

Genetic testing modalities and diagnostic trends in patients with multiple congenital anomalies in the neonatal intensive care unit

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Congenital anomalies are frequently observed in infants admitted to the Boston Children's Hospital (BCH) level IV NICU. Our objective was to examine the prevalence of multiple congenital anomalies (MCA) in admitted neonates that received genetics/metabolics services within their first two years of life, and assess the proportion with a genetic diagnosis. A retrospective analysis was conducted for all admissions from January 1, 2011 to December 31, 2014 for whom a genetics/metabolics consult was obtained before two years of age. Of 2,126 admissions, 449 patients met inclusion criteria. The majority (77%, 345/449) presented with at least one congenital anomaly, with 51% (231/449) having MCA. For MCA patients, chromosomal microarray analysis (CMA) was most commonly ordered (124/193, 64%), with a diagnostic yield of 19%. The overall diagnostic yield for patients presenting with MCA was 39% (89/231) and 29% (63/218) for non-MCA cases. The MCA population in a level IV NICU setting is enriched with genetic diagnoses. Although there is a range of testing methodologies utilized in the genetic workup of MCA patients, CMA was the most utilized modality. Understanding the current landscape of genetic evaluation as well as future technologies is critical to guide future practices in the diagnostic process for patients with MCA.