Investigation of the Splicing of BRCA2

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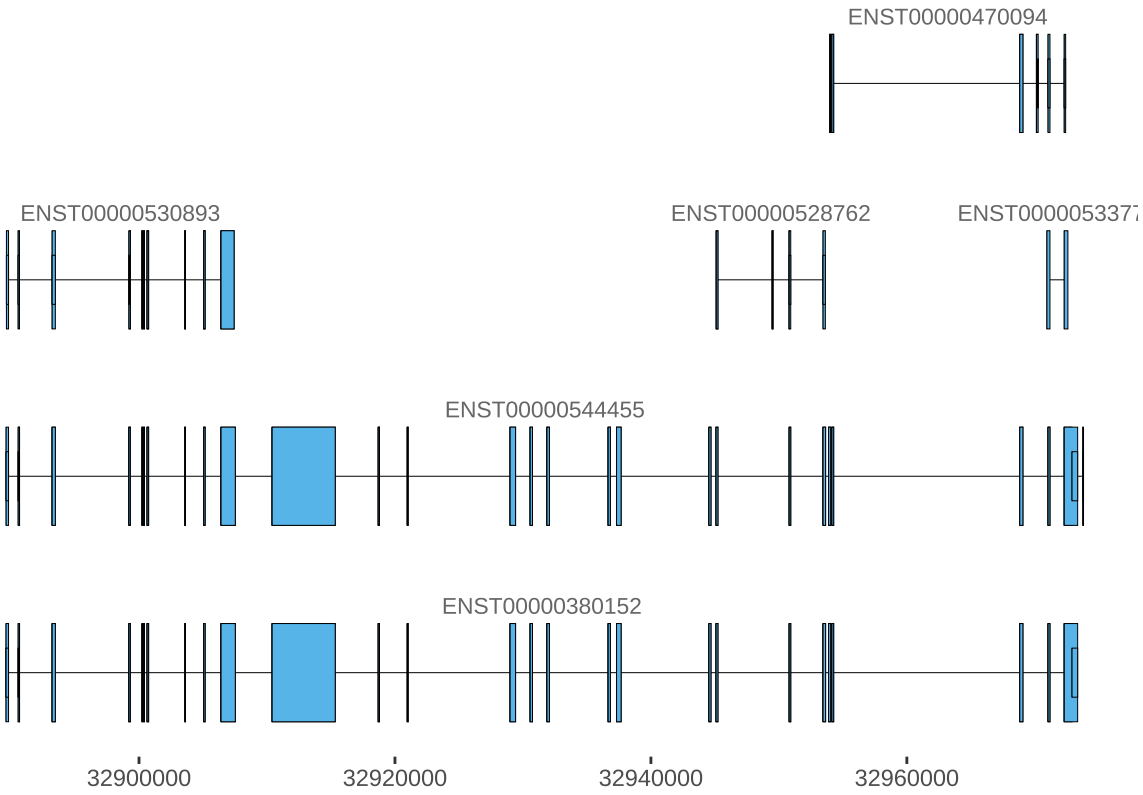
Introduction

BRCA2 is a human tumor suppressor gene (specifically, a caretaker gene), found in all humans; its protein, also called by the synonym breast cancer type 2 susceptibility protein, is responsible for repairing DNA. Inherited mutations in BRCA1 and this gene, BRCA2, confer an increased lifetime risk of developing breast or ovarian cancer. Both BRCA1 and BRCA2 are involved in the maintenance of genome stability, specifically the homologous recombination pathway for double-strand DNA repair. The largest exon in both genes is exon 11, which harbors the most important and frequent mutations in breast cancer patients. The BRCA2 gene was found on chromosome 13q12.3 in humans. The BRCA2 protein contains several copies of a 70 aa motif called the BRC motif, and these motifs mediate binding to the RAD51 recombinase which functions in DNA repair. BRCA2 is considered a tumor suppressor gene, as tumors with BRCA2 mutations generally exhibit loss of heterozygosity (LOH) of the wild-type allele. **This brief study will attempt to investigate large deletion and the exon skipping in BRCA2 splicing in different types of cancer**.

Retrive the exons of BRCA2

Initially, we need to retrieve the unmber of exons for BRCA2 and their positions based on the genome reference consortium human genome build 37 (GRCh37). We will use `BioMart` from `Bioconductor package` in `R` to map the start and the end position of each exon. The `BioMart` package enables retrieval of large amounts of data in a uniform way.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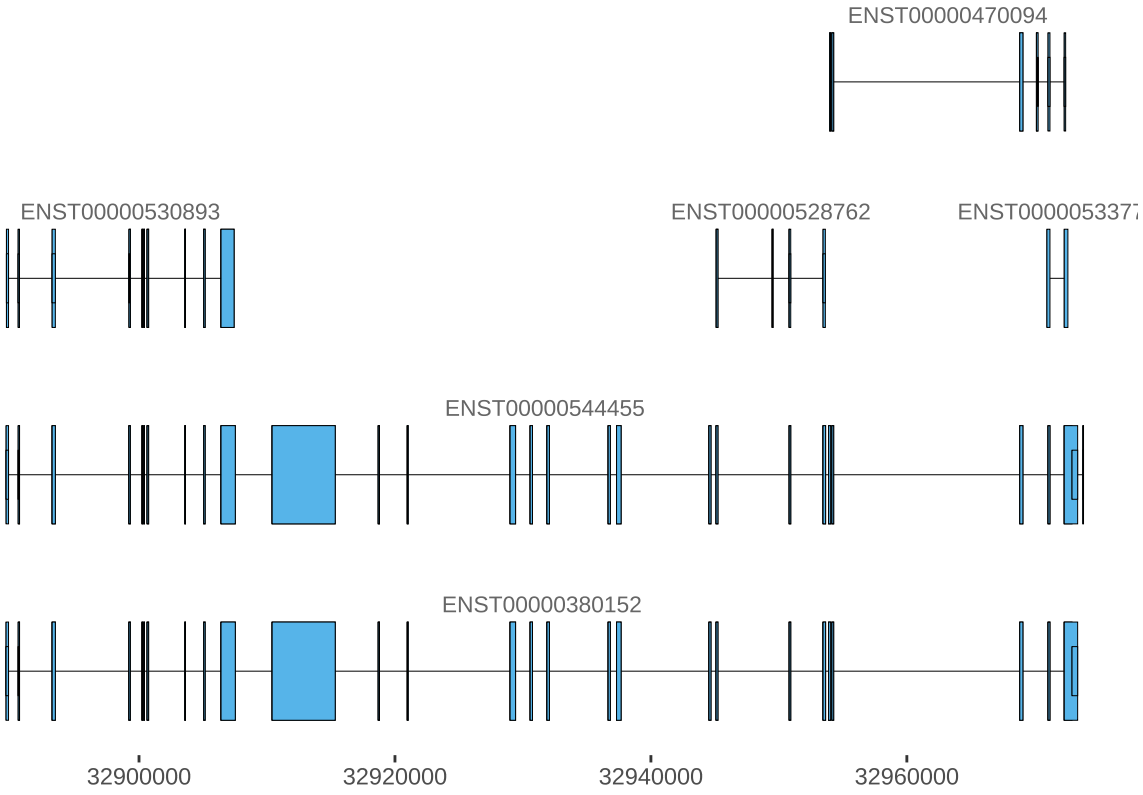
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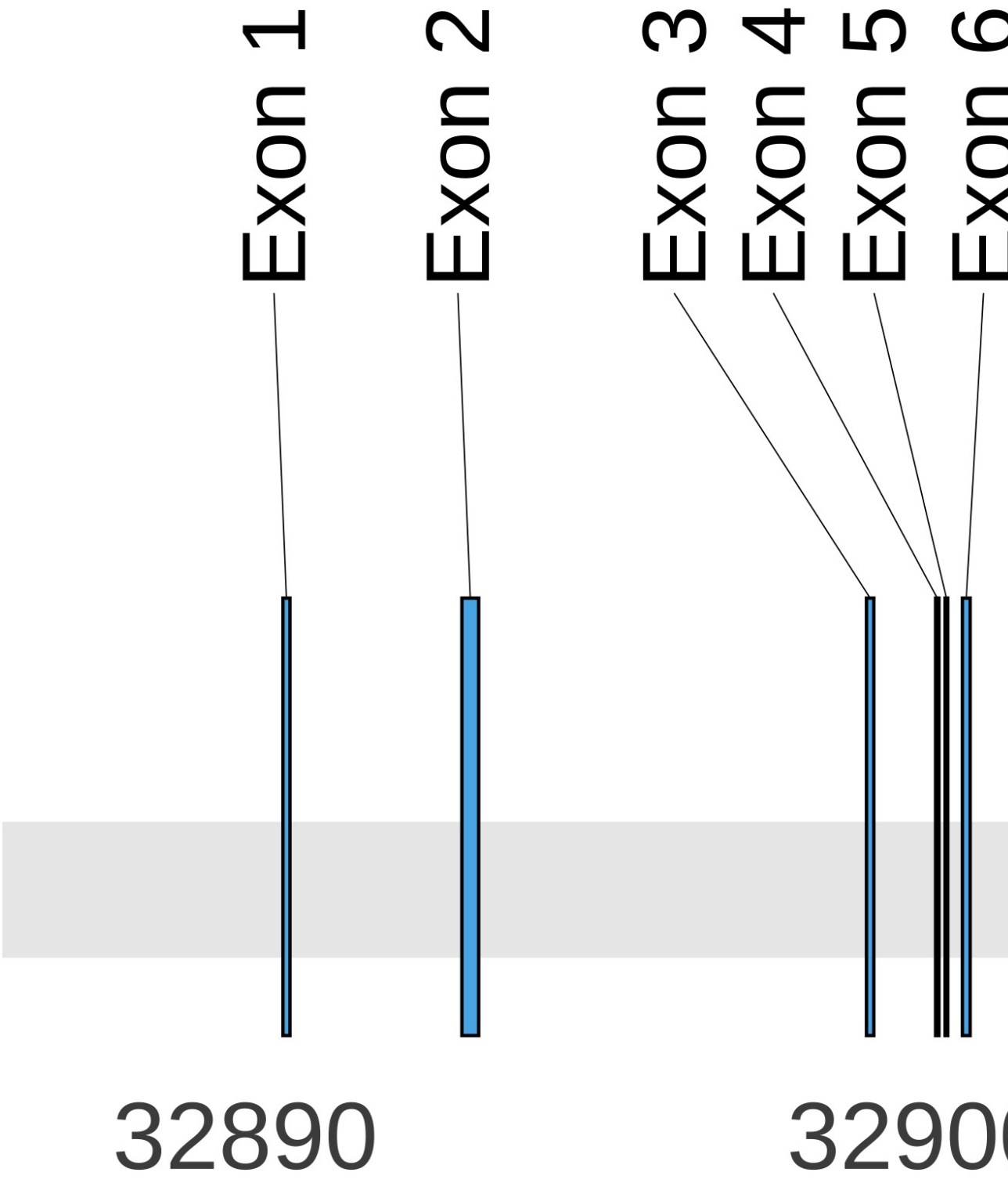

Figure 1: Mapping of the exons and introns in BRCA2 gene