Introduction to Genetics

What is genetics?

- The scientific study of heredity the process in which a parent passes certain **genes** onto their children."

What does that mean?

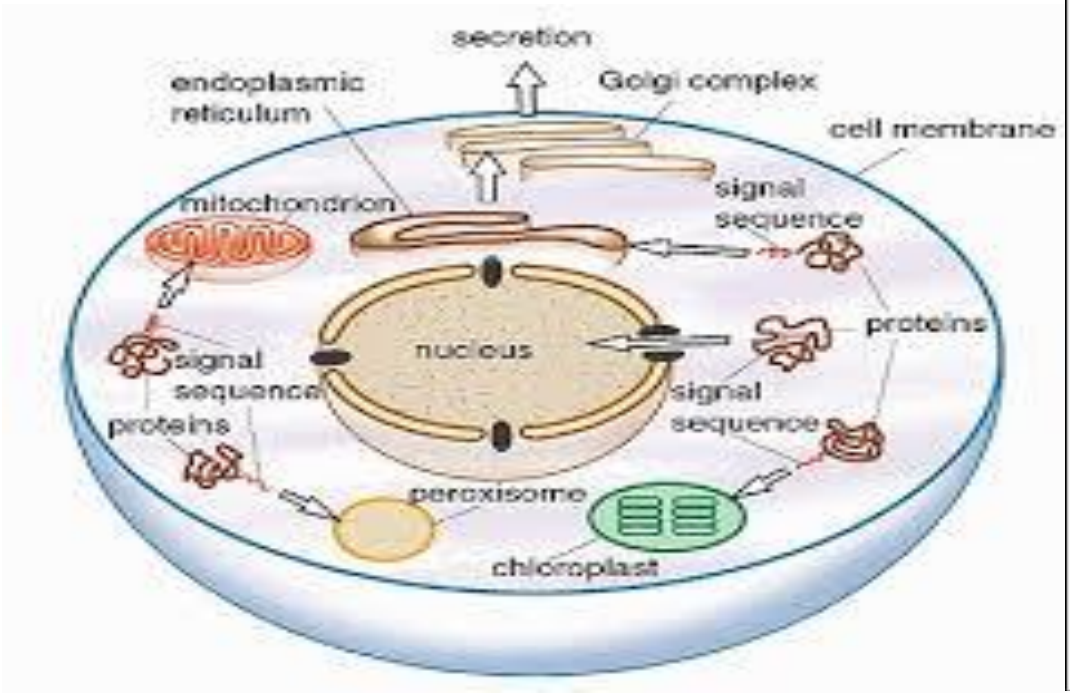
Children **inherit** their biological parents' genes that express specific **traits**, such as some physical characteristics and genetic disorders.

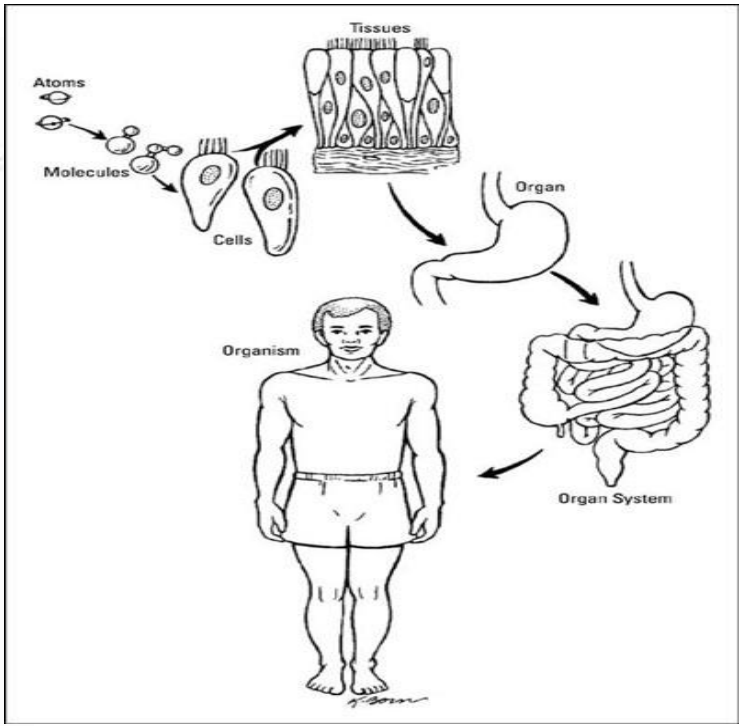
Table 1.1 Early concepts of heredity

Concept	Proposed	Correct or Incorrect
Pangenesis	Genetic information travels from different parts of the body to reproductive organs.	Incorrect
Inheritance of acquired characteristics	Acquired traits become incorporated into hereditary information.	Incorrect
Preformationism	Miniature organism resides in sex cells, and all traits are inherited from one parent.	Incorrect
Blending inheritance	Genes blend and mix.	Incorrect
Germ-plasm theory	All cells contain a complete set of genetic information.	Correct
Cell theory	All life is composed of cells, and cells arise only from cells.	Correct
Mendelian inheritance	Traits are inherited in accord with defined principles.	Correct

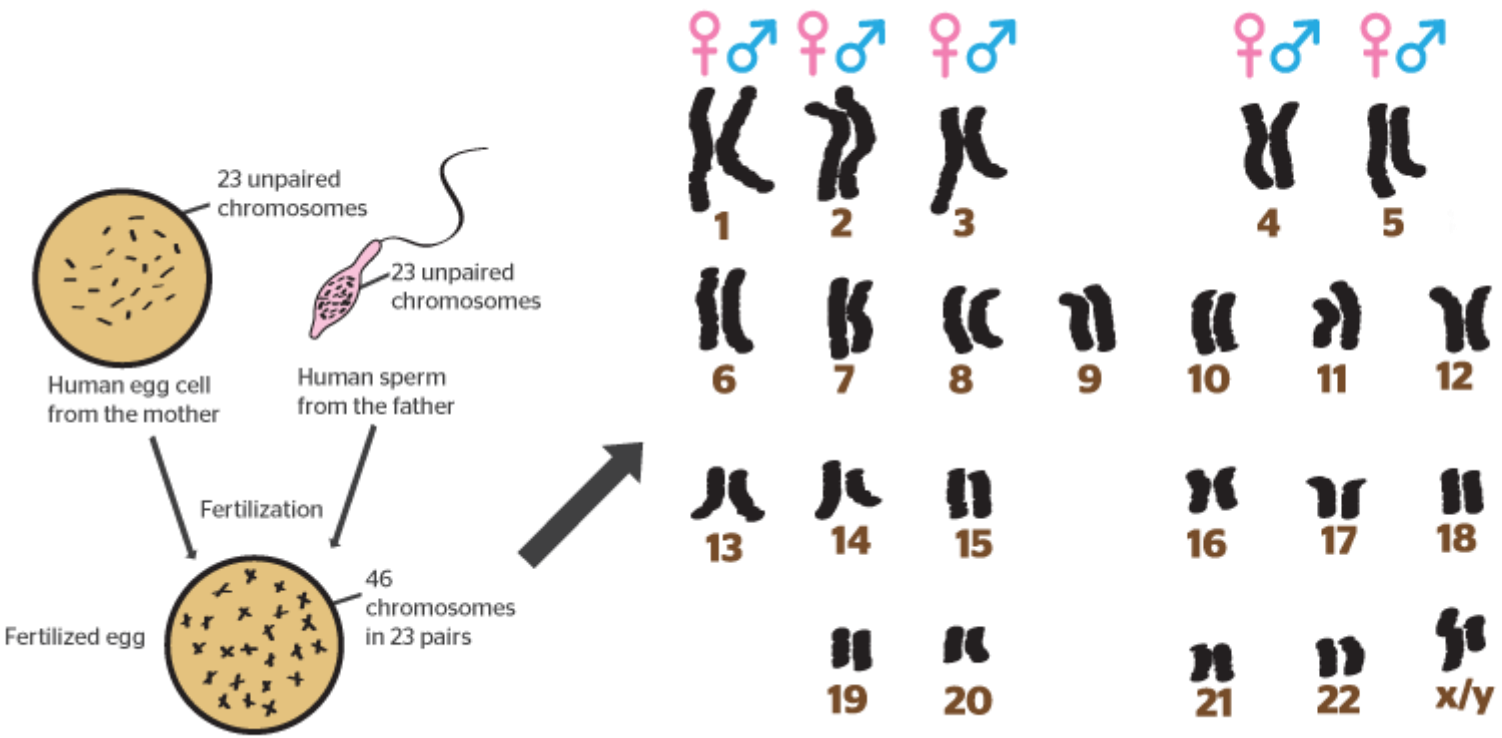
The cell: the smallest unit of life

All life is composed of cells, and cells arise only from cells

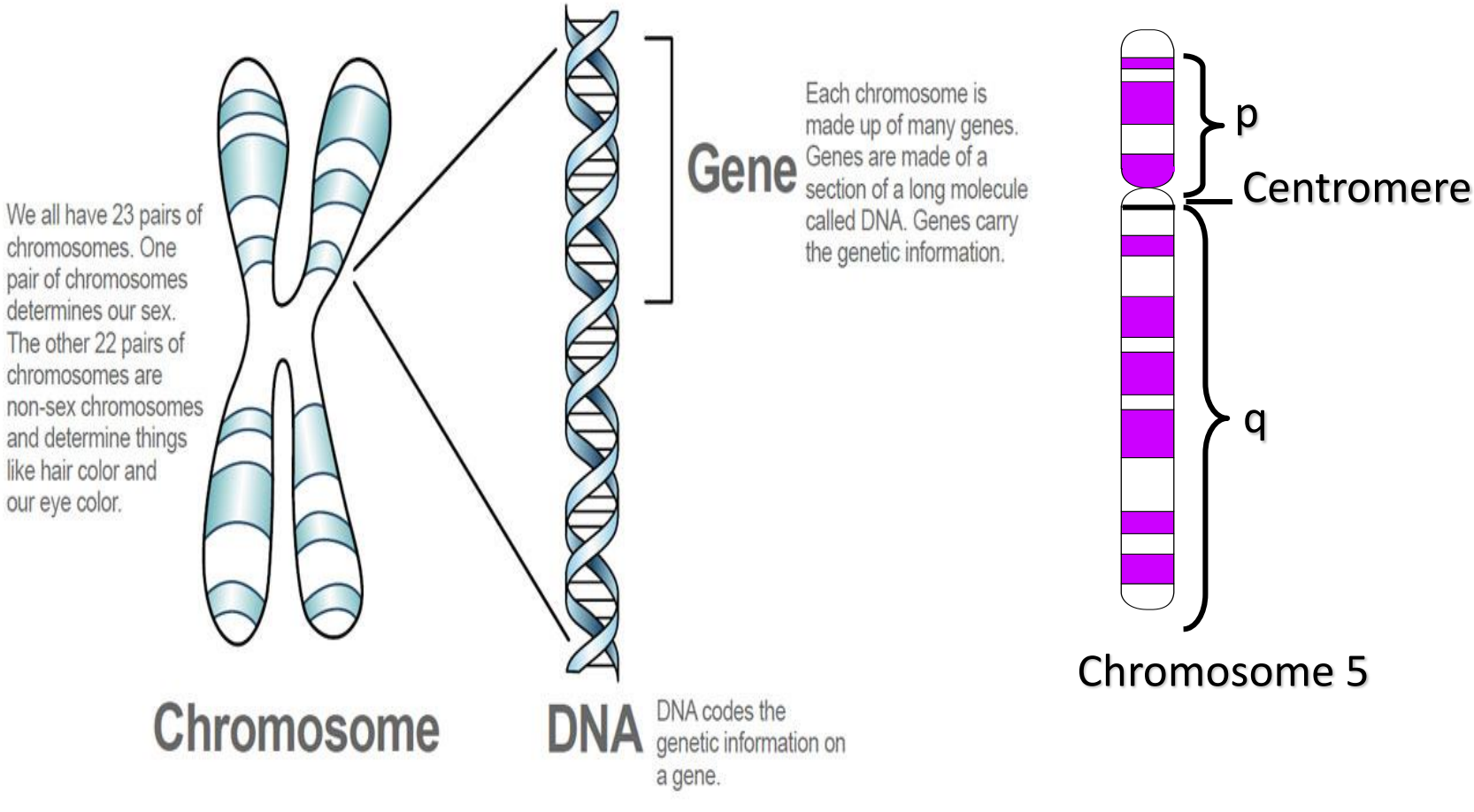




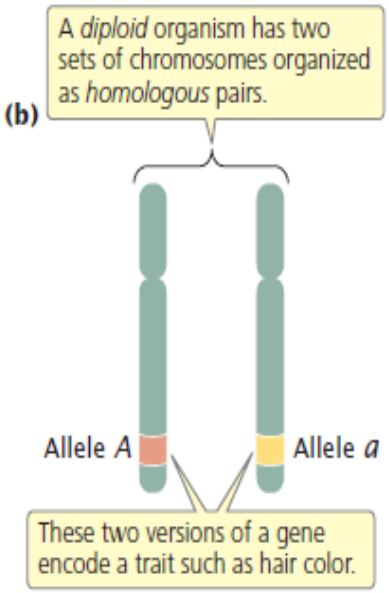
How many chromosome do we have?

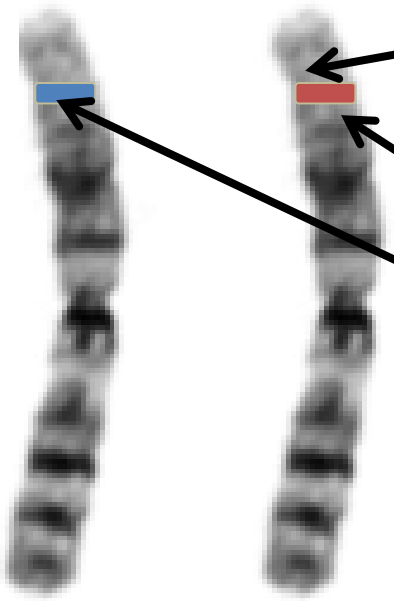


Chromosome number and morphology



A pair of homologous chromosomes

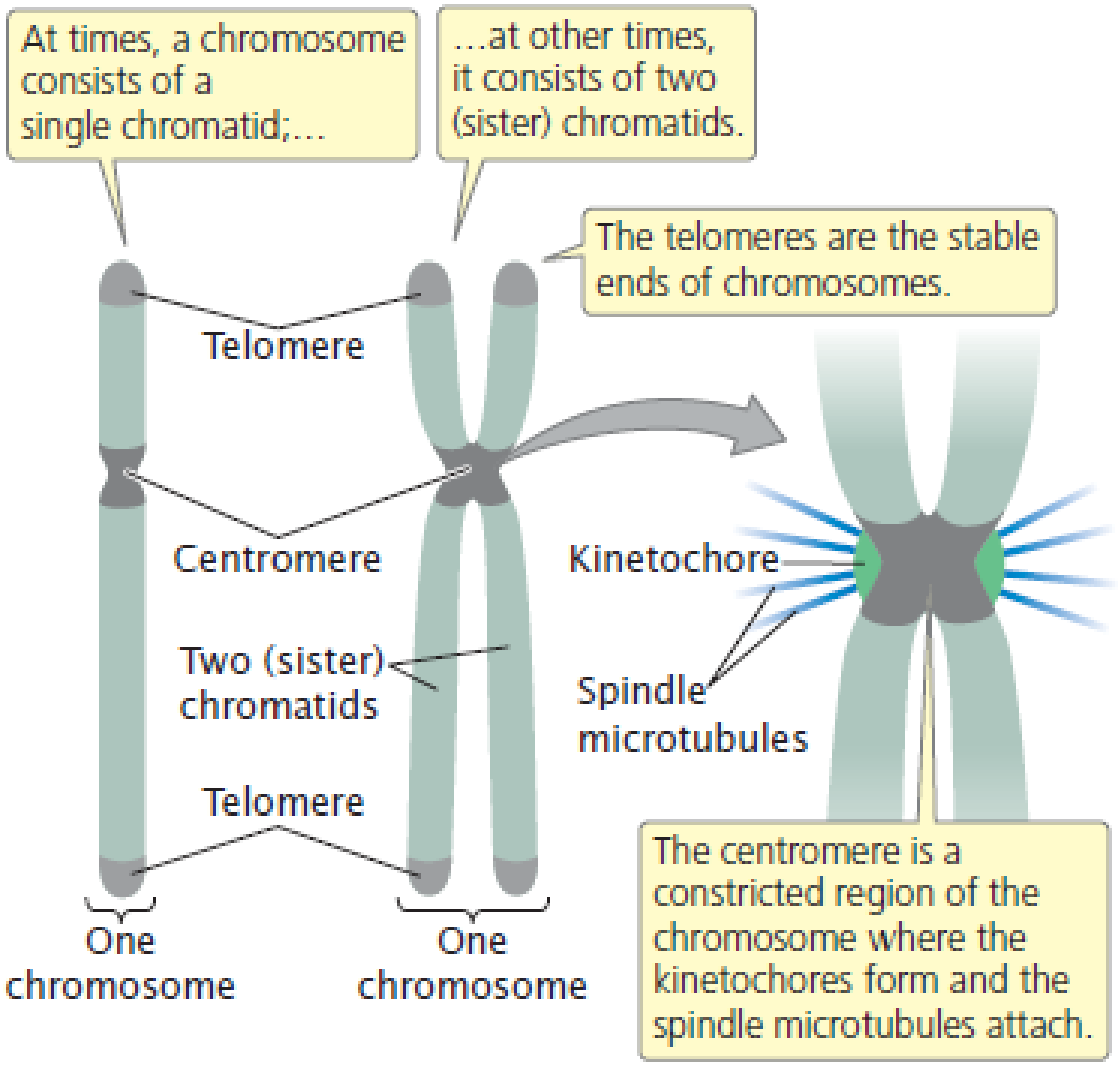




Locus (position of a gene)

Allele (alternative form of a gene)

A functional chromosome

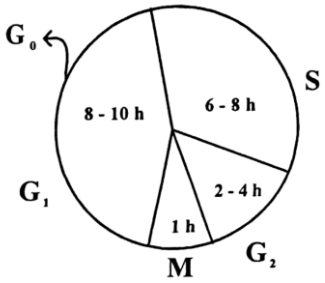


What is a Karyotype?

A display or photomicrograph of an individual's somatic-cell metaphase chromosomes that are arranged in a standard sequence (usually based on number, size, and type)

Preparing a karyotype

Cells are first cultured



Dividing cells

Colcemide

arrests the cells in metaphase

Harvest

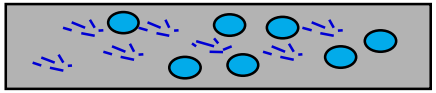
Hypotonic treatment

0.075 M KCl

Fixation

(M:A, 3:1)

Giemsa staining



Chromosomes can be photographed

Karyotype

How Do Scientists Identify Chromosomes?

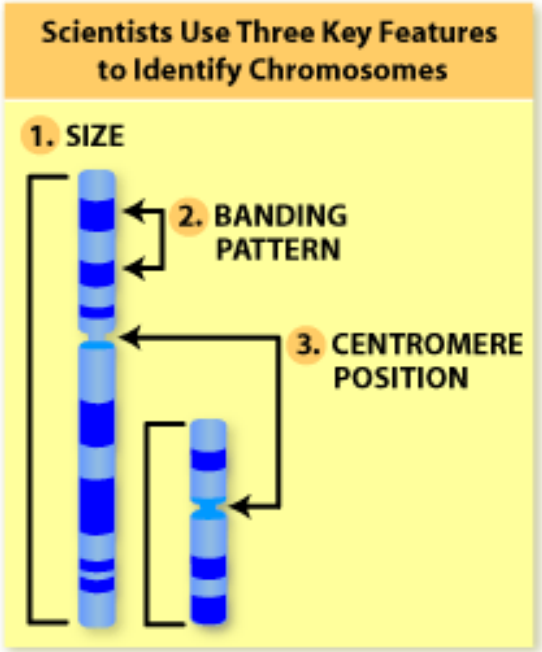
Three key features to identify their similarities and differences:

Size. This is the easiest way to tell two different chromosomes apart.

Banding pattern. The size and location of Giemsa bands on chromosomes make each chromosome pair unique.

Centromere position. Centromeres are regions in chromosomes that appear as a constriction.

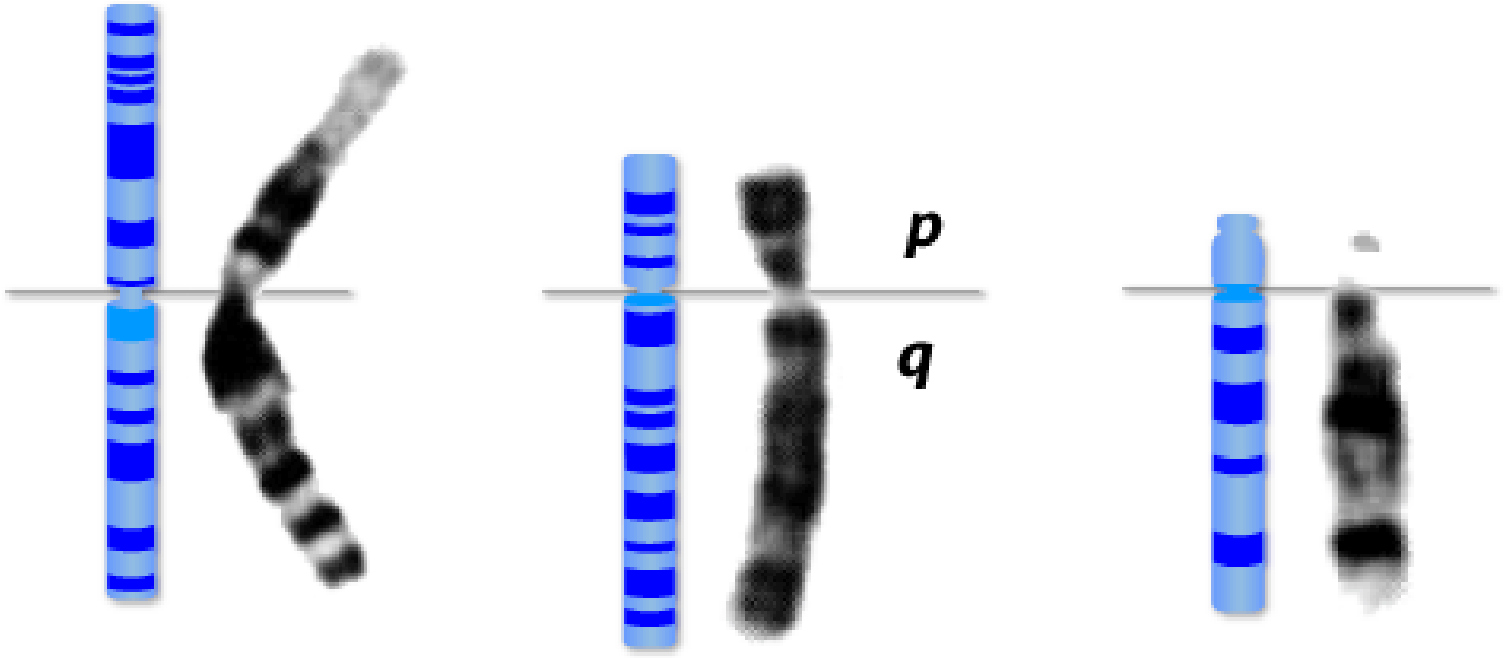
Using these key features, scientists match up the 23 pairs



In metacentric chromosomes, the centromere lies near the center of the chromosome.

Submetacentric, have a centromere that is off-center, so that one chromosome arm is longer than the other.

In acrocentric chromosomes, the centromere resides very near one end. In telocentric chromosome the centromere resides at end.

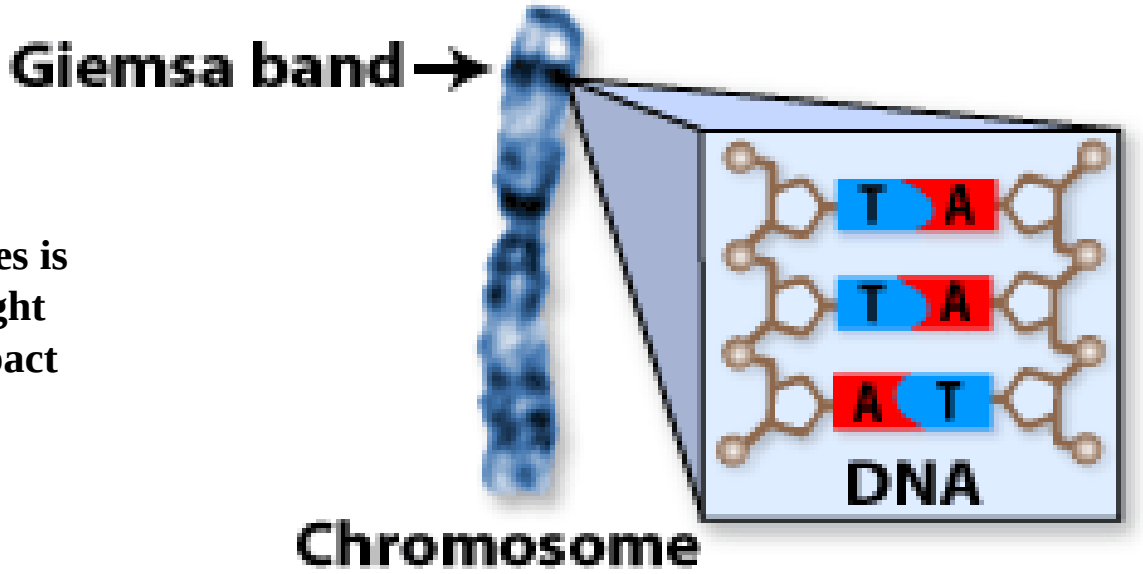


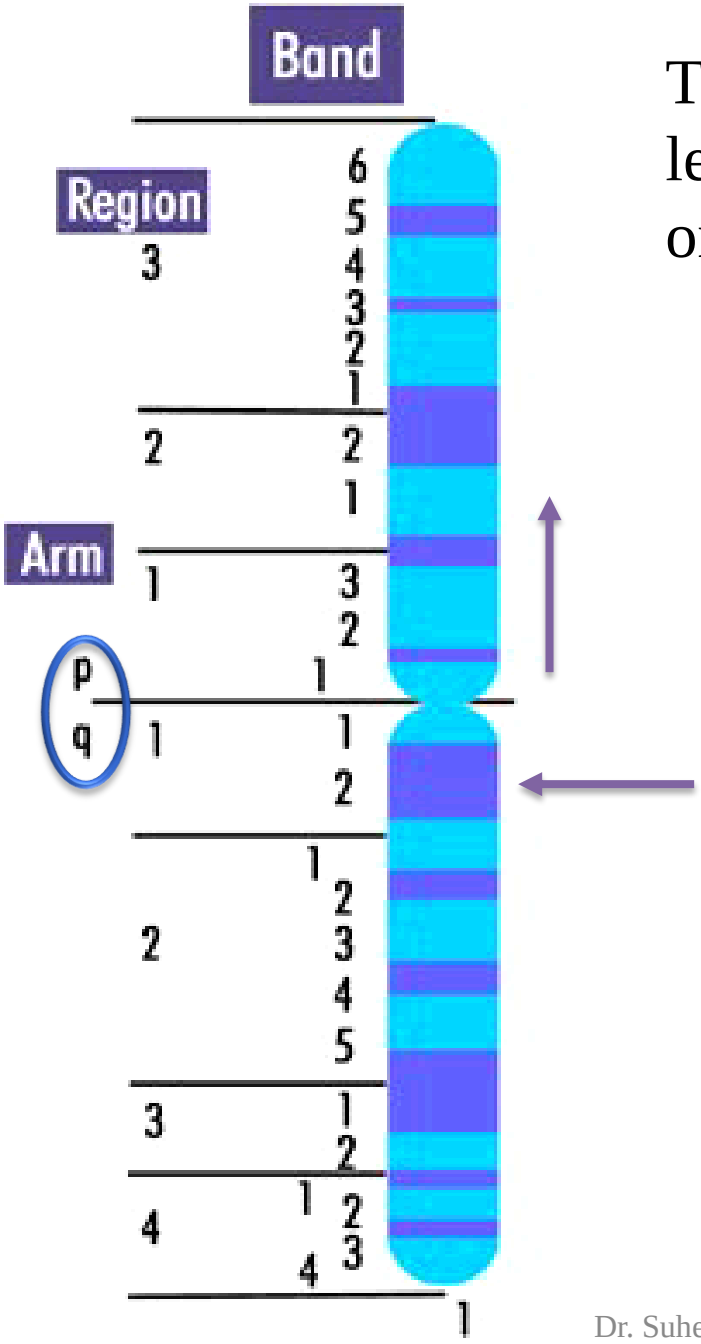
(13,14,15,21,22

G-Banding

Dye gives chromosomes a striped appearance because it stains the regions of DNA that are rich in adenine (A) and thymine (T) base pairs.

The of active genes is higher in the G-light regions=less compact





The combination of numbers and letters provide a gene's "address" on a chromosome

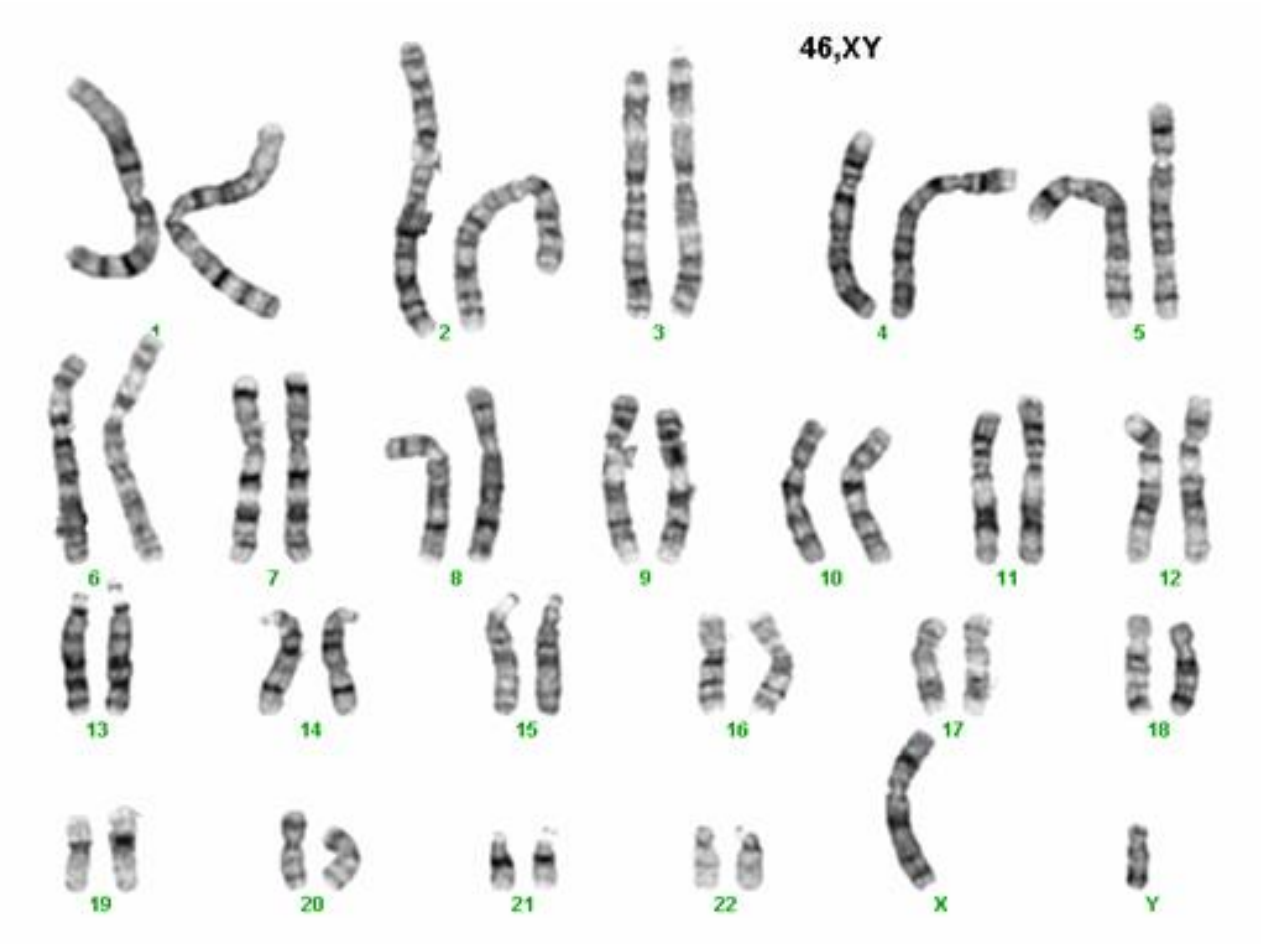
q12

Example: 14q21 represents position 21 on the long arm of chromosome 14. 14q21 is closer to the centromere than 14q22

Organize the chromosomes into a karyotype!

1	2	3	4	5
6	7	8	9	10
11	12	13	14	15
16	17	18	19	20
21	**22**	**x**	**y**	

Karyotype: Autosomes Vs sex chromosome



The Karyotype

A normal male chromosome pattern would be described as:

46,XY.

46 = total number of chromosomes
XY = sex chromosome constitution
(XY = male, XX = female).

Any further description would refer to any abnormalities or variants found

Indications for a karyotype

- Problems of early growth and development: **failure to thrive**, **developmental delay**, short stature
- Stillbirth and neonatal death
- **Fertility problems**: couples with a history of infertility or multiple pregnancy loss
- **Family history:** a known/suspected chr. abnormality in a first degree relative
- **Pregnancy in a woman of advanced age (>35 yrs)**

Genetic Concepts

- **Heredity** describes how some traits are passed from parents to their children.
- The traits are expressed by **genes**, which are small sections of DNA that are coded for specific traits.
- Genes are found on **chromosomes**.
- Humans have two sets of **23** chromosomes—one set from each parent.

Phenotype vs genotype

- **Genotype**
  - The genetic makeup
  - Symbolized with letters
  - Tt or TT, tt
  - Heterozygous
  - Homozygous

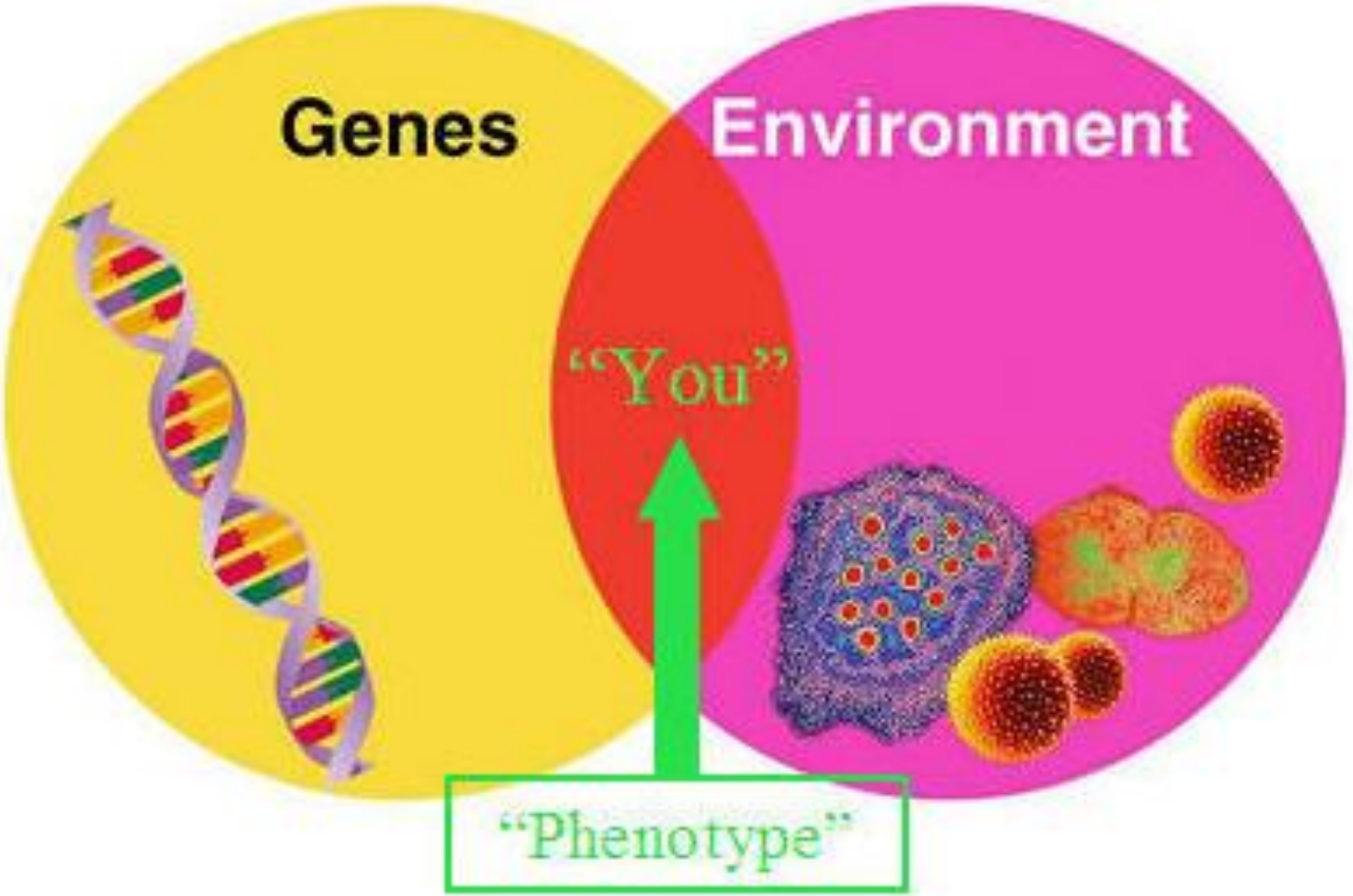
- **Phenotype**
- Physical appearance of the organism
- Expression of the trait
- shape, size, color, and behavior, ..Short, tall, yellow, smooth, etc.

- Many phenotypes are influenced by the environment

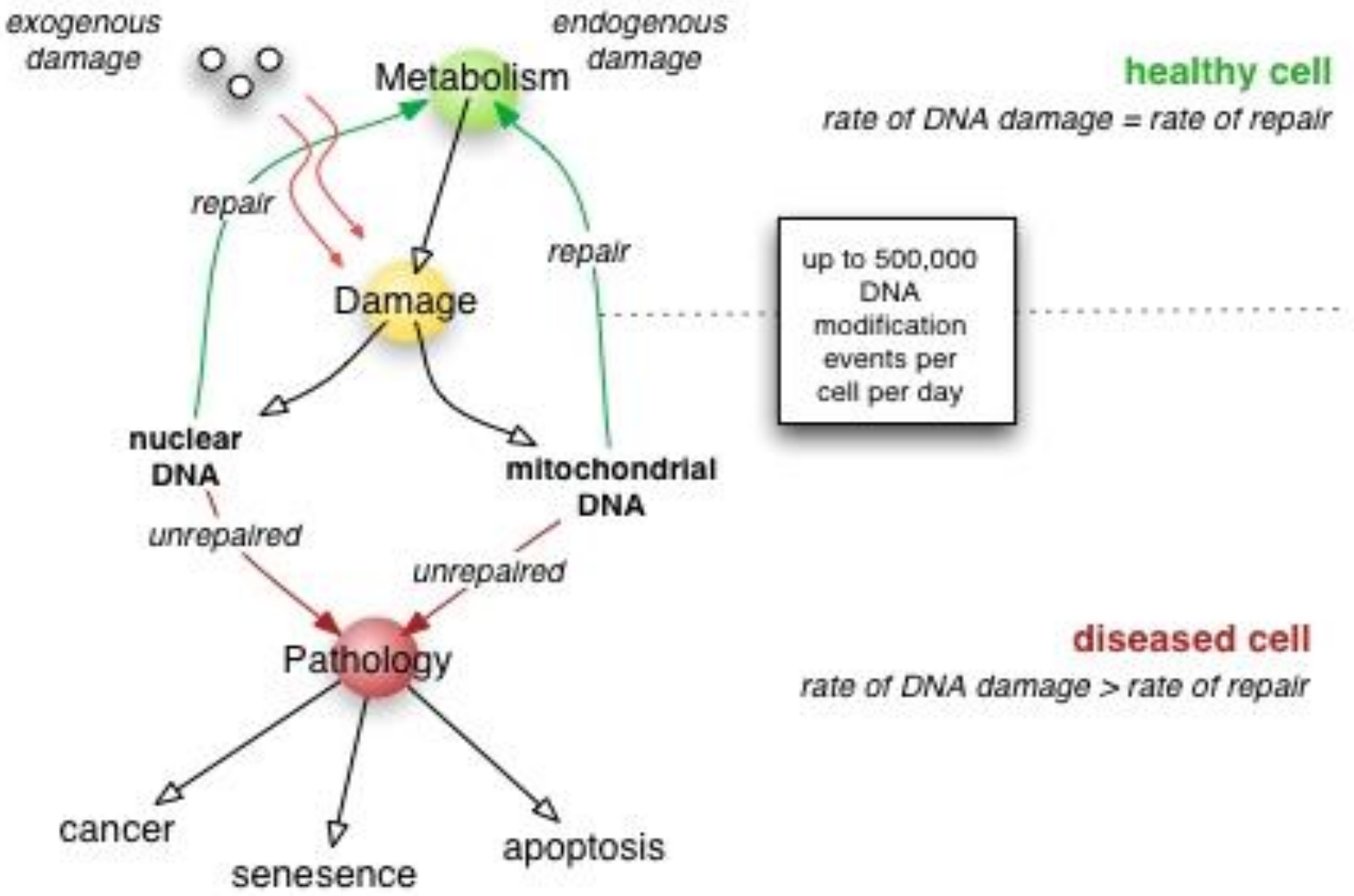
Phenotype = result from the interaction of its – genotype (total genetic makeup) with the environment.

The most common phenotype in a natural population is referred to:

Wildtype


Genes
Environment
"You"
"Phenotype"

Smoking
Radiation
viruses
Sun exposure
chemicals



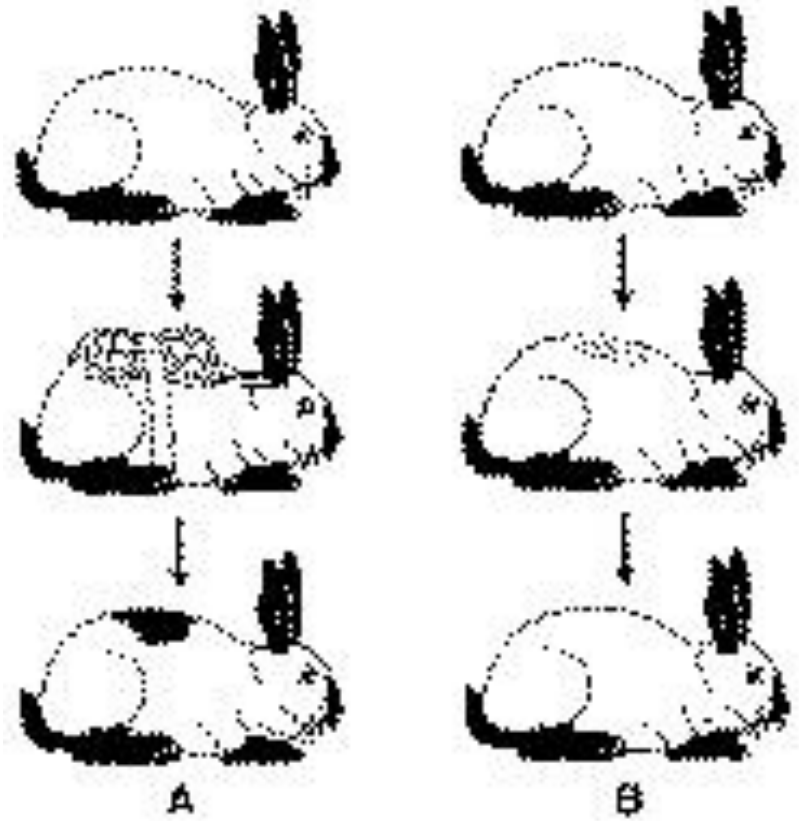
The Himalayan Rabbit

- This rabbit has white fur with black fur on its ears, nose and tail

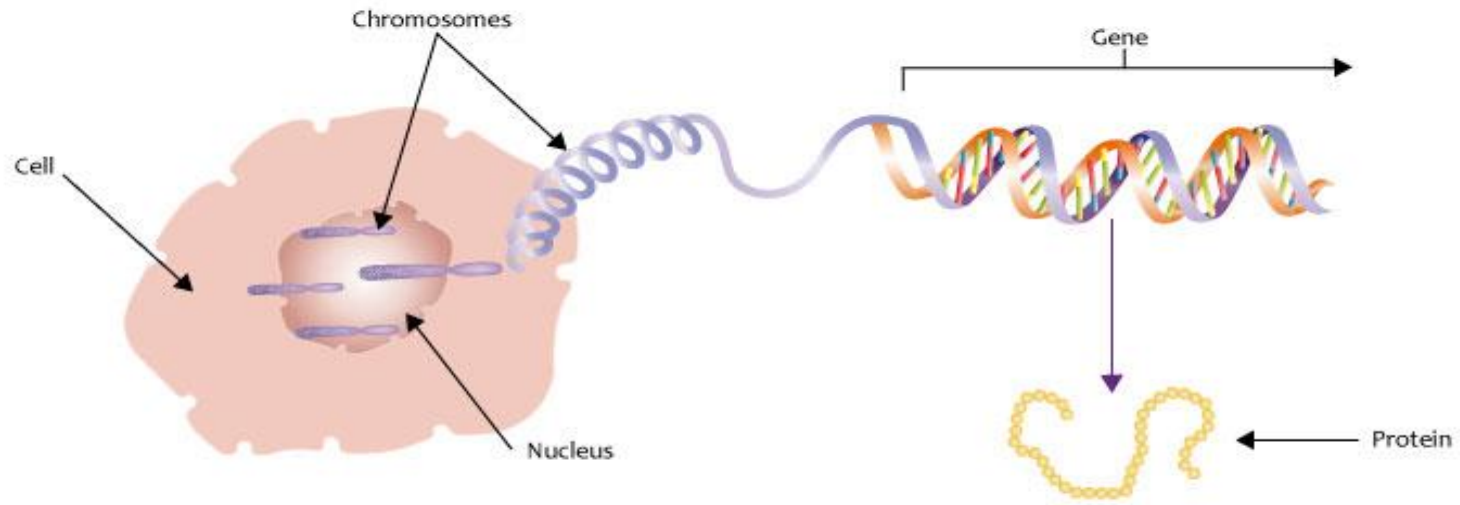
the Himalayan Rabbits carry “temperature sensitive tyrosinase genes” which controls fur pigmentation

The extremities are usually coldest due to less blood flow, therefore the temperature-induced gene is activated for these areas, producing a darker fur.

- Black pigment is deposited in fur when the temperature falls below 33^0C
- When hair is shaved and an ice pack is placed in the area, the new fur will grow in black



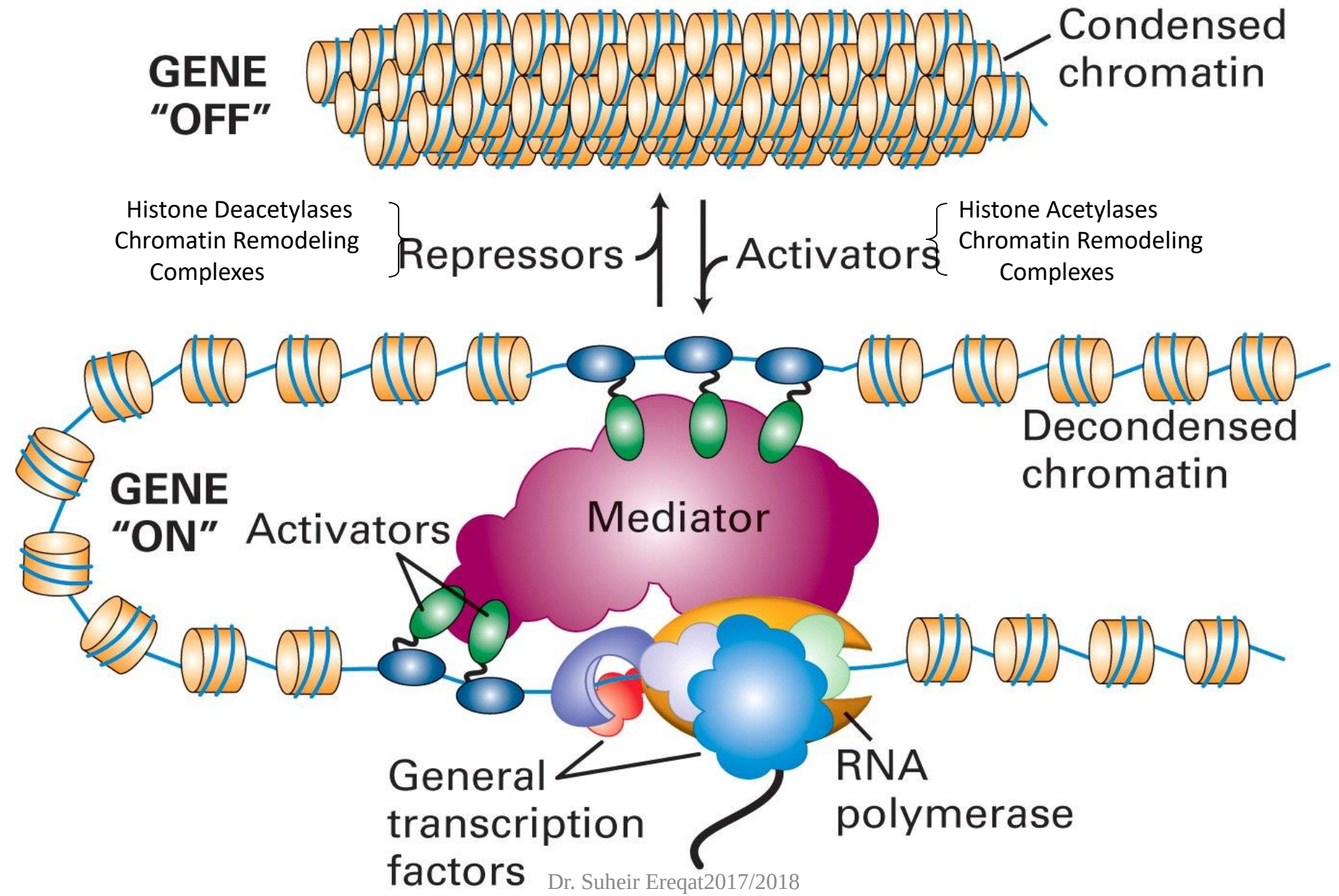
The gene: the hereditary unit that is transmitted from generation to next



What is the relationship between genes and traits?

Genes → Protein → Traits

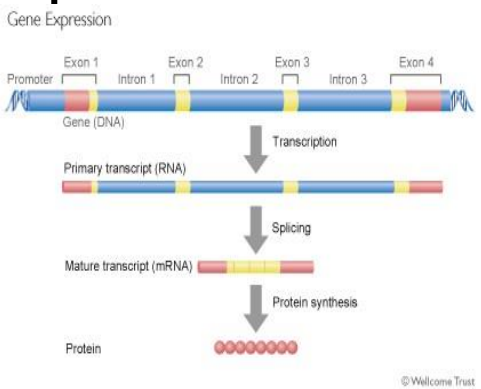
Each cell expresses, or turns on, only a fraction of its genes. The rest of the genes are repressed, or turned off. The process of turning genes on and off is known as **gene regulation**


GENE
"OFF"
Condensed
chromatin
Histone Deacetylases
Chromatin Remodeling
Complexes
Repressors
Activators
Histone Acetylases
Chromatin Remodeling
Complexes
Decondensed
chromatin
GENE
"ON"
Activators
Mediator
General
transcription
factors
RNA
polymerase

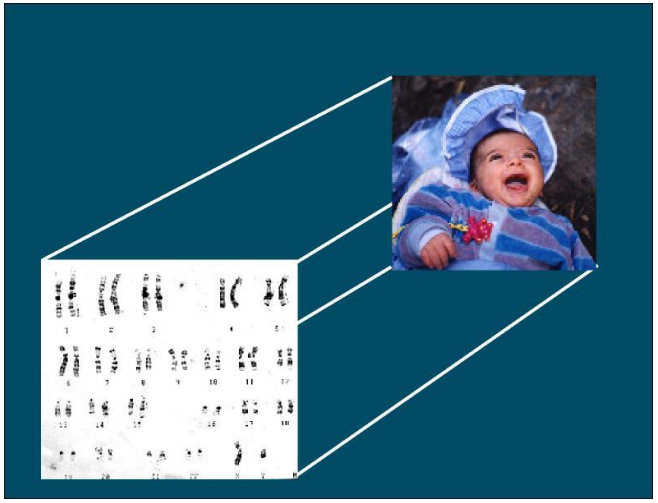
How many genes do we have ?

The answer to this question is almost meaningless because:

- Each gene can give rise to several proteins by alternative splicing
- And each protein can be modified in multiple ways by phosphorylation, methylation, acetylation, glycosylation etc.
- These modified proteins can further take part in different **protein complexes**.



All the cells in the organism have the same DNA



All genes in the human genome are not expressed in the same way!

Epigenetics

Heritable changes in gene expression that operate outside of changes in DNA itself

Glossary

Table 3.1 Summary of important genetic terms

Term	Definition
Gene	A genetic factor (region of DNA) that helps determine a characteristic
Allele	One of two or more alternate forms of a gene
Locus	Specific place on a chromosome occupied by an allele
Genotype	Set of alleles possessed by an individual organism
Heterozygote	An individual organism possessing two different alleles at a locus
Homozygote	An individual organism possessing two of the same alleles at a locus
Phenotype or trait	The appearance or manifestation of a character