AWESOME team



One project idea...topic modeling

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EREADME.md

ClusterDuck

Disease clustering from phenotypic literature data through Document Understanding, Comprehension and Knowledge (doi:)

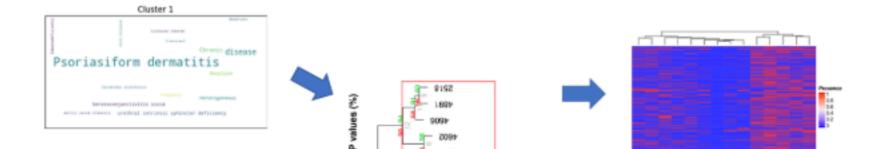


phenotypeXpression

esearchers and clinicians NPs.

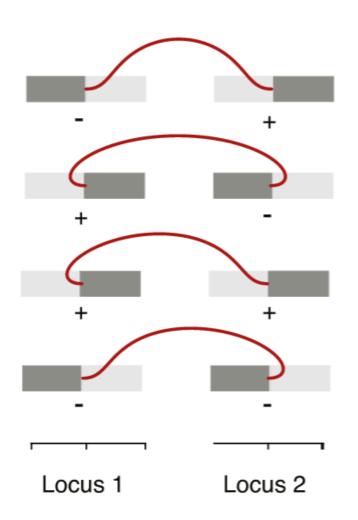
Subclassification of disease states based on the intersection of literature and expression

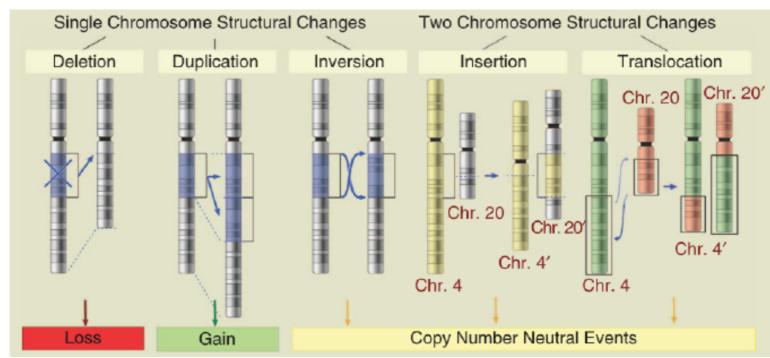
PhenotypeXpression (PhenoX) is a prototype precision medicine tool for clinicians to rapidly aggregate the publicly available gene expression data and literature available from the NCBI. It requires a minimum of resources and can quickly survey publicly available research. By combining the subtype HPO and DOID information with GEO dataset expression profiles, each sub-classification of a disease is given a characteristic fingerprint. This enables clinicians to (1) identify a set of genes that have altered expression in a phenotype and its related targets to aid in research, (2) use those target genes to inform treatment selection by affected pathway, and (3) rapidly identify public datasets that are available for comparison with their own gene expression data.



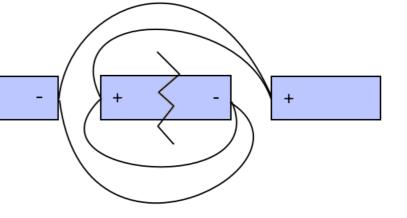
Structural Variations

- (-) left-side of breakpoint
- (+) right-side of breakpoint





- + deletion+ tan. dup.+ inversion
- inversion



Goal: Build R package to annotate SV BEDPEs

Inputs:
SV VCF/ BEDPE File
Window Size (length)
Reference genome (hg19/hg38)
Annotations:
Gencode, Ensembl
Input data: GTEx
RNAseq data

R package: using GRanges, data.table, annotatedb Outputs:
Annotated VCF/Bed
PE file with affected
Gene Expression data

Challenges: translocations? Gene fusions? Features?

