



Application Number / Title: 19348 - Control data-sets for congenital heart disease copy number variant analysis

Applicant PI: Professor Peter Gruber

Applicant Institution: University of Iowa, Cardiothoracic Surgery, 451 Newton Road, Gruber Lab, 4338 PBDB, Iowa City IA 52246, United States

Lead Collaborators: 1) Professor Stephen Gruber

Collaborating Institutions and Addresses: 1) University of Southern California, USC Norris Comprehensive Cancer Center, 1441 Eastlake Avenue, 8302L, Los Angeles CA 90033, United States

Funding body: Internally funded by University of Iowa's Carver College of Medicine

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

Application Lay Summary:

1a: We are studying the genetic basis of congenital heart disease. Specifically we are interested in genetic rearrangements that may occur in these children and adults.

1b: We are interested in improving the health of children and adults with congenital heart disease. It has been clearly show that children with identified genetic abnormalities have an increased risk of morbidity and mortality compared to those without obvious identifiable genetic abnormalities. However, no study to date has comprehensively looked at CNV (copy number variants) in this population. We are completing this study but need a control population in order to compare.

1c: We will use the data from the UK Biobank to compare to our other data. This comparison will allow us to determine which genetic abnormalities are distinct for the population with congenital heart disease compared to that without.

1d: 4,000 normal subjects without known congenital heart disease

The Scientific Team has agreed that it is acceptable to tweak the wording if necessary to rectify any words spelt incorrectly etcetera. The Applicant PI has been made aware.