The Bejerano Lab studies genome function in human and related species. We are deeply interested in the following broad questions: Mapping genome sequence (variation) to phenotype (differences) and extracting specific genetic insights from deep sequencing measurements. We take a particular interest in gene cis regulation. We use our joint affiliation to apply a combination of computational and experimental approaches. We collect large scale experimental data; write computational analysis tools; run them massively to discover the most exciting testable hypotheses; which we proceed to experimentally validate. We work in small teams, in house or with close collaborators of experimentalists and computational tool users who interact directly with our computational tool builders. Please see our research tab for more.
CURRENT RESEARCH AND SCHOLARLY INTERESTS

The Bejerano Lab is currently focused on the following topics:

1. Genotype - Phenotype relationships in humans.
   We are developing novel methods for linking human whole genome variation with human disease and trait variation. We apply these methods to multiple datasets in the contexts of prematurity, autism, heart disease and more [20, 29, 32, 34, 36, 38, 39, 43].

2. Genotype - Phenotype relationships between mammals.
   We develop novel methods to link trait evolution in the mammalian tree to whole genome evolution in over a hundred species. Application of these methods allow us to shed new light on human genome function, on human disease and on human evolution [29, 34, 35]. See our “Forward Genomics” web server.

3. Extracting genetic knowledge from high throughput genomic assays.
   High throughput genomic assays are most often used to make biochemical discoveries. We develop methods to extract genetic and developmental knowledge from these assays [27, 28, 31]. Through joint work with Sue McConnell we take special interest in the developing neocortex [29, 41]. Also see our popular GREAT web server for the cis-regulatory interpretation of high throughput genomic datasets.

4. Vertebrate transcription regulation.
Much of our work relies on our strong foundations in the study of vertebrate gene regulation [9-11, 14, 15, 18, 22, 25, 27, 29-33, 35, 38-42]. See our PRISM resource of predicted transcription factor functions and COMPLEX resource for predicted transcription factor dimers and complexes. Also see our zCNE resource of conserved non-coding (likely gene regulatory) sequences in the zebrafish genome.

5. Vertebrate genome evolution.

We are extremely well versed in human and vertebrate genome evolution [9-11, 14, 17, 18, 22, 23, 25, 26, 29, 33-35, 37, 39, 40]. Notably, we discovered ultraconservation and correctly postulated that many of these elements are developmental enhancers. We also showed that mammalian ultraconserved elements evolve under extreme purifying selection, and that they are almost never lost during mammalian evolution [9, 23, 25]. We also discovered the first developmental enhancers conserved between human and protostomes [33], attempted to group human conserved non-coding DNA into paralog families [10], and studied the co-option of mobile elements into cis-regulatory roles [18, 22, 26, 41].


We have done work in the field of evolutionary developmental biology [29, 33-35, 43], including a first survey of developmental enhancers (including a penile spine/vibrissae enhancer) uniquely lost in humans [29], fueled by our deep interest in phenotype - genotype relationships.

[For links to the references and more, please see our lab's website]

Teaching

COURSES

2017-18

• The Human Genome Source Code: BIOMEDIN 273A, CS 273A, DBIO 273A (Win)

2016-17

• A Computational Tour of the Human Genome: CS 273A (Aut)

2015-16

• A Computational Tour of the Human Genome: CS 273A (Aut)

2014-15

• A Computational Tour of the Human Genome: CS 273A (Aut)

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Solomon Endlich, Amir Marcovitz

Master's Program Advisor

Julia Daniel, Jonathan Deaton, Taide Ding, Samuel Premutico

Doctoral Dissertation Co-Advisor (AC)

Yosuke Tanigawa

Undergraduate Major Advisor

Sebastian Le Bras

Postdoctoral Research Mentor

Whitney Heavner
GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Biomedical Informatics (Phd Program)
- Biomedical Informatics (Masters 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

- 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*
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- Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. *Genetics in Medicine*
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- M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. *Nature genetics*
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- TBR1 regulates autism risk genes in the developing neocortex. *Genome research*
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- "Reverse Genomics" Predicts Function of Human Conserved Noncoding Elements. *Molecular Biology and Evolution*
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- Erosion of Conserved Binding Sites in Personal Genomes Points to Medical Histories. *PLoS computational biology*
  Guturu, H., Chinchali, S., Clarke, S. L., Bejerano, G.
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• Characterization of TCF21 Downstream Target Regions Identifies a Transcriptional Network Linking Multiple Independent Coronary Artery Disease Loci. *PLOS Genetics*
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• A family of transposable elements co-opted into developmental enhancers in the mouse neocortex. *Nature Communications*
  Notwell, J. H., Chung, T., Heavner, W., Bejerano, G.
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• Microbiota modulate transcription in the intestinal epithelium without remodeling the accessible chromatin landscape. *Genome research*
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• A Penile Spine/Vibrissa Enhancer Sequence Is Missing in Modern and Extinct Humans but Is Retained in Multiple Primates with Penile Spines and Sensory Vibrissae. *PLOS ONE*
  2013; 8 (12)

• Computational methods to detect conserved non-genic elements in phylogenetically isolated genomes: application to zebrafish. *Nucleic acids research*
  Hiller, M., Agarwal, S., Notwell, J. H., Parikh, R., Guturu, H., Wenger, A. M., Bejerano, G.
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• The Enhancer Landscape during Early Neocortical Development Reveals Patterns of Dense Regulation and Co-option. *PLoS genetics*
  Wenger, A. M., Clarke, S. L., Notwell, J. H., Chung, T., Tuteja, G., Guturu, H., Schaar, B. T., Bejerano, G.
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• A penile spine/vibrissa enhancer sequence is missing in modern and extinct humans but is retained in multiple primates with penile spines and sensory vibrissae. *PLoS one*
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Human-specific loss of an androgen receptor enhancer is associated with the loss of vibrissae and penile spines. 

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• Efficient exact p-value computation and applications to biosequence analysis Proceedings of the 7th annual international conference on research in computational molecular biology
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