

Clinical Genetics

AB019. Clinical chromosomal microarray analysis in Singapore

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Background: Chromosome microarray analysis (CMA) is recommended as the first-tier genetic test for children with intellectual disabilities, developmental delay, autism spectrum disorder and/or multiple congenital anomalies. Our laboratory has been offering CMA as a clinical test since February 2014. We aimed to describe the range of copy number variants detected and the detection rates of the test.

Methods: From February 2014 to June 2017, 510 patients underwent testing using Agilent Technologies 4x180 K SurePrint G3 Human CGH+SNP Platform. Analysis was performed using Cytogenomics software. Benign copy number variants (CNVs) and CNVs less than 300 kb in size

were not reported unless they contained genes known to be associated with disease.

Results: CNVs ranging in size from 0.009 to 155 Mb were found in 161 out of 510 (32%) samples. Eighty patients (17%) had pathogenic CNVs identified and variants of uncertain significance (VUS) were detected in 88 patients (17%). Pathogenic deletions exceeded duplications (71 vs. 24), but for CNVs classified as VUS, there were 31 deletions versus 49 duplications.

Conclusions: CMA is a useful diagnostic tool for delineating the cause of intellectual disability, developmental delay and multiple congenital anomalies in our patient population. The information obtained helps to guide the medical management of the patient and allows the medical team to provide recurrence risk assessment for the families.

Keywords: Chromosomal microarray; aCGH; developmental delay; intellectual disability; Singapore

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