**ACAT-V**

**ACAT-V: a fast gene- or set-based association test based on Cauchy distribution**

Aggregated Cauchy Association Test based on Variant-level p-values (ACAT-V) is a general and computationally efficient p-value combination test. It was originally proposed by [Liu et al, 2019](https://www.sciencedirect.com/science/article/pii/S0002929719300023). Only summary statistics are required for the test. We have implemented ACAT-V by efficient C and C++ code and made it compatible with the output from [fastGWA-GLMM](http://172.16.13.142/software/gcta/index.html" \l "fastGWA-GLMM). The test is very efficient. For example, to run a genome-wide analysis through 26,292 genes, the average runtime (#SNPs = ~12 million) is around 30 seconds for one trait. Credits: [Longda Jiang](mailto:longda.jiang@uq.edu.au), [Hailing Fang](mailto:fanghailing@westlake.edu.cn) and [Jian Yang](http://researchers.uq.edu.au/researcher/2713).

**References**

Liu, Y., Chen, S., Li, Z., Morrison, A. C., Boerwinkle, E., & Lin, X. (2019). ACAT: A fast and powerful p value combination method for rare-variant analysis in sequencing studies. The American Journal of Human Genetics, 104(3), 410-421.

Jiang L., Zheng Z., Yang J. (2021). FastGWA-GLMM: a generalized linear mixed model association tool for biobank-scale data, 12 February 2021, PREPRINT (Version 1) available at Research Square [https://doi.org/10.21203/rs.3.rs-128758/v1](https://europepmc.org/article/PPR/PPR283012)

--acat  
To perform ACAT-V test (based on GWAS results from fastGWA-GLMM).

--gene-list gene\_list.txt  
To input a gene list with gene start and end positions.

Input file format  
gene\_list.txt (columns are gene ID, chromosome, left- and right-side boundary of the gene region)

1 19774 19899 Gene1

1 34627 35558 Gene2

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Please click the link below to download the gene list file (provided by [Plink1.9](https://www.cog-genomics.org/plink/1.9/resources#genelist)).

Gene list (hg19): [glist-hg19.txt](http://172.16.13.142/software/gcta/res/glist-hg19.txt)

--snp-list gwas.fastGWA  
To input the GWAS summary statistics produced by [fastGWA-GLMM](http://172.16.13.142/software/gcta/index.html#fastGWA-GLMM).

--max-maf 0.01  
To specify the maximum minor allele frequency (MAF) allowed for a variant to be included in the ACAT-V test. Any variant with MAF larger than this value will be excluded.

--min-mac 20  
To specify the minimum minor allele count (MAC) allowed for a variant to be included in the ACAT-V test. Any variant with MAC smaller than this value will be excluded.

--wind 0  
To specify a flanking region of a gene (unit: kilobase). By default, the region to be tested is +/- 0kb of a gene.

Examples

# Gene-based ACAT-V test for rare variants

gcta64 --acat --snp-list assoc.fastGWA --gene-list gene\_list.txt --max-maf 0.01 --min-mac 20 --wind 0 --out test.acat.res

Output file format (columns are chromosome, gene ID, left- and right-side boundary of the gene region tested, number of qualified variants in the gene region, and ACAT-V test p-value)

CHR GENE START END SNP\_NUM P\_ACAT

1 FAM87B 752750 755214 2 0.772495

1 LINC01128 762970 794826 13 0.250037

1 LOC100130417 852952 854817 4 0.416386

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