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ASviewer : Visualizing the transcript structure and functional domains of alternatively-spliced genes



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Short Abstract: We present a novel viewer named ASviewer that visualizes the transcript structure and functional inference of alternatively spliced genes intuitively. Key ideas involve in clustering of overlapping exons and representing introns in arbitrary scales. ASviewer is available at <http://genome.ewha.ac.kr/ASviewer>.

Long Abstract:

Alternative splicing (AS) is a mechanism of creating complex transcripts from a given genomic locus by differential use of splice sites in eukaryote. Recent genome-wide analyses of alternative splicing have estimated that 40-74% of human genes have transcript forms that are alternatively spliced, suggesting that alternative splicing is one of the most important parts of the functional diversity of the human genome. According to the ECgene analysis, 44% of human multi-exon genes have more than one isoform and the average number of isoforms is 7.4. This number is likely to be an underestimate since the number of isoforms increases with more sequence data available. Furthermore, many groups have recently presented the gene model of alternative splicing across the whole genome substantially.

Representation of gene structures is one of the most serious aspects of any genome browser application as the gene-flow from genes to proteins is related with sights of biological research. Therefore, the viewer specialized for alternative splicing is required actually since one gene can create various proteins due to alternative splicing. In the sight of this, techniques for representing the key elements of gene structures and their encoded proteins are necessary to be developed. Feature elements of alternative splicing include exon skipping, alternative donor sites and alternative acceptor sites, and intron retention. Also CDS, untranslated regions(UTR), functional domains should be distinguishable on a gene structure. Even though the exon-intron composition of genes is concerning, biologists are more interested in the proteins that genes encode. Therefore, an alternative splicing viewer should merge information describing how the gene is translated into protein in the context of genomic position.

Global gene structure is not evident in the splicing graphs which are commonly used to show the AS events at the exon level. In contrast, genome browser provides an excellent interface to show the gene structure. Present splicing graphs and the genome browsers are not so proper for describing not only features of alternatively spliced gene structures but also their functional domain changes mainly due to the presence of longer length of introns than that of exons. In addition, most applications show them in the individual mRNA or protein sequence as in the Ensembl browser or UCSC proteome browser. The difficulties arise from the requirement of showing the overall gene structures and the relatively microscopic changes in the splice site usage simultaneously and their effects on functional domain changes.

In this paper, we present ASviewer, java application for describing alternatively spliced gene structures and their functional domains effectively. We created two simple but useful rules for presenting various alternative splicing events in order to overcome the weaknesses of present applications maximally. Additionally, to facilitate the estimation of the impact of alternative splicing on the protein function in particular with respect to the domain component of the protein, Pfam or CDD domains were incorporated onto our viewer. ASviewer is available at <http://genome.ewha.ac.kr/ASviewer>.