Phenotyping Genetic Risks of Type 2 Diabetes

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Introduction

❑ Type 2 diabetes mellitus affects the body's ability to process glucose(glucose tolerance) and response to insulin
❑ People have a varying genetic risk of developing type 2 Diabetes

❑ Data from transethnic and race-specific genome-wide association studies may be used to calculate a polygenic risk score for the development of type 2 Diabetes
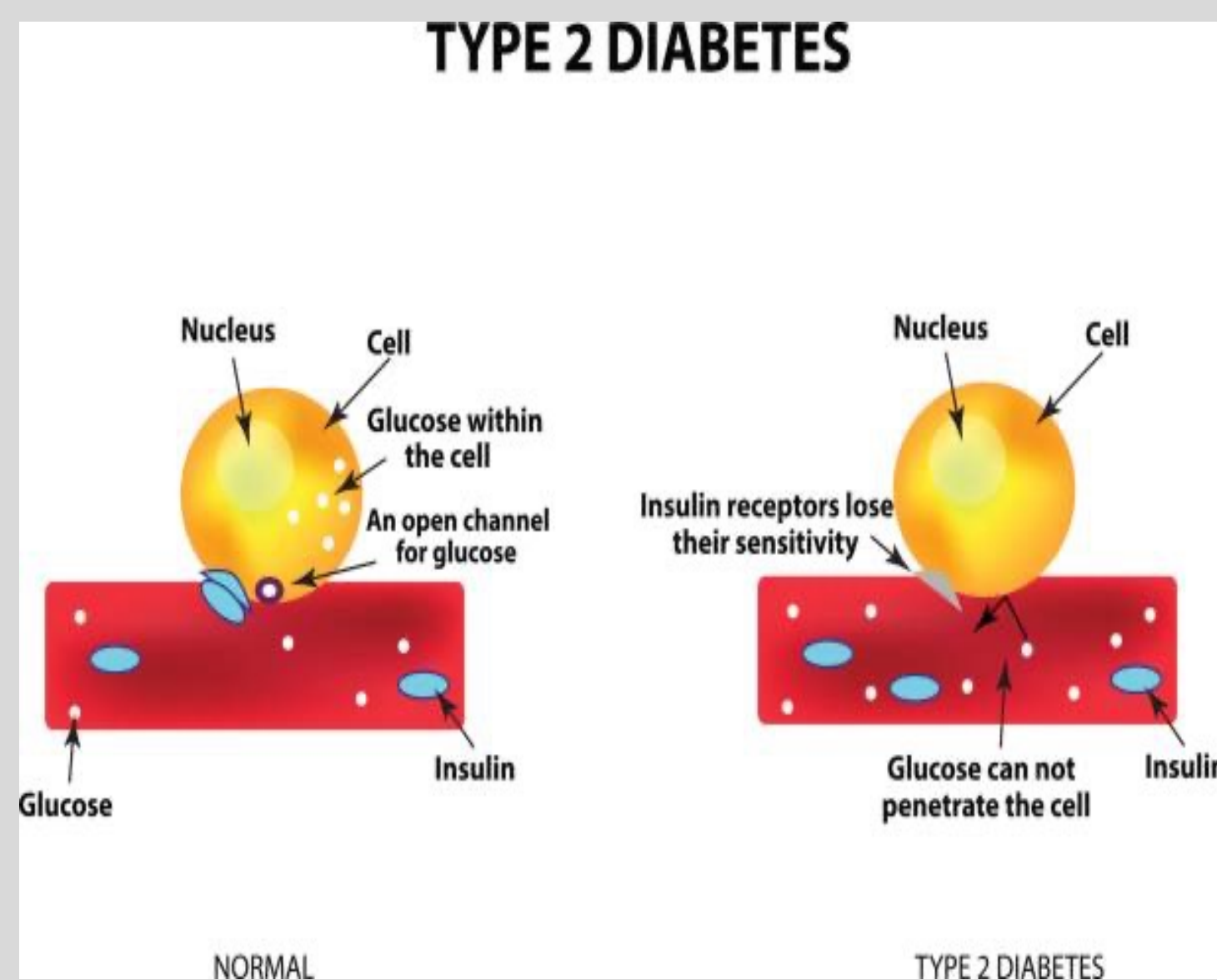
TYPE 2 DIABETES

Objectives

❑ To determine if overweight and obese individuals who do not have type 2 diabetes but have a high genetic risk for the disease will have greater insulin resistance and lower glucose tolerance than those who have a low genetic risk for the disease.
❑ To enroll 50 overweight and obese adults and 50 overweight and obese children with either high or low polygenic risk scores for type 2 diabetes mellitus but who do not have the disease.

Methods

❑ We will recruit individuals based on estimated Genetic risk from the Center for Applied Genomics Study at CHOP(children/teens) and Penn Medicine Biobank(adults).
❑ Recruitment techniques include calling, emailing and texting.
❑ Screening techniques consist of a review of electronic medical records, BMI, current medications, surgical history, and allergies.

Procedures

❑ After informed consent is obtained, An IV is placed in the individual's arm.
❑ Blood samples are collected twice, at specific time periods, before the oral glucose solution is ingested. The individual is given 5 minutes to ingest the oral glucose solution. After ingestion, blood samples are then collected nine times, at specific time periods.
❑ Dual-energy X-Ray absorptiometry(DXA)scan is done to measure body fat distribution.

Protocol

❑ Study protocol is cross-sectional.

❑ Study day is one-half day, and the main data point is the blood samples which are collected over 4 hours.

❑ Blood samples will be used to produce a glucose area under the curve (AUC) as a primary study endpoint

❑ Other measures, such as body fat distribution, insulinogenic index, and others may be used as secondary study endpoints.

❑ Standard graphing and screening techniques will be used to ensure data accuracy.

Recruitment and Results

❑ Recruitment is one of the most important parts of research, as it allows for participation in the study, as well as ensuring the sample size is large enough for a suitable data analysis.

❑ As of right now, 22 adults and 5 children have completed the study. We also have 2 adults and 1 child on the schedule for August and September

❑ Some of the challenges involved within the recruitment process are:
❖ Failure of prospective participants in remembering participation in previous study in which they agreed to be recontacted for future research.
❖ Prospective participants asking to be recontacted at a later date and time but not responding at that later date and time.
❖ Prospective participants agreeing to participate in the study but failing to show up on the day of the study.

Conclusions

❑ Due to the ongoing nature of the study the results and conclusions are pending.

❑ However, we expect individuals with a high genetic risk for type 2 diabetes will have a greater insulin resistance and lower glucose tolerance than those who have a low genetic risk for the disease.

References

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2. Valdez R. Detecting undiagnosed type 2 diabetes: family history as a risk factor and screening tool. Journal of diabetes science and technology 2009;3:722-6.