Elizabeth Fischman, August 2022

Defining clinical characteristics and hemodynamics of patients with Trisomy 18 undergoing congenital heart surgery

Background

Trisomy 18, also known as Edwards Syndrome, is the second most common trisomy condition. The prevalence is estimated to be 1 in 6,000 live births (Fick 2021). The anomalies most seen with Trisomy 18 are congenital heart disease (CHD) and limb, GI, eye, CNS, GU, and renal abnormalities (Baty 1994). CHD is by far the most common anomaly with rates as high as 85%-91% (Kosiv 2017; Fick 2021). Specifically, the most prevalent congenital heart lesions are atrial septal defect, ventricular septal defect, patent ductus arteriosus, and polyvalvular disease (Fick 2021).

Historically, children born with Trisomy 18 were only offered palliative and comfort care due to their expected early death. Until recently, central apneas were the leading cause of death Until recently, central apneas were the leading cause of death. However, with improving supportive care measures, CHD and heart failure have become a leading cause of death for these patients (Domingo 2018). The overall mortality, once believed to be less than 1 year, is now being reported in more recent studies to be up to the second decade of life (Rasmussen 2003).

Early studies failed to show improvement in outcomes following Congenital Heart Surgery (CHS), but, in more recent studies, survival rates have improved with palliative surgery (Fick 2021). The change in research outcomes has led to a paradigm shift within cardiology and cardiothoracic surgery to offer palliative surgery to these patients. The Journal of Pediatrics recently published an article reviewing trends in Trisomy 18 treatment from 1997 to 2016. Over the past 20 years, cardiac intervention increased 5-fold. Additionally, the overall inpatient mortality rate decreased in those with Trisomy 18 from 32% in 1997 to 21% in 2016 (Fick 2021). A smaller, 50-person follow-up study found the median survival of patients who had undergone CHS to be 16.2 years (95% CI: 12 to 20.4 years) (Peterson 2017). Although there has been improved mortality following CHS, the Trisomy 18 patient population continues to have increased overall mortality rates following CHS in comparison to the general population (Kosiv 2017).

Despite increases in surgical intervention in these patients, the qualifications and characteristics of patients being offered surgery has not been clearly defined. Furthermore, there appears to be differing opinions within pediatric subspecialities regarding CHS in these patients versus comfort care measures alone. This was seen in a recent study comparing opinions of neonatologists to pediatric pulmonologists, where it was found that neonatologists were significantly more likely to recommend comfort care only or palliative care (Hurley 2014). Furthermore, 54% neonatologists that responded to the survey believe that patients with Trisomy 18 would die prior to age one, even without the presence of CHD, compared to only 5% of pulmonologists. Investigating which parameters were determined appropriate for CHS within this unique patient population will be beneficial in disseminating information throughout the different pediatrics subspecialities. It may also be helpful for determining characteristics of patients that warrant transfer to pediatric cardiac surgical center.

In reviewing current studies evaluating surgical decision making, there is a particular gap in hemodynamic information for these patients (Carvajal 2020). A specific interest of this study is to further evaluate hemodynamics as it relates to the presence and degree of pulmonary hypertension with the Trisomy 18 and CHD population.

Study Aims

Primary Aim:

- Assess clinical characteristics and hemodynamic indicators in patients with Trisomy 18 and CHD who underwent cardiac surgery at NYP- Morgan Stanley Children's Hospital from 2012 to 2021.

Secondary Aim:

- Assessment of pre-operative catheterization parameters in patients who underwent CHS

Study Design

Category: A case series using retrospective chart review

Study Population: All children ages 0-18 with Trisomy 18 and CHD who had congenital heart surgery at NYP- Morgan Stanley Children's Hospital from 2012 to 2021

Study Variables:

- Gestational age at birth
- Birth weight
- Sex
- Weight at time of surgery
- Age at time of surgery
- Complexity of congenital heart disease defined by a predetermined scale
- Need for supplemental oxygen at time of surgery
- Need for pressure support ventilation at time of surgery
- Tracheostomy present at time of surgery
- Gastrostomy tube present at time of surgery
- Presence and degree of pulmonary hypertension at time of surgery
- Diuretic usage at time of surgery
- Inotropic agent usage at time of surgery
- Any history of neurosurgical procedure prior to time of surgery
- Presence of mosaicism
- Presence of extracardiac anomalies: limb deformities, oro-facial clefts, CNS, GI and Renal/urinary tract defects

Methodology:

- Chart review using ICD codes for Trisomy 18 and CHS

Statistical Analysis Plan:

- Descriptive statistics and incidence rates within study variables

Future Research:

- Comparison of neonatologist, pediatric cardiologist, pediatric cardiothoracic surgeon opinion on surgical decision making for real time cases being presented at cardiac conferences
- Comparison of study variables to patients who did not undergo CHS at a set time point
- Assess parental wishes and palliative care involvement
- Assessing predictors in 5-year survival following CHS using the same study variables

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