

Fundamental Molecular Biology

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Mutations: "Changes in the genetic material of a cell (or virus)" -Campbell et al.

- Russell suggested "a mutation is a heritable alteration in the genetic material"
- Not accepted as not all mutations are heritable e.g. cancer in the soma, not germ line

Background mutation (spontaneous) - very low rate

- 3 new mutations per 10^8 base pairs per generation
- 200 new mutations in each human child
- 20 billion new mutations per year in world population

Origin: Replication/repair errors

DNA damaging agents

Ionizing radiation

Gene-related sequence: an area of a gene that was once active but is now inactive

DNA damage - repair = net mutation

- Increase mutation = increase damage or decrease repair

Germline: 1 cell

- Passed on
- Low mutation rate (~200/gen)

Soma: Genetic dead-end

- Disposable to natural selection
- Higher mutation rate

UV light damage: Causes thymine dimers (T-T)

- Lead to frame-shift mutations if not repaired
- Bright sunlight- each skin cell suffers 50-100 new T-T dimers every second = overwhelms repairs => mutations

Xeroderma pigmentosum

- Mutations in genes for Excision Repair Machinery
- Extreme sensitivity to UV light
- Exposure = basal cell carcinoma
metastatic malignant melanoma
Squamous cell carcinoma
- Inherited