PREVALENCE OF EXTRACARDIAC BIRTH DEFECTS AND GENETIC CONDITIONS IN HYPOPLASTIC LEFT HEART SYNDROME AND ASSOCIATION WITH HOSPITAL UTILIZATION AND MORTALITY

Subhrajit Lahiri[1], Shaine Morris[2]

[1] Baylor College of Medicine, Department of Pediatrics, Cardiology

[2] Baylor College of Medicine, Pediatrics, Cardiology

Background: Patients with congenital heart disease have associated extracardiac birth defects and/or genetic conditions (BDGC), and the presence of these may influence therapies and outcomes. We aimed to evaluate the prevalence of BDGC in hypoplastic left heart syndrome (HLHS) and evaluate the associations between specific BDGC and hospital mortality and post-Stage I mortality

Materials/Methods: We included patients from the Pediatric Health Information System database from 1999-2018 with an ICD9 or ICD10 code for HLHS and admission <30 days of life. We queried all hospitalizations for 47 BDGC. We then evaluated hospital mortality and hospital mortality after Stage I surgery by specific BDGC. Stage I surgery was defined by ICD diagnostic code for HLHS and ICD procedure code for systemic to pulmonary artery shunt or right ventricle to pulmonary artery conduit, or pulmonary artery band placement in the neonatal period.

Results: A total of 5438 patients admitted during the neonatal period were included in the study, of which 1923 (35.3%) had a BD. Out of these, 4719 (87%) underwent Stage 1 intervention, of which 1597 (33.8%) had BD. 3410 (60%) were male. The most common genetic condition was Turner syndrome, and the most common birth defects were laryngeal defects, genitourinary defects, congenital hip dysplasia, and cystic kidney disease; Table 1 and Table 2 demonstrate the prevalence and mortality of the most common BDGC noted for all neonates, and neonates who underwent Stage 1 repair. The overall mortality was 19%(n=1045) and mortality of those undergoing Stage 1 was 7.8% (n=370). Presence of BD was found to be significantly associated with mortality in neonates (339,18%, p <0.001). The specific BDGCs associated with higher mortality than the non BDGC population were Turner syndrome, genitourinary anomalies, anencephaly, intestinal atresia and cystic diseases of kidney ($p<0.05$) for both groups.

Conclusions: : Laryngeal anomalies, genitourinary anomalies, congenital hip dysplasia and cystic kidney disease are the most common BDGC in patients with HLHS. The additional mortality associated with BDGC varies widely by the condition. Patients with BDGC who underwent Stage I palliation demonstrate better survival compared to the whole group– this may represent selection bias in which only the best candidates are put forward for surgery. Information obtained in this study may be useful for patient counseling, recommendations for screening for anomalies and genetic conditions, and perioperative management.