

2013

[Adiposity, inflammation, genetic variants and risk of post-menopausal breast cancer findings from a prospective-specimen-collection, retrospective-blinded-evaluation \(PRoBE\) design approach.](#)

Yan XS, Barnholtz-Sloan J, Chu X, Li L, Colonie R, Webster J, Smelser D, Patel N, Prichard J, Stark A. Springerplus. 2013 Nov 27; 2:638. doi: 10.1186/2193-1801-2-638. eCollection 2013. PMID: 24340245

2014

[Population risk factor estimates for abdominal aortic aneurysm from electronic medical records: a case control study.](#)

Smelser DT, Tromp G, Elmore JR, Kuivaniemi H, Franklin DP, Kirchner HL, Carey DJ. BMC Cardiovasc Disorder. 2014 Dec 4; 14:174. doi: 10.1186/1471-2261-14-174. PMID: 25475588

[Implementation of genomic medicine in a health care delivery system: a value proposition?](#)

Wade JE, Ledbetter DH, Williams MS. Am J Med Genet C Semin Med Genet. 2014 Mar;166C(1):112-6. doi: 10.1002/ajmg.c.31392. Epub 2014 Mar 11. PMID: 24619641

2015

[Contrasting Association Results between Existing PheWAS Phenotype Definition Methods and Five Validated Electronic Phenotypes.](#)

Leader JB, Pendergrass SA, Verma A, Carey DJ, Hartzel DN, Ritchie MD, Kirchner HL. AMIA Annu Symp Proc. 2015 Nov 5;2015:824-32. eCollection 2015. PMID: 26958218

[Your DNA is not your diagnosis: getting diagnoses right following secondary genomic findings.](#)

Murray MF. Genet Med. 2016 Aug;18(8):765-7. doi: 10.1038/gim.2015.134. Epub 2015 Oct 8. PMID: 26447529

2016

[How Geisinger made the case for an institutional duty to return genomic results to biobank participants.](#)

Faucett WA, Davis FD. Appl Transl Genom. 2016 Feb 1; 8:33-5. doi: 10.1016/j.atg.2016.01.003. eCollection 2016 Mar. PMID: 27047758

[Inactivating Variants in ANGPTL4 and Risk of Coronary Artery Disease.](#)

Dewey FE, Gusarova V, O'Dushlaine C, Gottesman O, Trejos J, Hunt C, Van Hout CV, Habegger L, Buckler D, Lai KM, Leader JB, Murray MF, Ritchie MD, Kirchner HL, Ledbetter DH, Penn J, Lopez A, Borecki IB, Overton JD, Reid JG, Carey DJ, Murphy AJ, Yancopoulos GD, Baras A, Gromada J, Shuldiner AR. N Engl J Med. 2016 Mar 24;374(12):1123-33. doi: 10.1056/NEJMoa1510926. Epub 2016 Mar 2. PMID: 26933753

[The Geisinger MyCode community health initiative: an electronic health record-linked biobank for precision medicine research.](#)

Carey DJ, Fetterolf SN, Davis FD, Faucett WA, Kirchner HL, Mirshahi U, Murray MF, Smelser DT, Gerhard GS, Ledbetter DH. Genet Med. 2016 Sep;18(9):906-13. doi: 10.1038/gim.2015.187. Epub 2016 Feb 11. PMID: 26866580

[Distribution and clinical impact of functional variants in 50,726 whole-exome sequences from the DiscovEHR study.](#)

Dewey FE, Murray MF, Overton JD, Habegger L, Leader JB, Fetterolf SN, O'Dushlaine C, Van Hout CV, Staples J, Gonzaga-Jauregui C, Metpally R, Pendergrass SA, Giovanni MA, Kirchner HL, Balasubramanian S, Abul-Husn NS, Hartzel DN, Lavage DR, Kost KA, Packer JS, Lopez AE, Penn J, Mukherjee S, Gosalia N, Kanagaraj M, Li AH, Mitnaul LJ, Adams LJ, Person TN, Praveen K, Marcketta A, Lebo MS, Austin-Tse CA, Mason-Suares HM, Bruse S, Mellis S, Phillips R, Stahl N, Murphy A, Economides A, Skelding KA, Still CD, Elmore JR, Borecki IB, Yancopoulos GD, Davis FD, Faucett WA, Gottesman O, Ritchie MD, Shuldiner AR, Reid JG, Ledbetter DH, Baras A, Carey DJ. *Science*. 2016 Dec 23;354(6319):aaf6814. doi: 10.1126/science.aaf6814. PMID: 28008009

[Recommendations for the integration of genomics into clinical practice.](#)

Bowdin S, Gilbert A, Bedoukian E, Carew C, Adam MP, Belmont J, Bernhardt B, Biesecker L, Bjornsson HT, Blitzer M, D'Alessandro LC, Deardorff MA, Demmer L, Elliott A, Feldman GL, Glass IA, Herman G, Hindorff L, Hisama F, Hudgins L, Innes AM, Jackson L, Jarvik G, Kim R, Korf B, Ledbetter DH, Li M, Liston E, Marshall C, Medne L, Meyn MS, Monfared N, Morton C, Mulvihill JJ, Plon SE, Rehm H, Roberts A, Shuman C, Spinner NB, Stavropoulos DJ, Valverde K, Waggoner DJ, Wilkens A, Cohn RD, Krantz ID. *Genet Med*. 2016 Nov;18(11):1075-1084. doi: 10.1038/gim.2016.17. Epub 2016 May 12. PMID: 27171546

[Genetic identification of familial hypercholesterolemia within a single U.S. health care system.](#)

Abul-Husn NS, Manickam K, Jones LK, Wright EA, Hartzel DN, Gonzaga-Jauregui C, O'Dushlaine C, Leader JB, Lester Kirchner H, Lindbuchler DM, Barr ML, Giovanni MA, Ritchie MD, Overton JD, Reid JG, Metpally RP, Wardeh AH, Borecki IB, Yancopoulos GD, Baras A, Shuldiner AR, Gottesman O, Ledbetter DH, Carey DJ, Dewey FE, Murray MF. *Science*. 2016 Dec 23;354(6319):aaf7000. doi: 10.1126/science.aaf7000. PMID: 28008010

2017

[Association of Rare and Common Variation in the Lipoprotein Lipase Gene with Coronary Artery Disease.](#)

Khera AV, Won HH, Peloso GM, O'Dushlaine C, Liu D, Stitzel NO, Natarajan P, Nomura A, Emdin CA, Gupta N, Borecki IB, Asselta R, Duga S, Merlini PA, Correa A, Kessler T, Wilson JG, Bown MJ, Hall AS, Braund PS, Carey DJ, Murray MF, Kirchner HL, Leader JB, Lavage DR, Manus JN, Hartzel DN, Samani NJ, Schunkert H, Marrugat J, Elosua R, McPherson R, Farrall M, Watkins H, Lander ES, Rader DJ, Danesh J, Ardissino D, Gabriel S, Willer C, Abecasis GR, Saleheen D, Dewey FE, Kathiresan S; Myocardial Infarction Genetics Consortium, DiscovEHR Study Group, CARDIoGRAM Exome Consortium, and Global Lipids Genetics Consortium. *JAMA*. 2017 Mar 7;317(9):937-946. doi: 10.1001/jama.2017.0972. PMID: 28267856

[Genome-wide association analyses for lung function and chronic obstructive pulmonary disease identify new loci and potential druggable targets.](#)

Wain LV, Shrine N, Artigas MS, Erzurumluoglu AM, Noyvert B, Bossini-Castillo L, Obeidat M, Henry AP, Portelli MA, Hall RJ, Billington CK, Rimington TL, Fenech AG, John C, Blake T, Jackson VE, Allen RJ, Prins BP; Understanding Society Scientific Group, Campbell A, Porteous DJ, Jarvelin MR, Wielscher M, James AL, Hui J, Wareham NJ, Zhao JH, Wilson JF, Joshi PK, Stubbe B, Rawal R, Schulz H, Imboden M, Probst-Hensch NM, Karrasch S, Gieger C, Deary IJ, Harris SE, Marten J, Rudan I, Enroth S, Gyllensten U, Kerr SM, Polasek O, Kähönen M, Surakka I, Vitart V, Hayward C, Lehtimäki T, Raitakari OT, Evans DM, Henderson AJ, Pennell CE, Wang CA, Sly PD, Wan ES, Busch R, Hobbs BD, Litonjua AA, Sparrow DW, Gulsvik A, Bakke PS, Crapo JD, Beaty TH, Hansel NN, Mathias RA, Ruczinski I, Barnes KC, Bossé Y, Joubert P, van den Berge M, Brandsma CA, Paré PD, Sin DD, Nickle DC, Hao K, Gottesman O, Dewey FE, Bruse SE, Carey DJ, Kirchner HL; Geisinger-Regeneron

DiscovEHR Collaboration, Jonsson S, Thorleifsson G, Jonsdottir I, Gislason T, Stefansson K, Schurmann C, Nadkarni G, Bottinger EP, Loos RJ, Walters RG, Chen Z, Millwood IY, Vaucher J, Kurmi OP, Li L, Hansell AL, Brightling C, Zeggini E, Cho MH, Silverman EK, Sayers I, Trynka G, Morris AP, Strachan DP, Hall IP, Tobin MD. *Nat Genet.* 2017 Mar;49(3):416-425. doi: 10.1038/ng.3787. Epub 2017 Feb 6. PMID: 28166213

[Adding Protective Genetic Variants to Clinical Reporting of Genomic Screening Results: Restoring Balance.](#)

Schwartz MLB, Williams MS, Murray MF. *JAMA.* 2017 Apr 18;317(15):1527-1528. doi: 10.1001/jama.2017.1533. PMID: 28288260

[Protein-Truncating Variants at the Cholesteryl Ester Transfer Protein Gene and Risk for Coronary Heart Disease.](#)

Nomura A, Won HH, Khera AV, Takeuchi F, Ito K, McCarthy S, Emdin CA, Klarin D, Natarajan P, Zekavat SM, Gupta N, Peloso GM, Borecki IB, Teslovich TM, Asselta R, Duga S, Merlini PA, Correa A, Kessler T, Wilson JG, Bown MJ, Hall AS, Braund PS, Carey DJ, Murray MF, Kirchner HL, Leader JB, Lavage DR, Manus JN, Hartze DN, Samani NJ, Schunkert H, Marrugat J, Elosua R, McPherson R, Farrall M, Watkins H, Juang JJ, Hsiung CA, Lin SY, Wang JS, Tada H, Kawashiri MA, Inazu A, Yamagishi M, Katsuya T, Nakashima E, Nakatochi M, Yamamoto K, Yokota M, Momozawa Y, Rotter JI, Lander ES, Rader DJ, Danesh J, Ardissino D, Gabriel S, Willer CJ, Abecasis GR, Saleheen D, Kubo M, Kato N, Ida Chen YD, Dewey FE, Kathiresan S. *Circ Res.* 2017 Jun 23;121(1):81-88. doi: 10.1161/CIRCRESAHA.117.311145. Epub 2017 May 15. PMID: 28506971

[Genetic and Pharmacologic Inactivation of ANGPTL3 and Cardiovascular Disease.](#)

Dewey FE, Gusarova V, Dunbar RL, O'Dushlaine C, Schurmann C, Gottesman O, McCarthy S, Van Hout CV, Bruse S, Dansky HM, Leader JB, Murray MF, Ritchie MD, Kirchner HL, Habegger L, Lopez A, Penn J, Zhao A, Shao W, Stahl N, Murphy AJ, Hamon S, Bouzelmat A, Zhang R, Shumel B, Pordy R, Gipe D, Herman GA, Sheu WHH, Lee IT, Liang KW, Guo X, Rotter JI, Chen YI, Kraus WE, Shah SH, Damrauer S, Small A, Rader DJ, Wulff AB, Nordestgaard BG, Tybjærg-Hansen A, van den Hoek AM, Princen HMG, Ledbetter DH, Carey DJ, Overton JD, Reid JG, Sasiela WJ, Banerjee P, Shuldiner AR, Borecki IB, Teslovich TM, Yancopoulos GD, Mellis SJ, Gromada J, Baras A. *N Engl J Med.* 2017 Jul 20;377(3):211-221. doi: 10.1056/NEJMoa1612790. Epub 2017 May 24. PMID: 28538136

[Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing.](#)

Haggerty CM, James CA, Calkins H, Tichnell C, Leader JB, Hartzel DN, Nevius CD, Pendergrass SA, Person TN, Schwartz M, Ritchie MD, Carey DJ, Ledbetter DH, Williams MS, Dewey FE, Lopez A, Penn J, Overton JD, Reid JG, Lebo M, Mason-Suares H, Austin-Tse C, Rehm HL, Delisle BP, Makowski DJ, Mehra VC, Murray MF, Fornwalt BK. *Genet Med.* 2017 Nov;19(11):1245-1252. doi: 10.1038/gim.2017.40. Epub 2017 May 4. PMID: 28471438

2018

[How powerful are summary-based methods for identifying expression-trait associations under different genetic architectures?](#)

Veturi Y, Ritchie MD. *Pac Symp Biocomput.* 2018; 23:228-239. PMID: 29218884

[Rare variants in drug target genes contributing to complex diseases, phenome-wide.](#)

Verma SS, Josyula N, Verma A, Zhang X, Veturi Y, Dewey FE, Hartzel DN, Lavage DR, Leader J, Ritchie MD, Pendergrass SA. *Sci Rep.* 2018 Mar 15;8(1):4624. doi: 10.1038/s41598-018-22834-4. PMID: 29545597

[Collective feature selection to identify crucial epistatic variants.](#)

Verma SS, Lucas A, Zhang X, Veturi Y, Dudek S, Li B, Li R, Urbanowicz R, Moore JH, Kim D, Ritchie MD. *BioData Min.* 2018 Apr 19;11:5. doi: 10.1186/s13040-018-0168-6. eCollection 2018. PMID: 29713383

[Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants.](#)

Buchanan AH, Manickam K, Meyer MN, Wagner JK, Hallquist MLG, Williams JL, Rahm AK, Williams MS, Chen ZE, Shah CK, Garg TK, Lazzeri AL, Schwartz MLB, Lindbuchler DM, Fan AL, Leeming R, Servano PO 3rd, Smith AL, Vogel VG, Abul-Husn NS, Dewey FE, Lebo MS, Mason-Suares HM, Ritchie MD, Davis FD, Carey DJ, Feinberg DT, Faucett WA, Ledbetter DH, Murray MF. *Genet Med.* 2018 Apr;20(5):554-558. doi: 10.1038/gim.2017.145. *Epub* 2017 Oct 26. PMID: 29261187

[Patient-Centered Precision Health In A Learning Health Care System: Geisinger's Genomic Medicine Experience.](#)

Williams MS, Buchanan AH, Davis FD, Faucett WA, Hallquist MLG, Leader JB, Martin CL, McCormick CZ, Meyer MN, Murray MF, Rahm AK, Schwartz MLB, Sturm AC, Wagner JK, Williams JL, Willard HF, Ledbetter DH. *Health Aff (Millwood).* 2018 May;37(5):757-764. doi: 10.1377/hlthaff.2017.1557. PMID: 29733722

[Profiling and Leveraging Relatedness in a Precision Medicine Cohort of 92,455 Exomes.](#)

Staples J, Maxwell EK, Gosalia N, Gonzaga-Jauregui C, Snyder C, Hawes A, Penn J, Ulloa R, Bai X, Lopez AE, Van Hout CV, O'Dushlaine C, Teslovich TM, McCarthy SE, Balasubramanian S, Kirchner HL, Leader JB, Murray MF, Ledbetter DH, Shuldiner AR, Yancopoulos GD, Dewey FE, Carey DJ, Overton JD, Baras A, Habegger L, Reid JG. *Am J Hum Genet.* 2018 May 3;102(5):874-889. doi: 10.1016/j.ajhg.2018.03.012. PMID: 29727688

[Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes.](#)

Gusarova V, O'Dushlaine C, Teslovich TM, Benotti PN, Mirshahi T, Gottesman O, Van Hout CV, Murray MF, Mahajan A, Nielsen JB, Fritsche L, Wulff AB, Gudbjartsson DF, Sjögren M, Emdin CA, Scott RA, Lee WJ, Small A, Kwee LC, Dwivedi OP, Prasad RB, Bruse S, Lopez AE, Penn J, Marcketta A, Leader JB, Still CD, Kirchner HL, Mirshahi UL, Wardeh AH, Hartle CM, Habegger L, Fetterolf SN, Tusie-Luna T, Morris AP, Holm H, Steinthorsdottir V, Sulem P, Thorsteinsdottir U, Rotter JI, Chuang LM, Damrauer S, Birtwell D, Brummett CM, Khera AV, Natarajan P, Orho-Melander M, Flannick J, Lotta LA, Willer CJ, Holmen OL, Ritchie MD, Ledbetter DH, Murphy AJ, Borecki IB, Reid JG, Overton JD, Hansson O, Groop L, Shah SH, Kraus WE, Rader DJ, Chen YI, Hveem K, Wareham NJ, Kathiresan S, Melander O, Stefansson K, Nordestgaard BG, Tybjaerg-Hansen A, Abecasis GR, Altshuler D, Florez JC, Boehnke M, McCarthy MI, Yancopoulos GD, Carey DJ, Shuldiner AR, Baras A, Dewey FE, Gromada J. *Nat Commun.* 2018 Jun 13;9(1):2252. doi: 10.1038/s41467-018-04611-z. PMID: 29899519

[Generation and Implementation of a Patient-Centered and Patient-Facing Genomic Test Report in the EHR.](#)

Goehring JM, Bonhag MA, Jones LK, Schmidlen T, Schwartz M, Rahm AK, Williams JL, Williams MS. *EGEMS (Wash DC).* 2018 Jun 26;6(1):14. doi: 10.5334/egems.256. PMID: 30094286

[Managing Secondary Genomic Findings Associated with Arrhythmogenic Right Ventricular Cardiomyopathy: Case Studies and Proposal for Clinical Surveillance.](#)

Haggerty CM, Murray B, Tichnell C, Judge DP, Tandri H, Schwartz M, Sturm AC, Matsumura ME, Murray MF, Calkins H, Fornwalt BK, James CM. *Circ Genom Precis Med.* 2018 Jul;11(7): e002237. doi: 10.1161/CIRCGEN.118.002237. PMID: 29997227

[Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals.](#)

Lee JJ, Wedow R, Okbay A, Kong E, Maghzian O, Zacher M, Nguyen-Viet TA, Bowers P, Sidorenko J, Karlsson Linnér R, Fontana MA, Kundu T, Lee C, Li H, Li R, Royer R, Timshel PN, Walters RK, Willoughby EA, Yengo L; 23andMe Research Team; COGENT (Cognitive Genomics Consortium); Social Science Genetic Association Consortium, Alver M, Bao Y, Clark DW, Day FR, Furlotte NA, Joshi PK, Kemper KE, Kleinman A, Langenberg C, Mägi R, Trampush JW, Verma SS, Wu Y, Lam M, Zhao JH, Zheng Z, Boardman JD, Campbell H, Freese J, Harris KM, Hayward C, Herd P, Kumari M, Lencz T, Luan J, Malhotra AK, Metspalu A, Milani L, Ong KK, Perry JRB, Porteous DJ, Ritchie MD, Smart MC, Smith BH, Tung JY, Wareham NJ, Wilson JF, Beauchamp JP, Conley DC, Esko T, Lehrer SF, Magnusson PKE, Oskarsson S, Pers TH, Robinson MR, Thom K, Watson C, Chabris CF, Meyer MN, Laibson DI, Yang J, Johannesson M, Koellinger PD, Turley P, Visscher PM, Benjamin DJ, Cesarini D. *Nat Genet.* 2018 Jul 23;50(8):1112-1121. doi: 10.1038/s41588-018-0147-3. PMID: 30038396

[Healthcare Utilization and Patients' Perspectives After Receiving a Positive Genetic Test for Familial Hypercholesterolemia.](#)

Jones LK, Kulchak Rahm A, Manickam K, Butry L, Lazzeri A, Corcoran T, Komar D, Josyula NS, Pendergrass SA, Sturm AC, Murray MF. *Circ Genom Precis Med.* 2018 Aug;11(8): e002146. doi: 10.1161/CIRCGEN.118.002146. PMID: 30354341

[A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort.](#)

Schwartz MLB, McCormick CZ, Lazzeri AL, Lindbuchler DM, Hallquist MLG, Manickam K, Buchanan AH, Rahm AK, Giovanni MA, Frisbie L, Flansburg CN, Davis FD, Sturm AC, Nicastro C, Lebo MS, Mason-Suares H, Mahanta LM, Carey DJ, Williams JL, Williams MS, Ledbetter DH, Faucett WA, Murray MF. *Am J Hum Genet.* 2018 Sep 6;103(3):328-337. doi: 10.1016/j.ajhg.2018.07.009. Epub 2018 Aug 9. PMID: 30100086

[Exome Sequencing-Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants.](#)

Manickam K, Buchanan AH, Schwartz MLB, Hallquist MLG, Williams JL, Rahm AK, Rocha H, Savatt JM, Evans AE, Butry LM, Lazzeri AL, Lindbuchler DM, Flansburg CN, Leeming R, Vogel VG, Lebo MS, Mason-Suares HM, Hoskinson DC, Abul-Husn NS, Dewey FE, Overton JD, Reid JG, Baras A, Willard HF, McCormick CZ, Krishnamurthy SB, Hartzel DN, Kost KA, Lavage DR, Sturm AC, Frisbie LR, Person TN, Metpally RP, Giovanni MA, Lowry LE, Leader JB, Ritchie MD, Carey DJ, Justice AE, Kirchner HL, Faucett WA, Williams MS, Ledbetter DH, Murray MF. *JAMA Netw Open.* 2018 Sep 7;1(5): e182140. doi: 10.1001/jamanetworkopen.2018.2140. PMID: 30646163

[Association Between Titin Loss-of-Function Variants and Early-Onset Atrial Fibrillation.](#)

Choi SH, Weng LC, Roselli C, Lin H, Haggerty CM, Shoemaker MB, Barnard J, Arking DE, Chasman DI, Albert CM, Chaffin M, Tucker NR, Smith JD, Gupta N, Gabriel S, Margolin L, Shea MA, Shaffer CM, Yoneda ZT, Boerwinkle E, Smith NL, Silverman EK, Redline S, Vasan RS, Burchard EG, Gogarten SM, Laurie C, Blackwell TW, Abecasis G, Carey DJ, Fornwalt BK, Smelser DT, Baras A, Dewey FE, Jaquish CE, Papanicolaou GJ, Sotoodehnia N, Van Wagoner DR, Psaty BM, Kathiresan S, Darbar D, Alonso A, Heckbert SR, Chung MK, Roden DM, Benjamin EJ, Murray MF, Lunetta KL, Lubitz SA, Ellinor PT; DiscovEHR study and the NHLBI Trans-Omics for Precision Medicine (TOPMed) Consortium. *JAMA.* 2018 Dec 11;320(22):2354-2364. doi: 10.1001/jama.2018.18179. PMID: 30535219

[Genetic identification of familial hypercholesterolemia within a single U.S. health care system.](#)

Abul-Husn NS, Manickam K, Jones LK, Wright EA, Hartzel DN, Gonzaga-Jauregui C, O'Dushlaine C, Leader JB, Lester Kirchner H, Lindbuchler DM, Barr ML, Giovanni MA, Ritchie MD, Overton JD, Reid JG, Metpally RP,

Wardeh AH, Borecki IB, Yancopoulos GD, Baras A, Shuldiner AR, Gottesman O, Ledbetter DH, Carey DJ, Dewey FE, Murray MF. *Science*. 2016 Dec 23;354(6319):aaf7000. doi: 10.1126/science.aaf7000. PMID: 28008010

[A Protein-Truncating HSD17B13 Variant and Protection from Chronic Liver Disease.](#)

Abul-Husn NS, Cheng X, Li AH, Xin Y, Schurmann C, Stevis P, Liu Y, Kozlitina J, Stender S, Wood GC, Stepanchick AN, Still MD, McCarthy S, O'Dushlaine C, Packer JS, Balasubramanian S, Gosalia N, Esopi D, Kim SY, Mukherjee S, Lopez AE, Fuller ED, Penn J, Chu X, Luo JZ, Mirshahi UL, Carey DJ, Still CD, Feldman MD, Small A, Damrauer SM, Rader DJ, Zambrowicz B, Olson W, Murphy AJ, Borecki IB, Shuldiner AR, Reid JG, Overton JD, Yancopoulos GD, Hobbs HH, Cohen JC, Gottesman O, Teslovich TM, Baras A, Mirshahi T, Gromada J, Dewey FE. *N Engl J Med*. 2018 Mar 22;378(12):1096-1106. doi: 10.1056/NEJMoa1712191. PMID: 29562163

[PheWAS and Beyond: The Landscape of Associations with Medical Diagnoses and Clinical Measures across 38,662 Individuals from Geisinger.](#)

Verma A, Lucas A, Verma SS, Zhang Y, Josyula N, Khan A, Hartzel DN, Lavage DR, Leader J, Ritchie MD, Pendergrass SA. *Am J Hum Genet*. 2018 Apr 5;102(4):592-608. doi: 10.1016/j.ajhg.2018.02.017. Epub 2018 Mar 29. PMID: 29606303

[Parental attitudes and expectations towards receiving genomic test results in healthy children.](#)

Kulchak Rahm A, Bailey L, Fultz K, Fan A, Williams JL, Buchanan A, Davis FD, Murray MF, Williams MS. *Transl Behav Med*. 2018 Jan 29;8(1):44-53. doi: 10.1093/tbm/ibx044. PMID: 29385584

How can we reach at-risk relatives? Efforts to enhance communication and cascade testing uptake. A mini-review (PubMed reference not available). Schwiter R, Rahm AK, Williams JL, Sturm AC. Accepted for publication. *Curr Genet Med Reports* 2018.

2019

[Systematic characterization of germline variants from the DiscovEHR study endometrial carcinoma population.](#)

Miller JE, Metpally RP, Person TN, Krishnamurthy S, Dasari VR, Shivakumar M, Lavage DR, Cook AM, Carey DJ, Ritchie MD, Kim D, Gogoi R; DiscovEHR collaboration. *BMC Med Genomics*. 2019 May 3;12(1):59. doi: 10.1186/s12920-019-0504-9. PMID: 31053132

[Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls.](#)

Flannick J, Mercader JM, Fuchsberger C, Udler MS, Mahajan A, Wessel J, Teslovich TM, Caulkins L, Koesterer R, Barajas-Olmos F, Blackwell TW, Boerwinkle E, Brody JA, Centeno-Cruz F, Chen L, Chen S, Contreras-Cubas C, Córdova E, Correa A, Cortes M, DeFronzo RA, Dolan L, Drews KL, Elliott A, Floyd JS, Gabriel S, Garay-Sevilla ME, García-Ortiz H, Gross M, Han S, Heard-Costa NL, Jackson AU, Jørgensen ME, Kang HM, Kelsey M, Kim BJ, Koistinen HA, Kuusisto J, Leader JB, Linneberg A, Liu CT, Liu J, Lyssenko V, Manning AK, Marcketta A, Malacara-Hernandez JM, Martínez-Hernández A, Matsuo K, Mayer-Davis E, Mendoza-Caamal E, Mohlke KL, Morrison AC, Ndungu A, Ng MCY, O'Dushlaine C, Payne AJ, Pihoker C; Broad Genomics Platform, Post WS, Preuss M, Psaty BM, Vasan RS, Rayner NW, Reiner AP, Revilla-Monsalve C, Robertson NR, Santoro N, Schurmann C, So WY, Soberón X, Stringham HM, Strom TM, Tam CHT, Thameem F, Tomlinson B, Torres JM, Tracy RP, van Dam RM, Vujkovic M, Wang S, Welch RP, Witte DR, Wong TY, Atzmon G, Barzilai N, Blangero J, Bonnycastle LL, Bowden DW, Chambers JC, Chan E, Cheng CY, Cho YS, Collins FS, de Vries PS, Duggirala R, Glaser B, Gonzalez C, Gonzalez ME, Groop L, Kooner JS, Kwak SH, Laakso M, Lehman DM, Nilsson P, Spector TD, Tai ES, Tuomi T, Tuomilehto J, Wilson JG, Aguilar-Salinas CA, Bottinger E, Burke B, Carey DJ, Chan JCN, Dupuis J, Frossard P, Heckbert SR, Hwang MY, Kim YJ, Kirchner HL, Lee JY, Lee J, Loos RJF, Ma RCW, Morris AD, O'Donnell CJ, Palmer CNA, Pankow J, Park KS, Rasheed A, Saleheen D, Sim X, Small KS, Teo YY, Haiman C, Hanis CL, Henderson BE,

Orozco L, Tusié-Luna T, Dewey FE, Baras A, Gieger C, Meitinger T, Strauch K, Lange L, Grarup N, Hansen T, Pedersen O, Zeitler P, Dabelea D, Abecasis G, Bell GI, Cox NJ, Seielstad M, Sladek R, Meigs JB, Rich SS, Rotter JI; DiscovEHR Collaboration; CHARGE; LuCamp; ProDiGY; GoT2D; ESP; SIGMA-T2D; T2D-GENES; AMP-T2D-GENES, Altshuler D, Burt NP, Scott LJ, Morris AP, Florez JC, McCarthy MI, Boehnke M. *Nature*. 2019 Jun;570(7759):71-76. doi: 10.1038/s41586-019-1231-2. Epub 2019 May 22. PMID: 31118516

[Genomics-First Evaluation of Heart Disease Associated with Titin-Truncating Variants.](#)

Haggerty CM, Damrauer SM, Levin MG, Birtwell D, Carey DJ, Golden AM, Hartzel DN, Hu Y, Judy R, Kelly MA, Kember RL, Lester Kirchner H, Leader JB, Liang L, McDermott-Roe C, Babu A, Morley M, Nealy Z, Person TN, Pulenthiran A, Small A, Smelser DT, Stahl RC, Sturm AC, Williams H, Baras A, Margulies KB, Cappola TP, Dewey FE, Verma A, Zhang X, Correa A, Hall ME, Wilson JG, Ritchie MD, Rader DJ, Murray MF, Fornwalt BK, Arany Z. *Circulation*. 2019 Jul 2;140(1):42-54. doi: 10.1161/CIRCULATIONAHA.119.039573. Epub 2019 Jun 20. PMID: 31216868

[Exome-Wide Rare Variant Analysis from the DiscovEHR Study Identifies Novel Candidate Predisposition Genes for Endometrial Cancer.](#)

Shivakumar M, Miller JE, Dasari VR, Gogoi R, Kim D. *Front Oncol*. 2019 Jul 5; 9:574. doi: 10.3389/fonc.2019.00574. eCollection 2019. PMID: 31338326

[Penetrance and Pleiotropy of Polygenic Risk Scores for Schizophrenia in 106,160 Patients Across Four Health Care Systems.](#)

Zheutlin AB, Dennis J, Karlsson Linnér R, Moscati A, Restrepo N, Straub P, Ruderfer D, Castro VM, Chen CY, Ge T, Huckins LM, Charney A, Kirchner HL, Stahl EA, Chabris CF, Davis LK, Smoller JW. *Am J Psychiatry*. 2019 Oct 1;176(10):846-855. doi: 10.1176/appi.ajp.2019.18091085. Epub 2019 Aug 16. PMID: 31416338

[Dissecting genetic factors affecting phenylephrine infusion rates during anesthesia: a genome-wide association study employing EHR data.](#)

Zhang Y, Poler SM, Li J, Abedi V, Pendergrass SA, Williams MS, Lee MTM. *BMC Med*. 2019 Aug 28;17(1):168. doi: 10.1186/s12916-019-1405-7. PMID: 31455332

[GSTM1 Copy Number Is Not Associated with Risk of Kidney Failure in a Large Cohort.](#)

Zhang Y, Zafar W, Hartzel DN, Williams MS, Tin A, Chang AR, Lee MTM. *Front Genet*. 2019 Aug 30; 10:765. doi: 10.3389/fgene.2019.00765. eCollection 2019. PMID: 31555322

[Patient assessment of chatbots for the scalable delivery of genetic counseling.](#)

Schmidlen T, Schwartz M, DiLoreto K, Kirchner HL, Sturm AC. *J Genet Couns*. 2019 Dec;28(6):1166-1177. doi: 10.1002/jgc4.1169. Epub 2019 Sep 24. PMID: 31549758

[Geisinger MyCode \(®\) detects BRCA2 mutation prior to abdominal panniculectomy allowing for DIEP flap breast reconstruction.](#)

Kauffman CA. *Case Reports Plast Surg Hand Surg*. 2019 Nov 6;6(1):145-147. doi: 10.1080/23320885.2019.1684824. eCollection 2019. PMID: 32002463

[Prevalence and Electronic Health Record-Based Phenotype of Loss-of-Function Genetic Variants in Arrhythmogenic Right Ventricular Cardiomyopathy-Associated Genes.](#)

Carruth ED, Young W, Beer D, James CA, Calkins H, Jing L, Raghunath S, Hartzel DN, Leader JB, Kirchner HL, Smelser DT, Carey DJ, Kelly MA, Sturm AC, Alsaïd A, Fornwalt BK, Haggerty CM. *Circ Genom Precis Med*. 2019 Nov;12(11):e002579. doi: 10.1161/CIRCGEN.119.002579. Epub 2019 Oct 22. PMID: 31638835

[ALG9 Mutation Carriers Develop Kidney and Liver Cysts.](#)

Besse W, Chang AR, Luo JZ, Triffo WJ, Moore BS, Gulati A, Hartzel DN, Mane S; Regeneron Genetics Center, Torres VE, Somlo S, Mirshahi T. *J Am Soc Nephrol*. 2019 Nov;30(11):2091-2102. doi: 10.1681/ASN.2019030298. Epub 2019 Aug 8. PMID: 31395617

[Quantifying the polygenic contribution to variable expressivity in eleven rare genetic disorders.](#)

Oetjens MT, Kelly MA, Sturm AC, Martin CL, Ledbetter DH. *Nat Commun*. 2019 Oct 25;10(1):4897. doi: 10.1038/s41467-019-12869-0. PMID: 31653860

[Rare Protein-Truncating Variants in APOB, Lower Low-Density Lipoprotein Cholesterol, and Protection Against Coronary Heart Disease.](#)

Peloso GM, Nomura A, Khera AV, Chaffin M, Won HH, Ardissino D, Danesh J, Schunkert H, Wilson JG, Samani N, Erdmann J, McPherson R, Watkins H, Saleheen D, McCarthy S, Teslovich TM, Leader JB, Lester Kirchner H, Marrugat J, Nohara A, Kawashiri MA, Tada H, Dewey FE, Carey DJ, Baras A, Kathiresan S. *Circ Genom Precis Med*. 2019 May;12(5):e002376. doi: 10.1161/CIRCGEN.118.002376. PMID: 30939045

[Finding missed cases of familial hypercholesterolemia in health systems using machine learning.](#)

Banda JM, Sarraju A, Abbasi F, Parizo J, Pariani M, Ison H, Briskin E, Wand H, Dubois S, Jung K, Myers SA, Rader DJ, Leader JB, Murray MF, Myers KD, Wilemon K, Shah NH, Knowles JW. *NPJ Digit Med*. 2019 Apr 11;2:23. doi: 10.1038/s41746-019-0101-5. eCollection 2019. PMID: 31304370

[A phenome-wide association study to discover pleiotropic effects of PCSK9, APOB, and LDLR.](#)

Safarova MS, Satterfield BA, Fan X, Austin EE, Ye Z, Bastarache L, Zheng N, Ritchie MD, Borthwick KM, Williams MS, Larson EB, Scrol A, Jarvik GP, Crosslin DR, Leppig K, Rasmussen-Torvik LJ, Pendergrass SA, Sturm AC, Namjou B, Shah AS, Carroll RJ, Chung WK, Wei WQ, Feng Q, Stein CM, Roden DM, Manolio TA, Schaid DJ, Denny JC, Hebbring SJ, de Andrade M, Kullo IJ. *NPJ Genom Med*. 2019 Feb 11;4:3. doi: 10.1038/s41525-019-0078-7. eCollection 2019. PMID: 30774981

[Hidden Burden of Electronic Health Record-Identified Familial Hypercholesterolemia: Clinical Outcomes and Cost of Medical Care.](#)

Patel P, Hu Y, Kolinovsky A, Geng Z, Ruhl J, Krishnamurthy S, deRichemond C, Khan A, Kirchner HL, Metpally R, Jones LK, Sturm AC, Carey D, Snyder S, Williams MS, Mehra VC. *J Am Heart Assoc*. 2019 Jul 2;8(13):e011822. doi: 10.1161/JAHA.118.011822. Epub 2019 Jun 29. PMID: 31256702

2020

[A genome-first approach to aggregating rare genetic variants in LMNA for association with electronic health record phenotypes.](#)

Park J, Levin MG, Haggerty CM, Hartzel DN, Judy R, Kember RL, Reza N; Regeneron Genetics Center, Ritchie MD, Owens AT, Damrauer SM, Rader DJ. *Genet Med*. 2020 Jan;22(1):102-111. doi: 10.1038/s41436-019-0625-8. Epub 2019 Aug 6. PMID: 31383942

[Healthcare Utilization and Costs after Receiving a Positive BRCA1/2 Result from a Genomic Screening Program.](#)

Hao J, Hassen D, Manickam K, Murray MF, Hartzel DN, Hu Y, Liu K, Rahm AK, Williams MS, Lazzeri A, Buchanan A, Sturm A, Snyder SR. *J Pers Med*. 2020 Feb 3;10(1):7. doi: 10.3390/jpm10010007. PMID: 32028596

[A genome-wide association study of polycystic ovary syndrome identified from electronic health records.](#)

Zhang Y, Ho K, Keaton JM, Hartzel DN, Day F, Justice AE, Josyula NS, Pendergrass SA, Actkins K, Davis LK, Velez Edwards DR, Holohan B, Ramirez A, Stanaway IB, Crosslin DR, Jarvik GP, Sleiman P, Hakonarson H, Williams

MS, Lee MTM. *Am J Obstet Gynecol*. 2020 Apr 11:S0002-9378(20)30428-2. doi: 10.1016/j.ajog.2020.04.004. Online ahead of print. PMID: 32289280

[Pediatric reporting of genomic results study \(PROGRESS\): a mixed-methods, longitudinal, observational cohort study protocol to explore disclosure of actionable adult- and pediatric-onset genomic variants to minors and their parents.](#)

Savatt JM, Wagner JK, Joffe S, Rahm AK, Williams MS, Bradbury AR, Davis FD, Hergenrather J, Hu Y, Kelly MA, Kirchner HL, Meyer MN, Mozersky J, O'Dell SM, Pervola J, Seeley A, Sturm AC, Buchanan AH. *BMC Pediatr*. 2020 May 15;20(1):222. doi: 10.1186/s12887-020-02070-4. PMID: 32414353

[Long overdue: including adults with brain disorders in precision health initiatives.](#)

Finucane BM, Myers SM, Martin CL, Ledbetter DH. *Curr Opin Genet Dev*. 2020 Jun 13;65:47-52. doi: 10.1016/j.gde.2020.05.001. Online ahead of print. PMID: 32544666

[Clinical outcomes of a genomic screening program for actionable genetic conditions.](#)

Buchanan AH, Lester Kirchner H, Schwartz MLB, Kelly MA, Schmidlen T, Jones LK, Hallquist MLG, Rocha H, Betts M, Schwiter R, Butry L, Lazzeri AL, Frisbie LR, Rahm AK, Hao J, Willard HF, Martin CL, Ledbetter DH, Williams MS, Sturm AC. *Genet Med*. 2020 Jun 30. doi: 10.1038/s41436-020-0876-4. Online ahead of print. PMID: 32601386

[Identification of Neuropsychiatric Copy Number Variants in a Health Care System Population.](#)

Martin CL, Wain KE, Oetjens MT, Tolwinski K, Palen E, Hare-Harris A, Habegger L, Maxwell EK, Reid JG, Walsh LK, Myers SM, Ledbetter DH. *JAMA Psychiatry*. 2020 Jul 22:e202159. doi: 10.1001/jamapsychiatry.2020.2159. Online ahead of print. PMID: 32697297

[Association of Bariatric Surgery With Rates of Kidney Function Decline Using Multiple Filtration Markers.](#)

Chang AR, Wood GC, Chu X, Surapaneni A, Grams ME. *JAMA Netw Open*. 2020 Sep 1;3(9):e2014670. doi: 10.1001/jamanetworkopen.2020.14670. PMID: 32886117

[Understanding the Return of Genomic Sequencing Results Process: Content Review of Participant Summary Letters in the eMERGE Research Network.](#)

Lynch JA, Sharp RR, Aufox SA, Bland ST, Blout C, Bowen DJ, Buchanan AH, Halverson C, Harr M, Hebring SJ, Henrikson N, Hoell C, Holm IA, Jarvik G, Kullo IJ, Kochan DC, Larson EB, Lazzeri A, Leppig KA, Madden J, Marasa M, Myers MF, Peterson J, Prows CA, Kulchak Rahm A, Ralston J, Milo Rasouly H, Scrol A, Smith ME, Sturm A, Stuttgen K, Wiesner G, Williams MS, Wynn J, Williams JL. *J Pers Med*. 2020 May 13;10(2):38. doi: 10.3390/jpm10020038. PMID: 32413979

[Predictive Utility of Polygenic Risk Scores for Coronary Heart Disease in Three Major Racial and Ethnic Groups.](#)

Dikilitas O, Schaid DJ, Kosel ML, Carroll RJ, Chute CG, Denny JA, Fedotov A, Feng Q, Hakonarson H, Jarvik GP, Lee MTM, Pacheco JA, Rowley R, Sleiman PM, Stein CM, Sturm AC, Wei WQ, Wiesner GL, Williams MS, Zhang Y, Manolio TA, Kullo IJ. *Am J Hum Genet*. 2020 May 7;106(5):707-716. doi: 10.1016/j.ajhg.2020.04.002. PMID: 32386537

In Press:

Mozersky J, Meyer MN, O'Dell SM, Rahm AK, Buchanan AH. Protecting research participants from psychosocial harms of genetic knowledge: are we compromising external validity of translational research? *Ethics & Human Research*. In press. PMID: pending.