

Resource Summary Report

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Genetic Testing Registry

RRID:SCR_005565

Type: Tool

Proper Citation

Genetic Testing Registry (RRID:SCR_005565)

Resource Information

URL: <http://www.ncbi.nlm.nih.gov/gtr/>

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Description: Central location for voluntary submission of genetic test information by providers including the test's purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts and credentials. GTR aims to advance the public health and research into the genetic basis of health and disease. GTR is accepting registration of clinical tests for Mendelian disorders, complex tests and arrays, and pharmacogenetic tests. These tests may include multiple methods and may include multiple major method categories such as biochemical, cytogenetic, and molecular tests. GTR is not currently accepting registration of tests for somatic disorders, research tests or direct-to-consumer tests.

Abbreviations: GTR

Synonyms: NIH Genetic Testing Registry, GTR: Genetic Testing Registry

Resource Type: data or information resource, data repository, storage service resource, database, service resource

Keywords: genetic, gene, clinical, genetic test, condition, phenotype, disease name, trait, drug, protein, analyte, disease, laboratory, molecular, clinical, genetics, people

Availability: The community can contribute to this resource

Resource Name: Genetic Testing Registry

Resource ID: SCR_005565

Alternate IDs: nlx_144654, OMICS_01541

Record Creation Time: 20220129T080231+0000

Record Last Update: 20260801T190256+0000

Ratings and Alerts

No rating or validation information has been found for Genetic Testing Registry.

No alerts have been found for Genetic Testing Registry.

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 [ASWG](#) .

Jia RY, et al. (2025) Effects of Rare Coding Variants in Severe Early-Onset Obesity Genes in the Population-Based UK Biobank Study. The Journal of clinical endocrinology and metabolism, 110(11), e3774.

Eadon MT, et al. (2025) Clinical Pharmacogenetics Implementation Consortium Guideline for NAT2 Genotype and Hydralazine Therapy. Clinical pharmacology and therapeutics, 118(6), 1430.

Duarte JD, et al. (2024) Clinical Pharmacogenetics Implementation Consortium Guideline (CPIC)??for CYP2D6, ADRB1, ADRB2, ADRA2C, GRK4, and GRK5 Genotypes and Beta-Blocker Therapy. Clinical pharmacology and therapeutics, 116(4), 939.

Robinson KM, et al. (2024) Clinical Pharmacogenetics Implementation Consortium Guideline for CYP2B6 Genotype and Methadone Therapy. Clinical pharmacology and therapeutics, 116(4), 932.

Peng W, et al. (2022) Clinical and genomic features of Chinese lung cancer patients with germline mutations. Nature communications, 13(1), 1268.

Huang J, et al. (2022) PAGEANT: personal access to genome and analysis of natural traits. Nucleic acids research, 50(7), e39.

Nogee LM, et al. (2020) Genetic testing for rare pediatric lung disorders: The promise and the pitfalls. Pediatric investigation, 4(1), 59.

Kalatharan V, et al. (2018) Opportunities and Challenges for Genetic Studies of End-Stage Renal Disease in Canada. Canadian journal of kidney health and disease, 5, 2054358118789368.

Gelb BD, et al. (2018) ClinGen's RASopathy Expert Panel consensus methods for variant interpretation. Genetics in medicine : official journal of the American College of Medical Genetics, 20(11), 1334.

Fowler SA, et al. (2018) Variation among Consent Forms for Clinical Whole Exome Sequencing. *Journal of genetic counseling*, 27(1), 104.

O'Daniel JM, et al. (2017) A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. *Genetics in medicine : official journal of the American College of Medical Genetics*, 19(5), 575.

Knowles L, et al. (2017) Paving the road to personalized medicine: recommendations on regulatory, intellectual property and reimbursement challenges. *Journal of law and the biosciences*, 4(3), 453.

Del Vecchio F, et al. (2017) Next-generation sequencing: recent applications to the analysis of colorectal cancer. *Journal of translational medicine*, 15(1), 246.

Medeiros BC, et al. (2017) Isocitrate dehydrogenase mutations in myeloid malignancies. *Leukemia*, 31(2), 272.

Ryan SE, et al. (2016) A real-time ARMS PCR/high-resolution melt curve assay for the detection of the three primary mitochondrial mutations in Leber's hereditary optic neuropathy. *Molecular vision*, 22, 1169.

Luzum JA, et al. (2016) Pharmacogenetic Risk Scores for Perindopril Clinical and Cost Effectiveness in Stable Coronary Artery Disease: When Are We Ready to Implement? *Journal of the American Heart Association*, 5(3), e003440.

Eustace Ryan S, et al. (2015) Development and validation of a novel PCR-RFLP based method for the detection of 3 primary mitochondrial mutations in Leber's hereditary optic neuropathy patients. *Eye and vision (London, England)*, 2, 18.

Riffel AK, et al. (2015) CYP2D7 Sequence Variation Interferes with TaqMan CYP2D6 (*) 15 and (*) 35 Genotyping. *Frontiers in pharmacology*, 6, 312.

Huang W, et al. (2015) Distribution of fragile X mental retardation 1 CGG repeat and flanking haplotypes in a large Chinese population. *Molecular genetics & genomic medicine*, 3(3), 172.

, et al. (2015) Database resources of the National Center for Biotechnology Information. *Nucleic acids research*, 43(Database issue), D6.