18 September, 2018

SNATCNV: Single Nucleotide Association Test for CNV analysis

Page: https://github.com/hamidrokny/SNATCNV

Input data:

SNATCNV takes CNV data in the following format:

Chr: Chromosome information of a CNV.

Start: Starting position of a CNV.

End Ending position of a CNV.

Patient_ID: Sample ID.

CNV_type: should be "Del" or "Dup".

case_control: should be "Case" or "Control".

Report_ID: "PMID for the CNV"; this filed can be "NA" as well.

Patient_gender: Can be "M" or "F"; if you have no gender information, put "NA".

Table.1 Example of the CNV dataset.

chr	start	end	Patient_ID	CNV_type	case_control	Report_ID	Patient_gender
chr1	10001	10077413	A12764	Del	Case	25503496	M
chr1	147198154	148498814	A12766	Dup	Case	22970919	F
chr2	51237623	52359612	A12766	Del	Case	27986125	F
chr2	234091785	239251404	A87021	Del	Control	22970919	M
chr6	7601267	7701391	A61230	Dup	Case	NA	NA
chr6	2045813	2145514	A61346	Del	Control	23970967	NA
chr11	19435001	19436039	A12317	Del	Case	22970919	F
chrX	26534242	27538142	A98125	Dup	Control	NA	M

Setting arguments

SNATCNV_indvbased:

CNV_type='Del'.

Can be specify by 'Del', 'Dup' or 'Both'.

CNV_size=10000.

Minimum length (base pair) for a CNV; all CNVs with less than this size will be excluded from the analysis.

GENOME_BUILD= 'GENOME_BUILD_19.

Can be specific by 'GENOME_BUILD_18' or GENOME_BUILD_19'.

PATH_INPUT='../input'.

Path to case CNVs.

PATH_OUTPUT='../output'.

Path to control CNVs.

FILE_CASE_NAME='ASD_cnv_case.txt'.

Filename for case CNVs.

FILE_CONTROL_NAME='ASD_control_case.txt'.

Filename for control CNVs.

NUMBER_OF_CHR=24.

Number of chromosome in the input file.

SNATCNV_permutation:

number_CASE_DEL=12721.

Number of case samples with deletion.

number_CASE_DUP=4441.

Number of case samples with duplication.

number_CONTROL_DEL=11924.

Number of control samples with deletion.

number_CONTROL_DUP=5015.

Number of control samples with duplication.

number_case=number_CASE_DUP.

If you want to run permutation to identify significant threshold for deleted regions, set this value to "number_CASE_DEL"; otherwise set it to "number_CASE_DUP".

number_control=number_CONTROL_DUP.

If you want to run permutation to identify significant threshold for deleted regions, set this value to "number_CONTROL_DEL"; otherwise set it to "number_CONTROL_DUP".

resultname='../permutation_ASD_del.txt'.

Path to save permutation output.

max_cnv=600.

Maximum number of case and control samples with CNV that overlap with the same position.

number_of_permutation=500000.

Number of permutation.

Contact

Hamid Alinejad-Rokny <h.alinejad@ieee.org>, Centre for Medical Research, The University of Western Australia, Perth, 6008, Australia.