

## **Postdoctoral positions available Individual Genome Interpretation**



**Research Group of Steven Brenner  
University of California, Berkeley**

We are seeking a postdoctoral researcher to perform analysis of individual genome sequences to diagnose newborns with rare immunological and metabolic disorders.

### **Project Background**

We are performing exome sequencing of DNA from blood spots from every child with a rare metabolic disease born in the state of California in the last 5 years. These children were identified by the California Department of Public Health Newborn Screening Program. The recent decline in cost of whole genome/ exome sequencing has now created a new opportunity to ask whether genome sequencing might be effective as a supplement and possible successor to the current newborn screening technology.

### **Job description**

A primary goal in the project is to evaluate how exomes compare with traditional MS/MS based newborn screening in terms of sensitivity and specificity and whether sequencing is a suitable supplement or successor. The exomes will be analyzed for variants whose frequency and pathogenicity need to be characterized. The exome sequencing data will then need to be combined with the clinical outcomes data in order to identify gene variants that precisely predict an individual newborn's clinical phenotypes.

Deducing the functional consequences of variants is a formidable challenge. Nearly 85% of all human variants are rare and are unevenly distributed among different ethnic groups. Most rare variants have no functional annotation and interpreting them requires a variety of imperfect in silico methods to assess whether a variant is deleterious. This is further complicated by the issue of genetic variant penetrance, where the same mutation could have variable phenotypic effect in different individuals. Tackling these problems will require integrating relevant biological and clinical data from multiple structured and unstructured sources, including but not limited to variant databases, genomic and other multi-omic datasets, biomedical and clinical literature, to create a platform that allows for precise genome interpretation in these metabolic disorders.

The ideal postdoctoral candidate will design methods for individual genome interpretation that tackle the aforementioned challenges as they pertain to rare metabolic conditions in newborns.

### **Position requirements**

Candidate should have Ph.D. (preferably in computational biology, genetics, computer science, molecular biology, biophysics, or a related field) with a strong publication record and compelling professional references. The ideal applicant will have strong biological insight and previous experience with solving biological problems with computers. The successful completion of this project requires programming skills, but may not require novel algorithmic development.

Excellent communication skills are required for effective interaction with the multidisciplinary cohort of researchers in our laboratory and with collaborators.

The Brenner lab is an interdisciplinary research group at the University of California, Berkeley. We are associated with the Center for Computational Biology, the Department of Plant and Microbial Biology, the Department of Bioengineering, the Department of Molecular and Cell Biology, the Biophysics Graduate Group, Lawrence Berkeley National Lab, and the Institute for Human Genetics as well as the Department of Bioengineering and Therapeutic Sciences at the University of California, San Francisco.

**To apply**

Interested applicants should have statement of interest, CV, transcript, and at least three letters of reference sent to [jobs@compbio.berkeley.edu](mailto:jobs@compbio.berkeley.edu).

For more information, see <http://compbio.berkeley.edu/jobs> listing.

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