

# Resource Summary Report

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## NHLBI Grand Opportunity Exome Sequencing Project

RRID:SCR\_010798

Type: Tool

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### Proper Citation

NHLBI Grand Opportunity Exome Sequencing Project (RRID:SCR\_010798)

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### Resource Information

**URL:** <https://esp.gs.washington.edu/>

**Proper Citation:** NHLBI Grand Opportunity Exome Sequencing Project (RRID:SCR\_010798)

**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:** 20260815T182408+0000

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## 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.

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## Data and Source Information

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

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## Usage and Citation Metrics

We found 35 mentions in open access literature.

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

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.

Zhunossova 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.

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.

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

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).

Zhunossova 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.

Ka S, et al. (2017) HLAScan: genotyping of the HLA region using next-generation sequencing data. *BMC bioinformatics*, 18(1), 258.