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Recent advancements in translation and clinical research in cancer

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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 genomic analysis, equipment and expertise required for carrying out large-scale sequence analysis studies. Our group foresaw the 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 and translational applications. Over the last couple of years, exome sequencing approach developed by us has identified disease causing variations from wide variety of phenotypes that has broadened understanding of the underlying biology linking mutations to human phenotypes. In addition, it fueled the development of essential tools for diagnosing, preventing and treating both rare and common diseases in the clinical setting. In the past five years, we have successfully applied exome analysis approach to complete whole-exome sequencing and analysis of over 75,000 samples of a wide spectrum of phenotypes to identify hundreds of novel genes. The presentation focuses on overview of translational research, biomarker discovery efforts and precision medicine efforts at Yale.

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