## Consider the gene ARL6:

You will first study its location in the human genome and its transcriptional structure, according to the Ensembl genome browser. Go to the browser at <a href="https://www.ensembl.org/">https://www.ensembl.org/</a> and look for the gene symbol ARL6 using the search box. Answer the following questions:

- Symbol and name of the gene
  - SYMBOL: HGNC:13210
  - NAME OF THE GENE: ARL6
- **Chromosomal location**. You must include the chromosome, start coordinate, end coordinate, and strand.
  - o <u>Chromosome 3: 97,764,521-97,801,229</u> forward strand.
  - o GRCh38:CM000665.2

Navigate to the *Location* tab:

• **Genomic context**. Closer upstream (closest to its 5' end) and downstream (closest to its 3' end) protein-coding genes, presence of non-protein coding genes between the nearest upstream and downstream protein-coding genes.

The protein-coding gene closer to the 5' end is the EPHA6-201 but in between there is a IncRNA. The protein-coding gene closer to the 3' end is the CRYBG3 but there is a IncRNA in between.

Return to the *Gene* tab. Regarding the transcriptional structure of the gene (consider only *GENCODE basic* transcripts):

- Number of different transcripts and their lengths (how many of them encode a protein?)
  - This gene has 8 transcripts:
    - ARL6-203 → 3988 bps
    - ARL6-201 → 1630 bps
    - ARL6-202 → 1590 bps
    - ARL6-208 →937 bps
    - ARL6-204 → 675 bps
    - ARL6-205 → 1705 bps
    - ARL6-207 → 851 bps
    - ARL6-206 → 553 bps
    - 5 of them are protein coding, and other 2 are protein coding CDS not defined.
- Number of different proteins

For all of this, there are 3 types of proteins.

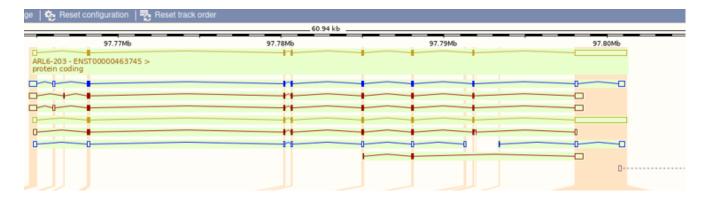
- o (length in amino acids)
- o ARL6-203 → 186aa
- o ARL6-201 → 186aa
- o ARL6-202 → 186aa
- o ARL6-208 →193aa
- $\circ$  ARL6-204  $\rightarrow$  54aa
- o ARL6-205 → 186aa
- Number of constitutive exons (present in all the transcripts) and alternative exons (only present in some transcripts)

There are 3 constitutive exons and there are 9 alternative exons

## • Alternative splicing mechanisms

С

Include a caption of the gene region as displayed in Ensembl and indicate consitutive and alternative exons, as well as alternative splicing mechanisms.



Highlight some other relevant information of the gene you may find in other databases discussed in this topic.

We have found 2 isoforms

To identify constitutive exons, we examine those present in all transcripts, while for alternative exons, we focus on those found only in specific transcripts. The gene exhibits alternative splicing through exon skipping and alternative 5' or 3' splice sites.

Regarding additional gene information from other databases, UniProt indicates 2 isoforms, with one being the canonical sequence; however, there are 3 computationally mapped isoforms. NCBI shows 2 current annotation releases and one outdated version.