

Accomplishments in Genomic Medicine

The **NHGRI Genomic Medicine Working Group** (GMWG) is compiling a list of interesting advances and helpful educational resources in genomic medicine. To submit notable accomplishments for consideration to the GMWG, please email: GMWG@nih.gov.


The GMWG published the following notable accomplishments:

- [2022 Genomic Medicine Year in Review](#)
- [2021 Genomic Medicine Year in Review](#)
- [2020 Genomic Medicine Year in Review](#)
- [2019 Genomic Medicine Year in Review](#)

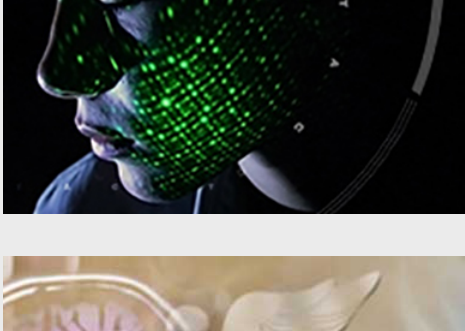
Search for specific publications by title, author, category and/or by date range. For an explanation about the selection criteria and categories, see the [list of categories and their definitions](#).

- 2023**
- January 04, 2023 - **Elective genomic testing: Practice resource of the National Society of Genetic Counselors**
First Author: Blout Zawatsky CL
Category: Educational Resource
- January 04, 2023 - **Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG)**
First Author: Dungan JS
Category: Educational Resource
- 2022**
- December 01, 2022 - **Genetic Testing to Inform Epilepsy Treatment Management From an International Study of Clinical Practice**
First Author: McKnight D
Category: Impact/Outcomes
- December 01, 2022 - **Maternal carrier screening with single-gene NIPS provides accurate fetal risk assessments for recessive conditions**
First Author: Hoskovec J
Category: Risk Assessment/Prediction
- November 23, 2022 - **Development of Competency-based Online Genomic Medicine Training (COGENT)**
First Author: Haga SB
Category: Educational Resource
- November 19, 2022 - **Workforce Considerations When Building a Precision Medicine Program**
First Author: Blout Zawatsky CL
Category: Educational Resource
- November 07, 2022 - **Systematic comparison of family history and polygenic risk across 24 common diseases**
First Author: Mars N
Category: Risk Assessment/Prediction , Other
- September 28, 2022 - **A Cross-Reference of PGx Drug-Gene Association Listings in FDA's Table of Pharmacogenetic Associations and CPIC Clinical Guidelines**
First Author: Manchester University
Category: Educational Resource , Pharmacogenomics
- September 15, 2022 - **Impact of integrating genomic data into the electronic health record on genetics care delivery**
First Author: Lau-Min KS
Category: Pilot Implementation
- September 15, 2022 - **Practice resource-focused revision: Standardized pedigree nomenclature update centered on sex and gender inclusivity: A practice resource of the National Society of Genetic Counselors**
First Author: Bennett RL
Category: Educational Resource
- September 07, 2022 - **Population genomic screening of young adults for familial hypercholesterolaemia: a cost-effectiveness analysis**
First Author: Marquina C
Category: Other
- September 01, 2022 - **Outcomes of Gene Panel Testing for Sensorineural Hearing Loss in a Diverse Patient Cohort**
First Author: Liao EN
Category: Impact/Outcomes , Health Disparities
- September 01, 2022 - **Addressing underrepresentation in genomics research through community engagement**
First Author: Lemke AA
Category: Educational Resource
- August 17, 2022 - **Discovering monogenic patients with a confirmed molecular diagnosis in millions of clinical notes with MonoMiner**
First Author: Wu DW
Category: Pilot Implementation
- August 12, 2022 - **Development and Validation of the PREMMplus Model for Multigene Hereditary Cancer Risk Assessment**
First Author: Yurgelun MB
Category: Educational Resource
- August 11, 2022 - **Environmental scan of family chart linking for genetic cascade screening in a U.S. integrated health system**
First Author: Haas CB
Category: Educational Resource
- August 10, 2022 - **Measures of Utility Among Studies of Genomic Medicine for Critically Ill Infants**
First Author: Callahan KP
Category: Educational Resource
- August 09, 2022 - **Pathogenic variants in arteriopathy genes detected in a targeted sequencing study: Penetrance and 1-year outcomes after return of results**
First Author: Sherafati A
Category: Risk Assessment/Prediction
- August 09, 2022 - **Use of eConsult to enhance genetics service delivery in primary care: a multimethod study**
First Author: Carroll JC
Category: Pilot Implementation
- August 04, 2022 - **The Potential Value-Based Price of a Multi-Cancer Early Detection Genomic Blood Test to Complement Current Single Cancer Screening in the USA**
First Author: Tafazzoli A
Category: Educational Resource
- August 01, 2022 - **Universal screening for familial hypercholesterolemia in 2 populations**
First Author: Sustar U
Category: Impact/Outcomes
- July 25, 2022 - **Genetic risk score enhances the risk prediction of severe obesity in adult survivors of childhood cancer**
First Author: Sapkota Y
Category: Risk Assessment/Prediction
- July 19, 2022 - **Recommendations for clinical interpretation of variants found in non-coding regions of the genome**
First Author: Ellingford JM
Category: Educational Resource
- July 15, 2022 - **Advancing precision medicine for ocular disorders: Diagnostic genomics to tailored therapies**
First Author: Panikker P
Category: Educational Resource
- July 13, 2022 - **Cost-effectiveness frameworks for comparing genome and exome sequencing versus conventional diagnostic pathways: A scoping review and recommended methods**
First Author: Ferket BS
Category: Educational Resource
- July 12, 2022 - **Effect of Pharmacogenomic Testing for Drug-Gene Interactions on Medication Selection and Remission of Symptoms in Major Depressive Disorder: The PRIME Care Randomized Clinical Trial**
First Author: Oslin DW
Category: Systematic Implementation , Pharmacogenomics
- July 07, 2022 - **Digital health-enabled genomics: Opportunities and challenges**
First Author: Bombard Y
Category: Educational Resource
- June 29, 2022 - **Development and validation of a trans-ancestry polygenic risk score for type 2 diabetes in diverse populations**
First Author: Ge T
Category: Educational Resource , Risk Assessment/Prediction
- June 16, 2022 - **Genome-wide polygenic score to predict chronic kidney disease across ancestries**
First Author: Khan A
Category: Risk Assessment/Prediction
- May 25, 2022 - **A RE-AIM Framework Analysis of DNA-Based Population Screening: Using Implementation Science to Translate Research into Practice in a Healthcare System**
First Author: Jones LK
Category: Systematic Implementation , Impact/Outcomes
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