Homozygous Loss

Homozygous loss refers to the **complete deletion or inactivation of both copies (alleles)** of a gene in a diploid organism. This term is commonly used in **genetics**, **molecular biology**, and **cancer genomics**.

What Does It Mean?

Humans typically have **two copies** of each gene (one from each parent). A **homozygous loss** means that **both copies** of a gene are **deleted, mutated, or otherwise inactivated**, resulting in **loss of gene function**.

Example

In cancer, **homozygous loss of the CDKN2A gene** is common and leads to the loss of tumor suppressor function (p16^INK4a^), promoting uncontrolled cell proliferation.

Key Points

Feature	**Explanation**
Zygosity	Homozygous = both alleles are affected
Mechanism	Large deletions, point mutations, LOH + mutation
Consequence	Often results in **loss of gene function**
Clinical relevance	Seen in tumor suppressor gene deletions (e.g., TP53, PTEN, RB1)
Detected by	Genomic techniques such as FISH, aCGH, NGS, or SNP arrays