

DNA Methylation and miRNA Regulation in Spermatogenesis: Epigenetic Mechanisms and Analytical Approaches in Male Infertility

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KEYWORDS Biomarkers. DNA Methylation. Epigenetics. Male Reproductive Health. MicroRNA. Spermatogenesis

ABSTRACT Male infertility is a complex disorder caused by genetic, epigenetic, environmental, and lifestyle factors. Among epigenetic mechanisms, DNA methylation and microRNA (miRNA) biogenesis serve critical roles in regulating gene expression during spermatogenesis and preserving sperm function. Differential methylation patterns in the MTHFR, MEST, and RHOX genes are linked to idiopathic male infertility, influencing sperm quality, motility, and function. Similarly, dysregulation of specific miRNAs, including the miR-34 family, has been linked to impaired sperm parameters and defective spermatogenesis. This review investigates how male fertility is influenced by miRNAs and DNA methylation and discusses advanced analysis techniques, including methylation-specific PCR, MethyLight, Oxford Nanopore sequencing, RT-qPCR, and sequencing-based approaches. These epigenetic mechanisms could be used as valuable potential therapeutic targets and biomarkers in male infertility.