# Resource Summary Report

Generated by dkNET on May 19, 2025

## Variant Reporter Software

RRID:SCR_002329
Type: Tool

### Proper Citation

Variant Reporter Software (RRID:SCR_002329)

### Resource Information

**URL:** http://www.thermofisher.com/order/catalog/product/4385261

**Proper Citation:** Variant Reporter Software (RRID:SCR_002329)

**Description:** THIS RESOURCE IS NO LONGER IN SERVICE, documented on April 28, 2017.

Software that performs comparative sequencing, also known as direct sequencing, medical sequencing, PCR sequencing and resequencing with DNA sequencing files. The software is designed for reference based and non-reference based analysis such as mutation detection and analysis, SNP discovery and validation and sequence confirmation.

**Resource Type:** software application, data analysis software, data processing software, sequence analysis software, software resource

**Keywords:** comparative sequencing, mutation detection, snp discovery, sequence confirmation

**Funding:**

**Availability:** THIS RESOURCE IS NO LONGER IN SERVICE

**Resource Name:** Variant Reporter Software

**Resource ID:** SCR_002329

**Alternate IDs:** OMICS_01818

**Old URLs:** http://www.lifetechnologies.com/order/catalog/product/4385261

**Record Creation Time:** 20220129T080212+0000

**Record Last Update:** 20250517T055528+0000

## Ratings and Alerts

No rating or validation information has been found for Variant Reporter Software.

No alerts have been found for Variant Reporter Software.

## Data and Source Information

**Source:** SciCrunch Registry

## Usage and Citation Metrics

We found 85 mentions in open access literature.

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


Kopernik A, et al. (2025) Sanger validation of WGS variants. Scientific reports, 15(1), 3621.

Kumar KS, et al. (2024) Mutation Analysis of Exon 1 in the Hemoglobin Subunit Beta (HBB) Gene in Beta-Thalassemia. Cureus, 16(7), e65198.

Raggio V, et al. (2023) Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. Human genomics, 17(1), 14.

Hong JH, et al. (2023) Lymphocyte activation gene (LAG)-3 is a potential immunotherapeutic target for microsatellite stable, programmed death-ligand 1 (PD-L1)-positive endometrioid endometrial cancer. Journal of gynecologic oncology, 34(2), e18.

Chiereghin C, et al. (2023) In-depth genetic and molecular characterization of diaphanous related formin 2 (DIAPH2) and its role in the inner ear. PloS one, 18(1), e0273586.

Torrado M, et al. (2022) Identification of an elusive spliceogenic MYBPC3 variant in an otherwise genotype-negative hypertrophic cardiomyopathy pedigree. Scientific reports, 12(1), 7284.

Lu H, et al. (2022) Association of the P441L KCNQ1 variant with severity of long QT syndrome and risk of cardiac events. Frontiers in cardiovascular medicine, 9, 922335.

Civeira-Marín M, et al. (2022) APOE Genotypes Modulate Inflammation Independently of Their Effect on Lipid Metabolism. International journal of molecular sciences, 23(21).

Bea AM, et al. (2021) ANGPTL3 gene variants in subjects with familial combined hyperlipidemia. Scientific reports, 11(1), 7002.

Ozi?b?o D, et al. (2021) TBC1D24 emerges as an important contributor to progressive postlingual dominant hearing loss. Scientific reports, 11(1), 10300.

Wang B, et al. (2021) The correlation of next-generation sequencing-based genotypic profiles with clinicopathologic characteristics in NPM1-mutated acute myeloid leukemia. BMC cancer, 21(1), 788.

Nadin-Davis SA, et al. (2021) Relationships between fox populations and rabies virus spread in northern Canada. PloS one, 16(2), e0246508.

Chiereghin C, et al. (2021) SLC22A4 Gene in Hereditary Non-syndromic Hearing Loss: Recurrence and Incomplete Penetrance of the p.C113Y Mutation in Northwest Africa. Frontiers in genetics, 12, 606630.

Foret JT, et al. (2021) Network Modeling Sex Differences in Brain Integrity and Metabolic Health. Frontiers in aging neuroscience, 13, 691691.

Huang J, et al. (2021) A hemizygous p.R204Q mutation in the ALAS2 gene underlies X-linked sideroblastic anemia in an adult Chinese Han man. BMC medical genomics, 14(1), 107.

Torrado M, et al. (2021) A cryptic splice-altering KCNQ1 variant in trans with R259L leading to Jervell and Lange-Nielsen syndrome. NPJ genomic medicine, 6(1), 21.

Previtera F, et al. (2021) Gene Polymorphism in Five Target Genes of Immunosuppressive Therapy and Risk of Development of Preeclampsia. Healthcare (Basel, Switzerland), 9(7).

Donnette M, et al. (2021) Pharmacokinetics and pharmacogenetics of liposomal cytarabine in AML patients treated with CPX-351. Journal of controlled release : official journal of the Controlled Release Society, 338, 244.

Mihalcea CE, et al. (2020) Analysis of TP53 gene and particular infrastructural alterations in invasive ductal mammary carcinoma. Romanian journal of morphology and embryology = Revue roumaine de morphologie et embryologie, 61(2), 441.

Chmielewski P, et al. (2020) Can Circulating Cardiac Biomarkers Be Helpful in the Assessment of LMNA Mutation Carriers? Journal of clinical medicine, 9(5).