Stanford



Gill Bejerano

Professor of Developmental Biology, of Computer Science and of Pediatrics (Genetics)

NIH Biosketch available Online

CONTACT INFORMATION

- **Alternate Contact**

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Bio

BIO

Gill Bejerano holds a B.Sc. In Mathematics, Physics and Computer Science, and a Ph.D in Computer Science (Machine Learning applications in Biology) from Hebrew University of Jerusalem. Gill got into genomics in 2003, started a wet lab in 2007, began analyzing patient genomes and medical records in 2014, got into cryptogenomics in 2017 and has become very interested in healthcare economics and patient risk management in 2021. He is recognized by multiple academic awards including two best paper and tomorrow's PI awards, Mallinckrodt, Sloan, Human Frontiers, Searle, Okawa, David and Lucile Packard, Microsoft and Sony Scholar awards. Gill has trained, collaborated and advised computational scientists, experimentalists, clinicians, and MBAs and has helped both start-ups and Fortune 500 companies.

ACADEMIC APPOINTMENTS

- Professor, Developmental Biology
- Professor, Computer Science
- Professor, Pediatrics - Medical Genetics
- Member, Bio-X
- Member, Cardiovascular Institute
- Faculty Affiliate, Institute for Human-Centered Artificial Intelligence (HAI)
- Member, Maternal & Child Health Research Institute (MCHRI)
- Member, Stanford Cancer Institute
- Member, Wu Tsai Neurosciences Institute

ADMINISTRATIVE APPOINTMENTS

- Member, Editorial Board, Gene, (2007-2008)
- Technical Advisory Board, Numenta, (2008- present)

HONORS AND AWARDS

- Rector Prize & Dean’s list for undergraduate achievements., Hebrew University (1993-1996)

- Intel award for achievements., Hebrew University (1996)
- Rector Prize & Dean’s list for graduate studies achievements., Hebrew University (1997-1999)
- Rachel & Salim Banin scholarship., Hebrew University (1999)
- Best paper by a young scientist award., RECOMB conference (1999)
- Levi Eshkol graduate studies fellowship., Hebrew University (1999-2002)
- Best paper by a young scientist award., RECOMB conference (2003)
- Junior Faculty Grant, Edward Mallinckrodt, Jr. Foundation (2007-2010)
- Tomorrow's Principal Investigator, Genome Technology Magazine (2008)
- Alfred P. Sloan Fellow, Alfred P. Sloan Foundation (2008-2010)
- Young Investigator Award, Human Frontier Science Program (2008-2011)
- Searle Scholar, Searle Scholars Program (2008-2011)
- Research Grant Award, Okawa Foundation (2008)
- Fellow, David & Lucile Packard Foundation (2008-2013)
- New Faculty Fellow, Microsoft Research (2009)

PROFESSIONAL EDUCATION

- Ph.D., Hebrew University , Computer Science (2004)
- B.Sc., Hebrew University , Physics, Mathematics, Computer Science (summa cum laude) (1997)

LINKS

- Bejerano Lab: http://bejerano.stanford.edu

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

The Bejerano lab interests evolve continuously. As of 2021 they span data science, genomic variants of large effect, cryptogenomics, machine learning with electronic health records and healthcare economics.

We have done seminal work and continue to play an active role in:

1. Automating monogenic patient diagnosis and reanalysis.
2. The genomic signatures of independent divergent and convergent trait evolution in mammals.
3. The logic of human gene regulation.
4. The reasons for sequence ultraconservation.
5. Cryptogenomics to bridge medical silos.
6. Cryptogenetics to debate social injustice.

We are also getting quite interested in:

7. Managing patient risk using machine learning.
8. Understanding the incentive structure of the US healthcare system.

Teaching

COURSES

2026-27

- Foundations of Computational Human Genomics: BMDS 173A, CS 173A, DBIO 173A (Aut)

2025-26

- Foundations of Computational Human Genomics: BMDS 173A, CS 173A, DBIO 173A (Aut)

2024-25

- Foundations of Computational Human Genomics: BIOMEDIN 173A, CS 173A, DBIO 173A (Aut)

2023-24

- Foundations of Computational Human Genomics: BIOMEDIN 173A, CS 173A, DBIO 173A (Win)

STANFORD ADVISEES

Master's Program Advisor

Arya Bakhtiar, Zhenghui Chen, Amy Tang, Artemis Xu, Lucy Yin

Undergraduate Major Advisor

Vincent Nguyen

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Data Science (Masters Program)
- Biomedical Data Science (PhD Program)
- Cardiovascular Medicine (Fellowship Program)
- Clinical Informatics (Fellowship Program)
- Developmental Biology (PhD Program)
- Developmental-Behavioral Pediatrics (Fellowship Program)
- Genetics (PhD Program)
- Human Genetics and Genetic Counseling (Masters Program)
- Medical Genetics (Fellowship Program)
- Molecular and Genetic Medicine (Fellowship Program)
- Neonatal-Perinatal Medicine (Fellowship Program)
- Neurosciences (PhD Program)

Publications

PUBLICATIONS

- **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

- **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)

- **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

- **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

- **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

- **Exome and genome sequencing in a heterogeneous population of patients with rare disease: Identifying predictors of a diagnosis.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Pucel, J., Briere, L. C., Reuter, C., Gochyyev, P., LeBlanc, K.
  2024: 101115

- **Recurring homozygous ACTN2 variant (p.Arg506Gly) causes a recessive myopathy.** *Annals of clinical and translational neurology*
  Donkervoort, S., Mohassel, P., O'Leary, M., Bonner, D. E., Hartley, T., Acquaye, N., Brull, A., Mozaffar, T., Saporta, M. A., Dyment, D. A., Sampson, J. B., Pajusalu, S., Austin-Tse, et al
  2024

- **Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care** *JOURNAL OF PEDIATRICS*
  Halley, M. C., Young, J. L., Tang, C., Mintz, K. T., Lucas-Griffin, S., Maghiro, A., Ashley, E. A., Tabor, H. K., Undiagnosed Diseases Network
  2023; 261

- **Whole-genome Comparisons Identify Repeated Regulatory Changes Underlying Convergent Appendage Evolution in Diverse Fish Lineages.** *Molecular biology and evolution*
  Chen, H. I., Turakhia, Y., Bejerano, G., Kingsley, D. M.
  2023; 40 (9)

- **Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care.** *The Journal of pediatrics*
  Halley, M. C., Young, J. L., Tang, C., Mintz, K. T., Lucas-Griffin, S., Maghiro, A. S., Ashley, E. A., Tabor, H. K.
  2023: 113537

- **Participation in a national diagnostic research study: assessing the patient experience.** *Orphanet journal of rare diseases*
  Rosenfeld, L. E., LeBlanc, K., Nagy, A., Ego, B. K., Undiagnosed Diseases Network, McCray, A. T., Acosta, M. T., Adam, M., Adams, D. R., Alvarez, R. L., Alvey, J., Amendola, L., Andrews, A., et al
  2023; 18 (1): 73

- **Analysis of structural variation among inbred mouse strains.** *BMC genomics*
  Arslan, A., Fang, Z., Wang, M., Tan, Y., Cheng, Z., Chen, X., Guan, Y., J Pisani, L., Yoo, B., Bejerano, G., Peltz, G.
  2023; 24 (1): 97

- **Whole-genome comparisons identify repeated regulatory changes underlying convergent appendage evolution in diverse fish lineages.** *bioRxiv : the preprint server for biology*
  Chen, H. I., Turakhia, Y., Bejerano, G., Kingsley, D. M.
  2023

- **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

- **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

- **WhichTF is functionally important in your open chromatin data?** *PLoS computational biology*
  Tanigawa, Y., Dyer, E. S., Bejerano, G.
  2022; 18 (8): e1010378

- **X-CAP improves pathogenicity prediction of stopgain variants.** *Genome medicine*
  Rastogi, R., Stenson, P. D., Cooper, D. N., Bejerano, G.
  2022; 14 (1): 81

- **Champagne: Automated whole-genome phylogenomic character matrix method using large genomic indels for homoplasy-free inference.** *Genome biology and evolution*
  Schull, J. K., Turakhia, Y., Hemker, J. A., Dally, W. J., Bejerano, G.
  2022

- **Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation.** *Journal of genetic counseling*
  Kohler, J. N., Kelley, E. G., Boyd, B. M., Sillari, C. H., Marwaha, S., Undiagnosed Diseases Network, Wheeler, M. T., Acosta, M. T., Adam, M., Adams, D. R., Agrawal, P. B., Alejandro, M. E., Alvey, J., et al
  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

- **Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain** *GENETICS IN MEDICINE*
  Marbach, F., Stoyanov, G., Erger, F., Stratakis, C. A., Settas, N., London, E., Rosenfeld, J. A., Torti, E., Haldeman-Englert, C., Sklirou, E., Kessler, E., Ceulemans, S., Nelson, et al
  2021

- **Avoiding genetic racial profiling in criminal DNA profile databases** *NATURE COMPUTATIONAL SCIENCE*
  Blindenbach, J. A., Jagadeesh, K. A., Bejerano, G., Wu, D. J.
  2021; 1 (4): 272-+

- **Avoiding genetic racial profiling in criminal DNA profile databases.** *Nature computational science*
  Blindenbach, J. A., Jagadeesh, K. A., Bejerano, G., Wu, D. J.
  2021; 1 (4): 272-279

- **Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Kobren, S. N., Baldridge, D., Velinder, M., Krier, J. B., LeBlanc, K., Esteves, C., Pusey, B. N., Zuchner, S., Blue, E., Lee, H., Huang, A., Bastarache, L., Bican, et al
  2021

- **The Effect of Population Structure on Murine Genome-Wide Association Studies.** *Frontiers in genetics*
  Wang, M., Fang, Z., Yoo, B., Bejerano, G., Peltz, G.
  2021; 12: 745361

- **A comparative genomics multitool for scientific discovery and conservation** *NATURE*
  Genereux, D. P., Serres, A., Armstrong, J., Johnson, J., Marinescu, V. D., Muren, E., Juan, D., Bejerano, G., Casewell, N. R., Chemnick, L. G., Damas, J., Di Palma, F., Diekhans, et al
  2020; 587 (7833): 240-+

- **A fully-automated method discovers loss of mouse-lethal and human-monogenic disease genes in 58 mammals.** *Nucleic acids research*
  Turakhia, Y., Chen, H. I., Marcovitz, A., Bejerano, G.
  2020

- **Morphogenesis is transcriptionally coupled to neurogenesis during peripheral olfactory organ development.** *Development (Cambridge, England)*
  Aguillon, R. n., Madelaine, R. n., Aguirrebengoa, M. n., Guturu, H. n., Link, S. n., Dufourcq, P. n., Lecaudey, V. n., Bejerano, G. n., Blader, P. n., Batut, J. n.

2020

- **Morphogenesis is transcriptionally coupled to neurogenesis during peripheral olfactory organ development.** *Development (Cambridge, England)*
  Aguillon, R., Madelaine, R., Aguirrebengoa, M., Guturu, H., Link, S., Dufourcq, P., Lecaudey, V., Bejerano, G., Blader, P., Batut, J.
  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)
- **Transcription factor expression defines subclasses of developing projection neurons highly similar to single-cell RNA-seq subtypes.** *Proceedings of the National Academy of Sciences of the United States of America*
  Heavner, W. E., Ji, S. n., Notwell, J. H., Dyer, E. S., Tseng, A. M., Birgmeier, J. n., Yoo, B. n., Bejerano, G. n., McConnell, S. K.
  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
- **A functional enrichment test for molecular convergent evolution finds a clear protein-coding signal in echolocating bats and whales.** *Proceedings of the National Academy of Sciences of the United States of America*
  Marcovitz, A., Turakhia, Y., Chen, H. I., Gloudemans, M., Braun, B. A., Wang, H., Bejerano, G.
  2019
- **ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis** *GENETICS IN MEDICINE*
  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., Network, et al
  2019; 21 (7): 1585–93
- **CRISPR/Cas9 Genome Engineering in Engraftable Human Brain-Derived Neural Stem Cells.** *iScience*
  Dever, D. P., Scharenberg, S. G., Camarena, J., Kildebeck, E. J., Clark, J. T., Martin, R. M., Bak, R. O., Tang, Y., Dohse, M., Birgmeier, J. A., Jagadeesh, K. A., Bejerano, G., Tsukamoto, et al
  2019; 15: 524–35
- **Darwin: A Genomics Coprocessor** *IEEE MICRO*
  Turakhia, Y., Bejerano, G., Dally, W. J.
  2019; 39 (3): 29–37
- **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; 51 (4): 755-+
- **LUNG DISEASE IN SYSTEMIC JIA: AN EMERGING PROBLEM LINKED WITH YOUNG AGE AND ANTI-IL-1/IL-6**
  Chen, G., Saper, V., Deutsch, G., Guillerman, R. P., Jagadeesh, K., Schulert, G., Canna, S., Lu, Y., Birgmeier, J., Leung, A., Grom, A., Bejerano, G., Davis, et al
  BMJ PUBLISHING GROUP.2019: A57
- **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
- **Components of genetic associations across 2,138 phenotypes in the UK Biobank highlight adipocyte biology.** *Nature communications*
  Tanigawa, Y. n., Li, J. n., Justesen, J. M., Horn, H. n., Aguirre, M. n., DeBoever, C. n., Chang, C. n., Narasimhan, B. n., Lage, K. n., Hastie, T. n., Park, C. Y., Bejerano, G. n., Ingelsson, et al
  2019; 10 (1): 4064

- **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

- **Emergent high fatality lung disease in systemic juvenile arthritis.** *Annals of the rheumatic diseases*
  Saper, V. E., Chen, G. n., Deutsch, G. H., Guillerman, R. P., Birgmeier, J. n., Jagadeesh, K. n., Canna, S. n., Schulert, G. n., Deterding, R. n., Xu, J. n., Leung, A. N., Bouzoubaa, L. n., Abulaban, et al
  2019

- **CLINPHEN EXTRACTS AND PRIORITIZES PHENOTYPES FROM MEDICAL RECORDS TO ACCELERATE GENOMIC DIAGNOSIS**
  Deisseroth, C. A., Birgmeier, J., Bodle, E. E., Kohler, J., Matalon, D., Nazarenko, Y., Genetti, C., Brownstein, C., Schmitz-Abe, K., Schoch, K., Cope, H., Signer, R., Ud, et al
  BMJ PUBLISHING GROUP.2019: 179

- **Darwin-WGA: A Co-processor Provides Increased Sensitivity in Whole Genome Alignments with High Speedup**
  Turakhia, Y., Goenka, S. D., Bejerano, G., Dally, W. J., IEEE
  IEEE.2019: 359–72

- **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

- **A sequence-based, deep learning model accurately predicts RNA splicing branchpoints.** *RNA (New York, N.Y.)*
  Paggi, J. M., Bejerano, G.
  2018

- **Independent erosion of conserved transcription factor binding sites points to shared hindlimb, vision and external testes loss in different mammals.** *Nucleic acids research*
  Berger, M. J., Wenger, A. M., Guturu, H., Bejerano, G.
  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

- **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 LOSS OF FUNCTION WNT5A MUTATIONS IN AN INFANT WITH SEVERE AND ATYPICAL MANIFESTATIONS OF ROBINOW SYNDROME AND UNAFFECTED PARENTS - A NEW LOCUS FOR AUTOSOMAL RECESSIVE DISEASE**
  Bernstein, J. A., Esplin, E. E., Jagadeesh, K. A., Birgmeier, J., Homeyer, M., Guturu, H., Wenger, A. M., Bejerano, G.
  WILEY.2018: 1504

- **A screen for deeply conserved non-coding GWAS SNPs uncovers a MIR-9-2 functional mutation associated to retinal vasculature defects in human** *Nucleic Acids Research*
  Madelaine, R., Notwell, J. H., Skariah, G., Halluin, C., Chen, C. C., Bejerano, G., Mourrain, P.
  2018; 1

- **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

- **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

- **Deriving genomic diagnoses without revealing patient genomes** *SCIENCE*
  Jagadeesh, K. A., Wu, D. J., Birgmeier, J. A., Boneh, D., Bejerano, G.
  2017; 357 (6352): 692-+

- **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
  2017; 54 (3): 157-165

- **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.
  2017; 19 (2): 209-214

- **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

- **MicroRNA-9 Couples Brain Neurogenesis and Angiogenesis.** *Cell reports*
  Madelaine, R. n., Sloan, S. A., Huber, N. n., Notwell, J. H., Leung, L. C., Skariah, G. n., Halluin, C. n., Paşca, S. P., Bejerano, G. n., Krasnow, M. A., Barres, B. A., Mourrain, P. n.
  2017; 20 (7): 1533–42

- **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

- **TBR1 regulates autism risk genes in the developing neocortex.** *Genome research*
  Notwell, J. H., Heavner, W. E., Darbandi, S. F., Katzman, S., McKenna, W. L., Ortiz-Londono, C. F., Tastad, D., Eckler, M. J., Rubenstein, J. L., McConnell, S. K., Chen, B., Bejerano, G.
  2016; 26 (8): 1013-1022

- **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

- **"Reverse Genomics" Predicts Function of Human Conserved Noncoding Elements** *MOLECULAR BIOLOGY AND EVOLUTION*
  Marcovitz, A., Jia, R., Bejerano, G.
  2016; 33 (5): 1358-1369

- **Erosion of Conserved Binding Sites in Personal Genomes Points to Medical Histories.** *PLoS computational biology*
  Guturu, H., Chinchali, S., Clarke, S. L., Bejerano, G.
  2016; 12 (2)

- **Changes in the enhancer landscape during early placental development uncover a trophoblast invasion gene-enhancer network.** *Placenta*
  Tuteja, G., Chung, T., Bejerano, G.
  2016; 37: 45-55

- **Mx1 and Mx2 key antiviral proteins are surprisingly lost in toothed whales** *PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA*

Braun, B. A., Marcovitz, A., Camp, J. G., Jia, R., Bejerano, G.
2015; 112 (26): 8036-8040

- **Characterization of TCF21 Downstream Target Regions Identifies a Transcriptional Network Linking Multiple Independent Coronary Artery Disease Loci** *PLOS GENETICS*
  Sazonova, O., Zhao, Y., Nuernberg, S., Miller, C., Pjanic, M., Castano, V. G., Kim, J. B., Salfati, E. L., Kundaje, A. B., Bejerano, G., Assimes, T., Yang, X., Quertermous, et al
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