Gene-centered View of Human Structural Variants -GeneVar

October 13, 2020

Methods

Module 1: SV<-> gene
In: dbVar + Gencode
Out TSV: ***var_id***, *gene_id*

Module 2: Gene impact
In: dbVar + Gencode
Out TSV: ***var_id***, *gene_id*, *elt_type*

Module 3: SV<-> clinical SNV/indels
In: dbVar + (ClinVar or custom variant set)
Out TSV: ***var_id***, *clinvar_pathogenic_snv*

Module 4: Allele frequency
In: dbVar + gnomAD-SV
Out TSV: ***var_id***, *af*

Module 5: SV<-> clinical SVs
In: dbVar + (ClinVar or ClinGen)
Out TSV: ***var_id***, *clinvar_pathogenic_sv*

Module 6: External resources
In: gene_id + OMIM + ...
Out TSV: ***gene_id***, *omim_url*

. . .

Module N: Awesome info
In: awesome DB
Out TSV: ***variant_id/gene_id***, *awesome_info*

TSV files

App locally or online
Status: *prototype*

Tools

- SnpEff
- AnnotSV
- bedtools
- Custom R or python scripts
- R-Shiny

Resources

- dbVar
- Gencode
- gnomAD-SV
- OMIM
- ClinVar
- ClinGen
- QC tracks from UCSC
- GWAS catalog SNVs
- ...

App prototype