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: 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: 20250331T061109+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 2118 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

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.

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.

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

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.

Shu Y, et al. (2025) Dias-Logan syndrome with a de novo p.Leu360Profs*212 heterozygous pathogenic variant of BCL11A in a Chinese patient: A case report. SAGE open medical case reports, 13, 2050313X251314069.

Anselmino N, et al. (2024) Integrative Molecular Analyses of the MD Anderson Prostate Cancer Patient-derived Xenograft (MDA PCa PDX) Series. Clinical cancer research : an official journal of the American Association for Cancer Research, 30(10), 2272.

Fu M, et al. (2024) Single-cell RNA sequencing in donor and end-stage heart failure patients identifies NLRP3 as a therapeutic target for arrhythmogenic right ventricular cardiomyopathy. BMC medicine, 22(1), 11.

Riedhammer KM, et al. (2024) Implication of transcription factor FOXD2 dysfunction in syndromic congenital anomalies of the kidney and urinary tract (CAKUT). Kidney international, 105(4), 844.

Kim YN, et al. (2024) Human epidermal growth factor receptor-2 expression and subsequent dynamic changes in patients with ovarian cancer. Scientific reports, 14(1), 7992.

Yao G, et al. (2024) Comparative analysis of the mutational landscape and evolutionary patterns of pancreatic ductal adenocarcinoma metastases in the liver or peritoneum. Heliyon, 10(15), e35428.

Cheong TC, et al. (2024) Mechanistic patterns and clinical implications of oncogenic tyrosine kinase fusions in human cancers. Nature communications, 15(1), 5110.

Li S, et al. (2024) Biallelic loss-of-function variants in GON4L cause microcephaly and brain structure abnormalities. NPJ genomic medicine, 9(1), 55.

Nokin MJ, et al. (2024) RAS-ON inhibition overcomes clinical resistance to KRAS G12C-OFF covalent blockade. Nature communications, 15(1), 7554.

Khan A, et al. (2024) Further delineation of Wiedemann-Rautenstrauch syndrome linked with POLR3A. Molecular genetics & genomic medicine, 12(3), e2274.

Han G, et al. (2024) An atlas of epithelial cell states and plasticity in lung adenocarcinoma. Nature, 627(8004), 656.

Wu H, et al. (2024) Characterization of novel PHEX variants in X-linked hypophosphatemic rickets and genotype-PHEX activity correlation. The Journal of clinical endocrinology and metabolism.

Vetri L, et al. (2024) Whole Exome Sequencing as a First-Line Molecular Genetic Test in Developmental and Epileptic Encephalopathies. International journal of molecular sciences, 25(2).

Antunes LN, et al. (2024) Genetic heterogeneity in autosomal recessive hearing loss: a survey of Brazilian families. Frontiers in genetics, 15, 1409306.

Fujii E, et al. (2024) Genomic profiles of Japanese patients with vulvar squamous cell carcinoma. Scientific reports, 14(1), 13058.

Di Nottia M, et al. (2024) Severe mitochondrial encephalomyopathy caused by de novo variants in OPA1 gene. Frontiers in genetics, 15, 1437959.