



Hong Kong College of Paediatricians
Annual Scientific Meeting



7th December 2013



2013

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08:50-18:30

Pao Yue Kong Auditorium & Lim Por Yen Lecture Theatre
Hong Kong Academy of Medicine Jockey Club Building
99 Wong Chuk Hang Road, Aberdeen, Hong Kong

Co-sponsored by:

Hong Kong Society of Paediatric Respiriology



Hong Kong Society for Paediatric Immunology & Infectious Diseases



Co-organized by:

Paediatric Neurology Association of Hong Kong



The Hong Kong Society of Child Neurology and Developmental Paediatrics



Appendix III

(Poster Presentations)

POSTER PRESENTATION 9:

Integration of chromosomal microarray into paediatric clinical care in Hong Kong

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Background and Aims

Chromosomal microarray (CMA) has emerged as a major tool to identify unbalanced chromosomal aberrations in children and is recommended as the first tiered investigation for intellectual disability, autism spectrum disorders and multiple congenital anomalies. While the clinical interpretation and genetic counseling remain as ongoing challenge, data about potential downstream benefits and harms of CMA is lacking, especially in paediatric population. Our objective is to evaluate the clinical impact of CMA on medical management in children.

Methods

In January 2011-May 2013, we performed high resolution CMA using the NimbleGen 135k oligonucleotide array on 330 children in a university-affiliated paediatric unit in Hong Kong at a research base. Patients who had received prenatal CMA test or failed to follow-up, and those with trisomy disorder were excluded. Cases with pathogenic/likely pathogenic CMA results were analyzed. By retrospective chart review, descriptive and multivariate analyses are performed to understand the association between CMA results and change in the medical management.

Results

Total 82 patients were reported to have abnormal CMA results. Pathogenic/likely pathogenic chromosomal aberrations accounted ~10% (32 patients with pathogenic and 2 patients with likely pathogenic changes), while the aberration with unclear/uncertain clinical significance accounted for the remaining 90%. CMA detects clinically significant submicroscopic (<5 MB) abnormality in 18 patients. Some syndromal disorders were initially missed on clinical assessment either due to atypical clinical feature or patient's young age, including two William syndrome, two DiGeorge syndrome, one Cri-du-Chat syndrome, and one Klinefelter syndrome. All clinical significant cases were followed with genetic counseling. Total 69 medical managements were prompted by pathogenic/likely pathogenic CMA in 34 patients. One family has withdrawn as the parents realized that "knowing more may not be better".

Conclusion

CMA findings can be medically actionable and/or have major implications for family members. The insights we have learned from some of our patients have wider implications for the medical community. The potential of CMA findings to impact, positively and negatively, on patients is tremendous and warrants careful evaluation. Our findings will be instructive in anticipating the impact of whole genomic analyses on medical management and downstream utilization of health services.