**Application number/Title:** 17731 - A phenome-wide association study of copy number variation

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**Funding body:** Internally funded by University of Minnesota

**Keywords provided by the Applicant PI to describe the research project:**
Copy_number_variation, deletions, duplications, cancer, cardiovascular_disease

**Application Lay Summary:**

1a: To identify changes in the DNA, particularly large deletions and duplications of the genome, that are related to complex diseases and intermediate biomarkers

1b: The identification of genetic risk factors that are related to disease will help reveal the underlying biology and thereby expose targets for environmental modification and/or novel drug development.

1c: We will use the raw Affymetrix Axiom data (.CEL files) and apply a principal components analysis on these intensities to correct for DNA quality and batch effects. We will then perform a joint calling across all samples to determine potential deletions and duplications using our soon to be open-source Genvisis software package, which will also assess the quality of these calls and filter them down to a set of high quality calls (~1 year to complete). We will then associate the high quality calls with the phenotypes available, and replicate any findings using external data sources (~1 additional year).

1d: Full cohort; all samples with high quality intensity data will be analyzed