Resource Summary Report

Generated by NIF on Apr 16, 2025

NGSrich

RRID:SCR_001333
Type: Tool

Proper Citation

NGSrich (RRID:SCR_001333)

Resource Information

URL: http://sourceforge.net/projects/ngsrich/

Proper Citation: NGSrich (RRID:SCR_001333)

Description: Software for target enrichment performance for next-generation sequencing.

Resource Type: software resource

Defining Citation: PMID:22290614

Keywords: standalone software, java, bio.tools

Funding:

Resource Name: NGSrich

Resource ID: SCR_001333

Alternate IDs: OMICS_03603, biotools:ngsrich

Alternate URLs: https://bio.tools/ngsrich

Record Creation Time: 20220129T080206+0000

Record Last Update: 20250410T064703+0000

Ratings and Alerts

No rating or validation information has been found for NGSrich.

No alerts have been found for NGSrich.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 10 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Barbosa-Gouveia S, et al. (2021) Utility of Gene Panels for the Diagnosis of Inborn Errors of Metabolism in a Metabolic Reference Center. Genes, 12(8).

Ahmed Z, et al. (2021) Genomics pipelines to investigate susceptibility in whole genome and exome sequenced data for variant discovery, annotation, prediction and genotyping. PeerJ, 9, e11724.

Roca I, et al. (2020) PattRec: An easy-to-use CNV detection tool optimized for targeted NGS assays with diagnostic purposes. Genomics, 112(2), 1245.

Barbosa-Gouveia S, et al. (2020) Identification of a Novel Variant in EARS2 Associated with a Severe Clinical Phenotype Expands the Clinical Spectrum of LTBL. Genes, 11(9).

Barbosa-Gouveia S, et al. (2019) Identification and Characterization of New Variants in FOXRED1 Gene Expands the Clinical Spectrum Associated with Mitochondrial Complex I Deficiency. Journal of clinical medicine, 8(8).

Fernández-Marmiesse A, et al. (2019) Septo-optic dysplasia caused by a novel FLNA splice site mutation: a case report. BMC medical genetics, 20(1), 112.

Brovkina OI, et al. (2018) The Ethnic-Specific Spectrum of Germline Nucleotide Variants in DNA Damage Response and Repair Genes in Hereditary Breast and Ovarian Cancer Patients of Tatar Descent. Frontiers in oncology, 8, 421.

Khaiboullina SF, et al. (2017) Cerebellar Atrophy and Changes in Cytokines Associated with the CACNA1A R583Q Mutation in a Russian Familial Hemiplegic Migraine Type 1 Family. Frontiers in cellular neuroscience, 11, 263.

Boichuk S, et al. (2017) A Novel Receptor Tyrosine Kinase Switch Promotes Gastrointestinal Stromal Tumor Drug Resistance. Molecules (Basel, Switzerland), 22(12).

Zong L, et al. (2015) Mutations in apoptosis-inducing factor cause X-linked recessive auditory neuropathy spectrum disorder. Journal of medical genetics, 52(8), 523.