Joel Krier, MD, MMSc

EDUCATION AND TRAINING

Medical Genetics Fellowship. Harvard Medical School (HMS) Genetics Training Program. Boston, MA. 07/2011-06/2014

Master's of Medical Science (MMSc). Biomedical Informatics. HMS. Boston, MA. 09/2012-05/2014

Master's Thesis: "A Matching Algorithm for Unsolved Genomic Sequencing Cases Based on Phenotypic and Genotypic Similarity".

Internal Medicine/Pediatrics Combined Residency. Rush University Medical Center. Chicago, IL. 07/2006-06/2010

MD. University of Pittsburgh School of Medicine. Pittsburgh, PA. 08/2002-05/2006

Awards: Dean's Merit Full-Tuition Scholarship (2002-2006); Gold Humanism Honors Society (2006).

BA Neuroscience, BM Saxophone Performance. Oberlin College and Conservatory of Music. Oberlin, OH. 08/1993-05/1998

Awards: Phi Beta Kappa (1997), Neuroscience Department Award (1998), Pi Kappa Lamda (National Music Honors Society, 1998), Swearer National Student Humanitarian Award for Excellence in Community Service (1996).

PROFESSIONAL EXPERIENCE

Chief. Department of Genetics and Genomics, Atrius Health/Harvard Vanguard Medical Associates. Boston, MA. 01/2022-

Roles and Responsibilities:

- Developing and executing strategic plan for clinical genetics/genomics services at Atrius Health including general pediatric and adult genetics, cancer genetics, reproductive genetics and genomic medicine.
- Sole MD-level clinical geneticist for Atrius Health System.
- Attending five half-day clinic sessions per week in general adult, prenatal and prenatal genetics

Clinical Chief. Division of Genetics, Department of Medicine. Brigham and Women's Hospital (BWH). Boston, MA. 07/2014-12/2021

Roles and Responsibilities:

- Developing and executing strategic plan for clinical genetics/genomics service.
- Budgetary planning and management.
- Attending two-to-four half-day clinic sessions per week and full-time staffing of inpatient consultations.
- Supervising four clinical geneticists, two genetic counselors, and clinical coordinator.
- BWH Rotation Director for HMS Genetics Training Program fellows.

Accomplishments/Awards:

- BWH Department of Medicine "Exemplary Service Award" (2021); among 12 recipients in the inaugural year of the award.
- Brigham Leadership Program (2015-2016): nominated by Division Chief and Department Chair and completed intensive year-long leadership development program jointly organized by BWH and the Harvard Business School.
- Developed new clinical models and services, including telemedicine visits (pre-pandemic), e-consults, and successfully launched two new clinic services in 2019: Preventative Genomics Clinic and Pharmacogenomics Clinic.

- Mentored and hired two former HMS combined program clinical genetics fellows, now attending clinical geneticists.
- Established Partners Healthcare's first infusion program for Morquio A syndrome.
- Developed inpatient protocol for recognition and management of idiopathic hyperammonemia in adults (Stergachis et al, 2020).

Director. Brigham Genomic Medicine (BGM)**.** Division of Genetics, Department of Medicine. Brigham and Women's Hospital (BWH). Boston, MA. 10/2016-

- BGM is a multidisciplinary novel gene/disease association discovery and rare disease genomic sequencing analysis and interpretation program.
- Roles include program administration, identifying appropriate unsolved cases from clinical settings, analysis of genomic sequencing data for unsolved rare disease cases, bioinformatics support, and oversight of BGM computational team.
- Dramatically increased rate of case analysis from pre-2016 vs. 2016-2021, culminating in a publication summarizing >40 likely monogenic diagnoses including multiple novel gene/disease associations in 2018 (Haghighi et al) and multiple subsequent published gene-disease association discoveries.
- Co-developed Anfisa, an open-source genomic variant filtration, annotation, and interpretation tool (2018-2019).
- Co-developing the "Clinical Genome Analysis Platform", an end-to-end computational platform for human genomic data analysis, filtration, annotation and reporting in collaboration with the HMS Dept. of Biomedical Informatics (2019-present).
    - Provided clinical and research vision for platform development and have served as lead clinical and BWH PI since inception of collaboration.
    - The development version of the platform is already being utilized by the BGM research operation, with version 1 to be released July, 2021.

Medical Director. Masters of Science Genetic Counseling Program. Massachusetts General Hospital (MGH) Institute of Health Professions. 10/2018-

- Program and curriculum development for newly accredited genetic counseling program, which graduated first class May, 2021.

Chief Resident, Adjunct Attending. Department of Pediatrics. Rush University Medical Center. 2010-2011

ACADEMIC APPOINTMENTS

Assistant Professor. Department of Medicine, Harvard Medical School.	7/2018-
Associate Professor (Adjunct). MGH Institute of Health Professions.	10/2018-
Instructor. Department of Medicine, Harvard Medical School.	07/2014-6/2018
Clinical Fellow. Department of Pediatrics, Harvard Medical School.	07/2011-06/2014
Instructor. Department of Pediatrics, Rush University Medical Center.	07/2010-06/2011

EDUCATIONAL ACTIVITIES

Course Co-Director, Advanced Clinical and Medical Genetics (2nd Year Master's level course), Genetic Counseling Master's Degree Program**,** MGH Institute of Health Professions.	2020-
Small Group Faculty Facilitator, Human Genetics, Advanced Integrative Science Course, Harvard Medical School.	2020-2021
Lecturer, Genetics Course, Harvard-MIT Program in Health Sciences and Technology (HST), "Clinical Genetic Testing: Prenatal/Postnatal Testing, Personalized Medicine, Pharmacogenetics."	2017-
Lecturer, Human Genetics, Advanced Integrative Science Course, Harvard Medical School, "Personalized Genetics."	2017
BWH Site Director, Clinical Preceptor, Lecturer. Harvard Medical School Genetics Training Program (Clinical and Laboratory Genetics Residency and Fellowship Program). Lecture: "Personalized Genetics."	2014-2021
Director, BWH Internal Medicine/Medical Genetics Residency Program. Harvard Medical School Genetics Training Program. - Led successful application for accreditation of 4-year combined program by American Board of Internal Medicine and American Board of Genetics and Genomics (2018).	2014-2021
Attending Physician. Inpatient Internal Medicine "Integrated Teaching Unit." BWH.	2015
Attending Physician. Inpatient Pediatrics Teaching Service. Rush University Medical Center.	2011
Clinic Preceptor. Internal Medicine-Pediatrics Outpatient Clinic. Rush University Medical Center.	2010-2011
Organizer, Presenter and Facilitator. Pediatrics Residency Educational Conferences and Grand Rounds. Rush University Medical Center.	2010-2011

RESEARCH ACTIVITIES

Past Funding

Center for Integrated Approaches to Undiagnosed Disease
NIH 1U01HG007690-01; PI: Joseph Loscalzo, MD, PhD, MA
Co-Investigator (2015-)

- Harvard Medical School's collaborative clinical site initiative for the NIH Undiagnosed Disease Network.
- Provide case assessment for the Harvard site and management of diagnostic evaluations at Brigham and Women's Hospital (enrollment phase initiated 8/2015).
- Through Brigham Genomic Medicine, lead the genomic sequencing re-analysis for cases unresolved via clinical exome or genome sequencing.
- Led formation of a multidisciplinary UDN evaluation clinic at BWH including Immunology, Cardiology, Endocrinology, and Neurology expertise.

Clinical Genome Analysis Platform
HMS/Blavatnik Institute
BWH Site Principal Investigator (2019-) (co-PI: Shamil Sunyaev, Peter Park)

- This project is focused on developing a cutting-edge analysis platform for genomic sequencing analysis and is a collaborative effort between Brigham Genomic Medicine and the HMS Department of Biomedical Informatics.

The MilSeq Project: Enabling Personalized Medicine
US Air Force FA8650-17-2-6704; PI: Robert Green, MD, MPH
Co-Investigator (2017-2019)

- This project is assessing the impact of implementing clinical genomic sequencing into the care of members of the U.S. Air Force
- Monitoring for safety concerns or miscommunications of genetic information in exome sequencing result disclosure session between primary care physicians and their patients.

SEQuencing a Baby for an Optimal Outcome (SEQaBOO)
NIH5R01DC015052-03; *PI: Cynthia Morton, PhD*
Co-Investigator (2018-2020)

- Adapted and validated Brigham Genomic Medicine's genomic sequencing analysis pipeline for use in CLIA-grade analysis for genetic variants associated with hearing loss.
- Provided conceptual design and data specifications for the development of a variant filtering and annotation tool utilized in the SEQaBOO project (and future BGM applications).

Brigham Genomic Medicine
Brigham Research Institute Director's Transformational Award; PI: Richard Maas, MD, PhD
Co-Investigator (2015-2017)

- Identified cases for genomic sequencing re-analysis from outpatient and inpatient settings.
- Performed and oversaw genomic sequencing analysis for unsolved rare disease cases from BWH, and served as a genomic sequencing analysis service for the Harvard Clinical site of the Undiagnosed Disease Network (NIH) and FaceBase consortium (NIH craniofacial development grant program).

The MedSeq Project: Integration of Whole Genome Sequencing into Clinical Medicine
NIH U01 HG006500; PI: Robert C. Green, MD, MPH
Co-Investigator (2014-2017), Postdoctoral Research Fellow (2012-14)

- Developed analytical approach and designed clinical genome reports, with a focus on pharmacogenomic variant analysis and reporting.
- Assisting primary care physician and cardiologist subjects with genomic report interpretation.
- Monitoring for safety issues and miscommunications of genetic information in genomic sequencing report disclosure sessions.

The ClinGen Project: The Clinical Genome Resource
NIH 1U41HG006834-01A1; PI: Heidi Rehm, PhD
Co-Investigator (2014-2016), Postdoctoral Research Fellow (2013-2014)

- The ClinGen Project's mission is to develop and curate authoritative information on genomic variants that are relevant to human disease and useful for clinical practice.
- Participation in Phenotype Working Group, which is focusing on improving and standardizing and phenotype data acquisition and curation for clinical genomic databases.
- Leading analysis of existing Mendelian disease-phenotype feature annotations in bioinformatics tools compared to expert-provided phenotypic feature lists.

T32 Postdoctoral Research Training Grant
NIH T32 GM007748-34; PI: Cynthia Morton, PhD
Postdoctoral Research Fellow (2012-2014); Research Mentor: Robert C. Green, MD, MPH

Unfunded Projects (Past)

The BabySeq Project
NIH U19 HD077670; PI: Robert Green, MD, MPH
Co-Investigator 2015-

- The BabySeq Project is exploring the utility of exome sequencing in sick and health newborns at Boston Children's Hospital and Brigham and Women's Hospital.
- Adapting the interpretive pipeline and reporting strategy developed for the MedSeq Project into a concise and understandable genomic newborn screening report.

Impact of Personal Genomics (PGEN) Study
NIH R01 HG005092; PI: Robert Green, MD, MPH
Co-Investigator 2015-2017

- Led analysis of how baseline characteristics for pursuing personal genomic testing, such as anxiety and motivation, associate with healthcare utilization after receiving personal genetic results.

CERTIFICATION AND LICENSURE

American Board of Medical Genetics (Clinical Genetics)	2013-
Massachusetts Medical License	2011-
American Board of Internal Medicine	2010-
American Board of Pediatrics	2010-
Illinois Medical License	2009-2011

COMMITTEE SERVICE

Advanced Labs Diagnostic Review Committee, Department of Pathology, BWH	2016-2021
Partners Healthcare Biobank Return of Results Taskforce	2015-2021

Admissions and Resident Performance Review Committee, Harvard Medical School 2015-2021

Genetics Training Program

Economics Committee, American College of Medical Genetics and Genomics 2011-2017

OTHER WORK EXPERIENCE

Clinical Research Assistant. Department of Neurology, Massachusetts General Hospital. Boston, MA. 2001-2002

Business Strategy Associate (Management Consultant). ZEFER, Inc. Boston, MA. 1999-2001

Research Associate (Management Consultant). Nextera, Inc. Boston, MA. 1998-1999

PUBLICATIONS

1. Mueller AA, Sasaki T, Keegan JW . . . **Krier JB**, Loscalzo J, Lederer JA, Rao DA. **High-dimensional immunophenotyping reveals immune cell aberrations in patients with undiagnosed inflammatory and autoimmune diseases**. J Clin Invest. 2023 Dec 15; 133 (24).

2. Bouzinier MA, Etin D, Trifonov SI, Evdokimova VN, Ulitin V, Shen J, Kokorev A, Ghazani AA, Chekaluk Y, Albertyn Z, Giersch A, Morton CC, Abraamyan F, Bendapudi PK, Sunyaev S, Undiagnosed Diseases Network, Brigham Genomic Medicine, SEQuencing A Baby For An Optimal Outcome, Quantori, Krier JB. **AnFiSA: An open-source computational platform for the analysis of sequencing data for rare genetic disease.** J Biomed Inform. 2022 Sep;133:104174.

3. Stergachis AB, **Krier JB**, Merugumala SK, Berry GT, Lin AP. **Clinical utility of brain MRS imaging of patients with adult-onset non-cirrhotic hyperammonemia**. Mol Genet Metab Rep. 2021 Jun. eCollection.

4. Shah SN, Gammal RS, Amato MG, Alobaidly M, Delos Reyes, D, Hasan S, Seger D, **Krier JB**, Bates DW. **Clinical utility of pharmacogenomic data collected by a health-system biobank to predict and prevent adverse drug events**. Drug Saf, 2021 May. 44(5):601-607.

5. Kobren SN, Baldridge D ,Velinder M, **Krier JB**, LeBlanc K, Esteves C, Pusey BN, Zuchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner D, Paul AJ, UDN; Sunyaev SR, Kohane IS. **Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases.** Genet Med, 2021 Feb 12. Online ahead of print.

6. Jiao X, Morleo M, Nigro V, Torella A, D'Arrigo S, Ciaccio C, Pantaleoni C, Gong P, Grand K, Sanchez-Lara PA, **Krier J**, Fieg E, Stergachis A, Wang X, Yang Z. **Identification of an identical de novo SCAMP5 missense variant in four unrelated patients with seizures and severe neurodevelopmental delay**. Front Pharmacol. 2020 Dec 18.

7. Sacoto MJG, Tchasovnikarova IA, Torti E, Forster C, Andrew EH, Anselm I, Baranano KW, Briere LC, Cohen JS, Craigen WJ, Cytrynbaum C, Eknilevitch N, Elrick M, Fatemi A, Fraser JL, GAllagner RC, Guerin A, Haynes D, High F, Inglese CN, Kiss C, Koenig MK, **Krier J,** Lindstrom K, Marble M, Meddaugh H, Moran ES, Morel C, Mu W, Muller EA, Nance J, Natowicz MR, Numis AL, Ostrem B, Pappas J, Stafstrom CE, Streff H, Sweetswer D, Szybowska M, Undiagnosed Disease Network, Walker M, Wang W, Weiss K, Weksberg R,

Wheeler PG, Yoon G, Kingston RE, Juusola J. **De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism**. Am J Hum Genet. 2020 Aug 6;107(2):352-363.

8. Stergachis AB, Mogensen KM, Khoury CC, Lin AP, Peake RW, Baker JJ, Barkoudah E, Sahai I, Sweetser D, Berry G, **Krier J**. **A retrospective study of adult patients with noncirrhotic hyperammonemia**. J Inherit Metab Dis. 26 Jul 2020.

9. Dutta D, Briere L, Kanca O, Marcogliese P, Walker M, High F, Vanderver A, **Krier J,** Carmichael N, Callahan C, Taft RJ, Simons C ,Helman G, Undiagnosed Disease Network, Wanger M, Yamamoto M, Sweetser D, Bellen H. **De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment.** Hum Molec Gen. 2020 May.

10. Cif L, Demailly D, Lin J-P, **Krier J** . . .Coubes P, Gorman KM, Kurian MA. **KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation**. Brain. 2020 Dec 5;143(11):3242-326.

11. Li GZ, Tio MC, Pak LM, **Krier J,** Tullius SG, Riella LV, Malek SY, Stergachis A. **Non-cirrhotic hyperammonemia after deceased donor kidney transplantation: a case report**. Am J Transplant. 2020.

12. Accogli A, Calabretta S, St-Onge J, Addour-Boudrahem N, Dionne-Laporte A, Joset P, Azzarello-Burri S, Rauch A, **Krier J**, Fieg E, Pallais JC, Undiagnosed Diseases Network, McConkie-Rosell A, McDonald M, Freedman SF, Rivière J-B, Lafond-Lapalm J, Simpson BN, Hopkin RJ, Trimouille A, Van-Gils J, Begtrup A, McWalter K, Delphine H, Keren B, Gnevieve D, Argilli E, Sherr EE, Severino M, Rouleau GA, Yam PT, Charron F, Srour M. **De novo pathogenic variants in N-cadherin cause a syndromic neurodevelopmental disorder with agenesis of the corpus callosum and axon pathfinding, cardiac, ocular and genital defects**. Am J Hum Genetics. 2020.

13. Reuter C, Kohler J, Bonner D, Zastrow D, Fernandez L, Dries A, Marwaha S, Davidson J, Brokamp E, Herzog M, Hong J, Macnamara E, Rosenfeld JA, Schoch K, Spillmann R, Loscalzo J, **Krier J**, Stoler J, Sweetser D, Palmer C, Phillips J, Shashi V, Adams D, Yang Y, Ashley E, Fisher P, Mulvihill J, Bernstein J, Wheeler MT. **Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing**. J Genet Couns. 2019 Sept 3.

14. Aday AW, **Krier JB**, Pallais JC, Fieg EL, MacRae CA, Loscalzo J; Members of the UDN. **The Undiagnosed Diseases Network as a tool for graduate medical education**. Am J Med. 2019 Jul 10.

15. Velilla J, Marchetti MM, Toth-Petroczy A, Grosgogeat C, Bennett AH, Carmichael N, Estrella E, Darras BT, Frank NY, **Krier J**, Gaudet R, Gupta VA. **Homozygous TRPV4 mutation causes congenital distal spinal muscular atrophy and arthrogryposis.** Neurol Genet. 2019 Mar. 7;5(2):e312.

16. Murry JB, Machini K, Ceyhan-Birsoy O, Kritzer A, Krier JB, Lebo MS, Fayer S, Genetti CA, VanNoy GE, Yu TW, Agrawal PB, Parad RB, Holm IA, McGuire AL, Green RC, Beggs AH, Rehm HL; BabySeq Project Team. **Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report**. Cold Spring Harb Mol Case Stud. 2018 Aug 1;4(4).

17. Holm IA, Agrawal PB, Ceyhan-Birsoy O, Christensen KD, Fayer S, Frankel LA, Genetti CA, **Krier JB**, LaMay RC, Levy HL, McGuire AL, Parad RB, Park PJ, Pereira S, Rehm HL, Schwartz TS, Waisbren SE, Yu TW; BabySeq Project Team, Green RC, Beggs AH. **The BabySeq project: implementing genomic sequencing in newborns.** BMC Pediatr. 2018 Jul 9;18(1):225.

18. Hart MR, Biesecker BB, Blout CL, Christensen KD, Amendola LM, Bergstrom KL, Biswas S, Bowling KM, Brothers KB, Conlin LK, Cooper GM, Dulik MC, East KM, Everett JN, Finnila CR, Ghazani AA, Gilmore MJ, Goddard KAB, Jarvik GP, Johnston JJ, Kauffman TL, Kelley WV, **Krier JB**, Lewis KL, McGuire AL,

McMullen C, Ou J, Plon SE, Rehm HL, Richards CS, Romasko EJ, Miren Sagardia A, Spinner NB, Thompson ML, Turbitt E, Vassy JL, Wilfond BS, Veenstra DL, Berg JS, Green RC, Biesecker LG, Hindorff LA. **Secondary findings from clinical genomic sequencing: prevalence, patient perspectives, family history assessment, and health-care costs from a multisite study.** Genet Med. 2018 Oct 5.

19. Haghighi A, **Krier JB,** Toth-Petroczy A, Cassa CA, Frank NY, Carmichael N, Fieg E, Bjonnes A, Mohanty A, Briere LC, Lincoln S, Lucia S, Gupta VA, Söylemez O, Sutti S, Kooshesh K, Qiu H, Fay CJ, Perroni V, Valerius J, Hanna M, Frank A, Ouahed J, Snapper SB, Pantazi A, Chopra SS, Leshchiner I, Stitziel NO, Feldweg A, Mannstadt M, Loscalzo J, Sweetser DA, Liao E, Stoler JM, Nowak CB, Sanchez-Lara PA, Klein OD, Perry H, Patsopoulos NA, Raychaudhuri S, Goessling W, Green RC, Seidman CE, MacRae CA, Sunyaev SR, Maas RL, Vuzman D; Undiagnosed Diseases Network, Brigham and Women's Hospital FaceBase Project, Brigham Genomic Medicine (BGM)**. An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery.** NPJ Genom Med. 2018 Aug 13;3:21.

20. Murry JB, Machini K, Ceyhan-Birsoy O, Kritzer A, **Krier JB**, Lebo MS, Fayer S, Genetti CA, VanNoy GE, Yu TW, Agrawal PB, Parad RB, Holm IA, McGuire AL, Green RC, Beggs AH, Rehm HL; BabySeq Project Team. **Reconciling newborn screening and a novel splice variant in BTD associated with partial biotinidase deficiency: a BabySeq Project case report.** Cold Spring Harb Mol Case Stud. 2018 Aug 1;4(4).

21. Holm IA, Agrawal PB, Ceyhan-Birsoy O, Christensen KD, Fayer S, Frankel LA, Genetti CA, **Krier JB**, LaMay RC, Levy HL, McGuire AL, Parad RB, Park PJ, Pereira S, Rehm HL, Schwartz TS, Waisbren SE, Yu TW; BabySeq Project Team, Green RC, Beggs AH. **The BabySeq project: implementing genomic sequencing in newborns**. BMC Pediatr. 2018 Jul 9;18(1):225.

22. Vassy JL, Christensen KD, Schonman EF, Blout CK, Robinson JL, **Krier J**, Diamond PM, Lebo M, Machini K, Azzariti DR, Dukhovny D, Bates D, MacRae CA, Murray MF, Rehm HL, McGuire AL, Green RC for the MedSeq Project. **The impact of genome sequencing on the primary care outcomes of healthy adult patients. A pilot randomized trial.** Ann Int Med. 2017;167(3):159-69.

23. Lin AE, Michot C, Cormier-Daire V, L'Ecuyer TJ, Matherne GP, Barnes BH, Humberson JB, Edmondson AC, Zackai E, O'Connor MJ, Kaplan JD, Ebeid MR, **Krier J**, Krieg E, Ghoshhajra B, Lindsay ME. **Gain-of-function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome.** Am J Med Genet A. 2016 Oct; 170(10):2617-31.

24. **Krier JB**, Kalia SS, Green RC. **Genomic sequencing in clinical practice: applications, challenges, and opportunities.** Dialogues Clin Neurosci. 2016 Sep; 18(3):299-312.

25. **Krier J***, Barfield R*, Green RC, Kraft P. **Reclassification of genetic-based risk predictions as GWAS data accumulate**. Genome Med. 2016 Feb 17; 8(1):20. (*Co-first authors)

26. Cantu S, **Krier J**, Hashemi N. **Hepatocyte Nuclear Factor 1a Mutation-associated MODY-3 and Familial Liver Adenomatosis**. J Clin Gastroenterol. 2016 Feb; 50(2):181-2. (letter to the editor)

27. Lin AE, Michot C, Cormier-Daire V, L'Ecuyer TJ, Matherne GP, Barnes BH, Humberson JB, Edmondson AC, Zackai E, O'Connor MJ, Kaplan JD, Ebeid MR, **Krier J**, Krieg E, Ghoshhajra B, Lindsay ME. **Gain-of-function mutations in SMAD4 cause a distinctive repertoire of cardiovascular phenotypes in patients with Myhre syndrome.** Am J Med Genet A. 2016 Oct; 170(10):2617-31.

28. Cipriano C, Brockman L, Romancik J, Hartemayer R, Ording J, Ginder C, **Krier J**, Gitelis S, Kent P. **The clinical significance of initial pulmonary micronodules in young sarcoma patients**. J Pediatr Hematol Oncol. 2015 Oct; 37(7):548-53.

29. Philippakis AA, Azzariti DR, Beltran S, Brookes AJ, Brownstein CA, Brudno M, Brunner HG, Buske OJ, Carey K, Doll C, Dumitriu S, Dyke SO, den Dunnen JT, Firth HV, Gibbs RA, Girdea M, Gonzalez M, Haendel MA, Hamosh A, Holm IA, Huang L, Hurles ME, Hutton B, **Krier JB**, Misyura A, Mungall CJ, Paschall J, Paten B, Robinson PN, Schiettecatte F, Sobreira NL, Swaminathan GJ, Taschner PE, Terry SF, Washington NL, Züchner S, Boycott KM, Rehm HL. **The matchmaker exchange: a platform for rare disease gene discovery**. 2015 Oct; 36(10):915-21.

30. Vassy JL, McLaughlin HL, MacRae CA, Seidman CE, Lautenbach D, **Krier JB**, Lane WJ, Kohane IS, Murray MF, McGuire AL, Rehm HL, Green RC. **A one-page summary report of genome sequencing for the healthy adult.** Public Health Genomics. 2015;18(2):123-9.

31. McLaughlin HM, Ceyhan-Birsoy O, Christensen KD, Kohane IS, **Krier J**, Lane WJ, Lautenbach D, Lebo MS, Machini K, MacRae CA, Azzariti DR, Murray MF, Seidman CE, Vassy JL, Green RC, Rehm HL; MedSeq Project. **A systematic approach to the reporting of medically relevant findings from whole genome sequencing.** BMC Med Genet. 2014 Dec 14;15(1):134.

32. Kong SK, Lee I, Leschiner I, **Krier J**, Kraft P, Rehm HL, Green RC, Kohane IS, MacRae CA, MedSeq Project. **Summarizing polygenic risks for complex diseases in a clinical whole genome report.** Genet Med. 2014 Oct 23.

33. Vassy JL, Lautenbach DM, McLaughlin HM, Kong SW, Christensen KD, **Krier J**, Kohane IS, Feuerman LZ, Blumenthal-Barby J, Roberts JS, Lehmann LS, Ho CY, Ubel PA, MacRae CA, Seidman CE, Murray MF, McGuire AL, Rehm HL, Green RC; MedSeq Project. **The MedSeq Project: a randomized trial of integrating whole genome sequencing into clinical medicine.** Trials. 2014 Mar 20;15:85.

34. **Krier JB**, Green RC. **The management of incidental findings from genomic sequencing.** Current Protocols in Human Genetics. 2013. Chapter 9: Unit 9.23.

35. Schnipper JL, Ackerman RH, **Krier JB**, Honour M. Diagnostic Yield and Utility of Neurovascular **Ultrasonography in the evaluation of patients with syncope.** Mayo Clinical Proceedings. 2005 Apr; 80 (4):480-8.

PLATFORM PRESENTATIONS AS PRESENTING OR SENIOR AUTHOR (NATIONAL MEETINGS)

1. Maas RL, Liao E, Stoler JM, Sanchez-Lara PA, Klein OD, Carmichael N, Fieg E, Perry HB, Sunyaev S, **Krier J,** NIDCR FaceBase Consortium, Brigham Genomic Medicine. **Rapid Identification and Validation of Human Craniofacial Development Genes**. American Society of Human Genetics Annual Meeting (Platform Presentation by Maas RL). October 2020.

2. Kerman B, Vassy JL, Rana HG, Fieg E, Green R, Blout CL, Gammal R, Zettler B, Krier J. **A provider facing clinical genetics electronic consultation service using a self-educated Primary Care Physician as "first responder": Impact, outcomes and provider satisfaction.** American Society of Human Genetics Annual Meeting (Platform Presentation by Kerman B). October 2020.

3. **Krier J**, Hamosh A, Schiettecatte F, Sobreira N. **Phenotype-based matching in mendeliangenomics.org.** American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. (Platform Presentation by **Krier J**) March, 2016. Tampa, FL.

4. **Krier J**, Blout C, Lautenbach D, Vassy J, Robinson J, Helm M, Lee K, Murray M, Green R. **Communication and management of genomic sequencing results by non-geneticist physicians**. American Society of Human Genetics, Annual Meeting (Platform Presentation by **Krier J**). October, 2015. Baltimore, MD.

5. Gallagher P*, **Krier J**, Carere DA, Chen C, Cupples L, Roberts J, Green R, PGen Study Group. **Healthcare utilization following personal genomic testing.** American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting (Platform Presentation by Gallagher P, medical student mentored by **Krier J**). March, 2015. Salt Lake City, UT. **Mentored medical student project.*

6. **Krier J,** McLaughlin HM, Lane WJ, Metterville D, Leshchiner I, Vassy JL, MacRae C, Lebo MS, Lautenbach D, Green RC, Rehm HL for The MedSeq Project. **The return of pharmacogenomic variants in the Medseq Project: reporting approach and physician response.** American Society of Human Genetics, Annual Meeting. (Platform Presentation by **Krier J**). October, 2013. Boston, MA.

POSTER PRESENTATIONS (NATIONAL MEETINGS, LAST 5 YEARS)

1. **Krier J**, Fieg E, Abraamyan F, Haghighi A, Undiagnosed Disease Network, Brigham Genomic Medicine, Sunyaev S, Maas RL; **Establishing molecular diagnoses for rare monogenic disease: Why do we fail?** American Society of Human Genetics Annual Meeting (Poster Presentation, Reviewer's Choice Designation). October 2020.

2. **Krier J,** Fieg E, Sunyaev S, Haghighi A, Maas R, Green, R. Workflow, **Implementation and remaining challenges for reanalysis of genomic sequencing data by a clinical genomics program**. American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. (Poster Presentation) April, 2019. Seattle, WA.

3. Baker J, Erickson T, Melo de Gusmao C, **Krier J**. **Oral chelation therapy to treat hypermanganesemia**. American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. (Poster Presentation) April, 2019. Seattle, WA.

4. Giersch A, Shen J, Chekaluk Y, Bouzinier M, Cohen M, Gregory K, Hochschild J, Johnson L, Kenna M, **Krier J**, McGrath L, Medina G, Ronner E, Sunyaev S, Morton C. **A genomic future for newborn screening: sequencing a baby for an optimal outcome.** American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. (Poster Presentation) April, 2019. Seattle, WA.

5. Green RC, Blout CL, Lebo MS, **Krier JB**, Smoller J, Karlson EW, Mahanta L, O'Brien K, Christensen KD, Boutin N, Weiss ST. **Returning unanticipated genomic results in a hospital-based research biobank**. American Society of Human Genetics. (Poster Presentation and Poster Talk) October, 2018. San Diego, CA.

6. Carmichael NC, Toth-Petroczy A, Haghighi A, Fieg E, Sunyaev S, Maas R, **Krier J. Successfully analyzed rare disease cases from Brigham Genomic Medicine: What case characteristics predispose to resolution?** American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting. (Poster Presentation) April, 2018. Charlotte, NC.

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