Belarussian State Pedagogical University named after M. Tank
Inclusive Education Institute
Correction and Development Technologies Department

Human Genetics Foundations

What is Mutation?

D. L. Nikolaev, associate professor
Mutations

- **Mutation** = change in DNA sequence
- Mutations can be caused by errors in replication, transcription, translation, cell division, or external agents.
- Mutations in **Reproductive Cells** can affect potential offspring (ex: inheritable genetic disorders)
- Mutations in **Body Cells** do not get passed onto offspring (ex: if an individual develops skin cancer)
Hugo de Vries
MUTATIONS

*Mutations are mistakes made in DNA.

*Mutations can be caused by either naturally occurring, random events, or by factors in the environment.

*Any environmental factor in the environment that causes a mutation is a mutagen. (UV light, radiation, chemicals, etc...)
What Causes Mutations?

- Can be caused by **mutagens** - a physical or chemical cause of mutation. Examples: UV light, radiation, drugs, and benzene.
- Mutagens are often also **carcinogens** – anything that causes cancer.
- Can be natural, random events.
  - Mutations occur in 1/100,000 DNA replications
- Mutations do not have to be bad (evolution)
- Mutations are **random** events that tend to be recessive so appear in a **low number** of the population
- Mutations are the source of new variation
- Variation is the differences between members of a species
- Mutagenic agents that can increase mutation rates

There are 2 main categories of mutagenic agents:
- Chemicals & Radiation

<table>
<thead>
<tr>
<th>Chemical Mutagenic Agents:</th>
<th>Radiation Mutagenic Agents:</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Mustard Gas</td>
<td>- X-rays</td>
</tr>
<tr>
<td>- Colchicine</td>
<td>- UV Light</td>
</tr>
<tr>
<td>- Caffeine</td>
<td>- Gamma Rays</td>
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<td>- Formaldehyde</td>
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Genetic Mutations of the Fruit Fly

- Wingless
- Sepia Eyed
- Bar-Eyed

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Mutations of the Fruit Fly
Reduction deformities
Reduction deformities
Types of Mutations

Normal gene

**AS THE MAN SAW THE DOG HIT THE CAN END ITIS**

Point mutation

**AS THE MAN SAW THE DOG HIT THE CAN END ITIS**

Deletion

**AS THE MAN SAW THE HIT THE CAN END ITIS**

Insertion

**AS THE MAN SAW THE FAT DOG HIT THE CAN END ITIS**

Frame Shift

**AS THE MAN SAW THE GCH ITT HEC ANE ND ITIS**
The highs and lows of mutation rates

The rate at which new mutations appear in a genome (sizes of circles) is inversely proportional to the so-called effective population size of the species. Microbes (right) have the largest populations and lowest mutation rates.

- African green monkey
- Human
- Gorilla
- Honey bee
- Mouse
- Butterfly
- Tetrahymena thermophila
- Paramecium tetraurelia
- Escherichia coli
- Pseudomonas aeruginosa

Effective population in thousands of individuals:
- 0
- 160
- 800
- 2000
- 50000
- 250000