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MedGenome study analyzing Wellderley data shows interesting trend on gene mutations associated with cardiac function



FOSTER CITY, CA, UNITED STATES, April 12, 2016 /EINPresswire.com/ — MedGenome will present results from its analysis of the whole genome sequence data from the Wellderly study (Scripps Translational Science Institute) at the Diabetes Summit 2016, from April 25-27th in Boston, MA.

The study titled "Whole Genome Sequencing data from the Wellderley study identifies rare variants in genes associated with diabetes and cardiomyopathy" involved the analysis of publicly available whole genome sequence data (WGS) of 454 healthy elderly Caucasian individuals (median age 85 years) from the Wellderly study. Genes known to be causative/predictive of diabetes and cardiovascular diseases were analyzed, and results were examined for any rare variants in this population. There were surprisingly few variants of relevance in the diabetic pathway. A significant finding was the discovery of a rare variant of Nebulin (NEB) which was present in about 60% of the individuals in this cohort. Nebulin is a multifunctional protein that binds and stabilizes actin allowing thin filaments to reach mature length. Mutations in NEB are known to be associated with recessive nemaline myopathy but better understanding of its possible protective role in preserving muscle function during aging can provide insights into cardiac conditions that arise due to a loss of muscle function.

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