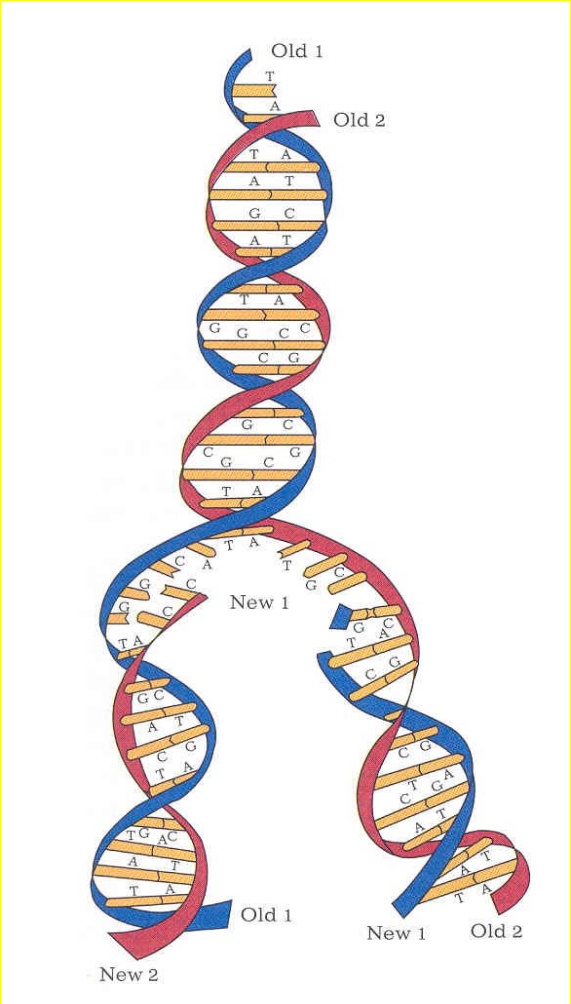
GENETIC DIVERSITY

UNITS OF HEREDITY



- **CHROMOSOMES**

  - threadlike structures in nucleus of all body cells

  - body cells have 46 (23 pairs)

  - are blueprints for maturation, growth, and cell functioning

  - abnormalities may result in death, or deficits in functioning

  - contain GENES and DNA (next…)

UNITS OF HEREDITY

- **DNA:** nitrogen-based molecules that make up GENES
- **GENES** (30,000+): units of DNA that are arranged on chromosomes **in pairs**

  - gene pairs determine **TRAITS**

  - gene pairs are a bit different for all except for identical twins, triplets, etc.

CHROMOSOMES

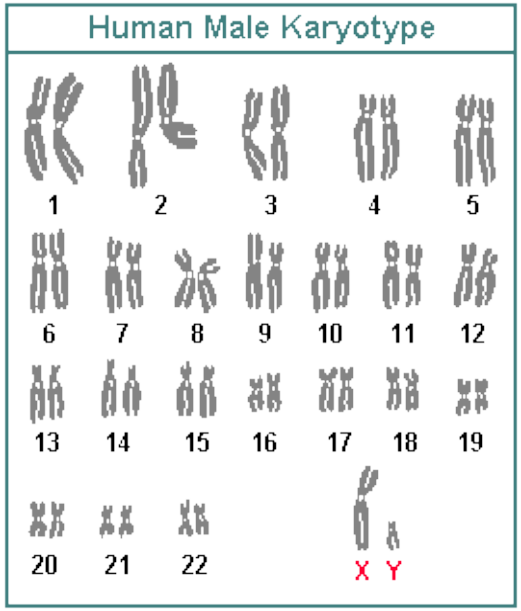
How does infant get 23 pairs?

- Mother contributes 23 singles in ovum (egg cell or female **GAMETE**)
- Father contributes another 23 singles (from a single sperm cell or male **GAMETE**)
- These fuse at fertilization to become 23 **pairs** in nucleus of **ZYGOTE**
- **A single Mom and Dad can produce about 8,000,000 possible viable combinations of genetic material**

CHROMOSOMES

<u>How does infant get GENDER?</u>

- Determined by 23rd chromosome pair
- XX = female
- XY = male
- Mother ALWAYS contributes X
- Father can contribute EITHER X or Y
- Father ALWAYS DETERMINES gender of baby



EFFECTS OF GENES

- ***ADDITIVE EFFECTS***

  - gene from Mom and gene from Dad AVERAGE OUT in the child (height?)

- ***NON-ADDITIVE EFFECTS***

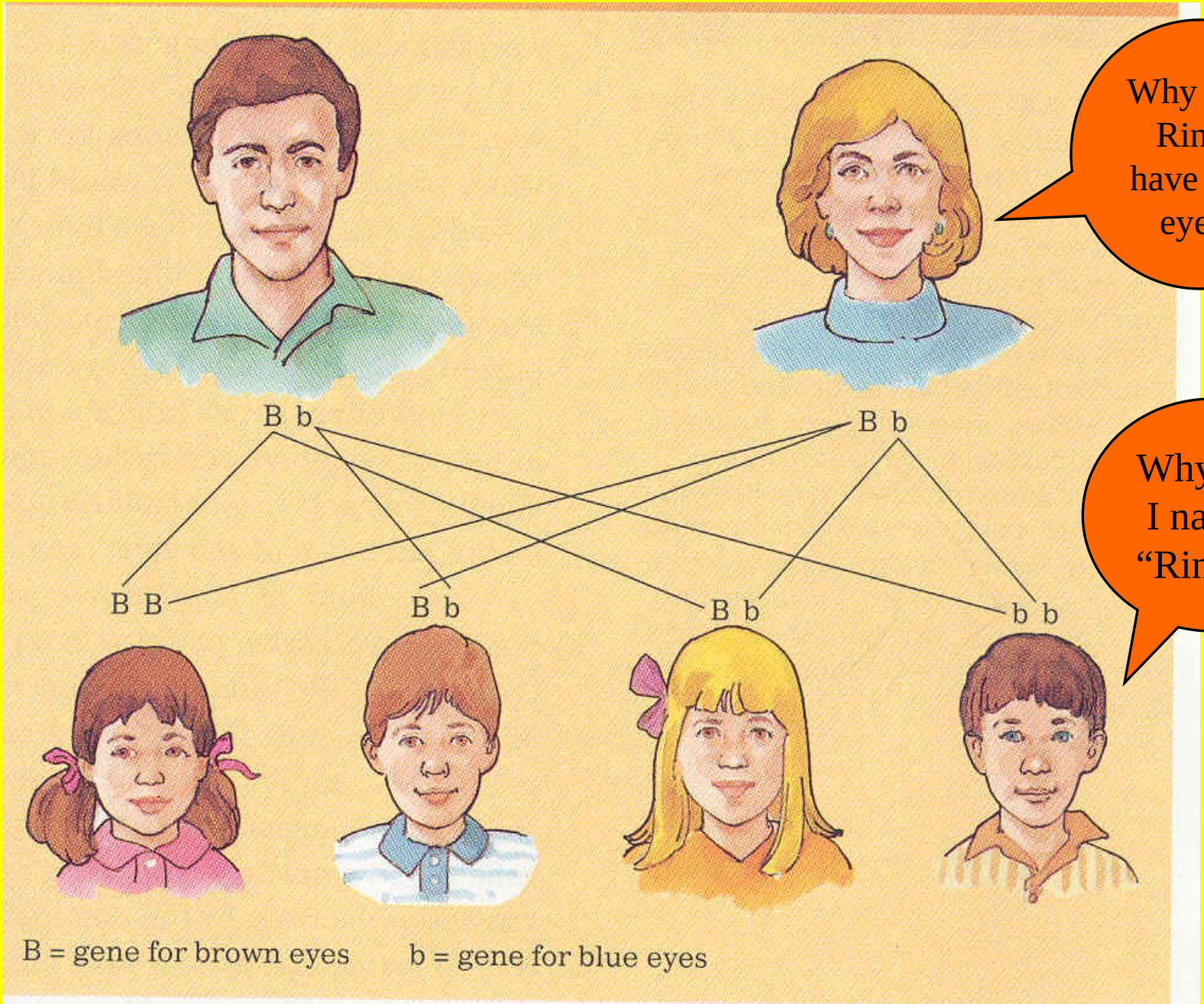
  - gene from one parent "wins out" over gene from other parent OR gene is the same from both parents (see next slide!)

NON-ADDITIVE GENES

- **DOMINANT VS. RECESSIVE**

  - Dominant gene is ALWAYS expressed

  - Recessive gene is typically ONLY expressed when paired with another recessive gene



Janie Johnny Joanie

NON-ADDITIVE GENE TRAITS

DOMINANT
Curly hair

RECESSIVE
straight hair

Free earlobe
→

attached lobes

NON-ADDITIVE GENE TRAITS

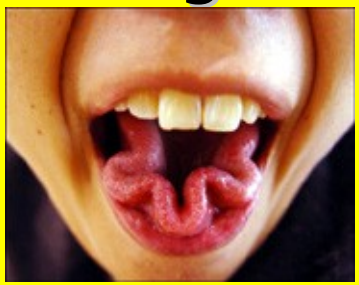
<u>DOMINANT</u>

Tongue straight

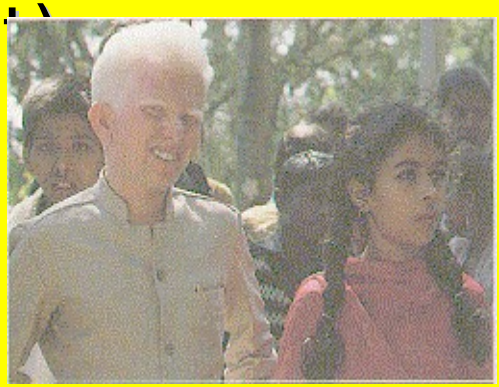
Colored skin pigmen

<u>RECESSIVE</u>

tongue curling



albinism (no



NON-ADDITIVE GENE TRAITS

<u>DOMINANT</u>

Dimples

Type A blood

Blood clots

<u>RECESSIVE</u>

no dimples

Type O

Hemophilia

What is a “CARRIER”?

- Person who has one recessive gene and one dominant gene for a trait
- The recessive gene is hidden (not expressed)

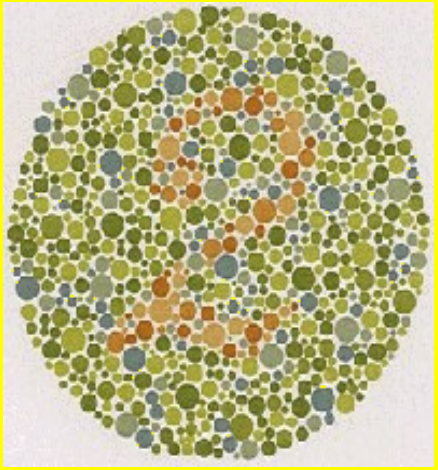
DANGEROUS GENES

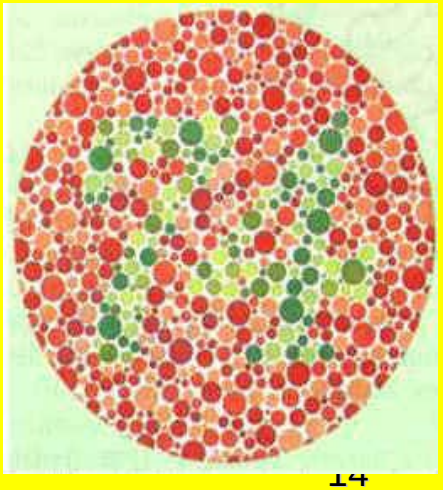
- Estimated that each person carries on average about 20 recessive genes that could cause serious abnormalities

- Fortunately, unlikely that you will procreate with someone who also carries those recessive genes

SEX-LINKED GENES

- FOUND ON 23rd CHROMOSOME PAIR ON X CHROMOSOMES (NOT Y)
- If RECESSIVE, will be expressed in **MALES**!!! Why? Because there is nothing to counteract it (is a single gene instead of a pair)
- Examples: some types of colorblindness; also may account for many forms of illness and mental retardation only seen in males





CHROMOSOMAL ABNORMALITIES

- Occurs when gamete formed through uneven division
- Gamete has wrong number of single chromosomes (NOT usual 23!)

CHROMOSOMAL ABNORMALITIES

- Scary news: this actually occurs in 50% of all ZYGOTES!!!!!
- But…most miscarry very early because infant is not viable
- **1/200 <u>births</u> has some form of chromosomal abnormality**
- **Characteristics of this are called SYNDROMES**

What are some SYNDROMES caused by chromosomal abnormalities?

- **DOWN SYNDROME (TRISOMY 21)**

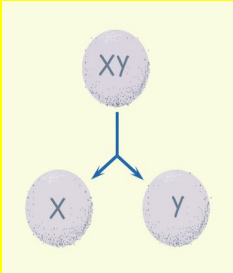
  - most often occurs from extra 21st chromosome

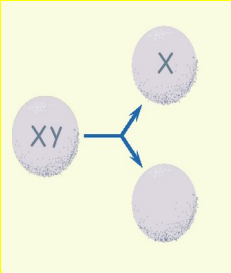
  (“3 CELLS on 21”)

  - easily recognizable from outward characteristics

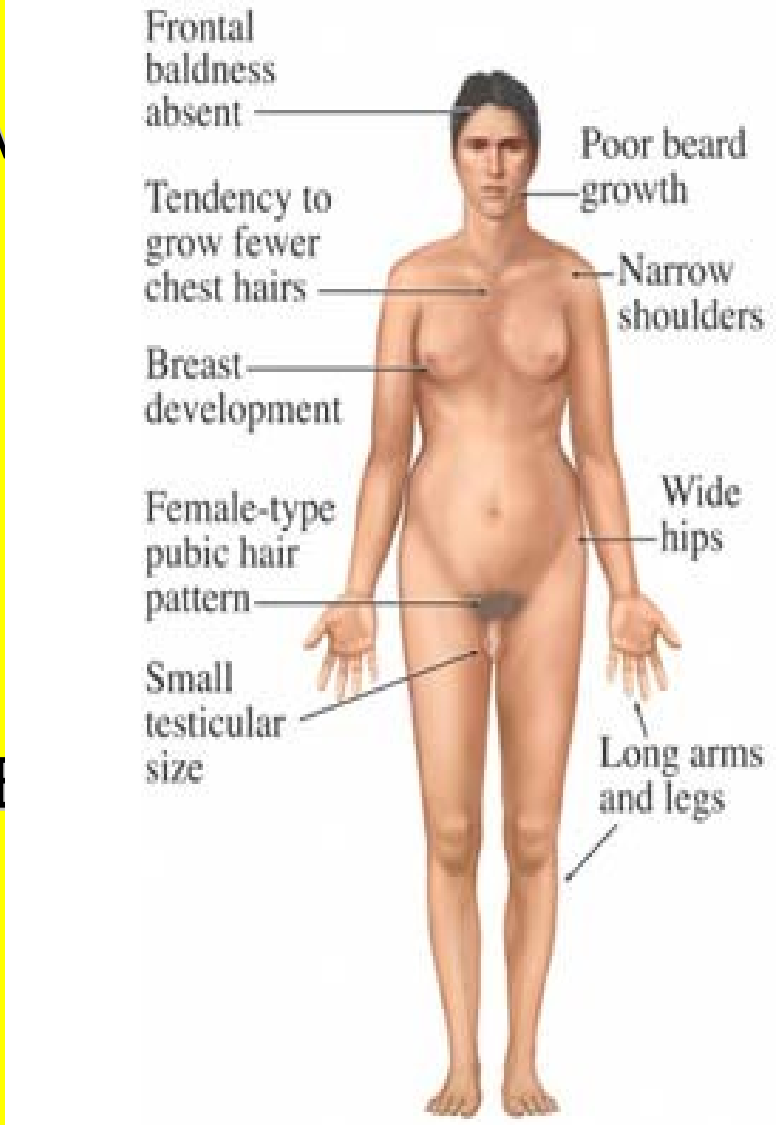
SYNDROMES Caused by SEX Chromosomal (23rd Pair) Abnormalities

- XO TURNER SYNDROM



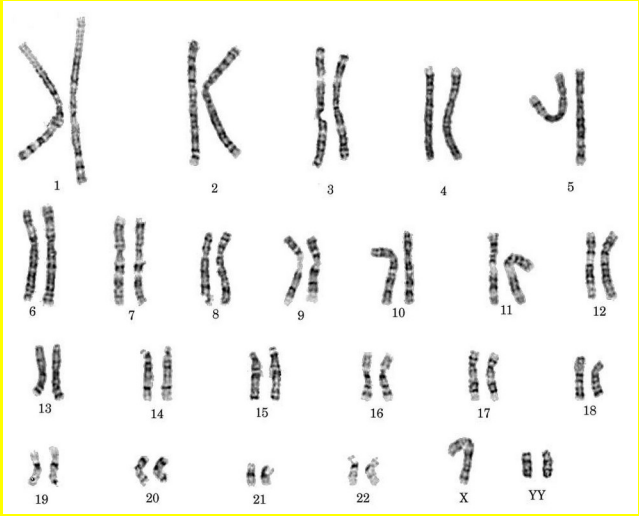


- XXY KLEINFELTER SYNDROME

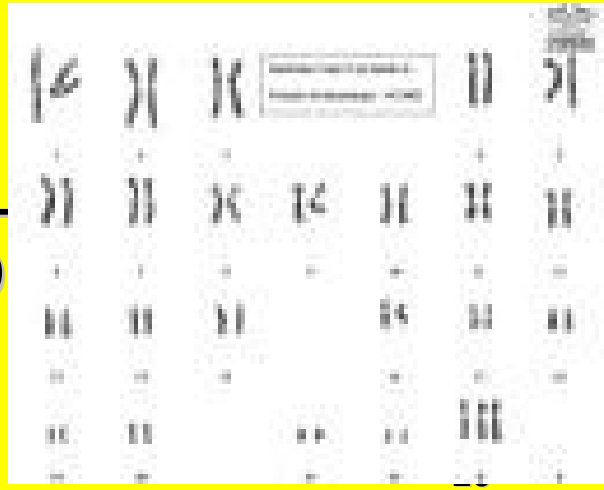


SYNDROMES Caused by SEX Chromosomal (23rd Pair) Abnormalities

- XYY “SUPERMALE” SYNDROME



- XXX, or XXXX “SUPERFEMAL SYNDRO



PREDICTING AND DETECTING GENETIC or CHROMOSOMAL ABNORMALITIES

<u>One option is GENETIC COUNSELING</u>

1. If already have child with abnormalities
2. If have relatives with abnormalities
3. If already had miscarriages
4. If parent age is high
5. If gene pool has had problems (Tay-Sachs in some of Jewish descent; Sickle-cell Anemia in some of African descent)

PREDICTING AND DETECTING GENETIC or CHROMOSOMAL ABNORMALITIES

- Carrier (parent) blood test
- Examples:
  - sickle-cell anemia (African: abnormality of red blood cell production)
  - Tay-Sachs (Jewish: abnormality of chromosome 15)
  - PKU (inability to inability of the body to utilize the essential amino acid, phenylalanine)
  - Hemophilia (inability of the blood to clot)