Special Issue

Alternative Splicing in Human Physiology and Disease

Message from the Guest Editors

Alternative splicing, first proposed by Gilbert in 1978, allows multi-exon genes to produce multiple splice variants. Several of the linear transcripts encode protein isoforms with distinct amino acid sequence, structure, and function(s). Alternatively spliced transcripts are generated from a single gene through selection of cassette exons, mutually exclusive exons, retained introns, alternative 3′ or 5′ splice sites, and/or usage of alternative promoters or polyadenylation sites. High-throughput sequencing has revolutionized transcriptomics, revealing that the post-transcriptional maturation of primary transcripts from more than 95% of human multi-exon genes involves alternative splicing.

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