OPEN ACCESS Freely available online

ISSN: 0974-8369
Biology and Medicine

Perspective

Epigenetic Dysregulation and its Impact on Developmental Diseases

Katelyn Elliot*

Department of Medicine, Virginia Commonwealth University, Virginia, United States of America

DESCRIPTION

Epigenetics refers to the study of changes in gene expression that do not involve alterations in the DNA sequence itself but rather involve chemical modifications to the DNA or histone proteins that affect chromatin structure. These modifications play an essential role in regulating gene activity and when dysregulated, they can lead to various developmental diseases. Epigenetic changes are essential during normal development, controlling processes such as cell differentiation, growth and organogenesis. However, when these mechanisms are disrupted, they can result in abnormal gene expression patterns that contribute to a wide range of diseases, particularly developmental disorders.

Epigenetic regulation occurs through several key mechanisms, including DNA methylation, histone modification and non-coding RNA molecules. DNA methylation involves the addition of a methyl group to the cytosine base of DNA, typically leading to gene silencing. Histone modifications, such as acetylation, methylation and phosphorylation, alter the chromatin structure, either promoting or inhibiting gene expression depending on the type of modification. Non-coding RNAs, such as microRNAs, play an impotant role in regulating gene expression at the post-transcriptional level. These processes are particularly important during early development when the differentiation of stem cells into specialized cell types occurs. The precise regulation of epigenetic modifications ensures that the right genes are activated or silenced at the appropriate times and in the correct cell types. For example, epigenetic reprogramming in early embryos is essential for establishing a functional and stable genome in all cells. Dysregulation of these processes can interfere with cellular differentiation and lead to developmental abnormalities.

Epigenetic dysregulation occurs when the normal patterns of DNA methylation, histone modifications or non-coding RNA expression are disrupted. Such disruptions can lead to altered gene expression patterns, resulting in diseases that affect development. These diseases can range from congenital disorders to neurodevelopmental and metabolic diseases. A few examples of developmental diseases that have been linked to epigenetic dysregulation include imprinting disorders, Rett syndrome and neurodevelopmental conditions such as autism and intellectual disabilities. One of the best-known examples of epigenetic dysregulation is seen in imprinting disorders, such as Prader-WilliS (PWS) and Angelman Syndrome (AS). These conditions arise due to defects in the epigenetic regulation of imprinted genes, which are expressed in a parent-of-origin-specific manner. In PWS, the paternal copy of a essential gene region on chromosome 15 is deleted or silenced, whereas in AS, the maternal copy is affected. These disorders illustrate how the misregulation of epigenetic marks on specific genes can lead to developmental abnormalities, including growth defects, intellectual disabilities and neurological problems.

Rett syndrome is a neurodevelopmental disorder primarily affecting girls and is characterized by a period of normal development followed by a regression in motor and cognitive functions. The disorder is caused by mutations in the *MECP2* gene, which encodes a protein involved in regulating DNA methylation. In Rett syndrome, mutations in the *MECP2* gene disrupt the normal epigenetic regulation of gene expression, leading to impaired neuronal development and function. Autism Spectrum Disorder (ASD) and Intellectual Disabilities (ID) have also been associated with epigenetic changes. Studies have shown that abnormal DNA methylation patterns in specific regions of the genome are often observed in individuals with ASD and ID. These changes can affect the expression of genes involved in brain development, synaptic plasticity and neuronal function. Furthermore, environmental factors such as maternal stress, diet and toxins during pregnancy can influence the epigenome of the developing fetus, increasing the risk of neurodevelopmental disorders.

Epigenetic modifications also play an essential role in organogenesis and their dysregulation can lead to congenital defects. For example, abnormalities in histone modifications and DNA methylation have been linked to congenital heart defects. These epigenetic alterations can affect the expression of genes critical for heart development, leading to malformed or

Correspondence to: Katelyn Elliot, Department of Medicine, Virginia Commonwealth University, Virginia, United States of America, E-mail: elli@katl.edu

Received: 23-Oct-2024, Manuscript No. BLM-24-27595; **Editor assigned:** 25-Oct-2024, PreQC No. BLM-24-27595 (PQ); **Reviewed:** 08-Nov-2024, QC No. BLM-24-27595; **Revised:** 15-Nov-2024, Manuscript No. BLM-24-27595 (R); **Published:** 22-Nov-2024, DOI: 10.35248/0974-8369.24.16.749

Citation: Elliot K (2024). Epigenetic Dysregulation and its Impact on Developmental Diseases. Bio Med. 16:749.

Copyright: © 2024 Elliot K. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original author and source are credited.

underdeveloped cardiac structures. Research has shown that changes in the expression of key transcription factors and signaling pathways involved in heart formation can be caused by disruptions in epigenetic regulation.