

# International Conference on **Transcriptomics**

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## **Recent advancement in Mendelian Genomics and data management/analysis challenges at the Yale Center for Genome Analysis**

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The Yale Center for Genome Analysis (YCGA) is located at 830 W. Campus Drive at Yale's West Campus and occupies approximately 7,000 sq. ft. of laboratory space and is equipped with all modern amenities. It is one of the premier genome centers in the country and is equipped with 8 Illumina HiSeq 2000, two Illumina HiSeq 2500, one each of MiSeq, PacBio-RS and Ion Torrent sequencing systems. The center delivers large and small-scale genomics projects, deploys further new sequencing technologies, develops new data collection and distribution strategies, and performs bioinformatics research and service support activities. In this short time period, YCGA has scaled up production to more than 10 terabases of DNA sequence per month and has successfully dealt with data analysis and management challenges. Since the inception in January 2010, YCGA has exemplified its scientific and institutional value through groundbreaking research projects that have shaped modern medicine and leveraged institutional assets in order to garner new funds and catalyze myriads of interdisciplinary collaborations. In the past three years, using YCGA's genomic resources, scientists have published more than 20 papers in Science, Nature, Cell and other high profile journals reporting basic understanding for long standing biological questions and new genes in disorders like hypertension, autism, several types of cancers, Gauche disease, skin disorders, brain malfunction disorders, and more. The presentation will focus on recent discoveries made at YCGA, its computer infrastructure and the current challenges and solutions developed for data analysis and management.

### **Biography**

Shrikant Mane is Senior Research Scientist in Genetics; Director, MBB Keck Biotech laboratory; Director, Yale Center for Genome Analysis at the Yale School.

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