

# Resource Summary Report

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## RefSeqGene

RRID:SCR\_013787

Type: Tool

### Proper Citation

RefSeqGene (RRID:SCR\_013787)

### Resource Information

**URL:** <http://www.ncbi.nlm.nih.gov/refseq/rsg/>

**Proper Citation:** RefSeqGene (RRID:SCR\_013787)

**Description:** A data set of defined genomic sequences used as reference standards for well-characterized genes. These standard nucleotide sequences serve as foundations for locating mutations, establishing conventions for numbering exons and introns, and defining the coordinates of other variations. Sequences are aligned to reference chromosomes. RefSeqGene is a subset of NCBI RefSeq.

**Synonyms:** NCBI RefSeqGene, RefSeqGene Project

**Resource Type:** information resource

**Defining Citation:** [PMID:18927115](#)

**Keywords:** data set, genomic sequences, reference standard

**Availability:** Free, Public

**Resource Name:** RefSeqGene

**Resource ID:** SCR\_013787

**License URLs:** <http://www.nlm.nih.gov/privacy.html>

**Record Creation Time:** 20220129T080318+0000

**Record Last Update:** 20260801T190454+0000

### Ratings and Alerts

No rating or validation information has been found for RefSeqGene.

No alerts have been found for RefSeqGene.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 53 mentions in open access literature.

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

Chen HH, et al. (2025) Population-specific polygenic risk scores for people of Han Chinese ancestry. *Nature*, 648(8092), 128.

DiPietro EE, et al. (2025) Genetics of ovulatory dysfunction and infertility: a scoping review and gene ontology analysis. *Frontiers in endocrinology*, 16, 1458711.

Burykina Y, et al. (2025) A Unique Case of a Child with Two Rare Hereditary Diseases: Familial Dilated Cardiomyopathy and Arterial Calcification. *International journal of molecular sciences*, 26(12).

Huang X, et al. (2025) Mutation spectra and genotype?phenotype analysis of congenital hypothyroidism in a neonatal population. *Biomedical reports*, 22(2), 30.

Lv W, et al. (2024) Extrachromosomal circular DNA orchestrates genome heterogeneity in urothelial bladder carcinoma. *Theranostics*, 14(13), 5102.

Pugacheva EM, et al. (2024) BORIS/CTCF epigenetically reprograms clustered CTCF binding sites into alternative transcriptional start sites. *Genome biology*, 25(1), 40.

Pranck?nien? L, et al. (2023) Microevolutionary processes analysis in the Lithuanian genome. *Scientific reports*, 13(1), 11941.

Ikeda S, et al. (2022) Disruption of piRNA machinery by deletion of ASZ1/GASZ results in the expression of aberrant chimeric transcripts in gonocytes. *The Journal of reproduction and development*, 68(2), 125.

Markova TV, et al. (2022) Clinical and genetic characterization of three Russian patients with pycnodysostosis due to pathogenic variants in the CTSK gene. *Molecular genetics & genomic medicine*, 10(5), e1904.

Chou KJ, et al. (2022) A new missense mutation of calcium sensing receptor with isoleucine replaced by serine at codon 857 leading to type V Bartter syndrome. *Experimental cell research*, 414(1), 113080.

Markova T, et al. (2021) Clinical and genetic characterization of autosomal recessive stickler syndrome caused by novel compound heterozygous mutations in the COL9A3 gene. *Molecular genetics & genomic medicine*, 9(3), e1620.

Erdoğan M, et al. (2021) The Genetic Analysis of Cystic Fibrosis Patients With Seven Novel Mutations in the CFTR Gene in the Central Anatolian Region of Turkey. *Balkan medical journal*, 38(6), 357.

Ziadi W, et al. (2021) STAT3 polymorphisms in North Africa and its implication in breast cancer. *Molecular genetics & genomic medicine*, 9(8), e1744.

Urnikyte A, et al. (2021) Genome-Wide Landscape of North-Eastern European Populations: A View from Lithuania. *Genes*, 12(11).

Penitenti F, et al. (2021) Clinical presentation, genotype-phenotype correlations, and outcome of pancreatic neuroendocrine tumors in Von Hippel-Lindau syndrome. *Endocrine*, 74(1), 180.

Hirsch D, et al. (2021) Molecular characterization of ulcerative colitis-associated colorectal carcinomas. *Modern pathology : an official journal of the United States and Canadian Academy of Pathology, Inc*, 34(6), 1153.

Liu W, et al. (2021) Analysis of STAG3 variants in Chinese non-obstructive azoospermia patients with germ cell maturation arrest. *Scientific reports*, 11(1), 10077.

Wu CY, et al. (2021) Lipopolysaccharide stimulation test on cultured PBMCs assists the discrimination of cryopyrin-associated periodic syndrome from systemic juvenile idiopathic arthritis. *Scientific reports*, 11(1), 11903.

Hirsch D, et al. (2020) Newly established gastrointestinal cancer cell lines retain the genomic and immunophenotypic landscape of their parental cancers. *Scientific reports*, 10(1), 17895.

Ahn YH, et al. (2020) Targeted Exome Sequencing Provided Comprehensive Genetic Diagnosis of Congenital Anomalies of the Kidney and Urinary Tract. *Journal of clinical medicine*, 9(3).