Stanford

Jon Bernstein

Associate Professor of Pediatrics (Genetics) at the Lucile Salter Packard Children's Hospital and, by courtesy, of Genetics

Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Pediatric Genetics**

  730 Welch Rd 2A

  Palo Alto, CA 94304

  Tel (650) 721-5804 **Fax** (650) 498-4555

ACADEMIC CONTACT INFORMATION

- **Administrative Contact - Academic**

  Stephanie Martinez

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Bio

CLINICAL FOCUS

- Clinical Genetics
- Autism
- Developmental Disorders
- Craniofacial Conditions
- Craniosynostosis
- Cleft Lip
- Cleft Palate
- Cardiovascular Genetics

ACADEMIC APPOINTMENTS

- Associate Professor - Med Center Line, Pediatrics - Medical Genetics
- Associate Professor - Med Center Line (By courtesy), Genetics
- Member, Bio-X
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

- Associate Director, Medical Genetics Residency Program, (2016- present)
- Director, Stanford Medical Genetics Residency Program, (2013-2016)

PROFESSIONAL EDUCATION

- Medical Education: Stanford University School of Medicine Registrar (2003) CA
- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2009)
- 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 interests include the genetics of autism and other developmental disorders. In collaboration with colleagues at Stanford, I am working to develop induced pluripotent stem cell (iPSC) models of genetic disorders associated with developmental disability. I am also engaged in the application of new technologies (Whole genome sequencing, Multi-omics profiling) for the diagnosis of developmental disorders.

CLINICAL TRIALS

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

Teaching

COURSES

2018-19

- Human Genetics: GENE 202 (Aut)

2017-18

- Human Genetics: GENE 202 (Aut)

2016-17

- Human Genetics: GENE 202 (Aut)

2015-16

- Human Genetics: GENE 202 (Aut)

Publications

PUBLICATIONS

- **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

- **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., Vargas, M., 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., Reuter, C. M., Fernandez, L., Grove, M. E., Fisk, D. G., Yang, Y., Eng, C. M., Ward, P. A., Bick, D., 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., Marwaha, S., Postolova, A., Kittle, J., Vasquez, R., Davidson, J., Kohler, J., Dries, A., Fernandez-Betancourt, L., Majcherska, M., Dearlove, J., Raghavan, S., Vogel, et al
  2019

- **Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts.** *Nature medicine*
  Frésard, L., Smail, C., Ferraro, N. M., Teran, N. A., Li, X., Smith, K. S., Bonner, D., Kernohan, K. D., Marwaha, S., Zappala, Z., Balliu, B., 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., Fisk, D. G., Rego, S., Dagan-Rosenfeld, O., Kohler, J. N., Reuter, C. M., Bonner, D., 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., Blumenfeld, Y. J., Cusmano, K., Hintz, S. R., Alcorn, D., Benitz, W. E., Berquist, W. E., Bernstein, J. A., Castillo, R. O., Concepcion, W., 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., Vandervore, L., van Slegtenhorst, M. A., Wilke, M., Kasteleijn, E., Lee, et al
  2018

- **Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations.** *Neuron*
  Tripathy, R., Leca, I., van Dijk, T., Weiss, J., van Bon, B. W., Sergaki, M. C., Gstrein, T., Breuss, M., Tian, G., Bahi-Buisson, 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., 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., Adams, D. R., Bacino, C. A., Bellen, H. J., Bernstein, J. A., Cheatle-Jarvela, A. M., Eng, C. M., Esteves, C., Gahl, W. A., Hamid, R., Jacob, H. J., Kikani, B., Koeller, et al

  2018

- **Volumetric Analysis of the Basal Ganglia and Cerebellar Structures in Patients with Phelan-McDermid Syndrome.** *Pediatric neurology*

  Srivastava, S., Scherrer, B., Prohl, A. K., Filip-Dhima, R., Kapur, K., Kolevzon, A., Buxbaum, J. D., Berry-Kravis, E., Soorya, L., Thurm, A., 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., Bodle, E. E., Kohler, J. N., Matalon, D. R., Nazarenko, Y., Genetti, C. A., Brownstein, C. A., Schmitz-Abe, K., Schoch, K., Cope, H., Signer, R., 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., Macnamara, E., Rosenfeld, J. A., Hanchard, N., Lewis, A. M., Brown, C. W., Marom, R., Shao, Y., Novacic, D., Wolfe, L., 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., Bruce, J. S., Bereknyei Merrell, S., 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., Esplin, E. D., Jagadeesh, K. A., Guturu, H., Wenger, A. M., Chaib, H., Buckingham, J. A., Bejerano, G., 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., DeFilippo, C., 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., Ge, X., Giordano, J., Wapner, R. J., Balci, T. B., Carter, M. T., Bernat, J. A., Moccia, A. N., Srivastava, A., 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., Sailani, M. R., Khodadadi, H., Teymurinezhad, A., Takahashi, S., Bernstein, J. A., Garshasbi, M., 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., Walkiewicz, M., Xiao, R., Magoulas, P., Pan, S., Beuten, J., He, W., Bernstein, J. A., Schaaf, C. P., Scaglia, F., Eng, et al
  2017

- **New insights into mitral valve dystrophy: a Filamin-A genotype-phenotype and outcome study.** *European heart journal*
  Le Tourneau, T., Le Scouarnec, S., Cueff, C., Bernstein, D., Aalberts, J. J., Lecointe, S., Mérot, J., Bernstein, J. A., Oomen, T., Dina, C., Karakachoff, M., Desal, H., 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., Timms, A. E., Gripp, K., Baker, L., Park, K., Collins, S., Cheng, C., Stewart, F., Mehta, S. G., Saggar, A., Sztriha, L., Zombor, M., 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., Jensik, P. J., Alaimo, J. T., Walkiewicz, M., Berger, S., Roeder, E., 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., Khan, T. N., Szafranski, P., Slattery, L., Streff, H., Vetrini, F., Bernstein, J. A., Brown, C. W., Rosenfeld, J. A., Rednam, S., Scollon, S., 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., van Woerden, G. M., Besnard, T., Proietti Onori, M., Latypova, X., Towne, M. C., Cho, M. T., Prescott, T. E., Ploeg, M. A., Sanders, S., Stessman, H. A., Pujol, A., 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., Simmons, J. M., Diekman, A. B., Franklin, D. S., Abali, E. E., Bateman, R. C., Fontes, J. D., Lindsley, J. E., Pearson, D., 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., Inlora, J., Lynch, L., Narasimha, A., Mazroui, S., Zia, A., Bernstein, J., Aryani, O., 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

- **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.
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