



Jon Bernstein

Professor of Pediatrics (Genetics) and, by courtesy, of Genetics
Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **Center for Academic Medicine- Genetics**

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ACADEMIC CONTACT INFORMATION

- **Administrative Contact - Academic**

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Bio

CLINICAL FOCUS

- Clinical Genetics and Genomics
- Rare Diseases
- Undiagnosed Diseases
- Autism
- Developmental Disorders
- Craniofacial Conditions
- Craniosynostosis
- Cleft Lip
- Cleft Palate

ACADEMIC APPOINTMENTS

- Professor - University Medical Line, Pediatrics - Medical Genetics
- Professor - University Medical Line (By courtesy), Genetics
- Member, Bio-X
- Faculty Affiliate, Institute for Human-Centered Artificial Intelligence (HAI)
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Stanford Medicine Children's Health Center for IBD and Celiac Disease

ADMINISTRATIVE APPOINTMENTS

- Chief, Division of Medical Genetics, Department of Pediatrics, (2016- present)
- Medical Director, Cleft and Craniofacial Center, (2014- present)
- Associate Director, Medical Genetics Residency Program, (2016- present)
- Director, Medical Genetics Residency Program, (2013-2016)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics and Genomics, American Board of Medical Genetics and Genomics (2020)
- Medical Education: Stanford University School of Medicine (2003) CA
- Board Certification: Pediatrics, American Board of Pediatrics (2006)
- Fellowship: Lucile Packard Children's Hospital (2008) CA
- Residency: Lucile Packard Children's Hospital (2006) CA
- PhD, Stanford University , Genetics (2003)

LINKS

- Stanford Medical Genetics: medicalgenetics.stanford.edu

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

My research is focused on the diagnosis, discovery and delineation of rare genetic conditions with a focus on neurodevelopmental disorders. This work includes the application of novel computational methods and multi-omics profiling (whole genome sequencing, long-read DNA sequencing, RNA sequencing, methylomics, metabolomics). I additionally participate in an interdisciplinary project to develop induced pluripotent stem cell and assembloid models of genetic neurodevelopmental disorders.

CLINICAL TRIALS

- Clinical and Genetic Evaluation of Individuals With Undiagnosed Disorders Through the Undiagnosed Diseases Network, Recruiting
- Mapping the Genotype, Phenotype, and Natural History of Phelan-McDermid Syndrome, Not Recruiting

Teaching

COURSES

2021-22

- Human Genetics: GENE 202 (Aut)

Publications

PUBLICATIONS

- **Bi-allelic LAMP3 variants in childhood interstitial lung disease: a surfactant-related disease.** *EBioMedicine*
Louvrier, C., Desroziers, T., Soreze, Y., Delgado Rodriguez, M., Thomas, L., Nau, V., Dastot-Le Moal, F., Bernstein, J. A., Cole, F. S., Damme, M., Fischer, A., Griese, M., Hinds, et al
2025; 113: 105626
- **Telehealth Is Effective in the Evaluation of Individuals With Undiagnosed Rare Disorders: An Undiagnosed Diseases Network Study.** *American journal of medical genetics. Part A*
Tan, Q. K., McConkie-Rosell, A., Mahoney, R., Spillmann, R. C., Schoch, K., Chanprasert, S., Acosta, M. T., Toro, C., Rosenfeld, J. A., Orengo, J. P., Scott, D. A., Granadillo, J. L., Sisco, et al
2024: e63956
- **Aortic Root Dilation and Genotype Associations in Phelan-McDermid Syndrome.** *American journal of medical genetics. Part A*
Gluckman, J., Levy, T., Friedman, K., Garcés, F., Filip-Dhima, R., Quinlan, A., Iannotti, I., Pekar, M., Hernandez, A. L., Nava, M. T., Kravets, E., Siegel, A., Bernstein, et al
2024: e63872
- **Large-scale mutational analysis identifies UNC93B1 variants that drive TLR-mediated autoimmunity in mice and humans.** *The Journal of experimental medicine*

- Rael, V. E., Yano, J. A., Huizar, J. P., Slayden, L. C., Weiss, M. A., Turcotte, E. A., Terry, J. M., Zuo, W., Thiffault, I., Pastinen, T., Farrow, E. G., Jenkins, J. L., Becker, et al
2024; 221 (8)
- **De novo variants in the RNU4-2 snRNA cause a frequent neurodevelopmental syndrome.** *Nature*
Chen, Y., Dawes, R., Kim, H. C., Ljungdahl, A., Stenton, S. L., Walker, S., Lord, J., Lemire, G., Martin-Geary, A. C., Ganesh, V. S., Ma, J., Ellingford, J. M., Delage, et al
2024
 - **The Undiagnosed Diseases Network: Characteristics of solvable applicants and diagnostic suggestions for non-accepted ones.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Mulvihill, J. J., Findley, L., Ni, W., Sinsheimer, J. S., Cole, F. S., Esteves, C., Bernstein, J. A., Newman, J. H., Wheeler, M. T., Mokry, J. R.
2024: 101203
 - **MRI in LARS1 deficiency-Spectrum, patterns, and correlation with acute neurological deterioration.** *Journal of inherited metabolic disease*
Hammann, N., Lenz, D., Bianzano, A., Husain, R. A., Forman, E., Bernstein, J. A., Dattner, T., Engelen, M., Hanson-Kahn, A. K., Isidor, B., Kotzaeridou, U., Tietze, A., Trollmann, et al
2024
 - **Implications of Provider Specialty, Test Type, and Demographic Factors on Genetic Testing Outcomes for Patients with Autism Spectrum Disorder.** *Journal of autism and developmental disorders*
Harrington, C. N., Morales, A., Bernstein, J. A., Calderwood, L.
2024
 - **Impact of genome build on RNA-seq interpretation and diagnostics.** *American journal of human genetics*
Ungar, R. A., Goddard, P. C., Jensen, T. D., Degalez, F., Smith, K. S., Jin, C. A., Bonner, D. E., Bernstein, J. A., Wheeler, M. T., Montgomery, S. B.
2024
 - **Loss of function of FAM177A1, a Golgi complex localized protein, causes a novel neurodevelopmental disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Kohler, J. N., Legro, N. R., Baldrige, D., Shin, J., Bowman, A., Ugur, B., Jackstadt, M. M., Shriver, L. P., Patti, G. J., Zhang, B., Feng, W., McAdow, A. R., Goddard, et al
2024: 101166
 - **Clinical, genetic, and cognitive correlates of seizure occurrences in Phelan-McDermid syndrome.** *Journal of neurodevelopmental disorders*
Levy, T., Gluckman, J., Siper, P. M., Halpern, D., Zweifach, J., Filip-Dhima, R., Holder, J. L., Trelles, M. P., Johnson, K., Bernstein, J. A., Berry-Kravis, E., Powell, C. M., Soorya, et al
2024; 16 (1): 25
 - **Immunological and hematological findings as major features in a patient with a new germline pathogenic CBL variant.** *American journal of medical genetics. Part A*
Stellacci, E., Carter, J. N., Pannone, L., Stevenson, D., Moslehi, D., Venanzi, S., Bernstein, J. A., Tartaglia, M., Martinelli, S.
2024: e63627
 - **Filamin A heart valve disease as a genetic cause of inherited bicuspid and tricuspid aortic valve disease.** *Heart (British Cardiac Society)*
Delwarde, C., Toquet, C., Boureau, A. S., Le Ruz, R., Le Scouarnec, S., Mérot, J., Kyndt, F., Bernstein, D., Bernstein, J. A., Aalberts, J. J., Le Marec, H., Schott, J. J., Roussel, et al
2023
 - **Updated consensus guidelines on the management of Phelan-McDermid syndrome.** *American journal of medical genetics. Part A*
Srivastava, S., Sahin, M., Buxbaum, J. D., Berry-Kravis, E., Soorya, L. V., Thurm, A., Bernstein, J. A., Asante-Otoo, A., Bennett, W. E., Betancur, C., Brickhouse, T. H., Passos Bueno, M. R., Chopra, et al
2023
 - **Alternative polyadenylation alters protein dosage by switching between intronic and 3'UTR sites.** *Science advances*
de Prisco, N., Ford, C., Elrod, N. D., Lee, W., Tang, L. C., Huang, K., Lin, A., Ji, P., Jonnakuti, V. S., Boyle, L., Cabaj, M., Botta, S., Ounap, et al
2023; 9 (7): eade4814
 - **Large 22q13.3 deletions perturb peripheral transcriptomic and metabolomic profiles in Phelan-McDermid syndrome.** *HGG advances*
Breen, M. S., Fan, X., Levy, T., Pollak, R. M., Collins, B., Osman, A., Tocheva, A. S., Sahin, M., Berry-Kravis, E., Soorya, L., Thurm, A., Powell, C. M., Bernstein, et al

2023; 4 (1): 100145

- **A concurrent dual analysis of genomic data augments diagnoses: experiences of two clinical sites in the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Spillmann, R. C., Tan, Q. K., Reuter, C., Schoch, K., Kohler, J., Bonner, D., Zastrow, D., Alkelai, A., Baugh, E., Cope, H., Marwaha, S., Wheeler, M. T., Bernstein, et al
2022
- **TCEAL1 loss-of-function results in an X-linked dominant neurodevelopmental syndrome and drives the neurological disease trait in Xq22.2 deletions.** *American journal of human genetics*
Hijazi, H., Reis, L. M., Pehlivan, D., Bernstein, J. A., Muriello, M., Syverson, E., Bonner, D., Estiar, M. A., Gan-Or, Z., Rouleau, G. A., Lyulcheva, E., Greenhalgh, L., Tessarech, et al
2022
- **A Cross-Sectional Study of the Neuropsychiatric Phenotype of CACNA1C-Related Disorder.** *Pediatric neurology*
Levy, R. J., Timothy, K. W., Underwood, J. F., Hall, J., Bernstein, J. A., Paşca, S. P.
2022; 138: 101-106
- **DPP9 deficiency: An inflammasomopathy that can be rescued by lowering NLRP1/IL-1 signaling.** *Science immunology*
Harapas, C. R., Robinson, K. S., Lay, K., Wong, J., Moreno Traspas, R., Nabavizadeh, N., Rass-Rothschild, A., Boisson, B., Drutman, S. B., Laohamonthonkul, P., Bonner, D., Xiong, J. R., Gorrell, et al
2022; 7 (75): eabi4611
- **2022 Association of Professors of Human and Medical Genetics (APHMG) consensus-based update of the core competencies for undergraduate medical education in genetics and genomics.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Massingham, L. J., Nunez, S., Bernstein, J. A., Gardner, D. P., Parikh, A. S., Strovel, E. T., Quintero-Rivera, F., Association of Professors of Human and Medical Genetics Course Directors Special Interest Group Medical Education Core Curriculum Workgroup, Anderson, H., Ashfaq, M., Bernstein, J., Burke, L., Cross, C., et al
2022
- **Discovering monogenic patients with a confirmed molecular diagnosis in millions of clinical notes with MonoMiner.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Wu, D. W., Bernstein, J. A., Bejerano, G.
2022
- **Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.** *Nature biotechnology*
Goenka, S. D., Gorzynski, J. E., Shafin, K., Fisk, D. G., Pesout, T., Jensen, T. D., Monlong, J., Chang, P. C., Baid, G., Bernstein, J. A., Christle, J. W., Dalton, K. P., Garalde, et al
2022
- **Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.** *Circulation. Genomic and precision medicine*
Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Bernstein, J. A., Ceresnak, S., Chang, P., Christle, et al
2022: CIRCGEN121003591
- **Perceived utility and disutility of genomic sequencing for pediatric patients: Perspectives from parents with diverse sociodemographic characteristics.** *American journal of medical genetics. Part A*
Halley, M. C., Young, J. L., Fernandez, L., Kohler, J. N., Undiagnosed Diseases Network, Bernstein, J. A., Wheeler, M. T., Tabor, H. K.
1800
- **Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder.** *American journal of human genetics*
Küry, S., Ebstein, F., Mollé, A., Besnard, T., Lee, M. K., Vignard, V., Hery, T., Nizon, M., Mancini, G. M., Giltay, J. C., Cogné, B., McWalter, K., Deb, et al
2022
- **TOWARDS TRANSCRIPTOMICS AS A PRIMARY TOOL FOR RARE DISEASE INVESTIGATION.** *Cold Spring Harbor molecular case studies*
Montgomery, S. B., Bernstein, J. A., Wheeler, M. T.
2022
- **Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.** *The New England journal of medicine*

- Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Baid, G., Bernstein, J. A., Ceresnak, S., Chang, et al
2022
- **Beyond race: Recruitment of diverse participants in clinical genomics research for rare disease.** *Frontiers in genetics*
Young, J. L., Halley, M. C., Anguiano, B., Fernandez, L., Bernstein, J. A., Wheeler, M. T., Tabor, H. K., Undiagnosed Diseases Network Consortium
2022; 13: 949422
 - **NSD1 mutations deregulate transcription and DNA methylation of bivalent developmental genes in Sotos syndrome.** *Human molecular genetics*
Brennan, K., Zheng, H., Fahrner, J. A., Shin, J. H., Gentles, A. J., Schaefer, B., Sunwoo, J. B., Bernstein, J. A., Gevaert, O.
2022
 - **Parent-reported measure of repetitive behavior in Phelan-McDermid syndrome.** *Journal of neurodevelopmental disorders*
Srivastava, S., Condry, E., Carmody, E., Filip-Dhima, R., Kapur, K., Bernstein, J. A., Berry-Kravis, E., Powell, C. M., Soorya, L., Thurm, A., Buxbaum, J. D., Sahin, M., Kolevzon, et al
2021; 13 (1): 53
 - **Variable clinical severity in TANGO2 deficiency: Case series and literature review.** *American journal of medical genetics. Part A*
Schymick, J., Leahy, P., Cowan, T., Ruzhnikov, M. R., Gates, R., Fernandez, L., Pramanik, G., Undiagnosed Diseases Network, Yarlagadda, V., Wheeler, M., Bernstein, J. A., Enns, G. M., Lee, C.
2021
 - **InpherNet accelerates monogenic disease diagnosis using patients' candidate genes' neighbors.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Yoo, B., Birgmeier, J., Bernstein, J. A., Bejerano, G.
2021
 - **Functional and structural analyses of novel Smith-Kingsmore Syndrome-Associated MTOR variants reveal potential new mechanisms and predictors of pathogenicity.** *PLoS genetics*
Besterman, A. D., Althoff, T., Elfferich, P., Gutierrez-Mejia, I., Sadik, J., Bernstein, J. A., van Ierland, Y., Kattentidt-Mouravieva, A. A., Nellist, M., Abramson, J., Martinez-Agosto, J. A.
2021; 17 (7): e1009651
 - **"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey.** *Journal of genetic counseling*
Deutch, N. T., Beckman, E., Halley, M. C., Young, J. L., Reuter, C. M., Kohler, J., Bernstein, J. A., Wheeler, M. T., Undiagnosed Diseases Network, Ormond, K. E., Tabor, H. K.
2021
 - **Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome.** *American journal of medical genetics. Part A*
Sheppard, S. E., Campbell, I. M., Harr, M. H., Gold, N., Li, D., Bjornsson, H. T., Cohen, J. S., Fahrner, J. A., Fatemi, A., Harris, J. R., Nowak, C., Stevens, C. A., Grand, et al
2021
 - **Functional and structural analysis of cytokine selective IL6ST defects that cause recessive hyper-IgE syndrome.** *The Journal of allergy and clinical immunology*
Chen, Y., Zastrow, D. B., Metcalfe, R. D., Gartner, L., Krause, F., Morton, C. J., Marwaha, S., Fresard, L., Huang, Y., Zhao, C., McCormack, C., Bick, D., Worthey, et al
2021
 - **Biallelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Melo, U. S., Bonner, D. n., Kent Lloyd, K. C., Moshiri, A. n., Willis, B. n., Lanoue, L. n., Bower, L. n., Leonard, B. C., Martins, D. J., Gomes, F. n., de Souza Leite, F. n., Oliveira, D. n., Kitajima, et al
2021
 - **Strong evidence for genotype-phenotype correlations in Phelan-McDermid syndrome: Results from the developmental Synaptopathies consortium.** *Human molecular genetics*
Levy, T., Foss-Feig, J. H., Betancur, C., Siper, P. M., Pilar Trelles-Thorne, M., Halpern, D., Frank, Y., Lozano, R., Layton, C., Britvan, B., Bernstein, J. A., Buxbaum, J. D., Berry-Kravis, et al

2021

- **"It seems like COVID-19 now is the only disease present on Earth": living with a rare or undiagnosed disease during the COVID-19 pandemic.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Halley, M. C., Stanley, T. n., Maturi, J. n., Goldenberg, A. J., Bernstein, J. A., Wheeler, M. T., Tabor, H. K.

2021

- **Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis and treatment suggestions.** *Journal of inherited metabolic disease*

Ng, B. G., Eklund, E. A., Shiryayev, S. A., Dong, Y. Y., Abbott, M., Asteggiano, C., Bamshad, M. J., Barr, E., Bernstein, J. A., Chelakkadan, S., Christodoulou, J., Chung, W. K., Ciliberto, et al

2020

- **Psychometric Study of the Social Responsiveness Scale in Phelan-McDermid Syndrome.** *Autism research : official journal of the International Society for Autism Research*

Gergoudis, K. n., Weinberg, A. n., Templin, J. n., Farmer, C. n., Durkin, A. n., Weissman, J. n., Siper, P. n., Foss-Feig, J. n., Del Pilar Trelles, M. n., Bernstein, J. A., Buxbaum, J. D., Berry-Kravis, E. n., Powell, et al

2020

- **Candidate variants in TUB are associated with familial tremor.** *PLoS genetics*

Sailani, M. R., Jahanbani, F. n., Abbott, C. W., Lee, H. n., Zia, A. n., Rego, S. n., Winkelmann, J. n., Hopfner, F. n., Khan, T. N., Katsanis, N. n., Müller, S. H., Berg, D. n., Lyman, et al

2020; 16 (9): e1009010

- **Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Lenz, D. n., Smith, D. E., Crushell, E. n., Husain, R. A., Salomons, G. S., Alhaddad, B. n., Bernstein, J. A., Bianzano, A. n., Biskup, S. n., Brennenstuhl, H. n., Caldari, D. n., Dikow, N. n., Haack, et al

2020

- **Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Schoch, K. n., Esteves, C. n., Bican, A. n., Spillmann, R. n., Cope, H. n., McConkie-Rosell, A. n., Walley, N. n., Fernandez, L. n., Kohler, J. N., Bonner, D. n., Reuter, C. n., Stong, N. n., Mulvihill, et al

2020

- **Combined Genome Sequencing and RNA Analysis Reveals and Characterizes a Deep Intronic Variant in IGHMBP2 in a Patient With Spinal Muscular Atrophy With Respiratory Distress Type 1.** *Pediatric neurology*

Bodle, E. E., Zhu, W. n., Velez-Bartolomei, F. n., Tesi-Rocha, A. n., Liu, P. n., Bernstein, J. A.

2020; 114: 16–20

- **Network Effects of the 15q13.3 Microdeletion on the Transcriptome and Epigenome in Human-Induced Neurons.** *Biological psychiatry*

Zhang, S. n., Zhang, X. n., Purmann, C. n., Ma, S. n., Shrestha, A. n., Davis, K. N., Ho, M. n., Huang, Y. n., Pattni, R. n., Wong, W. H., Bernstein, J. A., Hallmayer, J. n., Urban, et al

2020

- **Automated syndrome diagnosis by three-dimensional facial imaging.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Hallgrímsson, B. n., Aponte, J. D., Katz, D. C., Bannister, J. J., Riccardi, S. L., Mahasuwan, N. n., McInnes, B. L., Ferrara, T. M., Lipman, D. M., Neves, A. B., Spitzmacher, J. A., Larson, J. R., Bellus, et al

2020

- **AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature.** *Science translational medicine*

Birgmeier, J. n., Haeussler, M. n., Deisseroth, C. A., Steinberg, E. H., Jagadeesh, K. A., Ratner, A. J., Guturu, H. n., Wenger, A. M., Diekhans, M. E., Stenson, P. D., Cooper, D. N., Ré, C. n., Beggs, et al

2020; 12 (544)

- **De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation.** *American journal of human genetics*

Mao, D. n., Reuter, C. M., Ruzhnikov, M. R., Beck, A. E., Farrow, E. G., Emrick, L. T., Rosenfeld, J. A., Mackenzie, K. M., Robak, L. n., Wheeler, M. T., Burrage, L. C., Jain, M. n., Liu, et al

2020

- **Neuronal defects in a human cellular model of 22q11.2 deletion syndrome.** *Nature medicine*
Khan, T. A., Revah, O. n., Gordon, A. n., Yoon, S. J., Krawisz, A. K., Goold, C. n., Sun, Y. n., Kim, C. H., Tian, Y. n., Li, M. Y., Schaepe, J. M., Ikeda, K. n., Amin, et al
2020
- **Diffusion Tensor Imaging Abnormalities in the Uncinate Fasciculus and Inferior Longitudinal Fasciculus in Phelan-McDermid Syndrome.** *Pediatric neurology*
Bassell, J. n., Srivastava, S. n., Prohl, A. K., Scherrer, B. n., Kapur, K. n., Filip-Dhima, R. n., Berry-Kravis, E. n., Soorya, L. n., Thurm, A. n., Powell, C. M., Bernstein, J. A., Buxbaum, J. D., Kolevzon, et al
2020
- **De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Mirzaa, G. M., Chong, J. X., Piton, A., Popp, B., Foss, K., Guo, H., Harripaul, R., Xia, K., Scheck, J., Aldinger, K. A., Sajan, S. A., Tang, S., Bonneau, et al
2019
- **Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing.** *Journal of genetic counseling*
Reuter, C. M., Kohler, J. N., Bonner, D., Zastrow, D., Fernandez, L., Dries, A., Marwaha, S., Davidson, J., Brokamp, E., Herzog, M., Hong, J., Macnamara, E., Rosenfeld, et al
2019
- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review.** *American journal of medical genetics. Part A*
Kumar, A., Zastrow, D. B., Kravets, E. J., Belefard, D., Ruzhnikov, M. R., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Undiagnosed Diseases Network, Mackenzie, K. M., et al
2019
- **S-CAP extends pathogenicity prediction to genetic variants that affect RNA splicing.** *Nature genetics*
Jagadeesh, K. A., Paggi, J. M., Ye, J. S., Stenson, P. D., Cooper, D. N., Bernstein, J. A., Bejerano, G.
2019
- **Reanalysis of Clinical Exome Sequencing Data.** *The New England journal of medicine*
Liu, P., Meng, L., Normand, E. A., Xia, F., Song, X., Ghazi, A., Rosenfeld, J., Magoulas, P. L., Braxton, A., Ward, P., Dai, H., Yuan, B., Bi, et al
2019; 380 (25): 2478–80
- **AVADA: toward automated pathogenic variant evidence retrieval directly from the full-text literature.** *Genetics in medicine : official journal of the American College of Medical Genetics*
Birgmeier, J. n., Deisseroth, C. A., Hayward, L. E., Galhardo, L. M., Tierno, A. P., Jagadeesh, K. A., Stenson, P. D., Cooper, D. N., Bernstein, J. A., Haeussler, M. n., Bejerano, G. n.
2019
- **Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases.** *Acta neuropathologica*
Perenthaler, E. n., Nikoncuk, A. n., Yousefi, S. n., Berdowski, W. M., Alsagob, M. n., Capo, I. n., van der Linde, H. C., van den Berg, P. n., Jacobs, E. H., Putar, D. n., Ghazvini, M. n., Aronica, E. n., van IJcken, et al
2019
- **Genomics in medicine: a novel elective rotation for internal medicine residents.** *Postgraduate medical journal*
Geng, L. N., Kohler, J. N., Levonian, P. n., Bernstein, J. A., Ford, J. M., Ahuja, N. n., Witteles, R. n., Hom, J. n., Wheeler, M. n.
2019
- **Mutation update for the SATB2 gene.** *Human mutation*
Zarate, Y. A., Bosanko, K. A., Caffrey, A. R., Bernstein, J. A., Martin, D. M., Williams, M. S., Berry-Kravis, E. M., Mark, P. R., Manning, M. A., Bhambhani, V. n., Vargas, M. n., Seeley, A. H., Estrada-Veras, et al
2019
- **A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing.** *Journal of genetic counseling*
Zastrow, D. B., Kohler, J. N., Bonner, D. n., Reuter, C. M., Fernandez, L. n., Grove, M. E., Fisk, D. G., Yang, Y. n., Eng, C. M., Ward, P. A., Bick, D. n., Worthey, E. A., Fisher, et al

2019; 28 (2): 213–28

- **A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis.** *Journal of general internal medicine*
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