Stanford

Jon Bernstein

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

Pediatrics - Medical Genetics

CLINICAL OFFICE (PRIMARY)

- **Center for Academic Medicine- Genetics**
  453 Quarry Rd MC 5660
  Stanford, CA 94304
  Tel (650) 721-5804 **Fax** (650) 498-4555

ACADEMIC CONTACT INFORMATION

- **Administrative Contact - Academic**
  Stephanie Martinez
  Email smart545@stanford.edu
  Tel 650-498-4937

Bio

CLINICAL FOCUS

- Autism
- Developmental Disorders
- Undiagnosed Diseases
- Craniofacial Conditions
- Craniosynostosis
- Cleft Lip
- Cleft Palate
- Medical Biochemical Genetics

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 (2009)
- 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 of neurodevelopmental disorders. This work includes the application of novel computational methods and multi-omics profiling (whole genome sequencing, RNA sequencing, metabolomics). I additionally participate in an interdisciplinary project to develop induced pluripotent stem cell (iPSC) models of genetic neurodevelopmental disorders..

CLINICAL TRIALS

- Mapping the Genotype, Phenotype, and Natural History of Phelan-McDermid Syndrome, Recruiting

Teaching

COURSES

2021-22

- Human Genetics: GENE 202 (Aut)

2020-21

- Human Genetics: GENE 202 (Aut)

STANFORD ADVISEES

Page Goddard

Publications

PUBLICATIONS

- **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., Pa#ca, 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., Condy, 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*
  Deuitch, 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., Shiryaev, 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., Beleford, 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*
  Hom, J. n., Marwaha, S. n., Postolova, A. n., Kittle, J. n., Vasquez, R. n., Davidson, J. n., Kohler, J. n., Dries, A. n., Fernandez-Betancourt, L. n., Majcherska, M. n., Dearlove, J. n., Raghavan, S. n., Vogel, et al
  2019

- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*
  Frésard, L. n., Smail, C. n., Ferraro, N. M., Teran, N. A., Li, X. n., Smith, K. S., Bonner, D. n., Kernohan, K. D., Marwaha, S. n., Zappala, Z. n., Balliu, B. n., Davis, J. R., Liu, et al
  2019

- **Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students.** *Journal of genetic counseling*
  Grove, M. E., White, S. n., Fisk, D. G., Rego, S. n., Dagan-Rosenfeld, O. n., Kohler, J. N., Reuter, C. M., Bonner, D. n., Wheeler, M. T., Bernstein, J. A., Ormond, K. E., Hanson-Kahn, A. K.
  2019

- **High Frequency Actionable Pathogenic Exome Variants in an Average-Risk Cohort.** *Cold Spring Harbor molecular case studies*
  Rego, S., Dagan-Rosenfeld, O., Zhou, W., Sailani, M. R., Limcaoco, P., Colbert, E., Avina, M., Wheeler, J., Craig, C., Salins, D., Rost, H. L., Dunn, J., McLaughlin, et al
  2018

- **An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease.** *European journal of human genetics : EJHG*
  Chen, H. I., Jagadeesh, K. A., Birgmeier, J., Wenger, A. M., Guturu, H., Schelley, S., Bernstein, J. A., Bejerano, G.
  2018

- **Change in Prevalence of Orofacial Clefts in California between 1987 and 2010.** *American journal of medical genetics. Part A*
  Andrew, T., Yang, W., Bernstein, J. A., Shaw, G. M.
  2018

- **Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Jagadeesh, K. A., Birgmeier, J., Guturu, H., Deisseroth, C. A., Wenger, A. M., Bernstein, J. A., Bejerano, G.
  2018

- **Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Olahova, M., Yoon, W., Thompson, K., Jangam, S., Fernandez, L., Davidson, J. M., Kyle, J. E., Grove, M. E., Fisk, D. G., Kohler, J. N., Holmes, M., Dries, A. M., Huang, et al
  2018; 102 (3): 494–504

- **Prenatal treatment of ornithine transcarbamylase deficiency.** *Molecular genetics and metabolism*
  Wilnai, Y. n., Blumenfeld, Y. J., Cusmano, K. n., Hintz, S. R., Alcorn, D. n., Benitz, W. E., Berquist, W. E., Bernstein, J. A., Castillo, R. O., Concepcion, W. n., Cowan, T. M., Cox, K. L., Lyell, et al
  2018

- **MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance.** *American journal of human genetics*
  Dobyns, W. B., Aldinger, K. A., Ishak, G. E., Mirzaa, G. M., Timms, A. E., Grout, M. E., Dremmen, M. H., Schot, R. n., Vandervore, L. n., van Slegtenhorst, M. A., Wilke, M. n., Kasteleijn, E. n., Lee, et al
  2018

- **Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations.** *Neuron*
  Tripathy, R. n., Leca, I. n., van Dijk, T. n., Weiss, J. n., van Bon, B. W., Sergaki, M. C., Gstrein, T. n., Breuss, M. n., Tian, G. n., Bahi-Buisson, N. n., Paciorkowski, A. R., Pagnamenta, A. T., Wenninger-Weinzierl, et al
  2018

- **Ethical Issues in Contemporary Clinical Genetics.** *Mayo Clinic proceedings. Innovations, quality & outcomes*
  Braverman, G. n., Shapiro, Z. E., Bernstein, J. A.
  2018; 2 (2): 81–90

- **Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease.** *The New England journal of medicine*
  Splinter, K. n., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C. n., Gahl, W. A., Hamid, R. n., Jacob, H. J., Kikani, B. n., Koeller, et al
  2018

- **Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome.** *Pediatric neurology*
  Srivastava, S. n., Scherrer, B. n., Prohl, A. K., Filip-Dhima, R. n., Kapur, K. n., Kolevzon, A. n., Buxbaum, J. D., Berry-Kravis, E. n., Soorya, L. n., Thurm, A. n., Powell, C. M., Bernstein, J. A., Warfield, et al
  2018

- **ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Deisseroth, C. A., Birgmeier, J. n., Bodle, E. E., Kohler, J. N., Matalon, D. R., Nazarenko, Y. n., Genetti, C. A., Brownstein, C. A., Schmitz-Abe, K. n., Schoch, K. n., Cope, H. n., Signer, R. n., Martinez-Agosto, et al
  2018

- **De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features.** *American journal of human genetics*
  Tokita, M. J., Chen, C. A., Chitayat, D. n., Macnamara, E. n., Rosenfeld, J. A., Hanchard, N. n., Lewis, A. M., Brown, C. W., Marom, R. n., Shao, Y. n., Novacic, D. n., Wolfe, L. n., Wahl, et al
  2018

- **Exploring the Medical and Psychosocial Concerns of Adolescents and Young Adults With Craniofacial Microsomia: A Qualitative Study.** *The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association*
  Hamilton, K. V., Ormond, K. E., Moscarello, T. n., Bruce, J. S., Bereknyei Merrell, S. n., Chang, K. W., Bernstein, J. A.
  2018: 1055665618768542

- **Biallelic loss-of-function WNT5A mutations in an infant with severe and atypical manifestations of Robinow syndrome.** *American journal of medical genetics. Part A*
  Birgmeier, J. n., Esplin, E. D., Jagadeesh, K. A., Guturu, H. n., Wenger, A. M., Chaib, H. n., Buckingham, J. A., Bejerano, G. n., Bernstein, J. A.
  2018; 176 (4): 1030–36

- **A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network.** *The Journal of pediatrics*
  Reuter, C. M., Brimble, E. n., DeFilippo, C. n., Dries, A. M., Enns, G. M., Ashley, E. A., Bernstein, J. A., Fisher, P. G., Wheeler, M. T.
  2018

- **Genotype-phenotype correlations in individuals with pathogenic RERE variants.** *Human mutation*
  Jordan, V. K., Fregeau, B. n., Ge, X. n., Giordano, J. n., Wapner, R. J., Balci, T. B., Carter, M. T., Bernat, J. A., Moccia, A. N., Srivastava, A. n., Martin, D. M., Bielas, S. L., Pappas, et al
  2018

- **Isolated Congenital Anosmia and CNGA2 Mutation.** *Scientific reports*
  Sailani, M. R., Jingga, I., MirMazlomi, S. H., Bitarafan, F., Bernstein, J. A., Snyder, M. P., Garshasbi, M.
  2017; 7 (1): 2667-?

- **Assembly of functionally integrated human forebrain spheroids** *NATURE*
  Birey, F., Andersen, J., Makinson, C. D., Islam, S., Wei, W., Huber, N., Fan, H. C., Metzler, K. R., Panagiotakos, G., Thom, N., O'Rourke, N. A., Steinmetz, L. M., Bernstein, et al
  2017; 545 (7652): 54-?

- **Characterizing regression in Phelan McDermid Syndrome (22q13 deletion syndrome).** *Journal of psychiatric research*
  Reierson, G., Bernstein, J., Froehlich-Santino, W., Urban, A., Purmann, C., Berquist, S., Jordan, J., O'Hara, R., Hallmayer, J.
  2017; 91: 139-144

- **Association of AHSG with alopecia and mental retardation (APMR) syndrome.** *Human genetics*
  Reza Sailani, M., Jahanbani, F., Nasiri, J., Behnam, M., Salehi, M., Sedghi, M., Hoseinzadeh, M., Takahashi, S., Zia, A., Gruber, J., Lynch, J. L., Lam, D., Winkelmann, et al
  2017; 136 (3): 287-296

- **The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Ramoni, R. B., Mulvihill, J. J., Adams, D. R., Allard, P., Ashley, E. A., Bernstein, J. A., Gahl, W. A., Hamid, R., Loscalzo, J., McCray, A. T., Shashi, V., Tifft, C. J., Wise, et al
  2017; 100 (2): 185-192

- **Sleep Disturbances in Individuals With Phelan-McDermid Syndrome: Correlation With Caregivers' Sleep Quality and Daytime Functioning** *SLEEP*
  Bro, D., O'Hara, R., Primeau, M., Hanson-Kahn, A., Hallmayer, J., Bernstein, J. A.
  2017; 40 (2)

- **De novo and rare mutations in the HSPA1L heat shock gene associated with inflammatory bowel disease** *GENOME MEDICINE*
  Takahashi, S., Andreoletti, G., Chen, R., Munehira, Y., Batra, A., Afzal, N. A., Beattie, R. M., Bernstein, J. A., Ennis, S., Snyder, M.
  2017; 9

- **Identification of a novel mutation in APTX gene associated with Ataxia-oculomotor apraxia.** *Cold Spring Harbor molecular case studies*
  Inlora, J. n., Sailani, M. R., Khodadadi, H. n., Teymurinezhad, A. n., Takahashi, S. n., Bernstein, J. A., Garshasbi, M. n., Snyder, M. P.
  2017

- **Clinical and molecular characterization of de novo loss of function variants in HNRNPU.** *American journal of medical genetics. Part A*
  Leduc, M. S., Chao, H. T., Qu, C. n., Walkiewicz, M. n., Xiao, R. n., Magoulas, P. n., Pan, S. n., Beuten, J. n., He, W. n., Bernstein, J. A., Schaaf, C. P., Scaglia, F. n., Eng, et al
  2017

- **New insights into mitral valve dystrophy: a Filamin-A genotype-phenotype and outcome study.** *European heart journal*
  Le Tourneau, T. n., Le Scouarnec, S. n., Cueff, C. n., Bernstein, D. n., Aalberts, J. J., Lecointe, S. n., Mérot, J. n., Bernstein, J. A., Oomen, T. n., Dina, C. n., Karakachoff, M. n., Desal, H. n., Al Habash, et al
  2017

- **Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly.** *Brain : a journal of neurology*
  Alcantara, D. n., Timms, A. E., Gripp, K. n., Baker, L. n., Park, K. n., Collins, S. n., Cheng, C. n., Stewart, F. n., Mehta, S. G., Saggar, A. n., Sztriha, L. n., Zombor, M. n., Caluseriu, et al
  2017; 140 (10): 2610–22

- **Functional analysis of novel DEAF1 variants identified through clinical exome sequencing expands DEAF1-associated neurodevelopmental disorder (DAND) phenotype.** *Human mutation*
  Chen, L. n., Jensik, P. J., Alaimo, J. T., Walkiewicz, M. n., Berger, S. n., Roeder, E. n., Faqeih, E. A., Bernstein, J. A., Smith, A. C., Mullegama, S. V., Saffen, D. W., Elsea, S. H.
  2017

- **Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features.** *American journal of human genetics*
  Stankiewicz, P. n., Khan, T. N., Szafranski, P. n., Slattery, L. n., Streff, H. n., Vetrini, F. n., Bernstein, J. A., Brown, C. W., Rosenfeld, J. A., Rednam, S. n., Scollon, S. n., Bergstrom, K. L., Parsons, et al
  2017

- **De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability.** *American journal of human genetics*
  Küry, S. n., van Woerden, G. M., Besnard, T. n., Proietti Onori, M. n., Latypova, X. n., Towne, M. C., Cho, M. T., Prescott, T. E., Ploeg, M. A., Sanders, S. n., Stessman, H. A., Pujol, A. n., Distel, et al
  2017; 101 (5): 768–88

- **Teaching Biochemistry and Genetics to Students of Dentistry, Medicine, and Pharmacy 6th International Conference of the Association of Biochemistry Educators (ABE) Clearwater Beach, FL, USA, May 7-11, 2017.** *Medical science educator*
  Niederhoffer, E. C., Cline, S. D., Osheroff, N. n., Simmons, J. M., Diekman, A. B., Franklin, D. S., Abali, E. E., Bateman, R. C., Fontes, J. D., Lindsley, J. E., Pearson, D. n., Rubenstein, P. A., Slaughter, et al
  2017; 27 (4): 855–59

- **WISP3 mutation associated with Pseudorheumatoid Dysplasia.** *Cold Spring Harbor molecular case studies*
  Sailani, M. R., Chappell, J. n., Inlora, J. n., Lynch, L. n., Narasimha, A. n., Mazroui, S. n., Zia, A. n., Bernstein, J. n., Aryani, O. n., Snyder, M. P.
  2017

- **in a patient with a complex connective tissue phenotype.** *Cold Spring Harbor molecular case studies*

Zastrow, D. B., Zornio, P. A., Dries, A., Kohler, J., Fernandez, L., Waggott, D., Walkiewicz, M., Eng, C. M., Manning, M. A., Farrelly, E., Fisher, P. G., Ashley, E. A., Bernstein, et al

2017; 3 (1)

- **The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies.** *Nature genetics*

  Redin, C., Brand, H., Collins, R. L., Kammin, T., Mitchell, E., Hodge, J. C., Hanscom, C., Pillalamarri, V., Seabra, C. M., Abbott, M., Abdul-Rahman, O. A., Aberg, E., Adley, et al

  2017; 49 (1): 36-45

- **Tumor-induced Osteomalacia in a 3-Year-Old With Unresectable Central Giant Cell Lesions.** *Journal of pediatric hematology/oncology*

  Crossen, S. S., Zambrano, E., Newman, B., Bernstein, J. A., Messner, A. H., Bachrach, L. K., Twist, C. J.

  2016: -?

- **M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity.** *Nature genetics*

  Jagadeesh, K. A., Wenger, A. M., Berger, M. J., Guturu, H., Stenson, P. D., Cooper, D. N., Bernstein, J. A., Bejerano, G.

  2016

- **Chitayat syndrome: hyperphalangism, characteristic facies, hallux valgus and bronchomalacia results from a recurrent c.266A>G p.(Tyr89Cys) variant in the ERF gene.** *Journal of medical genetics*

  Balasubramanian, M., Lord, H., Levesque, S., Guturu, H., Thuriot, F., Sillon, G., Wenger, A. M., Sureka, D. L., Lester, T., Johnson, D. S., Bowen, J., Calhoun, A. R., Viskochil, et al

  2016

- **De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms** *AMERICAN JOURNAL OF HUMAN GENETICS*

  Weiss, K., Terhal, P. A., Cohen, L., Bruccoleri, M., Irving, M., Martinez, A. F., Rosenfeld, J. A., Machol, K., Yang, Y., Liu, P., Walkiewicz, M., Beuten, J., Gomez-Ospina, et al

  2016; 99 (4): 934-941

- **Impaired Health-Related Quality of Life in Children and Families Affected by Methylmalonic Acidemia.** *Journal of genetic counseling*

  Splinter, K., Niemi, A., Cox, R., Platt, J., Shah, M., Enns, G. M., Kasahara, M., Bernstein, J. A.

  2016; 25 (5): 936-944

- **Clinical and radiographic delineation of Bent Bone Dysplasia-FGFR2 type or Bent Bone Dysplasia with Distinctive Clavicles and Angel-shaped Phalanges.** *American journal of medical genetics. Part A*

  Krakow, D., Cohn, D. H., Wilcox, W. R., Noh, G. J., Raffel, L. J., Sarukhanov, A., Ivanova, M. H., Danielpour, M., Grange, D. K., Elliott, A. M., Bernstein, J. A., Rimoin, D. L., Merrill, et al

  2016; 170 (10): 2652-2661

- **Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers.** *Genetics in medicine*

  Wenger, A. M., Guturu, H., Bernstein, J. A., Bejerano, G.

  2016

- **Respiratory System Involvement in Costello Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*

  Gomez-Ospina, N., Kuo, C., Ananth, A. L., Myers, A., Brennan, M., Stevenson, D. A., Bernstein, J. A., Hudgins, L.

  2016; 170 (7): 1849-1857

- **Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism** *JAMA NEUROLOGY*

  Mirzaa, G. M., Campbell, C. D., Solovieff, N., Goold, C. P., Jansen, L. A., Menon, S., Timms, A. E., Conti, V., Biag, J. D., Olds, C., Boyle, E. A., Collins, S., Ishak, et al

  2016; 73 (7): 836-845

- **RASA1 somatic mutation and variable expressivity in capillary malformation/arteriovenous malformation (CM/AVM) syndrome.** *American journal of medical genetics. Part A*

  Macmurdo, C. F., Wooderchak-Donahue, W., Bayrak-Toydemir, P., Le, J., Wallenstein, M. B., Milla, C., Teng, J. M., Bernstein, J. A., Stevenson, D. A.

  2016; 170 (6): 1450-1454

- **Prenatally Diagnosed Cases of Binder Phenotype Complicated by Respiratory Distress in the Immediate Postnatal Period.** *Journal of ultrasound in medicine*

  Blumenfeld, Y. J., Davis, A. S., Hintz, S. R., Milan, K., Messner, A. H., Barth, R. A., Hudgins, L., Chueh, J., Homeyer, M., Bernstein, J. A., Enns, G., Atwal, P., Manning, et al

2016; 35 (6): 1353-1358

- **Clinical Course of Six Children With GNAO1 Mutations Causing a Severe and Distinctive Movement Disorder** *PEDIATRIC NEUROLOGY*
  Ananth, A. L., Robichaux-Viehoever, A., Kim, Y., Hanson-Kahn, A., Cox, R., Enns, G. M., Strober, J., Willing, M., Schlaggar, B. L., Wu, Y. W., Bernstein, J. A.
  2016; 59: 81-84

- **Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Petrovski, S., Kury, S., Myers, C. T., Anyane-Yeboa, K., Cogne, B., Bialer, M., Xia, F., Hemati, P., Riviello, J., Mehaffey, M., Besnard, T., Becraft, E., Wadley, et al
  2016; 98 (5): 1001-1010

- **Clinical, cytogenetic, and molecular outcomes in a series of 66 patients with Pierre Robin sequence and literature review: 22q11.2 deletion is less common than other chromosomal anomalies.** *American journal of medical genetics. Part A*
  Gomez-Ospina, N., Bernstein, J. A.
  2016; 170 (4): 870-880

- **Novel X-linked syndrome of cardiac valvulopathy, keloid scarring, and reduced joint mobility due to filamin A substitution G1576R.** *American journal of medical genetics. Part A*
  Atwal, P. S., Blease, S., Braxton, A., Graves, J., He, W., Person, R., Slattery, L., Bernstein, J. A., Hudgins, L.
  2016; 170 (4): 891-895

- **Clinical Delineation of the PACS1-Related Syndrome-Report on 19 Patients** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Schuurs-Hoeijmakers, J. H., Landsverk, M. L., Foulds, N., Kukolich, M. K., Gavrilova, R. H., Greville-Heygate, S., Hanson-Kahn, A., Bernstein, J. A., Glass, J., Chitayat, D., Burrow, T. A., Husami, A., Collins, et al
  2016; 170 (3): 670-675

- **A deleterious Nav1.1 mutation selectively impairs telencephalic inhibitory neurons derived from Dravet Syndrome patients.** *eLife*
  Sun, Y., Pasca, S. P., Portmann, T., Goold, C., Worringer, K. A., Guan, W., Chan, K. C., Gai, H., Vogt, D., Chen, Y. J., Mao, R., Chan, K., Rubenstein, et al
  2016; 5

- **Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases** *JOURNAL OF ALLERGY AND CLINICAL IMMUNOLOGY*
  Holzinger, D., Fassl, S. K., de Jager, W., Lohse, P., Roehrig, U. F., Gattorno, M., Omenetti, A., Chiesa, S., Schena, F., Austermann, J., Vogl, T., Kuhns, D. B., Holland, et al
  2015; 136 (5): 1337-1345

- **Prenatal hydrops foetalis associated with infantile free sialic acid storage disease.** *Journal of obstetrics and gynaecology*
  Chock, V. Y., MILAN, K. E., Folkins, A. K., Hazard, F. K., Bernstein, J. A., Hintz, S. R.
  2015; 35 (8): 850-852

- **Clinical reappraisal of SHORT syndrome with PIK3R1 mutations: towards recommendation for molecular testing and management.** *Clinical genetics*
  Avila, M., Dyment, D. A., Sagen, J. V., St-Onge, J., Moog, U., Chung, B. H., Mansour, S., Albanese, A., Garcia, S., Ortiz Martin, D., Lopez, A. A., Claudi, T., König, et al
  2015

- **Factors Associated with Uptake of Genetics Services for Hypertrophic Cardiomyopathy.** *Journal of genetic counseling*
  Khouzam, A., Kwan, A., Baxter, S., Bernstein, J. A.
  2015; 24 (5): 797-809

- **DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Ji, J., Lee, H., Argiropoulos, B., Dorrani, N., Mann, J., Martinez-Agosto, J. A., Gomez-Ospina, N., Gallant, N., Bernstein, J. A., Hudgins, L., Slattery, L., Isidor, B., Le Caignec, et al
  2015; 23 (11): 1473-1481

- **46,XY disorders of sex development and congenital diaphragmatic hernia: A case with dysmorphic facies, truncus arteriosus, bifid thymus, gut malrotation, rhizomelia, and adactyly** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Esplin, E. D., Chaib, H., Haney, M., Martin, B., Homeyer, M., Urban, A. E., Bernstein, J. A.
  2015; 167A (6): 1360-1364

- **Increased body mass in infancy and early toddlerhood in Angelman syndrome patients with uniparental disomy and imprinting center defects.** *American journal of medical genetics. Part A*
  Brennan, M., Adam, M. P., Seaver, L. H., Myers, A., Schelley, S., Zadeh, N., Hudgins, L., Bernstein, J. A.
  2015; 167A (1): 142-146
- **Cold-aggravated pain in humans caused by a hyperactive NaV1.9 channel mutant.** *Nature communications*
  Leipold, E., Hanson-Kahn, A., Frick, M., Gong, P., Bernstein, J. A., Voigt, M., Katona, I., Oliver Goral, R., Altmüller, J., Nürnberg, P., Weis, J., Hübner, C. A., Heinemann, et al
  2015; 6: 10049-?
- **Perinatal features of the RASopathies: Noonan syndrome, cardiofaciocutaneous syndrome and Costello syndrome.** *American journal of medical genetics. Part A*
  Myers, A., Bernstein, J. A., Brennan, M., Curry, C., Esplin, E. D., Fisher, J., Homeyer, M., Manning, M. A., Muller, E. A., Niemi, A., Seaver, L. H., Hintz, S. R., Hudgins, et al
  2014; 164A (11): 2814-2821
- **Inappropriate p53 activation during development induces features of CHARGE syndrome.** *Nature*
  Van Nostrand, J. L., Brady, C. A., Jung, H., Fuentes, D. R., Kozak, M. M., Johnson, T. M., Lin, C., Lin, C., Swiderski, D. L., Vogel, H., Bernstein, J. A., Attié-Bitach, T., Chang, et al
  2014; 514 (7521): 228-232
- **Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway.** *Genetics in medicine*
  Enns, G. M., Shashi, V., Bainbridge, M., Gambello, M. J., Zahir, F. R., Bast, T., Crimian, R., Schoch, K., Platt, J., Cox, R., Bernstein, J. A., Scavina, M., Walter, et al
  2014; 16 (10): 751-758
- **Clinical whole-exome sequencing: are we there yet?** *Genetics in medicine*
  Atwal, P. S., Brennan, M., Cox, R., Niaki, M., Platt, J., Homeyer, M., Kwan, A., Parkin, S., Schelley, S., Slattery, L., Wilnai, Y., Bernstein, J. A., Enns, et al
  2014; 16 (9): 717-719
- **A recurrent fibrillin-1 mutation in severe early onset Marfan syndrome.** *Journal of pediatric genetics*
  Sureka, D., Stheneur, C., Odent, S., Arno, G., Murphy, D., Bernstein, J. A.
  2014; 3 (3): 157-162
- **Clinical interpretation and implications of whole-genome sequencing.** *JAMA*
  Dewey, F. E., Grove, M. E., Pan, C., Goldstein, B. A., Bernstein, J. A., Chaib, H., Merker, J. D., Goldfeder, R. L., Enns, G. M., David, S. P., Pakdaman, N., Ormond, K. E., Caleshu, et al
  2014; 311 (10): 1035-1045
- **SHANK3 and IGF1 restore synaptic deficits in neurons from 22q13 deletion syndrome patients.** *Nature*
  Shcheglovitov, A., Shcheglovitova, O., Yazawa, M., Portmann, T., Shu, R., Sebastiano, V., Krawisz, A., Froehlich, W., Bernstein, J. A., Hallmayer, J. F., Dolmetsch, R. E.
  2013; 503 (7475): 267-271
- **ß-Galactosidosis in Patient with Intermediate GM1 and MBD Phenotype.** *JIMD reports*
  Moore, T., Bernstein, J. A., Casson-Parkin, S., Cowan, T. M.
  2013; 7: 77-79
- **Underutilization of Genetics Services for Autism: The Importance of Parental Awareness and Provider Recommendation** *JOURNAL OF GENETIC COUNSELING*
  Vande Wydeven, K., Kwan, A., Hardan, A. Y., Bernstein, J. A.
  2012; 21 (6): 803-813
- **ALX4 gain-of-function mutations in nonsyndromic craniosynostosis** *HUMAN MUTATION*
  Yagnik, G., Ghuman, A., Kim, S., Stevens, C. G., Kimonis, V., Stoler, J., Sanchez-Lara, P. A., Bernstein, J. A., Naydenov, C., Drissi, H., Cunningham, M. L., Kim, J., Boyadjiev, et al
  2012; 33 (12): 1626-1629
- **Spectrum of Mutations in the Renin-Angiotensin System Genes in Autosomal Recessive Renal Tubular Dysgenesis** *HUMAN MUTATION*
  Gribouval, O., Moriniere, V., Pawtowski, A., Arrondel, C., Sallinen, S., Saloranta, C., Clericuzio, C., Viot, G., Tantau, J., Blesson, S., Cloarec, S., Machet, M. C., Chitayat, et al

2012; 33 (2): 316-326

- **Analysis of the Alternative Splicing of an FGFR2 Transcript Due to a Novel 5 ' Splice Site Mutation (1084+1G > A): Case Report** *CLEFT PALATE-CRANIOFACIAL JOURNAL*
  Traynis, I., Bernstein, J. A., Gardner, P., Schrijver, I.
  2012; 49 (1): 104-108

- **Rapid Implementation of Inpatient Electronic Physician Documentation at an Academic Hospital** *APPLIED CLINICAL INFORMATICS*
  Hahn, J. S., Bernstein, J. A., MCKENZIE, R. B., King, B. J., Longhurst, C. A.
  2012; 3 (2): 175-185

- **Using iPSC-derived neurons to uncover cellular phenotypes associated with Timothy syndrome** *NATURE MEDICINE*
  Pasca, S. P., Portmann, T., Voineagu, I., Yazawa, M., Shcheglovitov, A., Pasca, A. M., Cord, B., Palmer, T. D., Chikahisa, S., Nishino, S., Bernstein, J. A., Hallmayer, J., Geschwind, et al
  2011; 17 (12): 1657-U176

- **Newborn with prenatally diagnosed choroidal fissure cyst and panhypopituitarism and review of the literature.** *AJP reports*
  Chitkara, R., Rajani, A., Bernstein, J., Shah, S., Hahn, J. S., Barnes, P., Hintz, S. R.
  2011; 1 (2): 111-114

- **Ectopia Lentis as the Presenting and Primary Feature in Marfan Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Zadeh, N., Bernstein, J. A., Niemi, A. K., Dugan, S., Kwan, A., Liang, D., Hyland, J. C., Hoyme, H. E., Hudgins, L., Manning, M. A.
  2011; 155A (11): 2661-2668

- **Horseshoe Kidney and a Rare TSC2 Variant in Two Unrelated Individuals With Tuberous Sclerosis Complex** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Niemi, A., Northrup, H., Hudgins, L., Bernstein, J. A.
  2011; 155A (10): 2534-2537

- **Familial Cardiac Valvulopathy Due to Filamin A Mutation** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Bernstein, J. A., Bernstein, D., Hehr, U., Hudgins, L.
  2011; 155A (9): 2236-2241

- **Using induced pluripotent stem cells to investigate cardiac phenotypes in Timothy syndrome** *NATURE*
  Yazawa, M., Hsueh, B., Jia, X., Pasca, A. M., Bernstein, J. A., Hallmayer, J., Dolmetsch, R. E.
  2011; 471 (7337): 230-U120

- **Clues to an Early Diagnosis of Kallmann Syndrome** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*
  Kaplan, J. D., Bernstein, J. A., Kwan, A., Hudgins, L.
  2010; 152A (11): 2796-2801

- **Two-Tier Approach to the Newborn Screening of Methylenetetrahydrofolate Reductase Deficiency and Other Remethylation Disorders with Tandem Mass Spectrometry** *JOURNAL OF PEDIATRICS*
  Tortorelli, S., Turgeon, C. T., Lim, J. S., Baumgart, S., Day-Salvatore, D., Abdenur, J., Bernstein, J. A., Lorey, F., Lichter-Konecki, U., Oglesbee, D., Raymond, K., Matem, D., Schimmenti, et al
  2010; 157 (2): 271-275

- **Index of suspicion.** *Pediatrics in review*
  Zadeh, N., Bernstein, J. A., Stiasny, D., Callaghan, M. U., Flores, C. E., Tytko, J. M., Mannarino, F. P., Moore, J.
  2010; 31 (4): 167-172

- **Improved physician work flow after integrating sign-out notes into the electronic medical record.** *Joint Commission journal on quality and patient safety / Joint Commission Resources*
  Bernstein, J. A., Imler, D. L., Sharek, P., Longhurst, C. A.
  2010; 36 (2): 72-78

- **Clinical and Molecular Heterogeneity in Patients with the CblD Inborn Error of Cobalamin Metabolism** *JOURNAL OF PEDIATRICS*
  Miousse, I. R., Watkins, D., Coelho, D., Rupar, T., Crombez, E. A., Vilain, E., Bernstein, J. A., Cowan, T., Lee-Messer, C., Enns, G. M., Fowler, B., Rosenblatt, D. S.
  2009; 154 (4): 551-556

- **GENOMIC ANALYSIS OF MRNA DECAY IN E. COLI WITH DNA MICROARRAYS** *RNA TURNOVER IN BACTERIA, ARCHAEA AND ORGANELLES*
  Lin, P., Singh, D., Bernstein, J. A., Lin-Chao, S.
  2008; 447: 47-?

- **Global analysis of Escherichia coli RNA degradosome function using DNA microarrays** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Bernstein, J. A., Lin, P. H., Cohen, S. N., Lin-Chao, S.
  2004; 101 (9): 2758-2763

- **Life after transcription - revisiting the fate of messenger RNA** *TRENDS IN GENETICS*
  Khodursky, A. B., Bernstein, J. A.
  2003; 19 (3): 113-115

- **Escherichia coli spotted double-strand DNA microarrays: RNA extraction, labeling, hybridization, quality control, and data management.** *Methods in molecular biology (Clifton, N.J.)*
  Khodursky, A. B., Bernstein, J. A., Peter, B. J., Rhodius, V., Wendisch, V. F., Zimmer, D. P.
  2003; 224: 61-78

- **Global analysis of mRNA decay and abundance in Escherichia coli at single-gene resolution using two-color fluorescent DNA microarrays** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*
  Bernstein, J. A., Khodursky, A. B., Lin, P. H., Lin-Chao, S., Cohen, S. N.
  2002; 99 (15): 9697-9702

- **RNase G complementation of me null mutation identifies functional interrelationships with RNase E in Escherichia coli** *MOLECULAR MICROBIOLOGY*
  Lee, K., Bernstein, J. A., Cohen, S. N.
  2002; 43 (6): 1445-1456

- **Use of traditional medicine in Mongolia: a survey** *COMPLEMENTARY THERAPIES IN MEDICINE*
  Bernstein, J. A., Stibich, M. A., LeBaron, S.
  2002; 10 (1): 42-45