

What are the genotypes? \_\_\_\_\_

6. If the smiley parents produced 1 circle eye(CC), 2 circle-star eye(CS) and 1 star eye(SS) offspring. What are the genotypes of the parents? Make a punnett square if needed

Parent genotypes: \_\_\_\_\_ and \_\_\_\_\_

What is the heterozygous phenotype? \_\_\_\_\_

7. Show the cross between a circle-star eyed and a circle eyed.

How many of the offspring are circle-eyed? \_\_\_\_\_

How many of the offspring are circle-star eyed? \_\_\_\_\_

How many are star eyed? \_\_\_\_\_

**Problem #1:**

In guinea pigs, black fur (B) is dominant. If a heterozygous black guinea pig is crossed with a white guinea pig what are the genotypes and the phenotypes of the offspring.

\_\_\_\_\_ X \_\_\_\_\_


Genotype:

Phenotype:

**Problem #2**

If two parents with dominant phenotypes produce an offspring with a recessive phenotype, what does this say about the parents? Explain.

**Problem #3**

Incomplete dominance problem

If you were to cross pink flowering plants with a white flowering plant, what would the phenotypes and genotypes be of the offspring?

\_\_\_\_\_ X \_\_\_\_\_


Genotype:

Phenotype:

**Problem #4**

Codominance Problem

A dog had offspring with the following phenotypes: 1/4 black, 2/4 black and white, 1/4 white puppies.

- Predict the genotypes of the parents? \_\_\_\_\_ and \_\_\_\_\_
- what is the phenotype of the heterozygous offspring? \_\_\_\_\_
- what are the genotypes of each offspring? \_\_\_\_\_

## Sex-Linked Traits

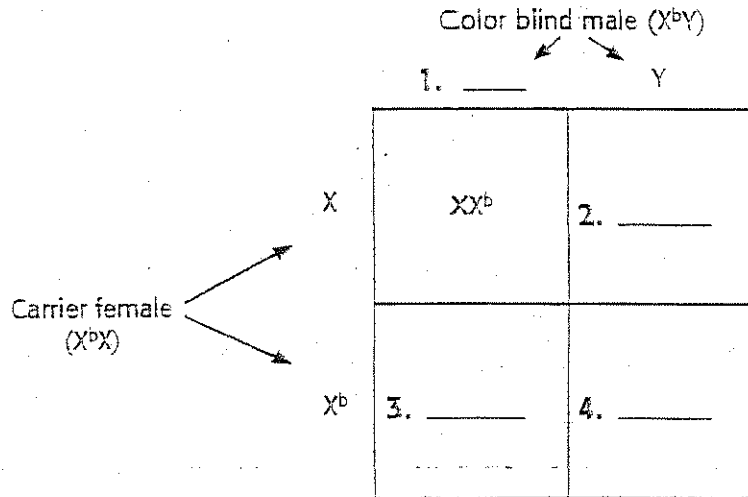
A pair of chromosomes known as sex chromosomes determines the sex of an organism. A male has one X-chromosome and one Y-chromosome. A female has two X-chromosomes. Although the sex chromosomes control the sex of an individual, there are non-sex related traits found on the X-chromosomes that are not found on the Y-chromosomes.

Trait	Possible Alleles
Normal male	XY
Colorblind male	$X^bY$
Carrier female	$X^bX$
Colorblind female	$X^bX^b$

A color-blind husband has children with his wife who carries the gene for color-blindness. What are the possible combinations of genes in their children?

Complete the missing parts of the Punnett square.

- Step 1** Place one of the allele sets from one parent plant on the left side of the Punnett square and the other parent allele set across the top of the square.
- Step 2** Match an allele from each parent into each of the sections of the Punnett square.



**Step 3** Count the outcomes as a fraction and a percentage.

Allele Combinations	Number	Male or Female?	Color-blind?	Fraction	Percentage
XY	1 section out of 4	Male	No	5.	6.
$X^bY$	1 section out of 4	7.	8.	9.	10.
$X^bX^b$	1 section out of 4	11.	12.	13.	14.
$XX^b$	1 section out of 4	Female	15.	16.	17.

18. A male with colorblind vision ( $X^bY$ ) marries a female who is a carrier of the gene for colorblindness ( $XX^b$ ). What are the possible combinations of genes for their children?

$xx$  = female, normal $XY$  = male, normal $x^Hx$  = female, carrier $x^HY$  = male, hemophiliac $x^Hx^H$  = female, hemophiliac

1. A woman who is a carrier marries a normal man. Show the cross. What is the probability that their children will have hemophilia? What sex will a child in the family with hemophilia be?
2. A woman who has hemophilia marries a normal man. How many of their children will have hemophilia, and what is their sex?
3. In cats, the gene for calico (multicolored) cats is codominant. Females that receive a B and an R gene have black and orange splotches on white coats. Males can only be black or orange, but never calico.  
Here's what a calico female's genotype would look like:  $X^B X^R$

Show the cross of a female calico cat with a black male? \_\_\_\_\_ x \_\_\_\_\_

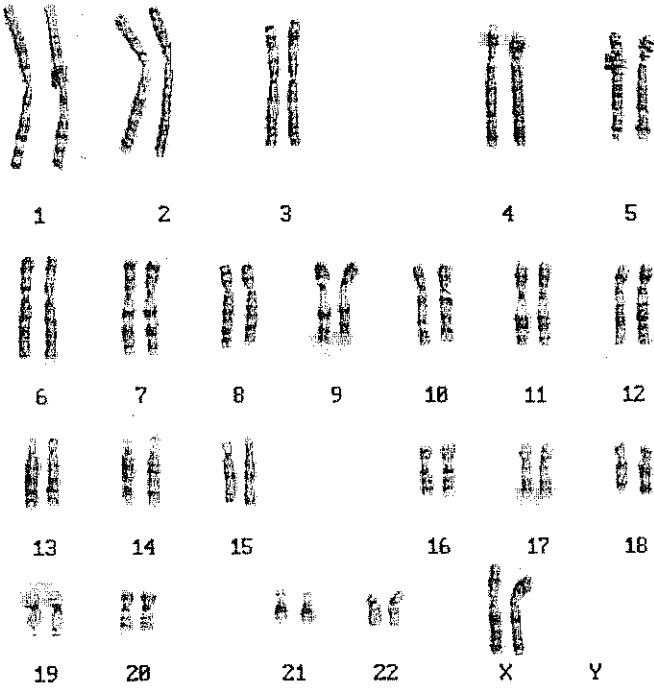
What percentage of the kittens will be black and male? \_\_\_\_\_  
 What percentage of the kittens will be calico and male? \_\_\_\_\_  
 What percentage of the kittens will be calico and female? \_\_\_\_\_

4. Show the cross of a female black cat with a male orange cat.

What percentage of the kittens will be calico and female? \_\_\_\_\_  
 What color will all the male cats be? \_\_\_\_\_

# Karyotype Interpretation

Patient A:



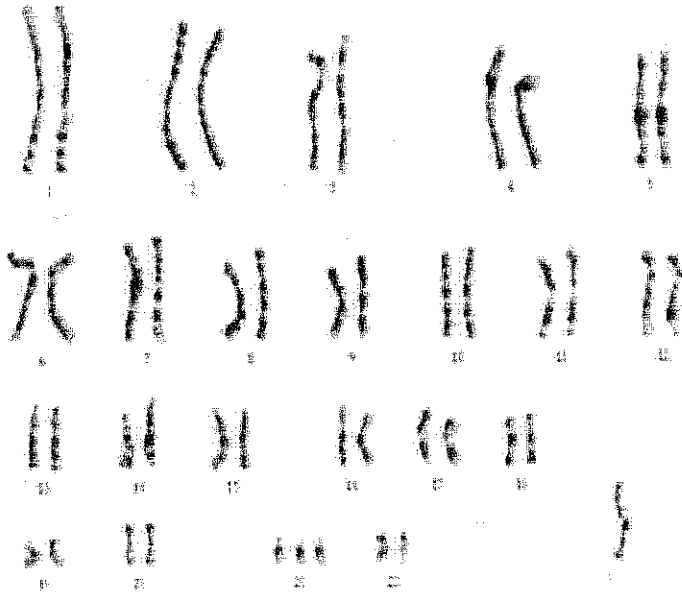
Patient A

Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

Patient B:



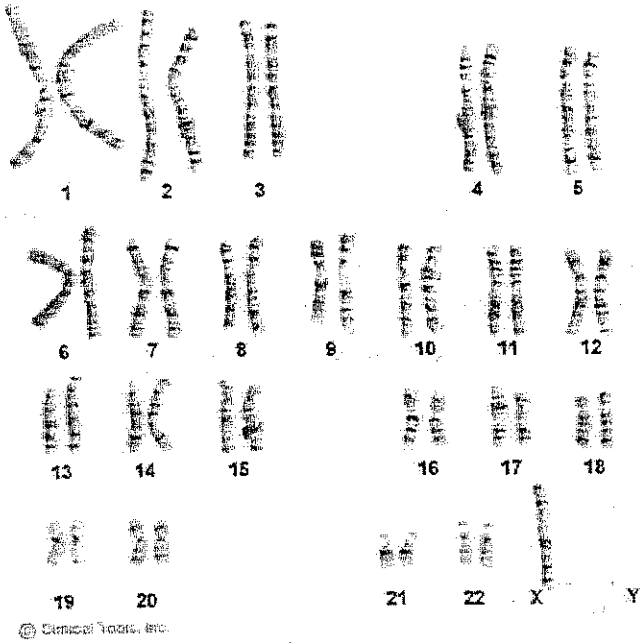
Patient B

Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

Patient C: Turner's Syndrome (1 in 2000)



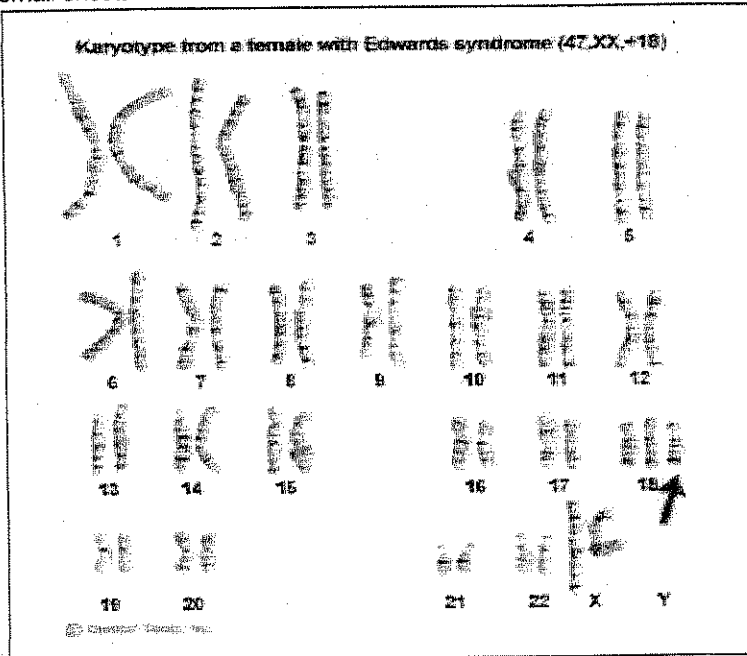
Patient C

Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

Patient D: Edward's Syndrome – only 1 in 8000 live births (fetus usually does not survive) – affects facial features, clenched fists, small chests



Patient D

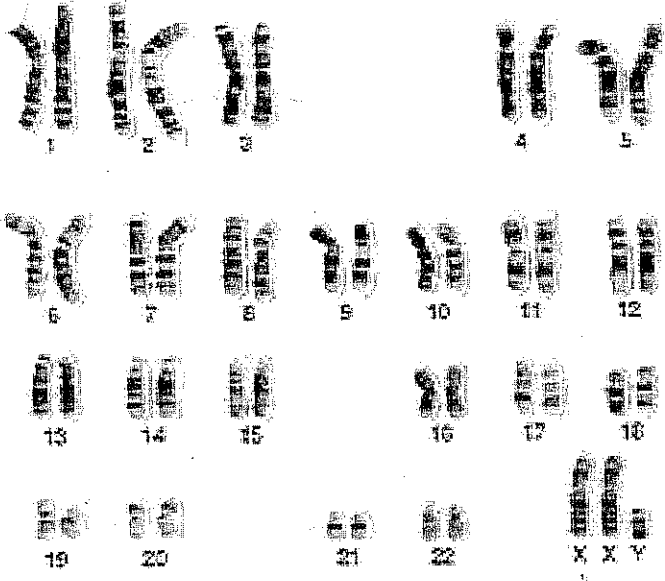
Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

Patient E: Klinefelter's Syndrome (1 in 500)

**Klinefelter's Syndrome**



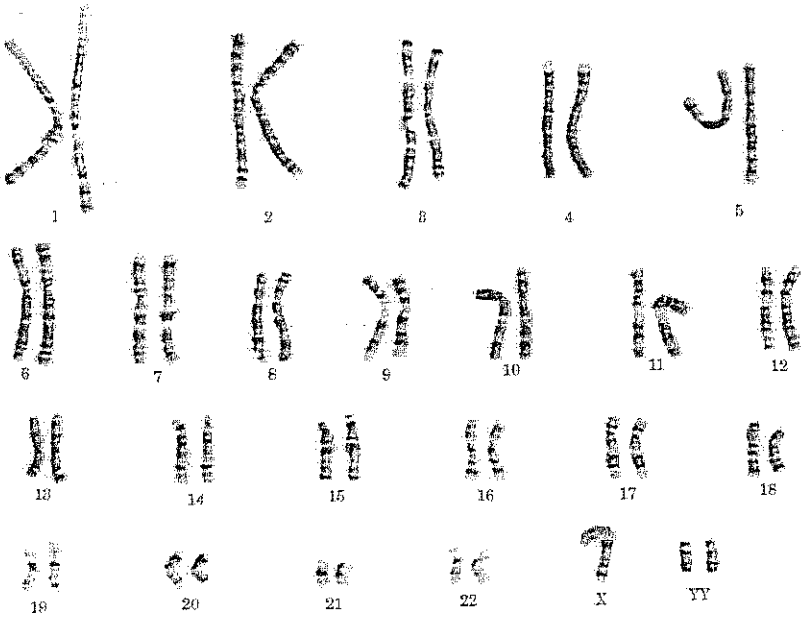
*Patient E*

Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

Patient F:



*Patient F*

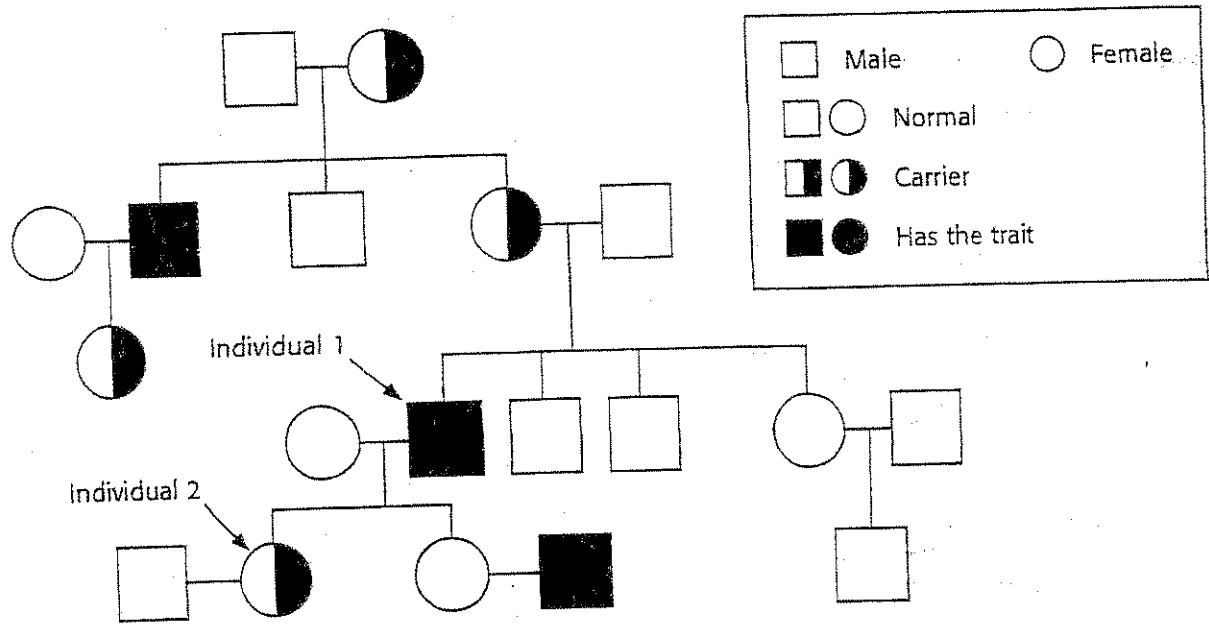
Pattern of Inheritance:

Chromosomal Abnormality:

Interpretation and Notation:

# Pedigrees

The pedigree chart below traces the inheritance of color blindness in a family. A pedigree is a chart that tracks which family members have a particular trait. A carrier is an individual that has the gene for the trait, but does not show that trait:



- How many males are shown in this chart? \_\_\_\_\_
- How many females? \_\_\_\_\_
- How many individuals are color blind? \_\_\_\_\_  
What is the sex of these individuals? \_\_\_\_\_
- How many individuals are carriers? \_\_\_\_\_  
What is the sex of the carriers? \_\_\_\_\_
- Are there any male carriers? \_\_\_\_\_ Any females with the trait? \_\_\_\_\_
- Individual 1 has the trait. What do you notice about this person's parents? \_\_\_\_\_  
\_\_\_\_\_
- Can a male inherit the trait if his mother is not a carrier? \_\_\_\_\_
- How can a female child be a carrier? \_\_\_\_\_  
\_\_\_\_\_
- Individual 2 marries a male who does not have the trait. Could this couple have a child that has the trait? \_\_\_\_\_