

Genomic Imprinting

1. What is Genomic Imprinting?

Genomic imprinting is an epigenetic process in which certain genes are expressed only from either the maternal or paternal allele. The other allele is silenced through DNA methylation and histone modifications. This parent-specific gene expression does not alter DNA sequence.

2. Why Imprinting Happens?

Helps regulate fetal growth, brain development, placental function. Parental conflict theory: paternal genes promote growth; maternal genes limit it to conserve resources.

3. Mechanism of Imprinting

- DNA Methylation silences genes.
- Histone modifications regulate activation.
- Imprinting control regions (ICRs) regulate parent-specific silencing.
- Steps: Erasure → Establishment → Maintenance.

Diagram 2: Imprinting Cycle

[DIAGRAM] Three boxes: 1. Erasure of old marks → 2. Addition of new parent-specific marks in sperm/egg → 3. Maintenance in embryo.

4. Types of Imprinting

- Maternal imprinting: maternal allele OFF.
- Paternal imprinting: paternal allele OFF.

5. Examples

- A. Igf2 (paternal active) – growth promoting.
- B. Igf2r (maternal active).
- C. Prader-Willi Syndrome (PWS): paternal deletion + maternal imprinting.
- D. Angelman Syndrome (AS): maternal deletion + paternal imprinting.
- E. Beckwith-Wiedemann Syndrome (BWS): excessive growth due to imprinting defects on chr 11p15.

Diagram 3: PWS vs AS

[DIAGRAM] Chromosome 15q11-13:

- PWS: paternal deleted + maternal imprinted → NO ACTIVE gene.
- AS: maternal deleted + paternal imprinted → NO ACTIVE UBE3A.

6. Key Features

- Epigenetic, not genetic.
- Parent-of-origin effect.
- Non-Mendelian.
- Reset every generation.
- Essential for development.

7. Summary

Imprinting = parent-specific monoallelic gene expression controlled by methylation and ICRs. Important disorders: PWS, AS, BWS.