Mutations from a population point of view, frequency of mutations

Mutation enriches the population with new alleles. Mutations are counteracted by selection and gene drift. If selection takes place and mutations occur at the same time, a mutation-selection equilibrium can be reached, when the frequency of individual alleles does not change.

- ***mutation intensity*** = relative frequency with which the mutation process repeats itself (mutation/gene/generation)
- μ: A → a (forward mutation)
- ν: a → A (backward mutation)
- **spontaneous mutation** - cause unknown, frequency 10^{-5} to 10^{-7}
- if mutation and reverse mutation take place at the same time, then the balance applies: $q_{equal} = \mu / (\mu + \nu)$
- **induced mutations** - it is not yet known how the frequency of mutations can be reduced, but it can be increased by mutagens that cause induced mutations

Mutagenic Factors

🔎 *For more information see Mutagens.*

1. **biological**
   - viruses
   - malfunctions of repair mechanisms
2. **chemical**
   - drugs (cytostatics, chemotherapy)
   - the mutagenicity of drugs is tested using the Ames test on the bacterium *Salmonella typhimurium*, which is auxotrophic and unable to multiply and survive on medium without the addition of histidine. On minimal medium with minimal histidine, where it does not multiply, it is tested whether the drug is mutagenic even outside of replication. After the addition of liver enzymes, mutagenicity after metabolic processing is tested.
   - acridine dyes, base analogues, alkylating, hydroxylating and deaminating agents
3. **physical**
   - **ionizing radiation** - Radiation dose is measured in Gy (Gray). For Drosophila and other simple organisms, the frequency of mutations is linearly dependent on dose. In mammals, the linear dependence is valid only for small doses, at higher doses the mutagenic effect is already lower than would correspond to a linear relationship. A high dose is also more mutagenic for mammals than a long-term low one. This can be explained by the effect of repair mechanisms. A doubling dose is one that doubles the amount of spontaneous mutations. In humans, it is estimated to be 0.5 Gy. Ionizing radiation primarily causes DNA breaks and, secondarily, chromosomal rearrangements.
   - **UV radiation** - dangerous mainly for unicellular organisms. In humans, it causes mutations only in the surface cells (epidermis). In particular, it induces the formation of thymine dimers.

Sources

Articles

- Mutation

References

- SOUKUPOVÁ, Milena – SOUKUP, František. *Kapitoly z lékařské biologie a genetiky II.* 1. edition. Karolinum, 1998. 98 pp. ISBN 80-7184-581-7.