

## **AnnotSV and knotAnnotSV: Human structural variations annotations, ranking and analysis**

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With the dramatic increase of pangenomic analysis, Human geneticists generate large amount of genomic data including millions of small variants (SNV/Indel) but also thousands of structural variations (SV) mainly from next-generation sequencing and array-based techniques. To help identifying human pathogenic SV, we have developed a webserver dedicated to their annotation and ranking (AnnotSV, PMID:29669011, PMID: 34023905) as well as their visualization and interpretation (knotAnnotSV, PMID: 34023905) at the following address: <https://www.lbgi.fr/AnnotSV/>. First, the available data sources in our annotation engine (AnnotSV) includes, among others, databases such as DGV, gnomAD, DDD, OMIM, intolerance score and known pathogenic SV (dbVar, ClinVar and ClinGen) as well as users own annotations (e.g. patient's SNV/indel...). Second, a phenotype driven analysis based on HPO and Exomiser has been implemented. Third, an automatic SV classification based on the latest ACMG recommendations (PMID:31690835) is available. Finally, knotAnnotSV displays the annotated SV in an interactive way including popups, filtering options, advanced colouring to highlight pathogenic SV and hyperlinks to the UCSC genome browser or other public databases. The annotation is available for the SV as a whole (full/compact mode) or divided for each overlapping gene (split/expanded mode). Output can be either visualized in a web browser directly or using a specific link, or downloaded as a tab separated file. To our knowledge, this makes our webserver the most comprehensive online SV annotation and interpretation tool.

This new version of the AnnotSV web server can be accessed at the following address: <https://lbgi.fr/AnnotSV/>. The underlying annotation engine (AnnotSV) has been upgraded to version 3 and is getting more and more citations since its publication in 2018 (80 in total, decomposed in 2 in 2018, 8 in 2019, 17 in 2020 and already 53 in 2021).

### **Biography**

Véronique Geoffroy professional interests are focused on Next Generation Sequencing data, especially the use of computational methods in human genetics and genomics. She strongly supports open sciences and she has developed under the GNU-GPL open source license, AnnotSV, a program designed for annotating and ranking Structural Variations.