Single-Cell Genomic Profiling of the Adult Human Heart and Transcriptomic Analysis of Differentially Expressed Genes (DEGs) in Myocardial Pathophysiology

Desai, Shanay (School: The Mississippi School for Mathematics and Science)

The prevalence of high mortality rates of cardiovascular diseases (CVD) in humans is contributed to the lack of detailed characterization of the human heart. Largely due to limited access in sampling and overwhelming extracellular diversity in myocytes, a clear transcriptomic profile of the human heart is difficult. Here, Next Generation Sequencing (NGS) techniques were utilized to create a genomic profile from an initial 34,543 young and adult Mus musculus mice cell dataset. With 2554 cells upregulated in the young phenotype (48.5% correlation area) and 2700 cells upregulated in the old phenotype (51.5% correlation area) and 2700 cells upregulated in the old phenotype (51.5% correlation area), a total of 255 significantly enriched genes (p<0.05) were tested for downstream analysis. Gene set enrichment analysis (GSEA) showed that histone H3 deacetylation, thyroid hormone receptor binding, and target of rapamycin (TOR) signaling were the most significantly enriched intracellular compositions and cell-to-cell signaling targeted pathways within the aged heart. Hdac8, Rps6kb1, Nr0b2, Thrap3, and Per3 were also uncovered as significant gene groups after performing comparative leading-edge analyses and enrichment map visualizations. As cardiac biologists begin to examine the heart at unprecedented biological levels, more is to be understood about the human heart. Beyond useful as a valuable resource tool, this study supplies insights into the cellular functionality of gene groups and potential therapeutic strategies useful to cardiovascular disease development, progression, and prevention.