<table>
<thead>
<tr>
<th><strong>Title</strong></th>
<th>Integration of chromosomal microarray into paediatric clinical care in Hong Kong</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Author(s)</strong></td>
<td>Tao, V; Chu, YWY; Chan, KY; Lau, EYL; Mok, GTK; Tso, W; Liu, A; Kan, A; Tang, MH; Lau, YL; Chung, BHY</td>
</tr>
<tr>
<td><strong>Citation</strong></td>
<td>The 2013 Annual Scientific Meeting of the Hong Kong College of Paediatricians (HKCPaed), Hong Kong, 7 December 2013. In ASM Programme Book, 2013, p. 37</td>
</tr>
<tr>
<td><strong>Issued Date</strong></td>
<td>2013</td>
</tr>
<tr>
<td><strong>URL</strong></td>
<td><a href="http://hdl.handle.net/10722/193308">http://hdl.handle.net/10722/193308</a></td>
</tr>
<tr>
<td><strong>Rights</strong></td>
<td>This work is licensed under a Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International License.</td>
</tr>
</tbody>
</table>
POSTER PRESENTATION 9:
Integration of chromosomal microarray into paediatric clinical care in Hong Kong
Tao V,1 Chu YWY,1 Chan KY,2 Lau EY,2 Mok G,1 Tso W,1 Liu A,1 Kan A,2 Tang MH,2 Lau YL,1 Chung BHY1,2
1Department of Paediatrics and Adolescent Medicine; 2Department of Obstetrics &
Gynaecology, LKS Faculty of Medicine, The University of Hong Kong, Hong Kong

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.