Stanford



Jon Bernstein

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

Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

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ACADEMIC CONTACT INFORMATION

- **Administrative Contact - Academic**
  Stephanie Martinez
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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-Univ Med Line, Pediatrics - Medical Genetics
- Professor-Univ Med 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, Recruiting

Publications

PUBLICATIONS

- **Genetic variants in recurrent euploid pregnancy loss.** *Genome medicine*
  Aminbeidokhti, M., Halstead, M., Rodriguez-Escriba, M., Tise, C. G., Bernstein, J. A., Cakmak, H., Pollard, E., Giudice, L. C., Merrion, K., Shaw, G. M., Edelman, A., Baldwin, M., Stevenson, et al
  2026; 18 (1)

- **MetaDome 2027: a comprehensively updated resource for aggregating missense variant evidence across homologous human protein domains.** *Nucleic acids research*
  Wiel, L., Ferraro, F., Yu, J., Zhen, J., Nachun, D., Mendez, R., Reuter, C. M., Cui, J. L., Bonner, D. E., Carter, J. N., Marwaha, S., van de Vorst, M., Emami, et al
  2026

- **Domain-associated phenotypic patterns with MACF1 variants.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Gogate, N., Jolly, A., Rosenfeld, J. A., Bahena-Carbajal, P., Carrasco, D., Bernstein, J. A., Bonner, D. E., Busa, T., Cristian, I., D'Souza, P., Friedman, J., Gorokhova, S., Haaf, et al
  2026: 102776

- **Arriving at a diagnosis: Effective strategies used by the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Hwang, S., Brown, R. M., Baldridge, D., Baldwin, E. E., Beggs, A. H., Bernstein, J. A., Blue, E., Borja, N. A., Botto, L. D., Briere, L. C., Cassini, T., Esteves, C., Fieg, et al
  2026: 102718

- **BHLHE22 monoallelic and biallelic variants cause a neurodevelopmental disorder with agenesis of the corpus callosum, intellectual disability, abnormal muscle tone and movement abnormalities.** *Journal of medical genetics*

Le, C., Kalayci, T., Uyguner, Z., Karaman, B., Demirören, T., Beck, B., Winnerling, N., George, E., DiTroia, S., Héron, D., Sabatier, I., Rodan, L. H., Girisha, et al

2026

- **Cognitive Decline, Neurologic Involvement, and Neonatal Crisis in ABCC9-Related Intellectual Disability and Myopathy Syndrome.** *Neurology. Genetics*

  Nagaraj, V., Thomas, Q. H., Nóbrega, P. R., Rodriguez Gil, J. L., Garavatti, E., Scala, M., Severino, M., Efthymiou, S., Amaral, S., Ng, T. C., Inder, T., Gao, J., Matreyek, et al

  2026; 12 (4): e200385

- **"It didn't feel like anything unusual because we had already been through so much": Disability-Related Research Experiences of Families with Children Enrolled in the Undiagnosed Diseases Network.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Mintz, K. T., Altamirano, E. N., Halley, M. C., Barton, K. S., Cho, M. K., Bernstein, J. A., Carter, J. N., Undiagnosed Diseases Network, Tabor, H. K., Quinlan, A., Elkadri, A., Beggs, A. H., La Spada, A. R., et al

  2026: 102663

- **Autonomous biomedical research with an artificial intelligence agent.** *Science (New York, N.Y.)*

  Huang, K., Zhang, S., Wang, H., Qu, Y., Lu, Y., Li, R., Roohani, Y., Qiu, L., Cao, S., Li, G., Zhang, J., Yin, D., Wierenga, et al

  2026: eadz4351

- **RNU4ATAC-opathy: Clinical, molecular and transcriptomic insights from a large cohort.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Matalon, D. R., Duker, A. L., Arriaga, T. M., Russell, K., Mendez, H. R., Bonner, D. E., Harley, M. E., Singer-Berk, M., Wojcik, M. H., Pais, L., DiTroia, S., O'Leary, M., Cassini, et al

  2026: 102633

- **Ensilication preserves high-molecular weight native DNA for clinical long-read sequencing.** *Genome biology*

  Ferrasse, A., Mendez, R., Gorzynski, J. E., Reuter, C., Carter, J. N., Blas, M., Bernstein, J. A., Wheeler, M. T., Banal, J. L., Ashley, E. A.

  2026; 27 (1)

- **Psychotic Features in Myhre Syndrome: Evidence for Broader Neuropsychiatric Surveillance.** *American journal of medical genetics. Part C, Seminars in medical genetics*

  Ebuen, M., Krishnan, V., Irby, K., Monsberger, R., Hopkin, R. J., Brand, M. R., Friedman, N., Thom, R. P., Reiss, A. L., Algaze, C., Bernstein, J. A., Alexander, S., Cormier-Daire, et al

  2026

- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*

  Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Andel, et al

  2026

- **Biallelic variants in the noncoding RNA gene RNU4-2 cause a recessive neurodevelopmental syndrome with distinct white matter changes.** *Nature genetics*

  Rius, R., Blakes, A. J., Chen, Y., De Jonghe, J., Lecoquierre, F., Dawes, R., Cogne, B., Kim, H. C., Alvi, J. R., Amblard, F., Ansari, M., Arlt, A., Austin-Tse, et al

  2026

- **Biallelic variants in RNU2-2 cause the most prevalent known recessive neurodevelopmental disorder.** *Nature genetics*

  Greene, D., Mendez, R., Lees, J., Barbosa, M., Bruselles, A., Chiriatti, L., Ferraro, F., Mancini, C., Schot, R., Sleutels, F., Bertini, E., Bonner, D. E., Bouman, et al

  2026

- **Biallelic Variants in RNU6ATAC Result in a Minor Spliceopathy Characterized by Transcriptome-Wide Minor Intron Retention Events and Short Stature with Variable Multisystem Manifestations.** *HGG advances*

  Mendez, R., Arriaga, T. M., Ma, J., Bonner, D. E., Emami, S., Levy, R. J., Alsagheir, A., Alhaddad, B., Bakur, K., Ungar, R. A., Matalon, D. R., Miller, A. M., Nguyen, et al

  2026: 100588

- **The Human Intolerome: a curated database to prioritize genomic variants in stillbirth, pregnancy loss, and neonatal death.** *Genetics in medicine : official journal of the American College of Medical Genetics*

Yatsenko, S. A., Nagasuri, A., Soman, V., Garakani, A., Aminbeidokhti, M., Chandran, U., Aarabi, M., Oskotsky, T., Bukhari, S. H., Hypes, C., Gu, Z., Monteiro, R., Smith, et al

2026: 102546

- **Genetic Diagnosis and Discovery Enabled by Large Language Models.** *Advanced science (Weinheim, Baden-Wurttemberg, Germany)*

  Tu, T., Saab, K., Liu, W., Fang, Z., Cheng, Z., Spasic, S., Djurisic, M., Mohri, H., Ren, W., Palepu, A., Gottweis, J., Karthikesalingam, A., Kulkarni, et al

  2026: e18656

- **Biallelic LAMP3 Variants in Five Families with Interstitial Lung Disease: Evidence of a Disease-Gene Association.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Keehan, L. A., Ono-Minagi, H., Hadhud, M., Rips, J., Hinds, D. M., Fischer, A. J., Bartlett, J. A., McCray, P. B., Qawasmi, N., Nathan, N., Louvrier, C., Desroziers, T., Damme, et al

  2026: 102531

- **DNASE1L3 Deficiency With Novel Missense Variant: Enzymatic and Plasma Fragmentomic Evidence of Pathogenicity and Partial Response to JAK Blockade.** *ACR open rheumatology*

  Leon Tenorio, A. A., Sugio, T., Cheng, J., Bonner, D. E., Esfahani, M. S., Kasinathan, S., Hsu, J. J., Moyer, A., Pimentel Vera, L., Carter, J., Reuter, C. M., Marwaha, S., Balboni, et al

  2026; 8 (2): e70184

- **Developmental convergence and divergence in human stem cell models of autism.** *Nature*

  Gordon, A., Yoon, S. J., Bicks, L. K., Martín, J. M., Pintacuda, G., Arteaga, S., Wamsley, B., Guo, Q., Elahi, L., Dolmetsch, R. E., Bernstein, J. A., O'Hara, R., Hallmayer, et al

  2026

- **De novo heterozygous variants of the RSF1 gene are responsible for a syndromic neurodevelopmental disorder.** *European journal of human genetics : EJHG*

  Jost, C., Busa, T., Wegner, D., Shinawi, M., Schaefer, E., Piton, A., Schluth-Bolard, C., Charles, P., Keren, B., Mayerhanser, K., Brunet, T., Schatz, U., Neil, et al

  2026

- **An encyclopedia of the cord blood metabolome reveals maternal-fetal interactions and disease risk.** *Cell reports. Medicine*

  Lancaster, S., Mataraso, S., Reiss, J. D., Contrepois, K., Trowbridge, C. A., Michael, B., Simms, I., Ellenberger, M., Gibson, M., Clary, M., McGuire, L., Wong, F., Canfield, et al

  2026: 102548

- **De novo variants in KDM2A cause a syndromic neurodevelopmental disorder.** *American journal of human genetics*

  Anderson, E. N., Drukewitz, S., Kour, S., Chimata, A. V., Rajan, D. S., Schönnagel, S., Stals, K. L., Donnelly, D., O'Sullivan, S., Mantovani, J. F., Tan, T. Y., Stark, Z., Zacher, et al

  2025

- **"Nobody listened to us for years": Parents' experiences of provider communication in the diagnostic odyssey.** *Molecular genetics and metabolism*

  Nguyen, M. M., Mahfoozi, S., Bonner, D., Martschenko, D. O., Giri, A., Tang, C., Bernstein, J. A., Wheeler, M. T., Halley, M. C.

  2025; 146 (4): 109283

- **GREGoR: accelerating genomics for rare diseases.** *Nature*

  Dawood, M., Heavner, B., Wheeler, M. M., Ungar, R. A., LoTempio, J., Wiel, L., Berger, S., Bernstein, J. A., Chong, J. X., Délot, E. C., Eichler, E. E., Lupski, J. R., Shojaie, et al

  2025; 647 (8089): 331-342

- **RAPSN-Associated Congenital Myasthenic Syndrome due to Biallelic Single Nucleotide Variants at the Same Position.** *Case reports in genetics*

  Keehan, L., Carter, J. N., Kravets, E., Wheeler, M. T., Bernstein, J. A., Maselli, R. A., Sampson, J. B., Bachir, S.

  2025; 2025: 1882021

- **Transcriptome-wide outlier approach identifies individuals with minor spliceopathies.** *American journal of human genetics*

  Arriaga, T. M., Mendez, R., Ungar, R. A., Bonner, D. E., Matalon, D. R., Lemire, G., Goddard, P. C., Padhi, E. M., Miller, A. M., Nguyen, J. V., Ma, J., Smith, K. S., Scott, et al

  2025

- **Genetic Subtypes of Phelan-McDermid Syndrome Exhibit Similar Rates of Change Despite Differences in Level of Impairment in Developmental Constructs.** *American journal on intellectual and developmental disabilities*
  Levy, T., Farmer, C., Srivastava, S., Johnson, K., Trayvick, J., Brune, C., Massa, A., Silver, H., Siper, P. M., Zweifach, J., Halpern, D., Foss-Feig, J. H., Bernstein, et al
  2025; 130 (5): 395-413

- **Which Score for What? Operationalizing Standardized Cognitive Test Performance for the Assessment of Change.** *American journal on intellectual and developmental disabilities*
  Farmer, C., Thurm, A., Das, T., Bebin, E. M., Bernstein, J. A., Berry-Kravis, E., Buxbaum, J. D., Eng, C., Frazier, T., Hardan, A. Y., Kolevzon, A., Krueger, D. A., Martinez-Agosto, et al
  2025; 130 (5): 344-361

- **Retrospective Reports of Skill Attainment and Loss in Phelan-McDermid Syndrome.** *American journal on intellectual and developmental disabilities*
  Farmer, C., Giserman-Kiss, I., Mohanty, E., Valluripalli Soorya, L., Sahin, M., Kolevzon, A., Buxbaum, J. D., Berry-Kravis, E., Powell, C. M., Bernstein, J. A., Thurm, A.
  2025; 130 (5): 362-379

- **Longitudinal Trajectory of Adaptive Skills in Phelan-McDermid Syndrome.** *American journal on intellectual and developmental disabilities*
  Srivastava, S., Johnson, K., Farmer, C., Levy, T., Thurm, A., Valluripalli Soorya, L., Filip-Dhima, R., Quinlan, A., Bernstein, J. A., Berry-Kravis, E., Powell, C. M., Buxbaum, J. D., Sahin, et al
  2025; 130 (5): 380-394

- **Characterizing Developmental and Behavioral Profiles in Developmental Synaptopathies to Inform Clinical Trial Endpoints.** *American journal on intellectual and developmental disabilities*
  Valluripalli Soorya, L., Brune, C. W., Farmer, C. A., Ocampo, E. V., Berger, N. I., Pearson, D. A., Busch, R. M., Klaas, P., Siper, P., Currans, K., Gulsrud, A. C., Phillips, J. M., Filip-Dhima, et al
  2025; 130 (5): 414-437

- **Genome Sequencing Uncovers Additional Findings in Phelan-McDermid Syndrome.** *American journal of medical genetics. Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics*
  Moses, R. G., Similuk, M., Hehn, A., Duncan, R., Pekar, M., Gordon-Lipkin, E., Acosta, M. T., Zeltser, D., Reynolds-Lallement, N., Soorya, L., Sahin, M., Levy, T., Kolevzon, et al
  2025: e33036

- **Protein-truncating variants and deletions of SHANK2 are associated with autism spectrum disorder and other neurodevelopmental concerns.** *Journal of neurodevelopmental disorders*
  Silver, H., Greenberg, R., Siper, P. M., Zweifach, J., Soufer, R., Sahin, M., Berry-Kravis, E., Soorya, L. V., Thurm, A., Bernstein, J. A., Kolevzon, A., Grice, D. E., Buxbaum, et al
  2025; 17 (1): 25

- **Integration of transcriptomics and long-read genomics prioritizes structural variants in rare disease.** *Genome research*
  Jensen, T. D., Ni, B., Reuter, C. M., Gorzynski, J. E., Fazal, S., Bonner, D., Ungar, R. A., Goddard, P. C., Raja, A., Ashley, E. A., Bernstein, J. A., Zuchner, S., Greicius, et al
  2025

- **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., Garces, 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., Baldridge, 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., Pașca, 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

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