PCGC CHD GENES STUDY
SUMMARY REPORT

The CHD GENES study enrollment started in October 2010 with a single pilot study. Full study enrollment started in November 2010 and continued through the second grant period end date (July 31, 2020). The study goal was to enroll 10,000 probands by the end of the first grant period, July 31, 2015 and 3250 probands by the end of the second grant period. This summary report discusses cumulative data from this study from 2010 through December 31, 2020.

ENROLLMENT

As of December 31, 2020,13,309 probands have been enrolled in CHD GENES across all centers. Fifty-six percent of enrollments are proband-parent trios, 23% are proband and one parent, and 21% proband-only. Any first-degree relative, as well as affected three-generation relatives of enrolled probands were eligible to enroll. To date, 18,195 relatives were enrolled including 17,872 (98%) parents and 323 (2%) other relatives.

PROBAND CHARACTERISTICS

Table 3A shows the proband sample has a mean age of 9.7±12.3 years and a median [inter quartile range] age of 4.4 [0.4, 15.5] years with 35% infants. Most patients in the sample are white (73%), 7% are black, and 6% Asian. Thirty-two percent (32%) of the probands (4172) have abnormal extracardiac findings. Clinical genetic testing was performed for 1,890 (45%) of probands that have abnormal extracardiac findings. For patients with extracardiac findings, abnormal Karyotype was found in 22%, microarray in 29%, and/or FISH testing in 21%.

Investigators used two methods to categorize cardiac diagnoses. First, subjects were placed into broad categories of high genetic interest on the enrollment form. Using these categories, 44% of the probands have a conotruncal defect, 20% have a left-sided obstructive lesion, 11% had atrial septal defect, and 7% had heterotaxy syndrome.

Additionally, detail cardiac diagnoses were coded using the Fyler system and have been completed for 13,214 (99%) of probands. The most common defect in probands is isolated ventricular septal defect (VSD, 28%), followed by atrial septal defect-secundum (ASD, 24%), and Tetralogy of Fallot (TOF, 16 %).

BIOREPOSITORY SPECIMENS

A total of 13,009 proband specimens (11,996 whole blood and 1,013 saliva) and 17,808 relative specimens (14,186 whole blood and 3,622 saliva) have been received by the repository.

For additional information and requests to access data via dbGAP:

https://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?study_id=phs001194.v2.p2

PCGC CHD GENES STUDY
PROBAND DEMOGRAPHICS AND ANATOMIC DIAGNOSIS
AS OF 12/31/2020

	Mean±SD / N(%)
Number of probands	13309
Age of consent (yrs)	
Mean±SD	9.7±12.3
Median (IQR)	4.4 (0.4 ,15.5)
Age group of consent (yrs)	
<1	4646 (34.9%)
1 - 12	4611 (34.6%)
13 - 18	1712 (12.9%)
19+	2340 (17.6%)
Race	
White	9765 (73.4%)
Black	967 (7.3%)
Asian	853 (6.4%)
Other	1254 (9.4%)
Unknown	470 (3.5%)
Hispanic	
Yes	3142 (23.6%)
No	9970 (74.9%)
Unknown	197 (1.5%)
Abnormal extracardiac findings	
Yes	4172 (31.5%)
No	9060 (68.5%)
If yes, clinical genetic testing performed*	1890 (45.3%)
Abnormal karyotype	
Yes	487 (22.4%)
No	1688 (77.6%)
Abnormal microarray results	
Yes	466 (29.0%)
No	1139 (71.0%)
Abnormal FISH results	
Yes	201 (20.5%)
No	781 (79.5%)

PCGC CHD GENES STUDY
PROBAND DEMOGRAPHICS AND ANATOMIC DIAGNOSIS
AS OF 12/31/2020

	Mean±SD / N(%)
Cardiac diagnosis	
Enrollment form	13309
Atrial septal defect	1401 (10.5%)
Conotruncal defect	5857 (44.0%)
Left-sided obstructive lesion	2625 (19.7%)
Heterotaxy syndrome	886 (6.7%)
Fyler codes	13214
Tetralogy of Fallot	2051 (15.5%)
Double outlet right ventricle	790 (6.0%)
Atrial septal defect - Secundum	3215 (24.3%)
Hypoplastic left heart syndrome	707 (5.4%)
Coarctation of the aorta	1587 (12.0%)
D-transposition of the great arteries	1195 (9.0%)
Isolated ventricular septal defect	3661 (27.7%)
Atrioventricular canal	1241 (9.4%)
Heterotaxy/Related Findings	931 (7.0%)