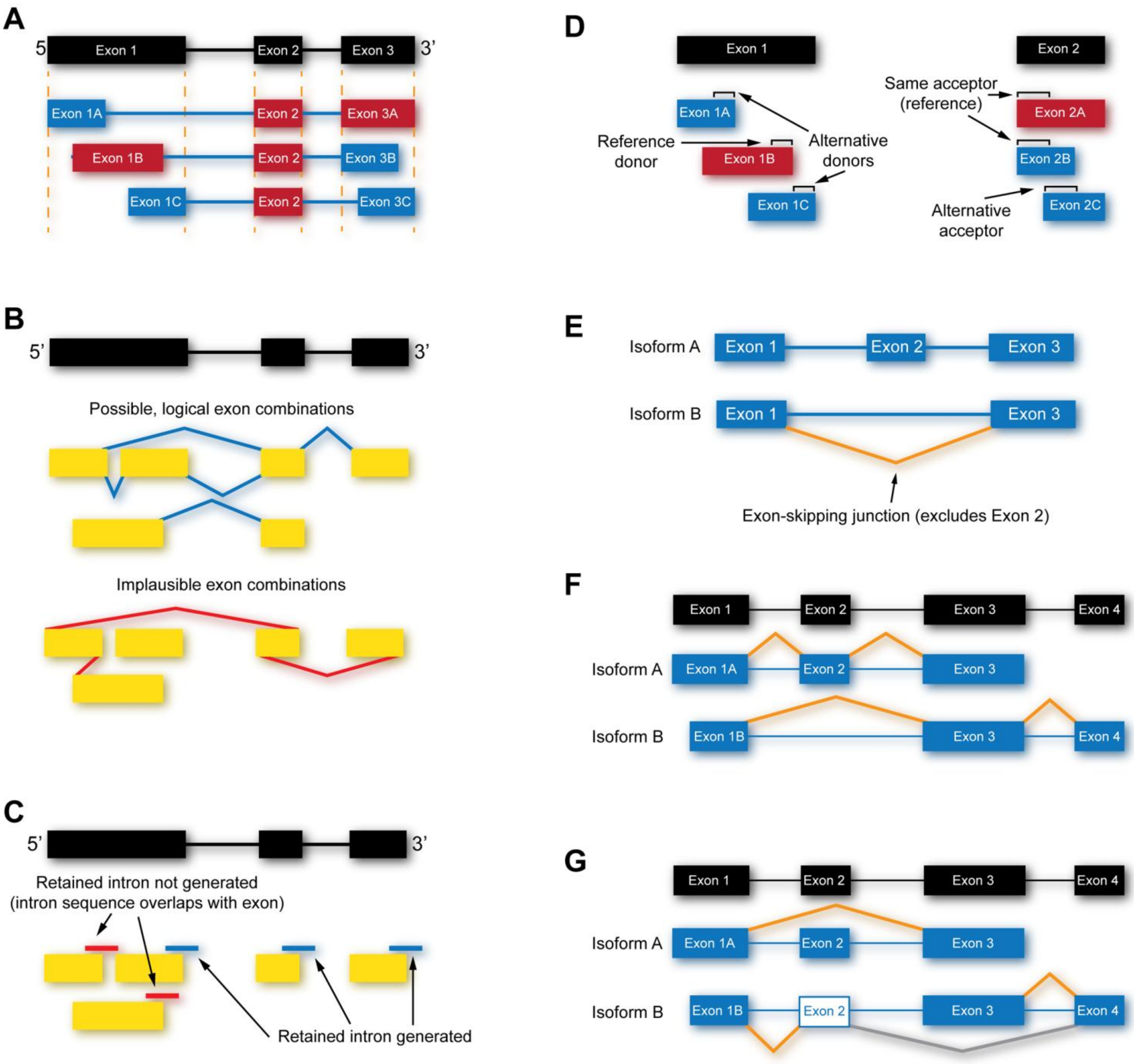

Supplemental Figure S7. Creation and annotation of alternative splicing events. (A) Overlapping exons per gene are grouped together and the longest exon per group is selected as the reference for that group. (B) All possible junctions are created as logical combinations of exons within a gene (blue junctions). Junctions are not made if they are not biologically plausible (e.g. between overlapping junction, junctions that loop back upon the gene; red junctions). (C) Intron retention events are created only from exon-intron boundaries where the intronic sequence does not overlap with known exonic regions. (D) Donor and acceptor sites for an exon (blue) are defined as alternative if their end (donor) or start (acceptor) positions differ from that on their reference exon (red). (E) Junctions are classified as exon skipping if they omit at least one known exon, regardless of whether the skipped exon appears in the same isoform as the exon-skipping junction. (F) Junctions are defined as “transcript annotated” if the donor/acceptor combination exists within at least one known isoform of a gene (orange junctions). (G) Junctions are defined as “unannotated” or novel if the donor/acceptor combination does not exist in any known isoform (grey junction). If the donor/acceptor combination does exist in at least one isoform then it is defined as “annotated” event the exon combination does not exist in an isoform (orange junctions; e.g. the junction between Exon-1B/Exon-2 is “annotated” as Exon-1B shares a donor site with Exon-1A, and the Exon-1A/Exon-2 junction exists in Isoform A even though Isoform B excludes Exon-2 (white exon).