

Gene control of differentiation in ontogeny

Gene control

Fertilization produces a zygote from the two gametes, and a morula from the grooving one. As the cells travel out, a blastula is formed and then a gastrula (three germ layers) is formed. This is followed by the development of the primitive streak, organogenesis and histogenesis.

During ontogeny, the following mechanisms apply:

- cell growth, cell division, migration cells,
- differentiation cells,
- apoptosis.

All cells in the body have the same set of genes, but differ in their expression.

Ontogenesis = sequence of events that is initiated by fertilization - **regulatory cascade** (local mediators, hormones, receptors, transcription factors, etc. are used here)

Morphogenes = control the differentiation and predetermination of cells, their action depends on the concentration of their products - they create a **concentration gradient** where the effect occurs only from places where the concentration reaches a certain threshold level. Their cascade is already activated before fertilization

Maternal Way Genes

- They are part of the mother's genome,
- expressed in follicular bb and their mRNA and proteins transported to the egg,
- in their mutations, the zygote dies independently of its genotype,
- they determine the anterioposterior and dorsoventral polarity of the embryo,
- this includes *bicoid* (determines where the front of the embryo is) and *nanos* (determines the back of the embryo),
- works in collaboration with **hunchback gene**.

Genes of the zygote

Segmentation genes

- In vertebrates, the segments are distinct only in the initial stages of development.

Gap genes

- E.g. *hunchback*, *knirps*, *giant* a *krüpl*,
- genes of maternal origin are activated,
- their mutation: developmental disorder of part of the segments,
- they are transcription factors,
- affect the basic differentiation of the embryo.

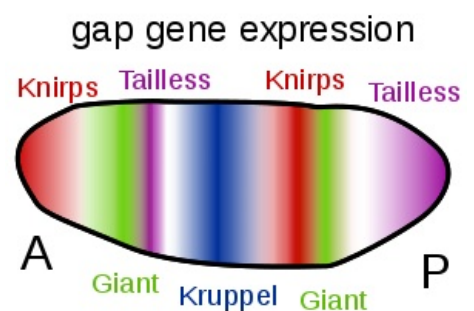
HOX genes

- They contain a homeodomain (homeobox)
- Their mutations can cause one organ to be confused with another,
- bithorax and antenapedia complex genes,
- mutation of the antenapedia gene in *Drosophila melanogaster* causes the development of a leg on the head instead of a antennae,
- in humans, HOX1 mutation causes **craniosynostosis**.

Pair-rule genes (PAX)

- They also contain a homeodomain (homeobox)
- They specify the nature of the segments,
- regulated by gap genes,
- expressed in **7 stripes** along the anterioposterior axis,
- they divide the embryo into **15 parasegments**,
- their mutations reduce the number of segments by half (fushi tarazu - development of odd parasegments, even-skipped - development of even parasegments),
- PAX3 mutation = **Waardenburg syndrome** (deafness, white strand of hair, iris heterochromia).

(Para)segment polarity genes



- They influence the anteroposterior polarity of parasegments, defined by pair-rule genes,
- the embryo is gradually divided into smaller and smaller developmental sections,
- e.g. engrailed gen.

Tissue-specific genes

- A cascade of hundreds of genes,
- eyeless gene - its mutation in *Drosophila* causes the development of a rudimentary eye, in mice microphthalmia and in humans aniridia,
- when the eyeless gene is linked to the promoter of a gene typical for another tissue, an eye develops in this tissue,
- with interspecies transfer of the eyeless gene, a species-specific eye is created → basic tissue-specific genes are developmentally old,
- other, developmentally younger and species-specific genes also decide on the definitive form of the organ.

The spectrum of genes that is expressed in a certain type of cells is determined by **RNA - DNA saturation**.

Links

Used literature

- KAPRAS, Jan – KOHOUTOVÁ, Milada. *Kapitoly z lékařské genetiky III.*. 1. edition. Karolinum, 2009. vol. 1. ISBN 80-246-0001-3.