Resource Summary Report

Generated by ASWG on May 2, 2025

GeneTests

RRID:SCR_010725
Type: Tool

Proper Citation

GeneTests (RRID:SCR_010725)

Resource Information

URL: http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab

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Description: The GeneTests Web site, a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, is available at no cost to all interested persons. By providing current, authoritative information on genetic testing and its use in diagnosis, management, and genetic counseling, GeneTests promotes the appropriate use of genetic services in patient care and personal decision making. At This Site: * GeneReviews: Expert-authored peer-reviewed disease descriptions * Laboratory Directory: International directory of genetic testing laboratories * Clinic Directory: International directory of genetics and prenatal diagnosis clinics * Educational Materials: Illustrated glossary, information on genetic services, PowerPoint presentations, annotated Internet resources We comply with the HONcode standard for trustworthy health information.

Abbreviations: GeneTests

Synonyms: GeneTests: Clinical Genetic Information Resource

Resource Type: portal, topical portal, analysis service resource, database, training material, biomaterial analysis service, narrative resource, data or information resource, material analysis service, service resource, production service resource

Funding: NCI ;
NHGRI 1 P41 LM/HG 06029;
NLM 1 P41 LM/HG 06029;
NLM contract N01-LM-4-3505;
NLM 5 P41 LM07242;
NLM 2 P41 LM 06001;

DOE DE-FG03-02ER63301/A00

Resource Name: GeneTests

Resource ID: SCR_010725

Alternate IDs: nlx_94696

Record Creation Time: 20220129T080300+0000

Record Last Update: 20250502T060009+0000

Ratings and Alerts

No rating or validation information has been found for GeneTests.

No alerts have been found for GeneTests.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 12 mentions in open access literature.

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

Chen X, et al. (2021) 11p11.12p12 duplication in a family with intellectual disability and craniofacial anomalies. BMC medical genomics, 14(1), 99.

Chen K, et al. (2020) Cbl Proto-Oncogene B (CBLB) c.197A>T Mutation Induces Mild Metabolic Dysfunction in Partial Type I Multiple Symmetric Lipomatosis (MSL). Diabetes, metabolic syndrome and obesity : targets and therapy, 13, 3535.

Shokeen Y, et al. (2018) Identification of Prognostic and Susceptibility Markers in Chronic Myeloid Leukemia Using Next Generation Sequencing. Ethiopian journal of health sciences, 28(2), 135.

Sanghvi RV, et al. (2018) Characterizing reduced coverage regions through comparison of exome and genome sequencing data across 10 centers. Genetics in medicine : official journal of the American College of Medical Genetics, 20(8), 855.

Di Resta C, et al. (2018) Integration of multigene panels for the diagnosis of hereditary retinal disorders using Next Generation Sequencing and bioinformatics approaches. EJIFCC, 29(1), 15.

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

Verhaart IEC, et al. (2017) A multi-source approach to determine SMA incidence and research ready population. Journal of neurology, 264(7), 1465.

Rappaport N, et al. (2017) Rational confederation of genes and diseases: NGS interpretation via GeneCards, MalaCards and VarElect. Biomedical engineering online, 16(Suppl 1), 72.

Mucciolo M, et al. (2016) Next Generation Sequencing Approach in a Prenatal Case of Cardio-Facio-Cutaneus Syndrome. International journal of molecular sciences, 17(6).

Pangalos C, et al. (2016) First applications of a targeted exome sequencing approach in fetuses with ultrasound abnormalities reveals an important fraction of cases with associated gene defects. PeerJ, 4, e1955.

Richards S, et al. (2015) Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in medicine : official journal of the American College of Medical Genetics, 17(5), 405.

Stewart A, et al. (2013) Genetic testing strategies in newly diagnosed endometrial cancer patients aimed at reducing morbidity or mortality from lynch syndrome in the index case or her relatives. PLoS currents, 5.