

Principal Investigator: Warram, James

Institution: Joslin Diabetes Center

Project Title: Search for Susceptibility Genes for Diabetic Nephropathy in Type 1 Diabetes

1. **Goal of study:** To identify susceptibility genes for diabetic nephropathy in type 1 diabetes.
2. **Design:** Case-control (parent-offspring trios available through NIDDK*).
3. **Race/ethnicity of participants:** European American.
4. **Country of origin of majority of study participants:** United States.
5. **Sample size:** 905 cases, 890 controls*, 10 pairs of parents of cases, 10 pairs of parents of controls (additional 564 pairs of parents available through NIDDK).
6. **Primary disease phenotypes and criteria for diagnosis:** Cases have type 1 diabetes and nephropathy defined as replacement therapy for end-stage renal disease (chronic dialysis or transplant) or persistent proteinuria evidenced by multiple measurements of the urinary albumin to creatinine ratio ≥ 300 μg albumin per mg creatinine. Controls have type 1 diabetes duration ≥ 15 years and normoalbuminuria evidenced by multiple measurements of the urinary albumin to creatinine ratio < 20 μg albumin per mg creatinine.
7. **Secondary phenotypes:** Creatinine and cystatin C in serum as quantitative measures of renal function (applies only to cases with proteinuria and controls).
8. **Matching variables:** None.
9. **Non-genetic risk factors, covariates, or confounders:** Hemoglobin A1c, a measure of glycemic control. (A low value in a control may indicate that genetic susceptibility has been masked.)
10. **Primary Institution or Study Site:** Section on Genetics and Epidemiology, Joslin Diabetes Center, Boston, MA.
11. **Lead Principal Investigator and contact information:**

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12. **Collaborating institutions or study sites:**

George Washington University Biostatistics Center, Washington, DC
Centers for Disease Control and Prevention, Atlanta, GA
University of Minnesota, Minneapolis, MN
Juvenile Diabetes Research Foundation, New York, NY

13. **Collaborating Co-Investigator and contact information:**

Andrzej S. Krolewski, MD, PhD
Contact information is the same as Lead PI

14. Study website URL: <https://www.gokind.org/access>

15. Key study publications:

Mueller PW, Rogus JJ, Cleary PA, et al. Genetics of Kidneys in Diabetes (GoKinD) Study: a genetics collection available for identifying genetic susceptibility factors for diabetic nephropathy in type 1 diabetes. *J Am Soc Nephrol* 17: 1782-1790, 2006.

* The case and control probands available in the GAIN dataset differ from those described in the GoKinD publication listed under #15. First, 27 cases and 47 controls were excluded from GAIN because of consent restrictions. They are available from NIDDK through a separate application process, along with the 564 pairs of parents. The second difference from the publication is due to the removal of individuals because of relatedness between probands. Finally, as stated in the publication, the numbers were preliminary, and a few additions and deletions to the collection were made after publication.

16. Data Use Limitations:

Cases and controls: Limited to genetic research on type 1 diabetes and its complications. Complications include nephropathy, cardiovascular disease, retinopathy, neuropathy, and mortality. Phenotypes related to diabetes and its complications, such as body mass index, blood pressure, lipids, and hemoglobin A1c, may also be studied.