

A1

Mutation

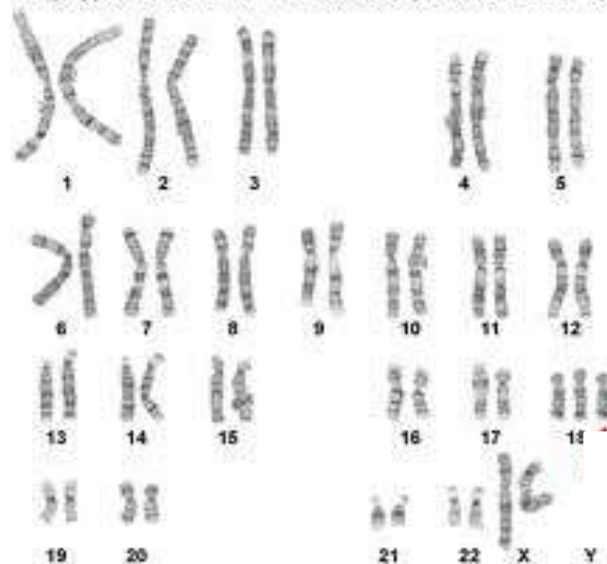
Learning Outcome:

1. A mutation is a spontaneous and random change in genetic material and can give rise to new alleles.
2. Mutations can be - advantageous, disadvantageous or neutral.
3. Environmental factors can increase the mutation rate.

Activity 1,
page 3

• Now let's discuss the differences you spotted.

Karyotype From a Female With Edwards Syndrome (47,XX,+18)



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Mutations

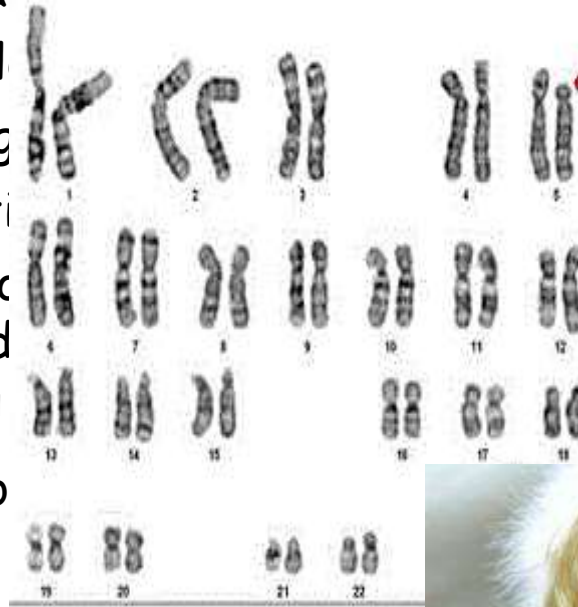
Edwards Syndrome:

- Extra chromosome 18
- Edwards syndrome occurs in around one in 6,000 live births and around 80 percent of those affected are female.
- The majority of fetuses with the syndrome die before birth.
- The syndrome has a very low rate of survival, resulting from heart abnormalities, kidney malformations, other internal organ disorders and some physical malformations .



Cri du Chat Syndrome

- Missing section of chromosome 5
- The syndrome gets its name from the characteristic cry of affected infants
- Feeding problems
- low birth weight
- severe cognitive delays;
- behavioral problems, tantrums, and aggression,
- unusual facial features
- excessive drooling;
- constipation;
- small head and jaw,
- wide eyes;
- skin tags in front of eyes.



swallowing;

delays;

hyperactivity, aggression,

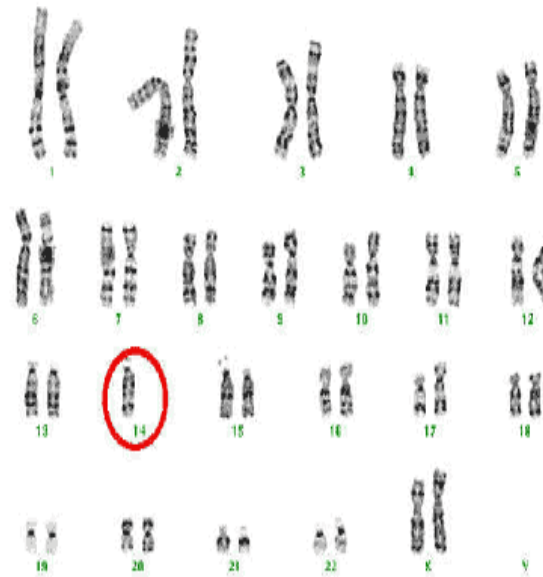
features that change over time;



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Mutations

- <http://ghr.nlm.nih.gov/chromosome/14/show/Conditions>



45,XX-14

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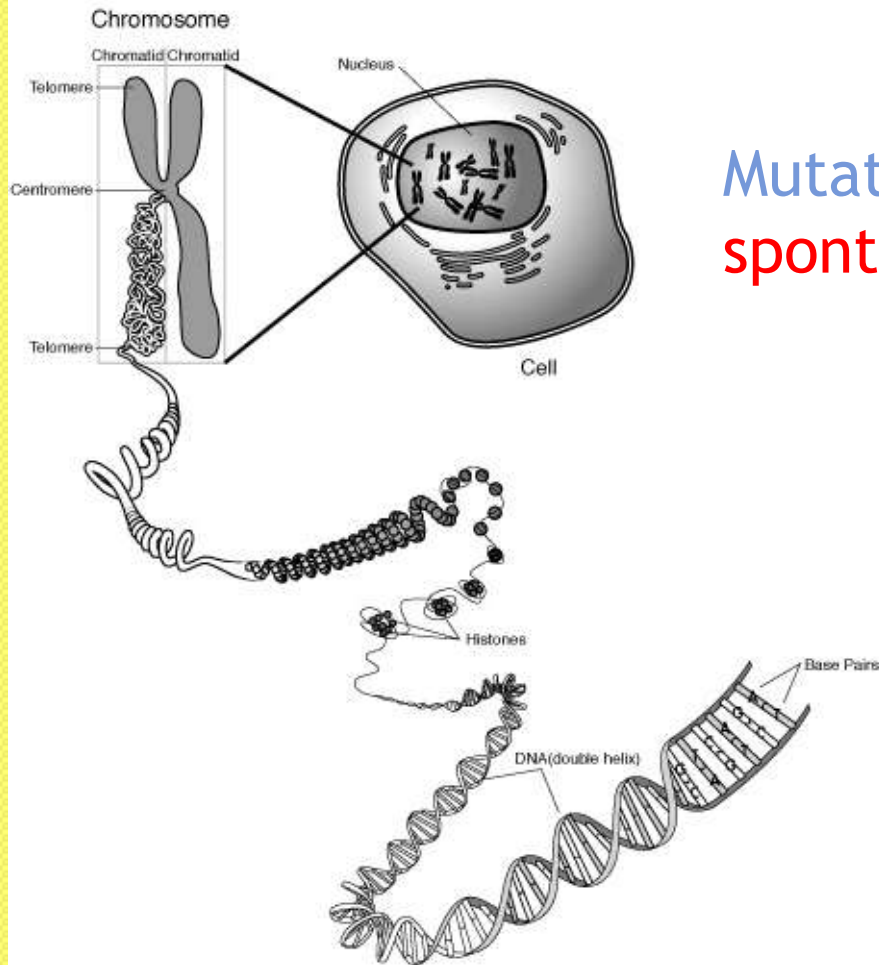


Mutations



A **mutation** is a sudden random change in the **number** or **structure** of an organisms chromosomes (**DNA**).

Mutations are usually **random**, **spontaneous** and occur very **rarely**.



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Disadvantageous Mutations

- Most mutations are unfortunately harmful as they disrupt the proper working of cells.
- Another example of the effects of a mutation in the human species is Down's Syndrome.
- Down's Syndrome is a syndrome that causes slowed growth, abnormal facial features, and mental retardation. It is caused by an extra chromosome 21.

Activity 2,
page 5



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Neutral Mutations

- Some mutations seem to be neutral- they don't seem to confer an advantage or a disadvantage on the organisms in which they occur.
- On Fair Isle an island with no land predators between Orkney and Shetland all of these different colours of rabbit occur.

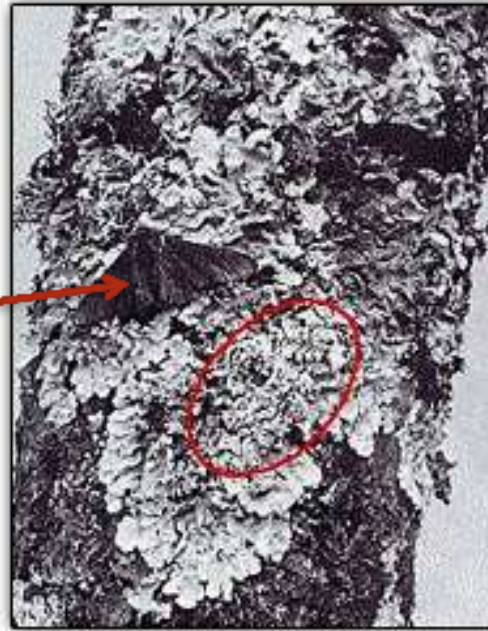
Activity 3,
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Advantageous Mutations

- A very few mutations are beneficial and improve survival chances.
- Without these mutations species would just stay the same. There would be no "survival of the fittest" and no evolution!



Pre-Industrial
Revolution



Pre-Industrial
Revolution

Mutant moth

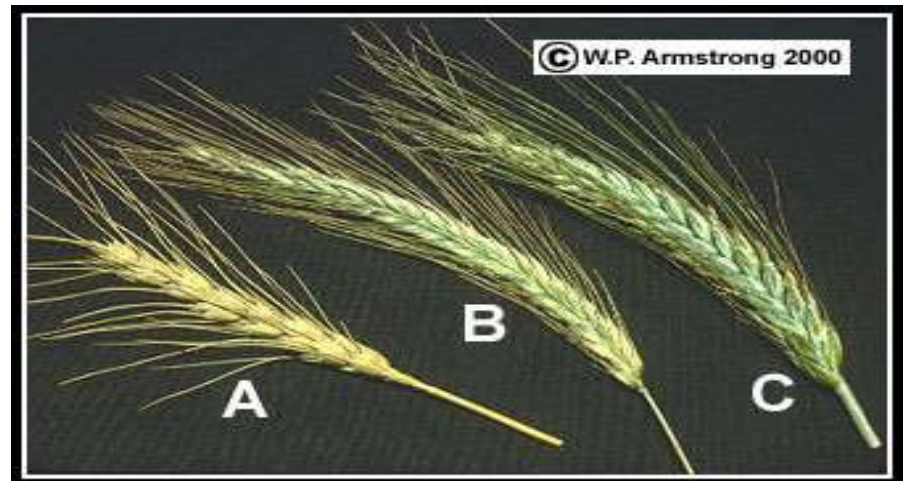


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Advantageous Mutations

Polyploidy

- Complete extra sets of chromosomes (DNA)
- Occurs in fruit such as strawberries and in cereal crops.
- Leads to increased fruit size and increased yield. Such plants are called **polyploids**.



Activity 4,
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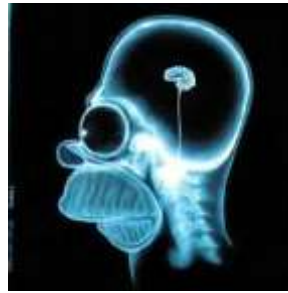


Mutagenic Agents



The rate of mutation can be increased by **mutagenic agents** such as :-

- **Irradiation** - gamma rays , X-rays and UV light



Fallujah, Iraq

- Depleted uranium weapons?



Dear 16-year-old me



Chernobyl



- Chemicals - Colchicine, mustard gas, benzene and agent orange.



Dulce et decorum est pro patria mori



Agent orange (Dioxin)



Benzene