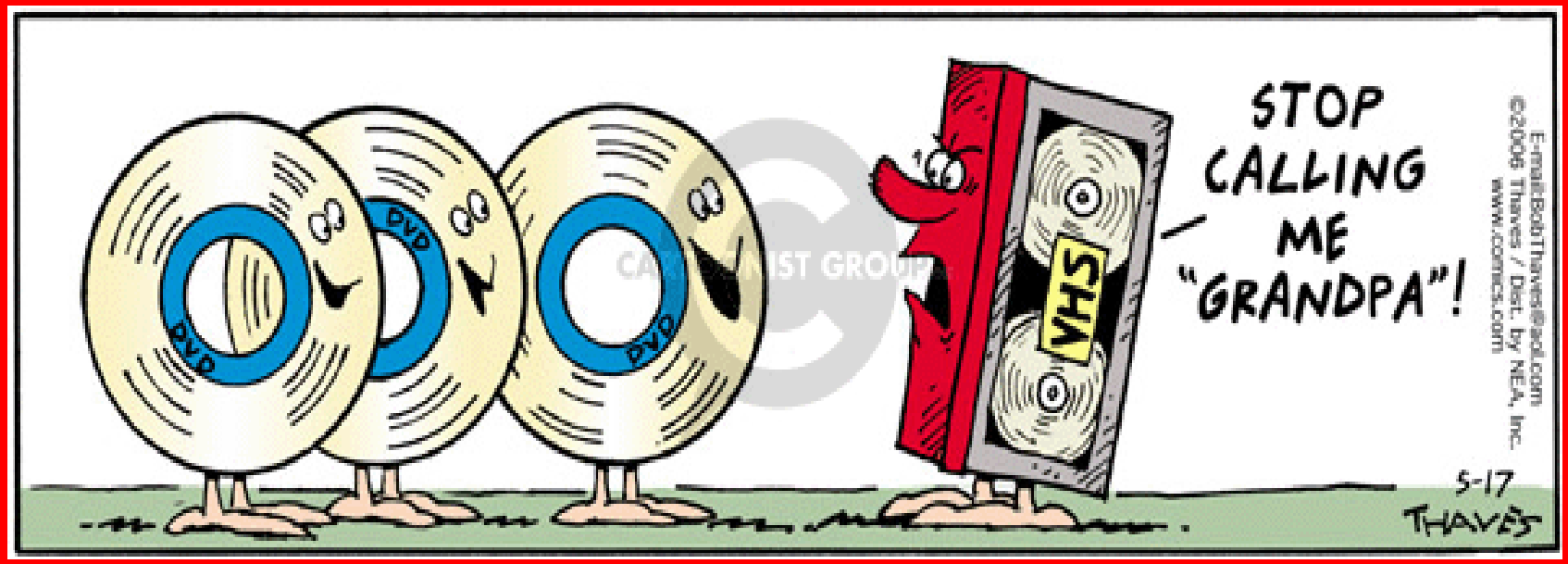
Chapter 14-2

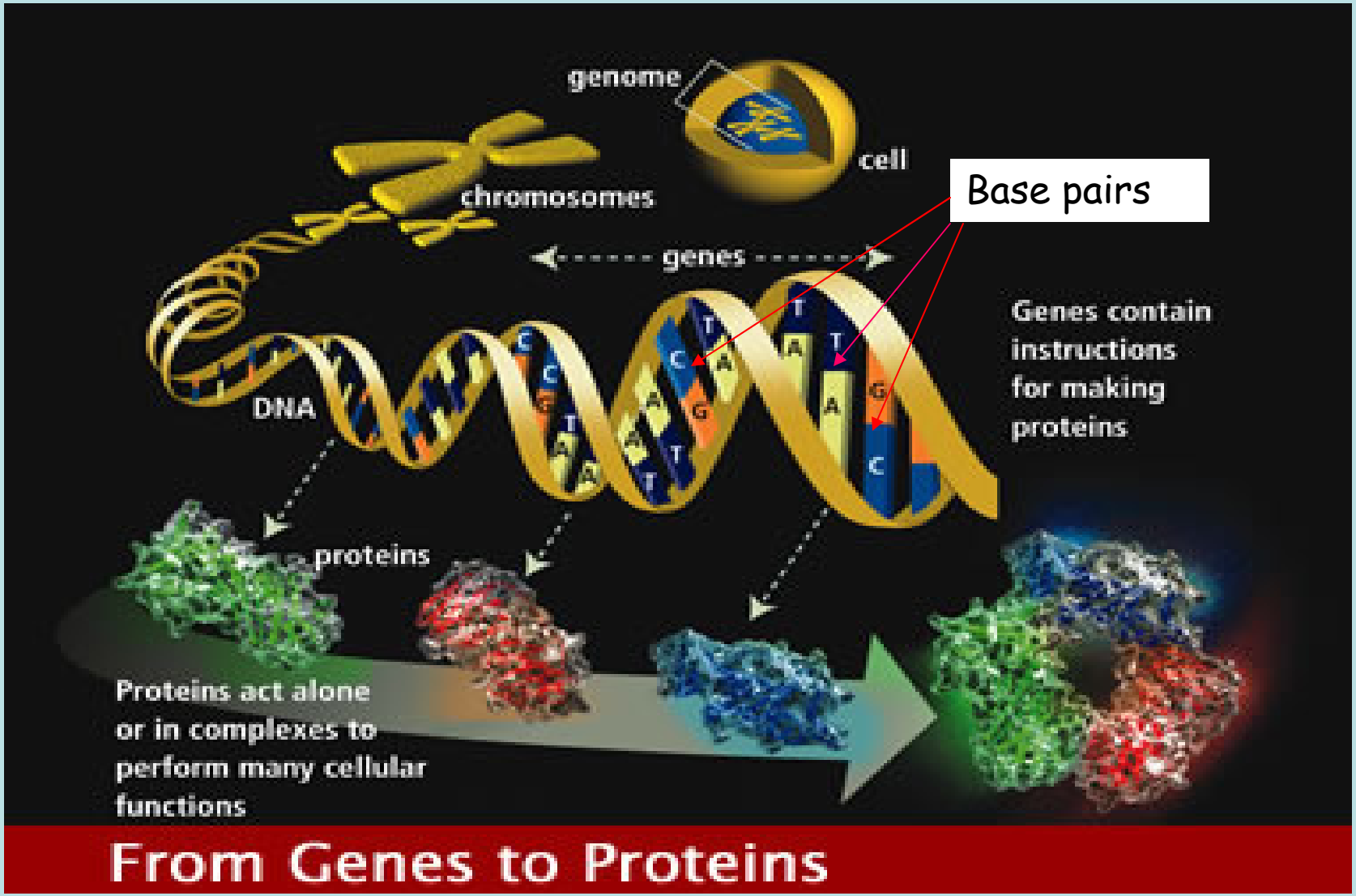
Human Chromosomes



I. Human Genes and Chromosomes

A. Average gene has 3000 nucleotide base pairs

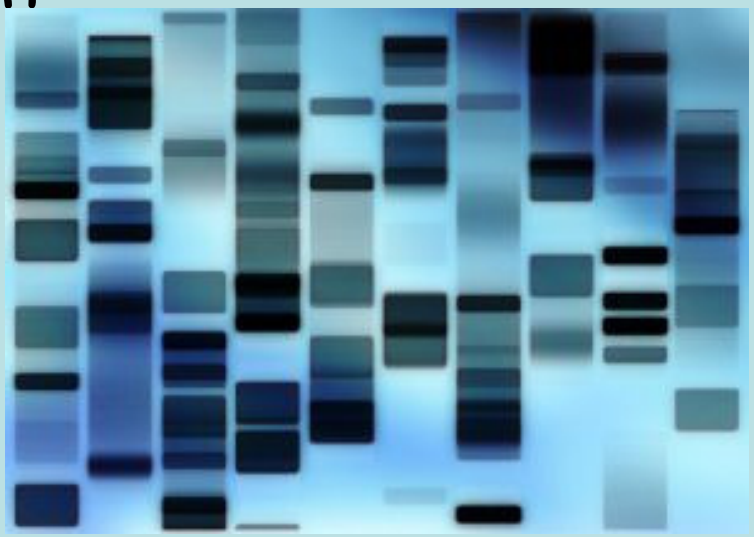
B. Largest more than 2 million nucleotide base pair



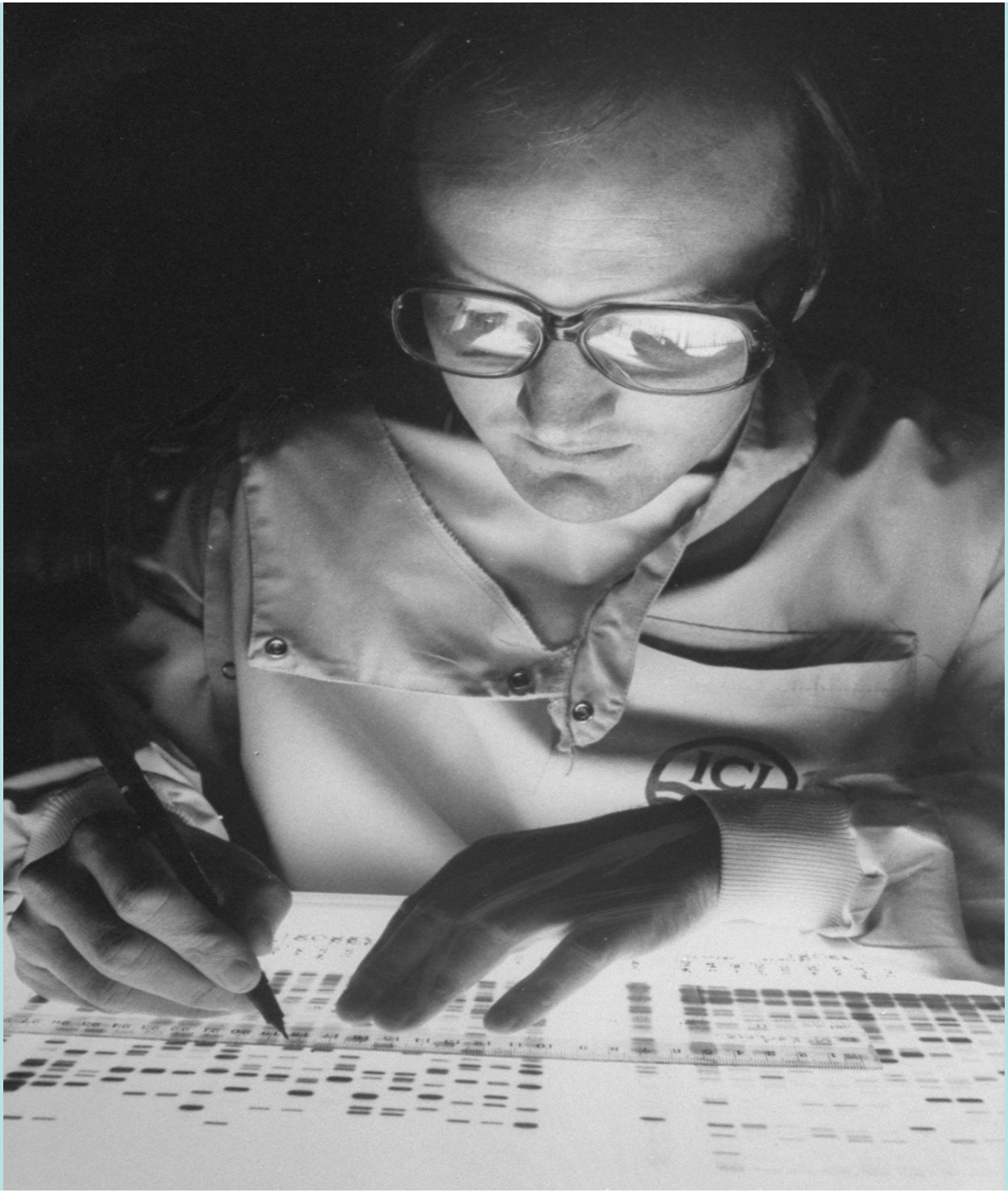
C. Portions of chromosome contain repeating segments of DNA-called "repeats"/"Junk".

1. Repeats are not genes (so they code for NOTHING)

2. "Junk" DNA is key in constructing humans "DNA fingerprint"



Scientist analyzing a DNA fingerprint



II. Human Genes

A. Human blood groups were one of the first identified by biologists.

B. Rh blood group is a single gene with two possible alleles (+ OR -)

1. Rh+/Rh+ = positive blood (ex. A+, B+, AB+ O+)

2. Rh+/Rh- = positive blood (ex. A+, B+, AB+ O+)

3. Rh-/Rh- = negative (ex. A-, B-, AB-, O-)

4. Called Rh factor because it was discovered in Rhesus monkeys ☺

III. Gene Mutations

A. Gene disorders occur when there is a small change in the DNA of ONE gene, this can affect the protein being made.

This **could** cause serious genetic disorders.

1. **Deletion of 3 bases** in a gene on chromosome #7 causes a protein to fold improperly causing cystic fibrosis.

2. **Substitution of a base** in a gene on chromosome #11 produces wrong protein causing low O_2 levels, proteins stick together causing sickle cell anemia.
    a. Individual heterozygous for sickle cell are resistant to malaria.

IV. Autosomal Disorders (mutations of **single genes** on an autosome)

D/R	Name	Symptoms	Chrom#
D	Polydactyly	Extra fingers/toes	Not known
Co	Sickle Cell	Misshapen/"sickled red blood cells	11
D	Huntington's	Mental deterioration, uncontrolled movements. First symptoms around age 30.	17
R	Cystic Fibrosis	nonfunctional enzyme for break down excess mucus in lungs, respiratory infections, cirrhosis of liver	7
R	Tay Sach	Lipid build up in brain, death in early childhood	15
D	Achondroplasia	Form of dwarfism	4
R	Galactosemia	Accumulation of galactose(sugar) mental impairment, liver damage	4
D	Hypercholest-erolemia	Excess cholesterol-heart disease	19

D/R	Name	Symptoms	Chrom#
R	Albinism	Lack of pigment in hair, skin & eyes	15
R	PKU	Accumulation of phenylalanine. Severe mental impairment. Can be prevented with early detection.	12

Ch 14-2, Part 2 Notes

Sex-Linked Disorders

V. Sex Linked Genes- Reproductive and sexual development genes are located on sex chromosomes (X-female) (Y-male)

A. Sex-Linked disorders

1. Carried on the X sex chromosome ONLY. NEVER on Y chromosome.

B. Can be recessive or dominant

1. Recessive sex-linked disorders are expressed in males more often than females

    a. A female requires a recessive allele on BOTH X chromosomes to be affected. If she only has one she is a carrier.

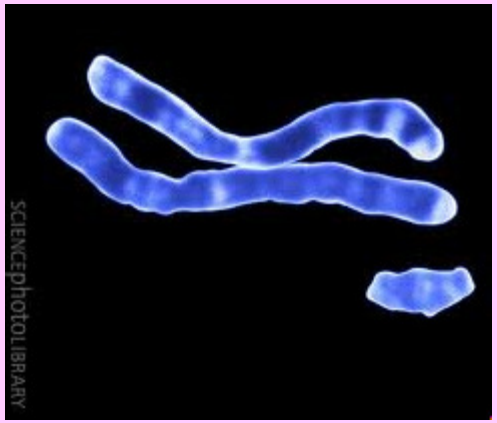
    b. A male only requires one recessive allele, since he only has one X, to be affected

    c. A man can never be a carrier. Either affected or not.

C. Dominant sex –linked disorders are expressed in males and females equally.

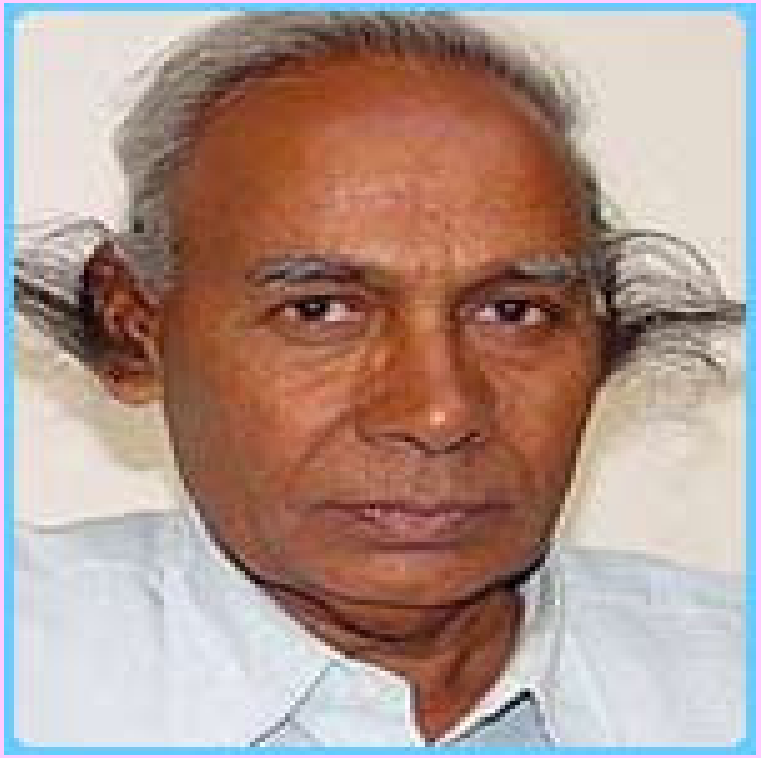
1. If either has at least one dominant allele they are affected.

D. There are more than 100 genes on X chromosome



E. Y chromosome much smaller and has only a few traits like……

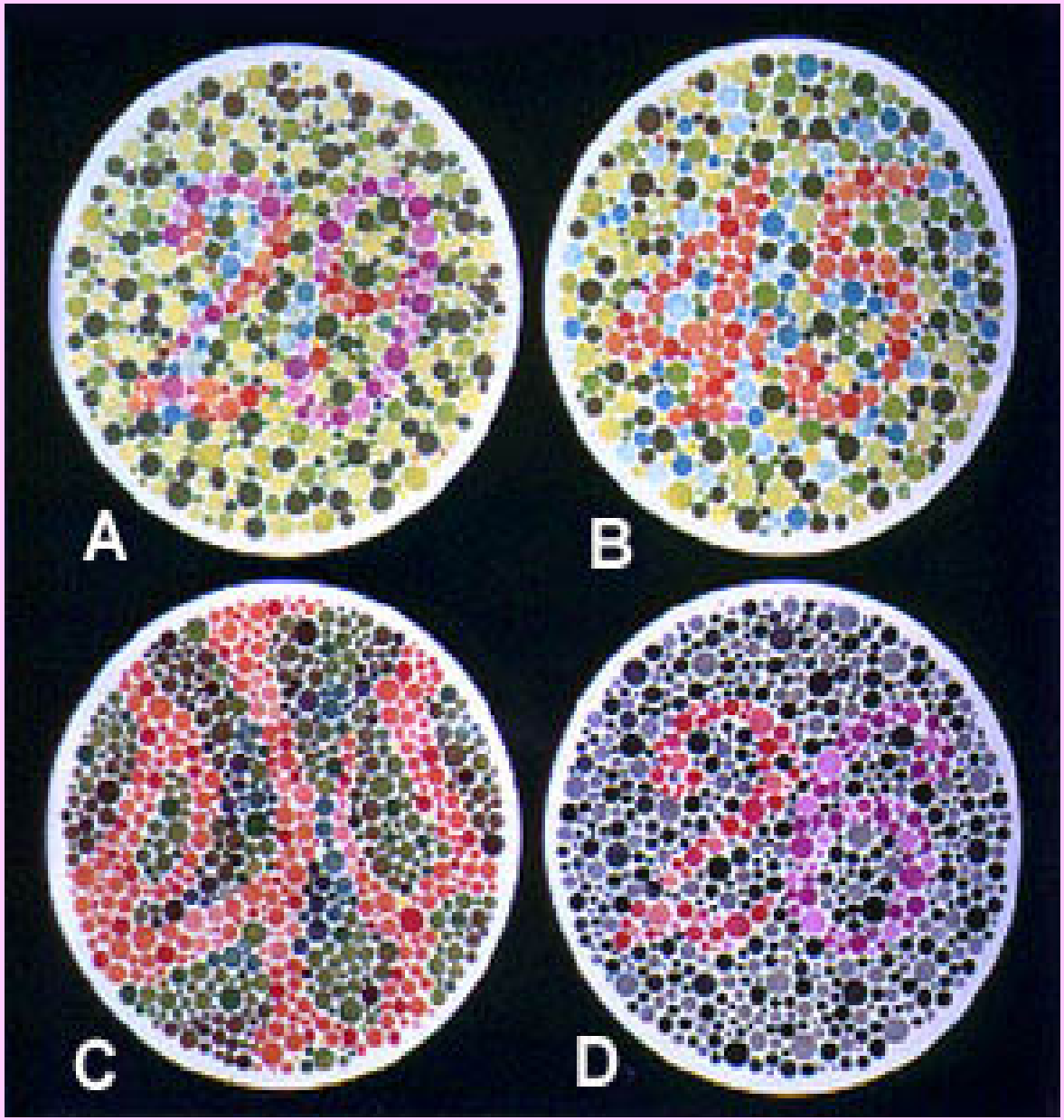
HAIRY EARS!!!! ☺



Hey all you Y chromosome holders,
Look what you have to look
Forward to. At least you won't
Need ear muffs in the winter ☺

VI. Sex Linked Disorders- affect one gene, on the X-sex chromosome.

HEMOPHILIA	Recessive-individual lacks a protein for normal blood clotting. Clotting protein now available. X^hX^h Hemophiliac X^HX^h Carrier X^HX^H Normal clotting X^hY- Hemophiliac X^HY-Normal clotting
COLORBLINDNESS	Recessive- inability to see certain colors. (4 different patterns exist) X^bX^b Colorblind X^BX^b Carrier X^BX^B Normal vision X^bY- Colorblind X^BY-Normal vision



4 Sex-Linked Traits:

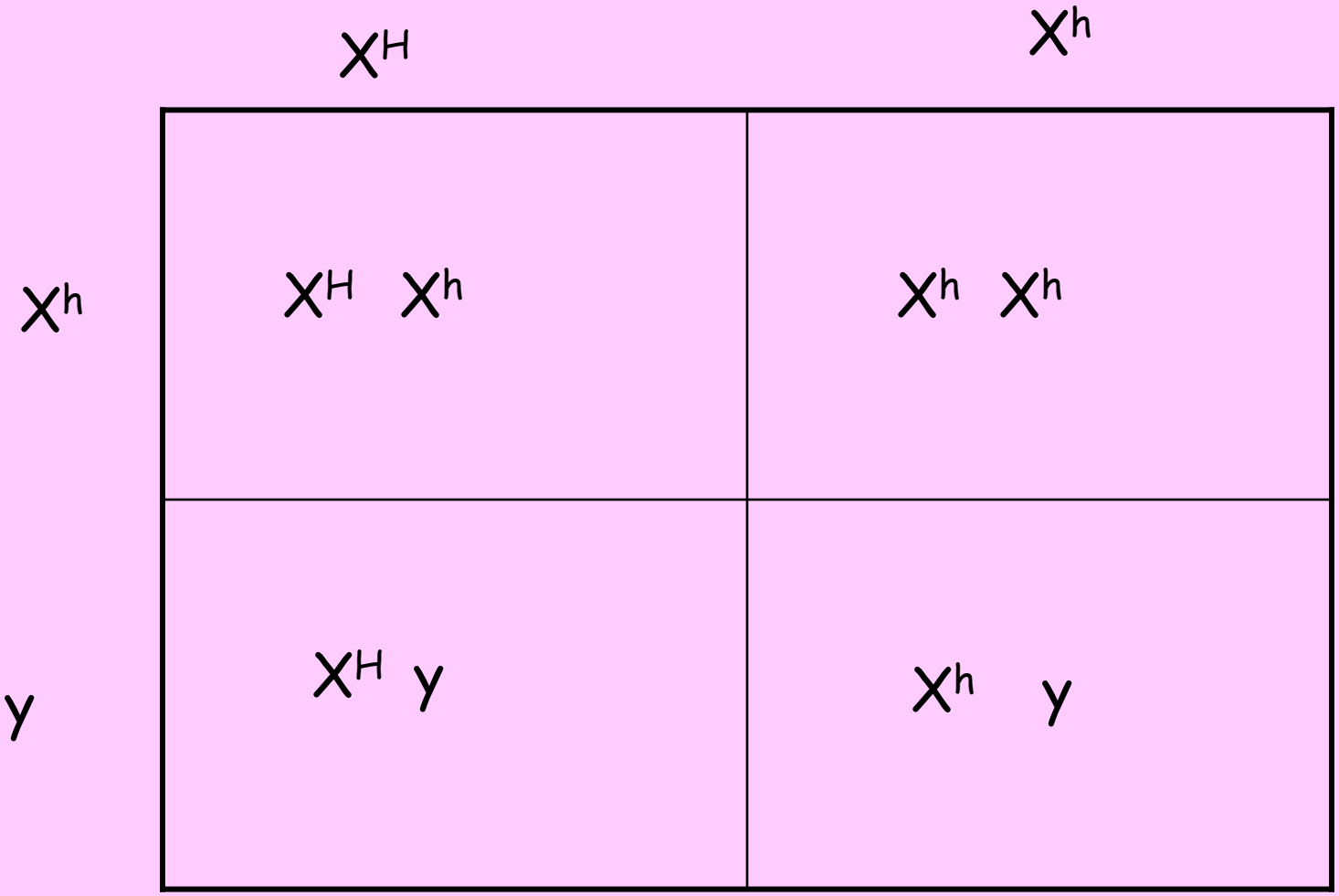
1. Normal Color Vision:
A: 29, B: 45, C: --, D: 26

2. Red-Green Color-Blind:
A: 70, B: --, C: 5, D: --

3. Red Color-blind:
A: 70, B: --, C: 5, D: 6

4. Green Color-Blind:
A: 70, B: --, C: 5, D: 26

Cross a hemophiliac carrier female with a hemophiliac male



Phenotype ratio:

Sex Linked Disorders Cont.

DUCHENNE MUSCULAR DYSTROPHY	Recessive-progressive weakening and loss of skeletal muscle. Occurs later in childhood.
HYPERTRICHOSIS	Dominant-Extreme hairiness on face. X^TX^T Hypertrichosis X^TX^t Hypertrichosis X^tX^t Normal X^TY- Hypertrichosis X^tY-Normal

Lionel- 5 years old- (1907 photograph)

[How stuff works Video: One Step Beyond hypertrichosis]

VII. Chromosomal Disorders- <u>All or parts of</u> (affects more than one gene!)
an autosome or sex chromosome are affected.

Can occur in two ways

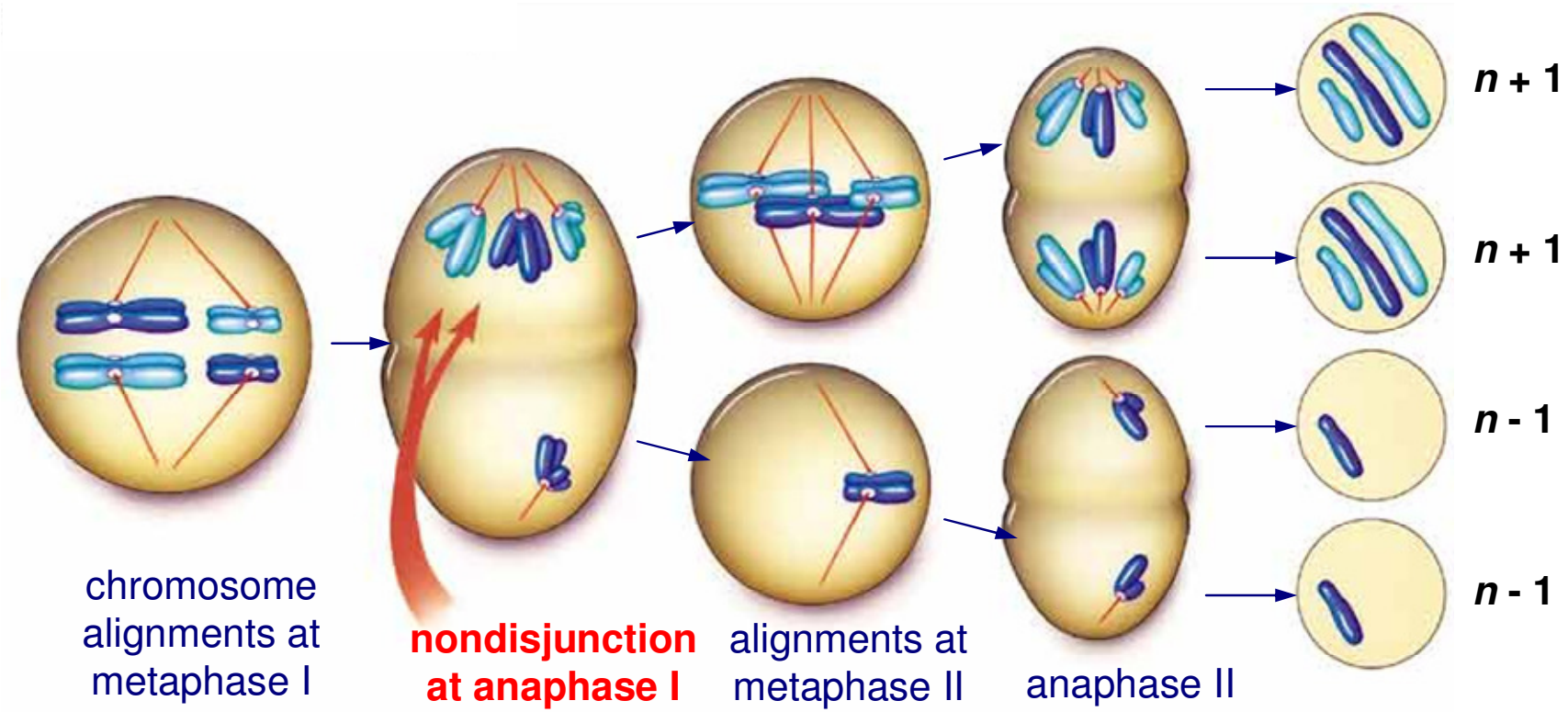
A. Nondisjunction- failure of homologous chromosomes to separate during meiosis results in:

1. <u>Trisomy</u>- three chromosomes at one location instead of 2

2. <u>Mono</u>somy-one chromosome at one location instead of 2

B. Deletions of parts of a chromosome

Nondisjunction



B. **Chromosomal Disorders-** affects all or sections of chromosome

Disorder	Description
Turner's (female)	**Monosomy** of sex chromosome. Genotype **XO**. Results in sterility
Klinefelter's (Male)	**Trisomy** of sex chromosome. Genotype **XXY**. Results in sterility and female characteristics.
Down's Syndrome (M & F)	**Trisomy** of autosome **#21**. Delayed cognitive and physical development. Heart defects.
Edward's Syndrome (M & F)	**Trisomy** on autosome **#18**. Many physical and mental Defects . Death in 1st year of life.
Patau Syndrome (M & F)	**Trisomy** of autosome **#13**. Many physical and mental defects. Death in 1st year of life.
Cri-du-chat (M & F)	**Deletion** of autosome **#5**. Physical and mental defects. Affects larynx. Baby's cry sounds like a cat, feeding problems, poor growth, hyperactivity.

Chromosomal Disorders Cont…

Triple X (XXX) Female	No unusual physical features. Usually taller and thinner than average. Higher risk of learning disabilities. In rare cases, severe mental impairment
Jacobs Syndrome (XYY) male	Learning problems. Tall, acne, aggressive, delayed emotional development.

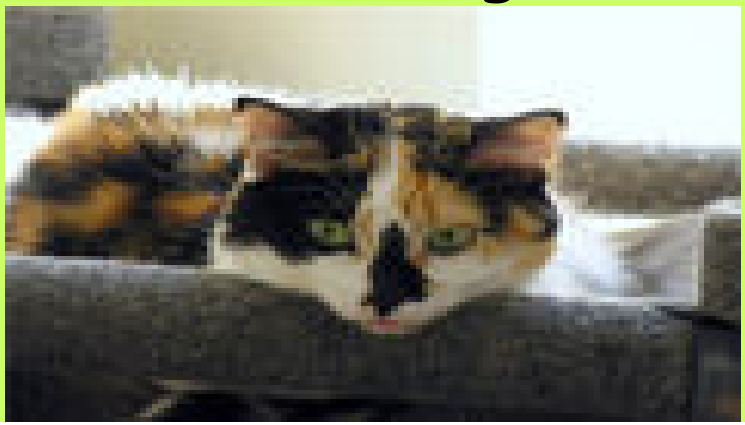
VIII. Chromosome Inactivation

A. In females: one of the X chromosomes is randomly turned off, so genes are not duplicated.

B. Turned off X, forms Barr body(appears as a dark spot in nucleus)

C. Same process in other mammals

1. Calico cats are always female!
2. The 3 colors in cat's fur result from different X's turning off forming Barr body.



Blood Type	Antigens (proteins on outside of cell that let's your cells know if they belong)
A (AA or AO)	Has A antigens on cell (will fight any blood cells with B antigens)
B (BB orBO)	Has B antigens on cell (will fight any blood cells with A antigens)
AB (AB)	Has both A and B antigens on cell. Will not fight any blood cells. ***Universal Acceptor***
O (OO)	Has no antigens. Will fight blood with A or B antigens. **Universal donor**