

Resource Summary Report

Generated by [ASWG](#) on Aug 4, 2026

SCORE-SEQ

RRID:SCR_013121

Type: Tool

Proper Citation

SCORE-SEQ (RRID:SCR_013121)

Resource Information

URL: <http://dlin.web.unc.edu/software/SCORE-Seq/>

Proper Citation: SCORE-SEQ (RRID:SCR_013121)

Description: A command-line program for detecting disease associations with rare variants in sequencing studies. The mutation information is aggregated across multiple variant sites of a gene through a weighted linear combination and then related to disease phenotypes through appropriate regression models. The weights can be constant or dependent on allele frequencies and phenotypes. The association testing is based on score-type statistics. The allele-frequency threshold can be fixed or variable. Statistical significance can be assessed by using asymptotic normal approximation or resampling. The current release covers binary and continuous traits with arbitrary covariates under case-control and cross-sectional sampling. (entry from Genetic Analysis Software)

Synonyms: SCORE-type tests for detecting disease associations with rare variants in SEQuencing Studies

Resource Type: software resource, software application

Keywords: gene, genetic, genomic

Resource Name: SCORE-SEQ

Resource ID: SCR_013121

Alternate IDs: nlx_154611

Record Creation Time: 20220129T080314+0000

Record Last Update: 20260801T191101+0000

Ratings and Alerts

No rating or validation information has been found for SCORE-SEQ.

No alerts have been found for SCORE-SEQ.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 6 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Fu X, et al. (2020) Association Between Lifelong Premature Ejaculation and Polymorphism of Tryptophan Hydroxylase 2 Gene in the Han Population. Sexual medicine, 8(2), 223.

Meyer CG, et al. (2016) TLR1 Variant H305L Associated with Protection from Pulmonary Tuberculosis. PloS one, 11(5), e0156046.

Tang ZZ, et al. (2015) Meta-analysis for Discovering Rare-Variant Associations: Statistical Methods and Software Programs. American journal of human genetics, 97(1), 35.

Xing C, et al. (2014) Performance of statistical methods on CHARGE targeted sequencing data. BMC genetics, 15, 104.

Ayers KL, et al. (2013) Identification of grouped rare and common variants via penalized logistic regression. Genetic epidemiology, 37(6), 592.

Bacanu SA, et al. (2012) Comparison of statistical tests for association between rare variants and binary traits. PloS one, 7(8), e42530.