Genomic imprinting

As diploid organisms, humans carry in the genetic equipment of their cells two corresponding sets of chromosomes, one inherited from the father (paternal) and one from the mother (maternal). Under normal circumstances, the paternal and maternal copies of a particular gene have the same potential for expression in any human cell. **Genomic imprinting** is a process that fundamentally changes this potential, because as a result, *the activity of a certain gene is limited to one chromosome depending on which parent it was inherited from*.

Parental sex and genomic imprinting

In most cases, genomic imprinting means that only the maternal or paternal allele is expressed (is active), while the other is silenced. Genomic imprinting occurs equally in offspring of both sexes, so for example a gene active only on the maternal chromosomes will be silenced on the paternal chromosomes in all daughters and sons.

Imprinting control region

This phenomenon physiologically affects only a few hundred of the more than 20,000 human genes. In most cases, these genes are not found in isolation in the genome, but on the contrary, they are clustered, and thus it is possible to identify entire chromosomal regions with imprinted genes. Here we find a fundamental structure for the actual implementation of imprinting, the so-called **imprinting control region (ICR)**. ICRs are amenable to epigenetic modification and usually one of the parent ICRs is *methylated.* Imprinting must be distinguished from other forms of gene expression depending on the sex of the parents, which arose as a result of their different genetic contribution - this includes genes on the Y chromosome or matroclinic (mitochondrial) inheritance.

Genomic imprinting study

Much of our current knowledge of imprinting comes from studies in model organisms, primarily mice. In those who had a complete set of chromosomes, but one pair of chromosomes came from a single parent (uniparental disomy), developmental abnormalities and the occurrence of various diseases were often described. It was interesting that this only applied to some chromosomes, in many cases uniparental disomy was done without obvious consequences for its carrier. A recent map of the number of imprinted regions identified so far within the genomes of various organisms (including humans) is available here (https://www.geneimprint.com/site/genes-by-species).

Diseases caused by genomic imprinting disorders

Diseases that arise on the basis of an imprinting disorder have a number of causes at the molecular level, e.g. (micro)deletion, point mutations or uniparental disomy. Diseases with a proven role for imprinting disorders include **Prader–Willi syndrome**, **Angelman syndrome**, **Beckwith–Wiedemann syndrome**, **Silver-Russell syndrom**, **transient neonatal diabetes mellitus** and others.

Links

Related Articles

- Gene imprinting and human pathologies
- Angelman Syndrome
- Beckwith-Wiedemann syndrome
- Prader-Willi syndrome
- Uniparental disomy