

# Genetic maps and their meaning

They determine the **mutual position** of polymorphic marks (markers) based on the **frequency of recombination** and at the same time inform us about the **probability of recombination** - thanks to this, we can more accurately **estimate the results of analyzes**, that use linkage (e.g. indirect DNA diagnostics)

- **polymorphism** = the existence of more distinguishable alleles (variants) in the population

**We distinguish two types of maps:**

- **Physical map (physical)** - absolute values of the position of gene loci in bp (base pairs).
- **Genetic map** - relative position of the gene locus according to the frequency of recombination.
  - uses binding;
  - in cM (centiMorgans);
  - 1cM = 1% chance that recombination will occur.

There are now **3 genome-wide genetic (linkage) maps available for humans**

- **2 maps** (Genethon and Marshfield) are based on **CEPH** (Centre d'Études du Polymorphisme Humaine du Paris)
- map from **deCODE** on 146 families **from Iceland**

## Conversion of genetic map to physical

- **difficulties in converting a genetic map to a physical one:**
  1. there is no linear relationship between bp and cM;
  2. the frequency of recombination as well as the variability of this frequency is greater in women;
  3. centromeres are "recombination deserts" - recombinations occur here very rarely;
  4. towards the telomeres, the frequency of recombination increases;
  5. hot spots for recombination are pseudoautosomal regions at both ends of the Y and X chromosomes.
- there are 3 whole-genome genetic maps available on the internet

## Three-point attempt

- **Three-point experiment:** We have locus A, B and C. We know the position of loci A and C. We want to find B. If B is between A and C, then recombination between A and B occurs with probability  $x$ , recombination between B and C with probability  $y$ . The probability of double recombination (ie between A and B and between B and C) is considerably lower and is equal to the product of the probabilities  $x$  and  $y$ . We therefore determine the correct order of loci A, B and C with the help of double recombinants, of which there must be the least number.

Genetic maps inform us about the *probability of recombination* - thanks to which we can more accurately estimate the results of analyzes that use linkage:

1. **indirect DNA diagnostics;**
  2. **linkage disequilibrium;**
  3. **selection sweep, background selection = genetic drift.**
- **Selection sweep** = if some random trait arises that is preferred, then the neighborhood of its gene locus is transmitted in linkage with it until recombination occurs. The neighborhood of this locus is therefore also indirectly preferred.
  - **Background selection** = if selection takes place against a certain allele of a locus, then it also affects the surroundings of this locus, because it is linked to it.

## Links

- Mapping the genome
- The genetic code

## References

- OTOVÁ, Berta. *Lékařská biologie a genetika I. díl*. 1. edition. Praha : Karolinum, 2008. 123 pp. ISBN 978-80-246-1594-3.