Structural variant report

Up to one A4 page of text (font 12 Arial, spacing 1.5).

Pick one variant from
/mnt/practical2022/part3/final_report_tsv/report.SV.mRNA.overlap.chr.tsv

Please note the ID of the variant and its coordinates and send it to me including your name in the email.

You can download the reference sequence, gff3 file, transcript sequences and BAM files (available after the end of practical) from here:

https://osf.io/b7uxe/

1. Short introduction - what are structural variants. Why are they important?
2. How was the structural variant identified?
3. Describe the structural variant. Include an image from IGV.
4. What are the possible consequences of this structural variant? Does it interrupt a gene, which part of the gene?

Please include at least 5-10 references.

Questions for Parts 2 & 3

1. Please include question numbers.
2. Please answer in full sentences.
3. Please use your own words in explanations.
4. There is no need to reference the Q&A part of report.

Tips

Visualizing bam files in IGV:

1. Download bam and bai files with your variant ID.
2. First upload fa and fai files (highlight both)
3. Then upload gff3 file
4. Then upload bam and bai files (highlight both)

Performing BLAST search

1. IDs of genes overlapping SVs can be found in SVs.genes.xlsx
2. Sequences can be obtained from Express617_v1_cds.fa
3. BLAST search can be performed using this website
   https://blast.ncbi.nlm.nih.gov/Blast.cgi