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Recent advancements in Mendelian genomics and data management at the Yale Center for genome analysis

Shrikant Mane
Yale University, USA

The Yale Center for Genome Analysis (YCGA) is a state-of-the-art DNA Sequencing Center launched in 2010 to provide an open access centralized facility for services, equipment and expertise required for carrying out large-scale sequence analysis studies. Since its inception in 2010, YCGA has emerged as one of the leaders in the field of identification of disease associated genetic factors. Our group foresaw scientific opportunities for the development and use of exome sequencing in Mendelian genetics and was the first to develop the method for exome capture on the Nimblegen/Roche platform. We were also the first to demonstrate the biological utility of exome sequencing for clinical diagnostic applications. Currently, YCGA is a part of the NHGRI (National Human Genome Research Institute) supported Yale Center for Mendelian Genomics that uses NGS and computational approaches to discover the genes and variants that underlie Mendelian conditions. In the last four years, the use of Nextgen sequencing has led to the publications of >175 articles in peer reviewed journals, including >40 in high profile journals such as *Science, Nature, Cell, New England Journal of Medicine* and *Nature Genetics* reporting new variants in various disorders, including hypertension, autism, several types of cancers, Gaucher disease, skin disorders, and cortical malfunctions, all using exome analysis. The presentation will focus on recent discoveries in Mendelian disorders made at YCGA, its computer infrastructure and the current challenges and solutions developed for data analysis and management.

shrikant.mane@yale.edu