

An **enhancer** is a **cis-regulatory DNA element** that increases the transcription of associated genes, often functioning at a **distance** from the gene it regulates. Unlike promoters, which are located immediately upstream of genes, enhancers can be **located upstream, downstream, or even within introns** of their target genes.

Key Characteristics of Enhancers

1. **Distance-Independent Action** – Enhancers can function **kilobases to megabases** away from the genes they regulate.
2. **Orientation Independence** – They can work in **both forward and reverse** orientations.
3. **Tissue-Specific and Developmental Regulation** – Enhancer activity varies across **different cell types, tissues, and developmental stages**.
4. **Binding Sites for Transcription Factors (TFs)** – Enhancers contain multiple **binding sites** for TFs that help recruit the transcriptional machinery to the promoter of the target gene.
5. **Chromatin Accessibility** – Enhancers are often found in **open chromatin regions**, marked by **histone modifications** like **H3K27ac (active enhancers)** and **H3K4me1 (primed enhancers)**.

How Enhancers Work

- **Transcription Factor Binding**: Enhancers bind TFs, which recruit co-activators and the basal transcription machinery.
- **Chromatin Looping**: Enhancers interact with promoters through **chromatin looping**, bringing regulatory elements into close proximity.
- **Mediator and Cohesin Complexes**: These protein complexes help facilitate the interaction between enhancers and promoters.

Types of Enhancers

1. **Constitutive Enhancers** – Active in multiple cell types.
2. **Cell-Type Specific Enhancers** – Only active in certain tissues or conditions.
3. **Super-Enhancers** – Clusters of highly active enhancers that drive **high expression of key genes**, often associated with cell identity and disease states like cancer.

Enhancers and Disease - **Mutations or dysregulation** of enhancers can lead to **misexpression of genes**, contributing to diseases such as:

1. **Cancer** (e.g., MYC oncogene activation by super-enhancers)
2. **Neurological disorders** (e.g., enhancer mutations affecting neurodevelopmental genes)
3. **Autoimmune diseases** (e.g., enhancer variants linked to immune gene regulation)

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Last update: **2025/02/06 08:18**

