Resource Summary Report

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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/

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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: biotools:esp, nlx_156901, biotools:exome_variant_server

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

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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 2231 mentions in open access literature.

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

Brantley KD, et al. (2026) Tumor genomics in patients younger than 40 years of age with metastatic breast cancer. NPJ precision oncology, 10(1).

Aslam F, et al. (2026) Molecular characterization of recessively inherited ataxic and neuropathic disorders in consanguineous Pakistani families. Scientific reports, 16(1), 6529.

Van Mackelenbergh MT, et al. (2026) Tumor mutations predict HER2-targeted therapy resistance in primary HER2-positive breast cancer. NPJ breast cancer, 12(1).

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.

Nasri M, et al. (2026) Identification of a novel ACADSB variant for the presymptomatic diagnosis of 2-Methylbutyryl-CoA dehydrogenase deficiency through newborn screening in Iran. Orphanet journal of rare diseases, 21(1), 13.

Li HH, et al. (2026) Generation of a PKP2 heterozygous knockout pig model of arrhythmogenic cardiomyopathy. Zoological research, 47(1), 139.

Ferrer M, et al. (2026) Clinical and molecular characterization of a novel pathogenic AIFM1 E336K mutation connecting mitochondrial dysfunction and neurodegeneration. Cell communication and signaling : CCS, 24(1).

Wu H, et al. (2026) Treatment with Minicircle DNA Expressing a FGF23 Fragment in a Clinically relevant Mouse Model of X-Linked Hypophosphatemic Rickets. Advanced science (Weinheim, Baden-Wurttemberg, Germany), 13(4), e08870.

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.

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.

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.

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).

Zheng W, et al. (2025) An intronic micro-deletion impacts the transcription and translation of PKD1 gene. Frontiers in genetics, 16, 1707053.

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.

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.

Wang Y, et al. (2025) Identification of candidate genes associated with bipolar disorder by whole-exome sequencing of a Chinese multi-affected pedigree. BMC psychiatry, 25(1), 612.

Perrone S, et al. (2025) How to Read a Next-Generation Sequencing Report for AML and MDS? What Hematologists Need to Know. Journal of clinical medicine, 14(24).