University of Utah
UNDERGRADUATE RESEARCH JOURNAL

CHARACTERIZING THE ROLE OF CD320 IN VITAMIN B12 UPTAKE IN IPSC'S

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The primary function of the gene, CD320, is to facilitate the uptake of vitamin B12 into cells. It is a cell surface receptor that specifically binds to a transcobalamin-vitamin B12 complex and moves it into a cell. It is ubiquitously expressed across all cell types. Vitamin B12 deficiency is linked to a number of health issues including heart disease. A study of a pedigree of one family with a history of structural heart defects shows that affected family members have damaging gene variants in 11 genes. Of these genes, CD320 is the only strong candidate for congenital heart disease. This study attempts to examine the interaction of CD320 with vitamin B12 using human induced pluripotent stem cells (IPSC's). The first part of the study is focused towards validating the existence of a CRISPR CD320 knockout line. Tests such as western blots to quantify protein expression and qPCR to quantify mRNA were carried out for this purpose. This study also focuses on creating and validating a vitamin B12 Elisa assay to study the effects of CD320 on vitamin B12 uptake. Once both aspects of this study are validated, experiments can be run to determine and quantify the effects that CD320, and absence thereof, can have on cellular vitamin B12 uptake as well as further downstream targets.