2017 Internship Project & Abstract - Rachel Fehrman

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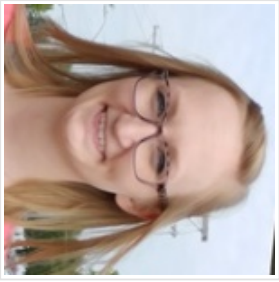
Determining the Genetic Requirements of Glycosuria Using Phenome-Wide Association Studies



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Research area: Genetics

Background: Renal glycosuria is characterized by excess excretion of glucose in the urine despite normal blood-glucose levels. Phenome-Wide Association Studies have identified variants in the SLC5A2 gene that lead to the development of glycosuria. Recent diabetic therapeutics have also targeted the protein product of SLC5A2 via inhibition. Through a combination of urine dipstick tests and genotyping, some individuals with high glucose-urine values have not been diagnosed with glycosuria, while others that have been diagnosed do not have one of the known variants. Because of this finding, we sought to identify the genetic requirements of glycosuria in order to study the impact these variations have on SLC5A2 inhibitors for the treatment of diabetes.

Methods: Marshfield Clinic Research Institute's Personalized Medicine Research Project combines genotypic data with electronic health records. Through this, we were able to obtain DNA from 14 individuals diagnosed with glycosuria. Polymerase Chain Reaction was used to amplify the exons, or protein coding regions, of the SLC5A2 gene in all individuals. The gene was then sequenced via Sanger Sequencing to identify novel variants that may be driving this phenotype.

Results: Preliminary results indicate that novel variants exist in the coding region of SLC5A2 that may predispose individuals to glycosuria.

Conclusions: Further sequencing will be done to validate results. These genetic variations will be used in various biological assays to help understand how they affect diabetic therapeutics targeted for SLC5A2.