

Eligibility Criteria

Did subject meet all Inclusion Criteria:

1	Subjects in whom WES or WGS has already been performed, either during the CHD GENES study or, for new centers (Utah or USCF/Gladstone), after trios in existing biobanks undergo analysis by WES or WGS during the PCGC2 grant cycle	Provided by ACC
2	Presence of deleterious mutations (damaging de novo mutations or stringently defined deleterious missense mutations) identified on sequencing (Cases) OR absence of such known deleterious mutations (Controls)	Provided by ACC
3	Males or females, age ≥8 years	<input type="checkbox"/> Yes <input type="checkbox"/> No
4	Diagnosis of CHD	<input type="checkbox"/> Yes <input type="checkbox"/> No
5	Informed consent obtained	<input type="checkbox"/> Yes <input type="checkbox"/> No

Did subject meet any Exclusion Criteria:

1	History of cardiac transplant	<input type="checkbox"/> Yes <input type="checkbox"/> No
2	A cardiac surgical procedure within 6 months of enrollment	<input type="checkbox"/> Yes <input type="checkbox"/> No
3	Known genetic syndrome due to a pathogenic variant identified in a gene associated with abnormalities of the brain structure or function, structural heart disease, and potentially other associated features.	<input type="checkbox"/> Yes <input type="checkbox"/> No
4	Presence of CNV known to be clinically pathogenic. Variants will be classified as pathogenic using accepted types of variant evidence (e.g., population data, computational data, functional data, segregation data) as detailed in the American College of Medical Genetics and Genomics “ACMG Standards and Guidelines for the interpretation of sequence variants” (Richards et al, GIM 2015).	Provided by ACC
5	Overwhelming acquired brain injury, such as a major stroke or severe ischemic injury, that would overshadow the effect of a genetic mutation on outcome in the opinion of the center investigator	<input type="checkbox"/> Yes <input type="checkbox"/> No
6	Lack of ability to communicate in English or Spanish	<input type="checkbox"/> Yes <input type="checkbox"/> No

Did subject meet any MRI Exclusion Criteria:

1	Contraindication to having brain MRI scan	<input type="checkbox"/> Yes <input type="checkbox"/> No
2	Claustrophobia or inability to lie still while in the MRI scanner for the required time (sedation will not be allowed)	<input type="checkbox"/> Yes <input type="checkbox"/> No
3	Pregnancy	<input type="checkbox"/> Yes <input type="checkbox"/> No

PI Signature: _____

Date: _____