



# MUTATIONS

Changes in DNA that affect genetic information

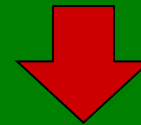
# Mutations

Changes to DNA are called mutations

- change the DNA
- changes the mRNA
- may change protein
- may change trait

DNA

TACGCACATTTACGTAC

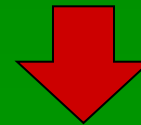


mRNA

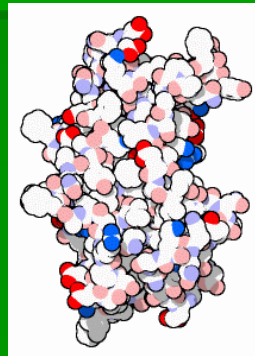
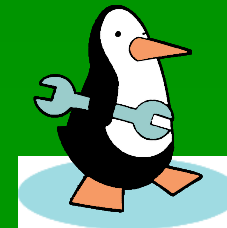
AUGCGUGUAAAUGCAUGC



protein



trait



# Gene Mutations

- **Point Mutations – changes in one or a few nucleotides**

- **Substitution**

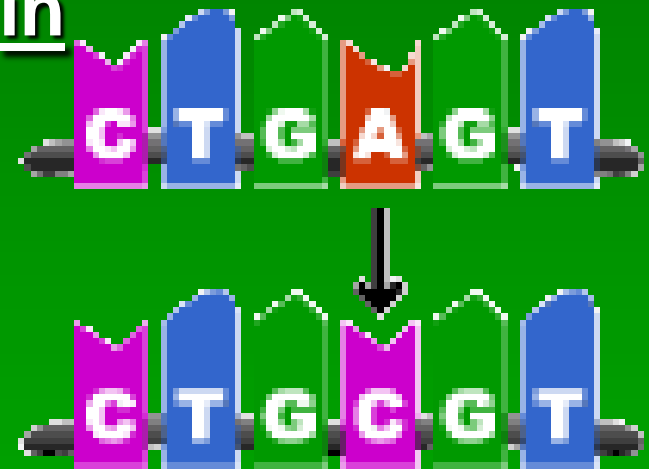
- THE FAT CAT ATE THE RAT
- THE FAT HAT ATE THE RAT

- **Insertion**

- THE FAT CAT ATE THE RAT
- THE FAT CAT XLW ATE THE RA

- **Deletion**

- THE FAT CAT ATE THE RAT
- THE FAT ATE THE RAT



Point mutation

# Gene Mutations

- Frameshift Mutations – shifts the reading frame of the genetic message so that the protein may not be able to perform its function.

- Insertion

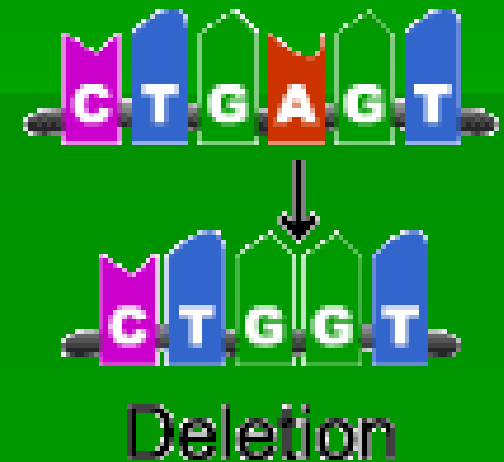
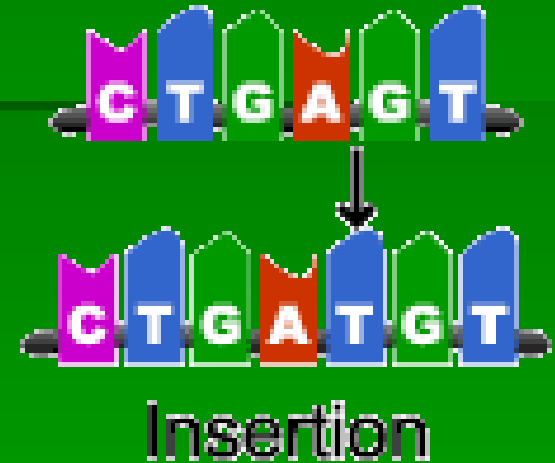
- THE FAT CAT ATE THE RAT
- THE FAT HCA TAT ETH ERA T

H

- Deletion

- THE FAT CAT ATE THE RAT
- TEF ATC ATA TET GER AT

H



# Chromosome Mutations

- Changes in number and structure of entire chromosomes
- Original Chromosome    ABC \* DEF
- Deletion                    AC \* DEF
- Duplication                ABBC \* DEF
- Inversion                  AED \* CBF
- Translocation  
                                  ABC \* JKL  
                                  GHI \* DEF

# Significance of Mutations

- **Most are neutral**
  - Eye color
  - Birth marks
- **Some are harmful**
  - Sickle Cell Anemia
  - Down Syndrome
- **Some are beneficial**
  - Sickle Cell Anemia to Malaria
  - Immunity to HIV

# What Causes Mutations?

- There are two ways in which DNA can become mutated:
  - Mutations can be inherited.
    - Parent to child
  - Mutations can be acquired.
    - Environmental damage
    - Mistakes when DNA is copied

# Chromosome Mutations

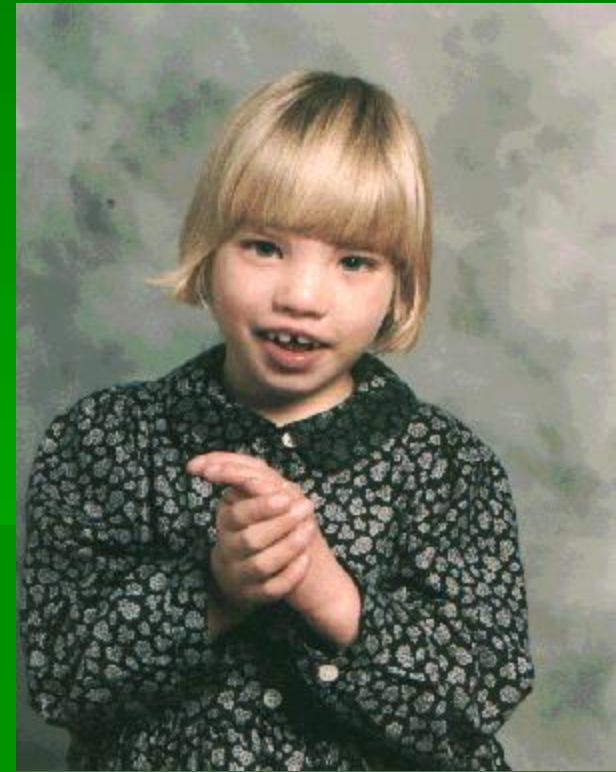
- **Down Syndrome**
  - Chromosome 21 does not separate correctly.
  - They have 47 chromosomes in stead of 46.
  - Children with Down Syndrome develop slower, may have heart and stomach illnesses and vary greatly in their degree of inteligence.





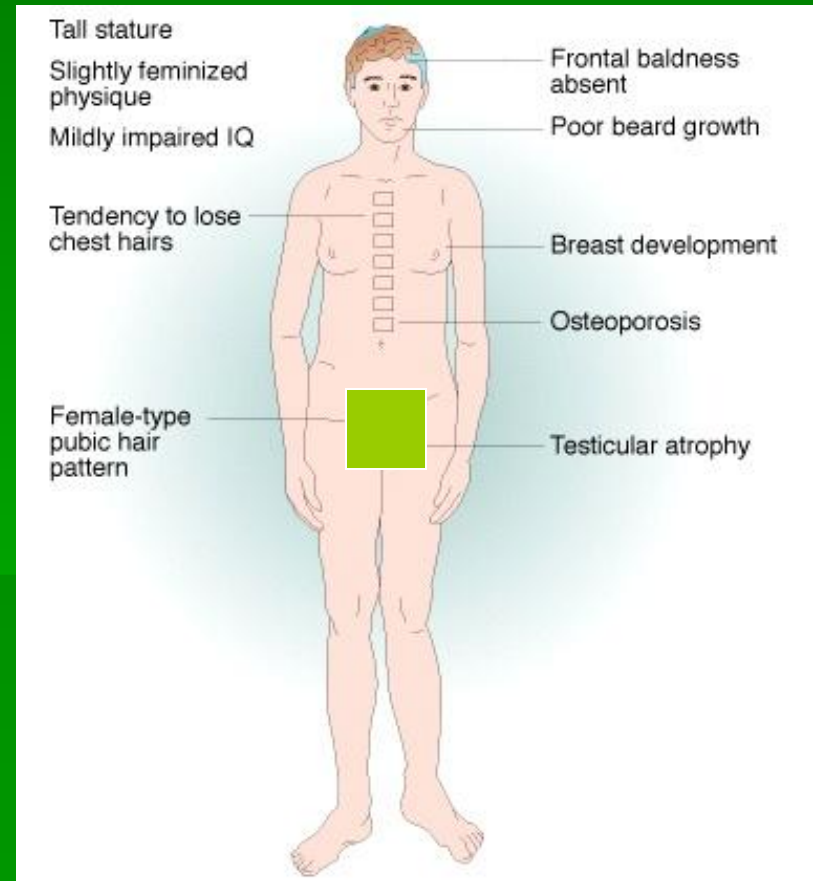
# Chromosome Mutations

- Cri-du-chat
  - Deletion of material on 5<sup>th</sup> chromosome
  - Characterized by the cat-like cry made by cri-du-chat babies
  - Varied levels of mental handicaps



# Sex Chromosome Abnormalities

- Klinefelter's Syndrome
  - XXY, XXYY, XXXY
  - Male
  - Sterility
  - Small testicles
  - Breast enlargement



# Sex Chromosome Abnormalities

- XYY Syndrome
  - Normal male traits
  - Often tall and thin
  - Associated with antisocial and behavioral problems



# Sex Chromosome Mutations

- Turner's Syndrome
  - X
  - Female
  - sex organs don't mature at adolescence
  - sterility
  - short stature



# Sex Chromosome Mutations

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- XXX
  - Trisomy X
  - Female
  - Little or no visible differences
  - tall stature
  - learning disabilities
  - limited fertility

# Gigantism

**Giant and midget in circus sideshow**



Circus World - Baraboo, WI



# Some mutations even make it on SNL!



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"YOUR CASE WOULD MAKE A GREAT MEDICAL JOURNAL ARTICLE,  
BUT FRANKLY, THE NATIONAL ENQUIRER PAYS MORE."