

# Resource Summary Report

Generated by [RRID](#) on Aug 6, 2026

## [NGSrich](#)

RRID:SCR\_001333

Type: Tool

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### Proper Citation

NGSrich (RRID:SCR\_001333)

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### 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

**Availability:** Free, Available for download, Freely available

**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:** 20260801T190139+0000

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### Ratings and Alerts

No rating or validation information has been found for NGSrich.

No alerts have been found for NGSrich.

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### Data and Source Information

## Usage and Citation Metrics

We found 10 mentions in open access literature.

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

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.