**Mutations - changes in the genetic code**

Point mutation: a single point in the sequence is changed. For instance a G is changed to an A.

Compare this to reading a sentence

Cats eat big rat. --> point mutation --> Cats eat big hat

The sentence is still almost recognizeable, the protein changed in this case might still function.

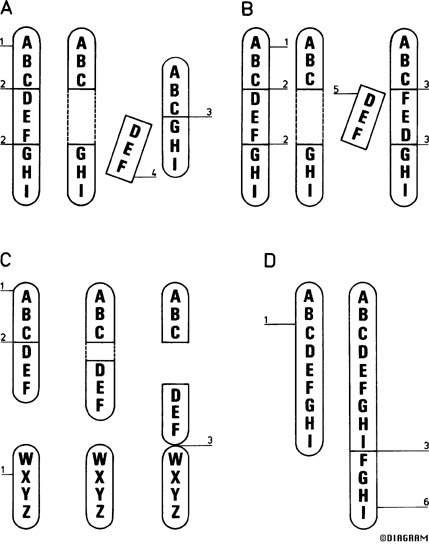
**FRAMESHIFT MUTATION** - a point mutation that involves a substitution or deletion, which results in a a shift in the reading frame.

Cats eat big rat --> add an additional letter A, and the reading frame changes

Aca tse atb igr at - this protein does not resemble the original and will probably not be functional

**Chromosomal Mutations**

Deletion (A)  
Inversion (B)  
Translocation (C)  
Duplication (D)





The Scottish Fold cat is a breed created from a single mutation that controls ear shape in cats.

**Human Chromosomes**

Karyotype = picture or pattern of chromosomes arranged in homologous pairs & organized by size (See fig. 14-1 p. 341)

Humans have 46 chromosomes

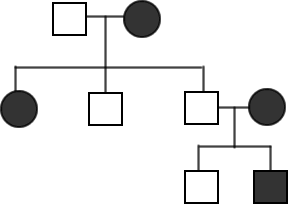
2 of these are sex chromosomes  
XX = female XY = male

The other 44 chromosomes are known as autosomes

**Human Traits**

To study inheritance, biologists use pedigree charts

Shows relationships within a family



Many traits are strongly influenced by environmental factors (ex: height,personality)

**Human Genes = Human genome**

Our complete set of genetic info   
Includes tens of thousands of genes

Genes controlling blood types were 1st genes to be identified  
Most common ones are the ABO & the Rh blood groups ( + and - )

**Human Genetic Disorders – Your task will be to write a quick description of each of these disorders with a focus on the cause, effects, and changes to life. You do not need to write more than 2 or 3 sentences for each one.**

**Recessive**

Phenylketonuria (PKU)  
Tay - Sachs disease recessive  
Albinism

**Dominant**

Achondroplasia  
Huntington's disease

**Codominant**

Sickle-cell disease   
heterozygous is resistant to malaria