Intron retention: What that means for the biologist

Submission: 24-12-2023 Revision: 13-01-2024 Publication: 01-02-2024

Access this article online

Website: http://nepjol.info/index.php/AJMS

DOI: 10.3126/ajms.v15i2.61029

E-ISSN: 2091-0576

P-ISSN: 2467-9100

Introns are non-coding segments within eukaryotic genes that are transcribed along with exons.[1] They are subsequently removed during splicing, and their presence allows for generating diverse mRNA isoforms through the process called alternative splicing.[2] The functional significance of introns going beyond mere "junk DNA" is questionable until recently; scientists identified that these pieces of junk DNA could hang around with the functional mRNA escaping splicing.[3] Intron retention is a type of alternative splicing, a process that occurs during the maturation of messenger RNA (mRNA) in eukaryotic cells.[4] In eukaryotes, genes typically consist of exons (coding regions) and introns (non-coding regions). After transcription, the precursor mRNA (pre-mRNA) undergoes splicing, where introns are removed, and exons are joined together to form the mature mRNA. Intron retention, however, deviates from this typical splicing process. Instead of being entirely removed from the pre-mRNA, the mature mRNA may retain one or more introns.[5,6] This results in the inclusion of intronic sequences within the final mRNA transcript. The retained introns may contain premature stop codons or regulatory elements that can influence gene expression and protein function. Intron retention is a form of alternative splicing because it generates multiple mRNA isoforms from a single gene. This diversity in mRNA transcripts can produce different protein isoforms with distinct functions or regulatory properties. The regulation of intron retention is complex and involves various cellular factors, including splicing machinery components and RNA-binding proteins.[7] Research on intron retention has revealed its importance in various biological processes, including development, cell growth and differentiation, and diseases like cancer.[8,9] Aberrant intron retention has been associated with certain genetic disorders and cancers. Understanding the mechanisms and functional consequences of intron retention is crucial for unraveling the complexity of gene regulation and its impact on cellular processes.[10] With an increasing output of data from high throughput assays line RNA Seq, scRNA-Seq, etc., it is high time data scientists look into these signatures in the context of disease pathology.[11] This may well be the tip of the iceberg.

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Source of Support: Nil, **Conflicts of Interest:** None declared.