Baylor Collegeof Medicine



The Inpatient Cardiovascular Genetics Consult Team A Quality Improvement Initiative

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BACKGROUND Texas Children's Hospital Inpatient Cardiovascular Genetics (CVG) service was established in July 2021 • Goal: improve the timeliness, appropriateness, and ethical ordering of genetic testing for children with congenital heart disease (CHD) • Team roles Rounding with the CICU once weekly – Performing consultations - Facilitating informed consent for genetic testing **HYPOTHESIS** Establishment of the CVG service decreased errors in obtaining appropriate genetic testing FIGURE 1: CHD Testing Algorithm **CONGENITAL HEART DISEASE TESTING ALGORITHM** No Strong suspicion o yes additional aneuploidy (T21, Karyotype T13, T18, Turner)? testing Chromosomal microarray No Hypoplastic left yes additional Rapid FISH for T21, T13, heart syndrome testing

T18, XY/XX

Chromosomal microarray

Other major

congenital heart

disease?

yes

Consider CHD panel or

whole exome sequencing

Solved?

No

additional

testing

MATERIALS AND METHODS

Inclusion	oritoria
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- Age < 30 days
- Born Aug Oct
 - Pre-CVG team (2019, 2020)
 - Post-CVG team (2021)
- Critical CHD
- Admission to Cardiac Intensive Care Unit

- Assessment of genetic testing appropriateness [Figure 1]
 - Implementation of adequate testing
- Reduction in unnecessary testing
- Demographics
 - Racial/Ethnic Groups: White, Black, Hispanic, Other
 - Primary Language: English, Spanish, Other

FIGURE 2: Genetic Testing Pre- and Post-CVG Team



2021 (n = 37) After initiation of the CVG team...

- Overall appropriate testing increased: 57.8% to 100% (p<0.001) [Figure 2]
- Adequate testing increased: 84.4 to 100% (p = 0.011)

RESULTS

- Inappropriate testing decreased: 26.7% to 0% (p < 0.001)
- Yield of the algorithm in determining an etiologic diagnosis: 26%
- 22q11.2 deletion (n=6, 5%)
- Trisomy 21 (n=4, 3%)
- Turner syndrome (n=4, 3%)
- Yield of sequencing 14% (variants in CHD7, KMT2D, ACTC1, DNAH5, NODAL)
- No difference in appropriate testing before or after CVG inpatient team initiation among racial/ethnic groups or English vs Spanish speaking groups, but improvements noted in all groups post-CVG team

CONCLUSIONS

Implementation of a CVG inpatient consult team can significantly improve appropriate and timely ordering of genetic testing

- Optimizing test appropriateness may portend a future cost benefit as inappropriate testing is decreased and diagnostic yield is increased
- Differences in racial/ethnic groups and primary language may be revealed with larger sample size
- User-friendly universal protocols for genetic testing based on CHD lesion may be beneficial for hospitals without a CVG inpatient team and are currently being developed by our team

