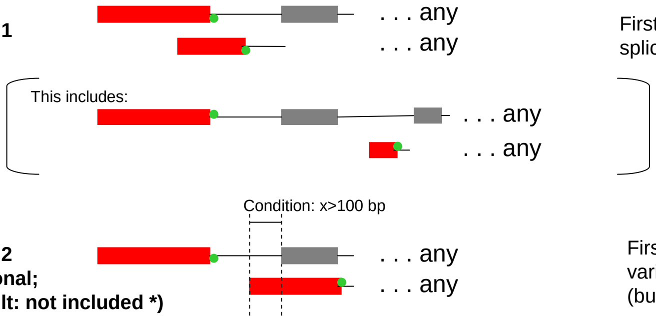
What constitutes an ‘alternative TSS’?

Case 1



. . . any

. . . any

This includes:

. . . any

. . . any

Condition: x>100 bp

. . . any

. . . any

First exon shows novel splice structure (likely real)

Case 2
(optional;
default: not included *)

First exon is significant variation on existing exon (but may also be artifact)

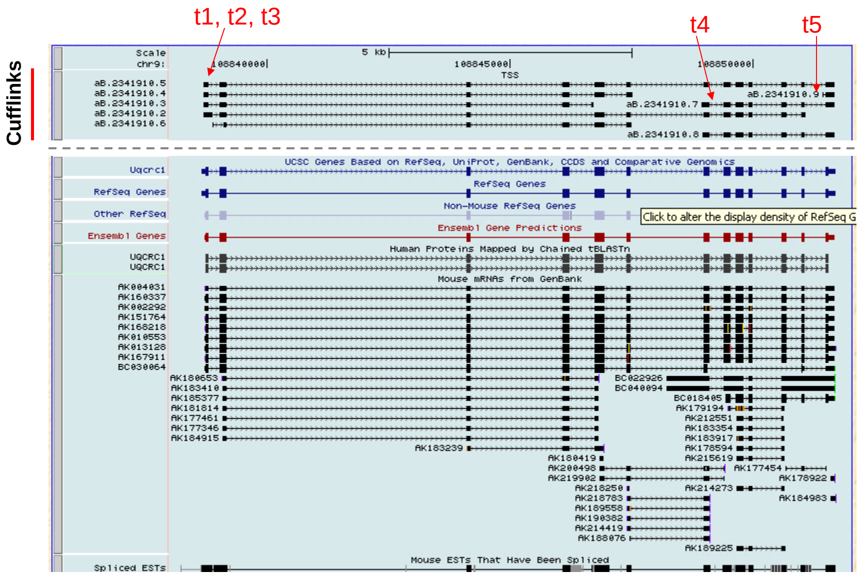
- Genes with a single TSS: by default, longest form included for multi-exon transcripts only
- Case 2 events account for ~40% of the cases (genes with a single TSS not included)
- Event “signature” is the in-boundary of the first exon (see below). Therefore, different events may lead to the same transcription start site in genomic coordinates:

Event 1001 . . . any

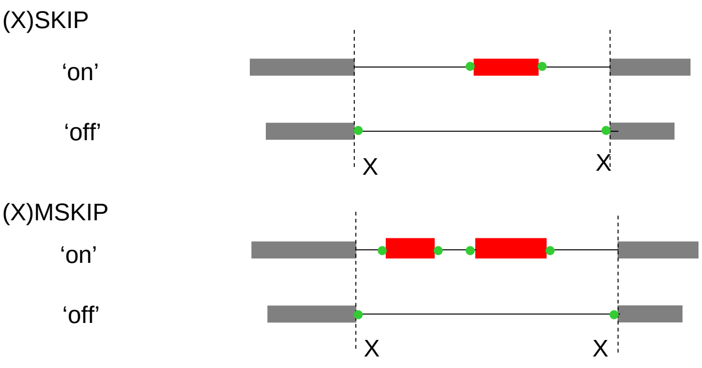
Event 1002 . . . any

Different events, same genomic TSS

Example 1: Alternative TSS at *Uqcrc1*

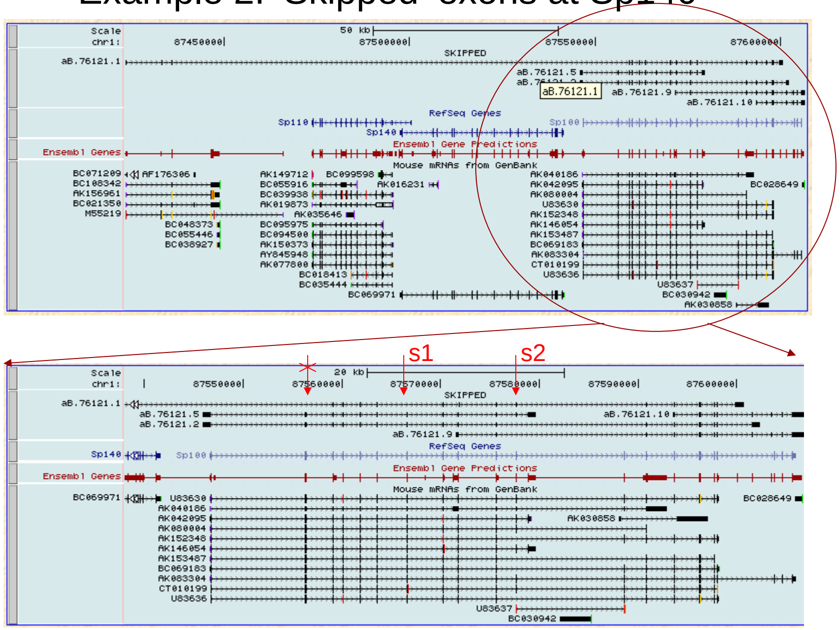


What constitutes a ‘skipped’ (cassette) exon?

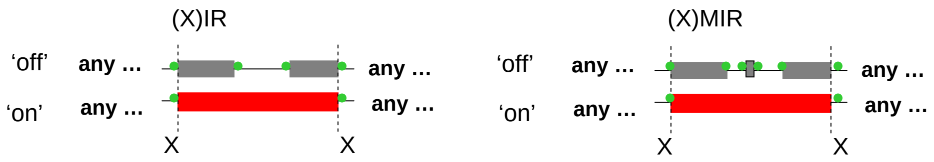


- Transfrags must belong to the same gene, have the same orientation
- Both transfrags must contain >=3 exons
- X – exact boundary match for SKIP, approximate (simple exon overlap) for XSKIP
- ● - event “pattern” or “signature” (column 7)

Example 2: ‘Skipped’ exons at Sp140

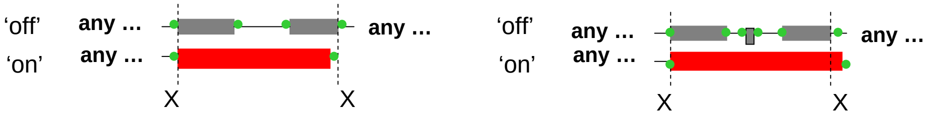


What constitutes a ‘retained intron’?

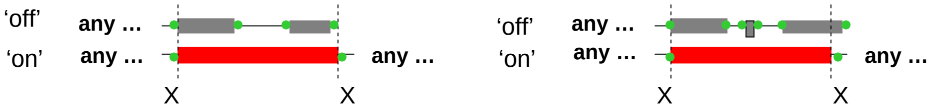


What if the event involves a terminal exon?

1. Long (‘on’) exon? No – APA!



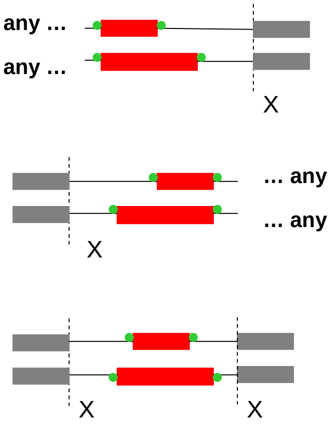
2. Short (‘off’) exon? Yes - IR!



- Transfrags must belong to the same gene, have the same orientation
- <u>Both</u> transfrags must contain >=3 exons

What constitutes an ‘alternative exon end’?

(X)AE



3’ end (5’ exon end must match)

5’ end (3’ exon end must match)

Both ends

- Transfrags must belong to the same gene, have the same orientation
- <u>Both</u> transfrags must contain >=3 exons; only internal exons can be AE

Relationship to a reference gene (column 19)