Resource Summary Report

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NHLBI Grand Opportunity Exome Sequencing Project

RRID:SCR_010798
Type: Tool

Proper Citation

NHLBI Grand Opportunity Exome Sequencing Project (RRID:SCR_010798)

Resource Information

URL: https://esp.gs.washington.edu/

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Description: Project focused on understanding the contribution of rare genetic variation to heart, lung and blood disorders through the sequencing of well-phenotyped populations.

Abbreviations: NHLBI GO ESP, GO ESP

Synonyms: NHLBI Grand Opportunity Exome Sequencing Project (ESP), NHLBI GO Exome Sequencing Project (ESP)

Resource Type: knowledge environment

Keywords: next-generation sequencing, protein coding region, human, genome, phenotype, exome sequencing

Funding: NHLBI RC2 HL-103010;
NHLBI RC2 HL-102923;
NHLBI RC2 HL-102924;
NHLBI RC2 HL-102925;
NHLBI RC2 HL-102926

Resource Name: NHLBI Grand Opportunity Exome Sequencing Project

Resource ID: SCR_010798

Alternate IDs: OMICS_00277

Record Creation Time: 20220129T080300+0000

Record Last Update: 20260905T132647+0000

Ratings and Alerts

No rating or validation information has been found for NHLBI Grand Opportunity Exome Sequencing Project.

No alerts have been found for NHLBI Grand Opportunity Exome Sequencing Project.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 35 mentions in open access literature.

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

Su J, et al. (2025) Unmasking a Recessive Allele by a Rare Interstitial Deletion at 10q26.13q26.2: Prenatal Diagnosis of MMP21 -Related Disorder and Further Refine INSYN2A Involvement in the Postnatal Cognitive Phenotype. Molecular genetics & genomic medicine, 13(2), e70082.

Ren Y, et al. (2024) Preliminary Study on Clinical Characteristics and Pathogenesis of IQSEC2 Mutations Patients. Pharmacogenomics and personalized medicine, 17, 289.

Zhunussova G, et al. (2023) Determination of genetic predisposition to early breast cancer in women of Kazakh ethnicity. Oncotarget, 14, 860.

Wei X, et al. (2022) An Investigation of the Etiologies of Non-Immune Hydrops Fetalis in the Era of Next-Generation Sequence-A Single Center Experience. Genes, 13(12).

Wang Q, et al. (2022) Case report: A de novo RASopathy-causing SHOC2 variant in a Chinese girl with noonan syndrome-like with loose anagen hair. Frontiers in genetics, 13, 1040124.

Chen G, et al. (2022) A 6.3 Mb maternally derived microduplication of 20p13p12.2 in a fetus with Brachydactyly type D and related literature review. Molecular cytogenetics, 15(1), 6.

Liu M, et al. (2022) Clinical characteristics and genetics of ten Chinese children with PRRT2-associated neurological diseases. Frontiers in pediatrics, 10, 997088.

Zhou X, et al. (2021) Value of Exome Sequencing in Diagnosis and Management of Recurrent Non-immune Hydrops Fetalis: A Retrospective Analysis. Frontiers in genetics, 12, 616392.

Lee M, et al. (2021) A New Human Leukocyte Antigen Typing Algorithm Combined With Currently Available Genotyping Tools Based on Next-Generation Sequencing Data and Guidelines to Select the Most Likely Human Leukocyte Antigen Genotype. Frontiers in immunology, 12, 688183.

Luo X, et al. (2021) Mechanisms of Congenital Myasthenia Caused by Three Mutations in the COLQ Gene. Frontiers in pediatrics, 9, 679342.

Su J, et al. (2021) Novel compound heterozygous frameshift variants in WDR81 associated with congenital hydrocephalus 3 with brain anomalies: First Chinese prenatal case confirms WDR81 involvement. Molecular genetics & genomic medicine, 9(4), e1624.

Alharatani R, et al. (2020) Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human molecular genetics, 29(11), 1900.

Ávila-Arcos MC, et al. (2020) Population History and Gene Divergence in Native Mexicans Inferred from 76 Human Exomes. Molecular biology and evolution, 37(4), 994.

Zhang S, et al. (2019) Novel genotypes and phenotypes among Chinese patients with Floating-Harbor syndrome. Orphanet journal of rare diseases, 14(1), 144.

Thormann A, et al. (2019) Flexible and scalable diagnostic filtering of genomic variants using G2P with Ensembl VEP. Nature communications, 10(1), 2373.

Thakur N, et al. (2019) Impacts of single nucleotide polymorphisms in three microRNAs (miR-146a, miR-196a2 and miR-499) on the susceptibility to cervical cancer among Indian women. Bioscience reports, 39(4).

Zhunussova G, et al. (2019) Mutation Spectrum of Cancer-Associated Genes in Patients With Early Onset of Colorectal Cancer. Frontiers in oncology, 9, 673.

, et al. (2018) A Supplement to TRANSFUSION Abstract Presentations from the AABB Annual Meeting Boston, MA, October 13-16, 2018. Transfusion, 58 Suppl 2(Suppl 2), 6A.

Moosa S, et al. (2017) Novel compound heterozygous mutations in TELO2 in a patient with severe expression of You-Hoover-Fong syndrome. Molecular genetics & genomic medicine, 5(5), 580.

Rochman M, et al. (2017) Profound loss of esophageal tissue differentiation in patients with eosinophilic esophagitis. The Journal of allergy and clinical immunology, 140(3), 738.