Resource Summary Report

Generated by NIF on Jul 31, 2026

NHLBI Exome Sequencing Project (ESP)

RRID:SCR_012761
Type: Tool

Proper Citation

NHLBI Exome Sequencing Project (ESP) (RRID:SCR_012761)

Resource Information

URL: http://evs.gs.washington.edu/EVS/

Proper Citation: NHLBI Exome Sequencing Project (ESP) (RRID:SCR_012761)

Description: The goal of the project is to discover novel genes and mechanisms contributing to heart, lung and blood disorders by pioneering the application of next-generation sequencing of the protein coding regions of the human genome across diverse, richly-phenotyped populations and to share these datasets and findings with the scientific community to extend and enrich the diagnosis, management and treatment of heart, lung and blood disorders. The groups participating and collaborating in the NHLBI GO ESP include: Seattle GO - University of Washington, Seattle, WA Broad GO - Broad Institute of MIT and Harvard, Cambridge, MA WHISP GO - Ohio State University Medical Center, Columbus, OH Lung GO - University of Washington, Seattle, WA WashU GO - Washington University, St. Louis, MO Heart GO - University of Virginia Health System, Charlottesville, VA ChargeS GO - University of Texas Health Sciences Center at Houston

Abbreviations: EVS

Synonyms: Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP)

Resource Type: data or information resource, database

Keywords: bio.tools, FASEB list

Funding: NHLBI

Resource Name: NHLBI Exome Sequencing Project (ESP)

Resource ID: SCR_012761

Alternate IDs: nlx_156901, biotools:esp, biotools:exome_variant_server

Alternate URLs: https://bio.tools/esp, https://bio.tools/exome_variant_server

Record Creation Time: 20220129T080312+0000

Record Last Update: 20260729T044310+0000

Ratings and Alerts

No rating or validation information has been found for NHLBI Exome Sequencing Project (ESP).

No alerts have been found for NHLBI Exome Sequencing Project (ESP).

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 2137 mentions in open access literature.

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

Xu H, et al. (2026) CUL1 variants cause severe neurodevelopmental disorders: Insights from human genetics and a zebrafish model of microcephaly. HGG advances, 7(1), 100542.

Salvador L, et al. (2025) DICER1 in pediatric and adult cancer predisposition populations: Prevalence, phenotypes, and mosaicism. Genetics in medicine : official journal of the American College of Medical Genetics, 27(5), 101385.

Mir R, et al. (2025) A rare likely pathogenic HLA-DRB1 variant with compromised immunity in severe COVID-19 patient and in-hospital mortality. Personalized medicine, 1.

Ji F, et al. (2025) Liver-specific gene PGRMC1 blocks c-Myc-induced hepatocarcinogenesis through ER stress-independent PERK activation. Nature communications, 16(1), 50.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Lu Y, et al. (2025) A novel variant in the MAP3K1 genomic locus reveals abnormal cell apoptosis as a potential pathogenic mechanism in 46, XY disorders of sex development. Molecular medicine reports, 32(2).

Ahmad R, et al. (2025) Identification of a Novel GRM1 Frameshift Variant in Two Pakistani Families Broadens the Genetic Landscape of Ultra-Rare Spinocerebellar Ataxia Type 13. Cerebellum (London, England), 24(5), 145.

Miyakoshi J, et al. (2025) PD-L1 phenotype classification based on expression in tumor and immune cells as a potential biomarker for optimizing anti-PD-1/CTLA-4

immunotherapies in NSCLC. Journal for immunotherapy of cancer, 13(12).

Liu Z, et al. (2025) Compound heterozygosity of a De novo 16q24.1 deletion and missense mutation in COX4I1 leads to developmental regression, intellectual disability, and seizures. Epilepsia open, 10(3), 942.

Zhao R, et al. (2025) A de novo SALL4 mutation causes unilateral renal agenesis by misregulating genes involved in kidney development. Orphanet journal of rare diseases, 20(1), 289.

Liu Y, et al. (2025) 18 individual genes underwent variant screening in a northwest Chinese group comprised 83 probands diagnosed with early-onset high myopia. PloS one, 20(9), e0329472.

Dong Y, et al. (2025) Multi-omics analysis reveals the association between intratumoral bacteria and somatic mutational signatures in oral squamous cell carcinoma. Journal of translational medicine, 24(1), 23.

LaBelle JJ, et al. (2025) Dissecting the immune landscape in pediatric high-grade glioma reveals cell state changes under therapeutic pressure. Cell reports. Medicine, 6(5), 102095.

Bazazzadegan N, et al. (2025) A Novel Candidate Gene MACF1 is Associated with Autosomal Dominant Non-syndromic Hearing Loss in an Iranian Family. Archives of Iranian medicine, 28(1), 63.

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

Sazhenova EA, et al. (2025) Genetic variants of the DLK1, KISS1R, MKRN3 genes in girls with precocious puberty. Vavilovskii zhurnal genetiki i selektsii, 29(2), 301.

Chen YL, et al. (2025) NDUFB7 mutations cause brain neuronal defects, lactic acidosis, and mitochondrial dysfunction in humans and zebrafish. Cell death discovery, 11(1), 82.

Dong J, et al. (2025) A Case Study Identified a New Mutation in the TTN Gene for Inherited Hypertrophic Cardiomyopathy. International journal of general medicine, 18, 447.

Thomas HB, et al. (2025) Bi-allelic variants in MRPL49 cause variable clinical presentations, including sensorineural hearing loss, leukodystrophy, and ovarian insufficiency. American journal of human genetics, 112(4), 952.

Pensato V, et al. (2025) Exploring NEK1 genetic variability in Italian amyotrophic lateral sclerosis patients. Journal of neurology, 272(7), 469.