



**Application number/Title:** 40709- Exploring the effects of genome structural variation on human health and disease

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**Keywords provided by the Applicant PI to describe the research project:**

GWAS, heritability, structural variation

**Application Lay Summary:**

Genetic variation is known to be an important contributor to nearly all health-related traits. However, the precise genetic variants that affect most diseases and traits are largely unknown, in part because many forms of genetic variation are difficult to ascertain. We will analyze a particular class of understudied genetic variants known as structural variants, which affect large segments of DNA. We will develop methods for detecting these variants in UK Biobank genotyping data, and we will then examine their effects on a broad spectrum of health-related traits by studying UK Biobank genotype and phenotype data for N=500,000 individuals. For variants of particular interest, we will further investigate how these variants have been passed down across generations. We anticipate that this multi-year project will identify many genome structural variants likely to have causal effects on health-related traits (e.g., diabetes, cardiovascular disease, and cancer).