

Complex Congenital Heart Disease: Rates of Genetic Abnormalities and Effects on Growth

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Infants with congenital heart disease (CHD) requiring surgery in the neonatal period frequently experience growth delay and failure to thrive that is multifactorial. This has been well studied in infants with hypoplastic left heart syndrome (HLHS), with improved growth demonstrated when standardized protocols for nutrition are followed. Genetic abnormalities occur in 5-15% of infants with complex CHD. Stunted growth occurs in many genetic syndromes. We hypothesized that those with complex CHD and identified genetic abnormalities will have a slower rate of growth than those with normal genetics. A retrospective chart review was performed of all patients who underwent a RACHS IV-VI procedure in the neonatal period between 1/1/2006 and 5/1/2017 at the University of Iowa, excluding those without genetic testing performed. Of the 30 patients analyzed to date, 10 patients had identified copy number variants (CNVs), one had cri-du-chat and one had DiGeorge anomaly. This rate of 40% with genetic abnormalities is higher than the previously published rates of 5-15% described for HLHS patients. Those with identified genetic abnormalities have consistently higher rates of weight and height z scores less than -2.00 at birth, discharge from the hospital, and follow-up close to 6 months of age. They also had higher rates of mortality by 6 months. Three patients without any identified genetic abnormalities discharged on oral feeding alone, whereas the remainder of the patients had to supplement with NG or G tube. From preliminary data on this small sample size, we concluded a large percentage of those with complex CHD and genetic abnormalities have growth concerns as early as birth and continuing into at least the first 6 months of life and are more likely to need supplemental methods of feeding as infants. We plan to further assess these parameters in more patients and at more frequent time points.

	Birth Weight z score <-2	Birth Height z score <-2	Discharge Weight z score <-2	Discharge Height z score <-2	Follow Up Weight z score <-2	Follow Up Height z score <-2	Discharge Feeds PO Only	Death
Without genetic abnormalities	11.1% (2/18)	0% (0/17)	30.8% (4/13)	27.3% (3/11)	31.3% (5/16)	18.2% (2/11)	21.4% (3/14)	16.7% (3/18)
With genetic abnormalities	16.7% (2/12)	20% (2/10)	33.3% (3/9)	40% (2/5)	40% (4/10)	25% (2/8)	0% (0/9)	41.7% (5/12)