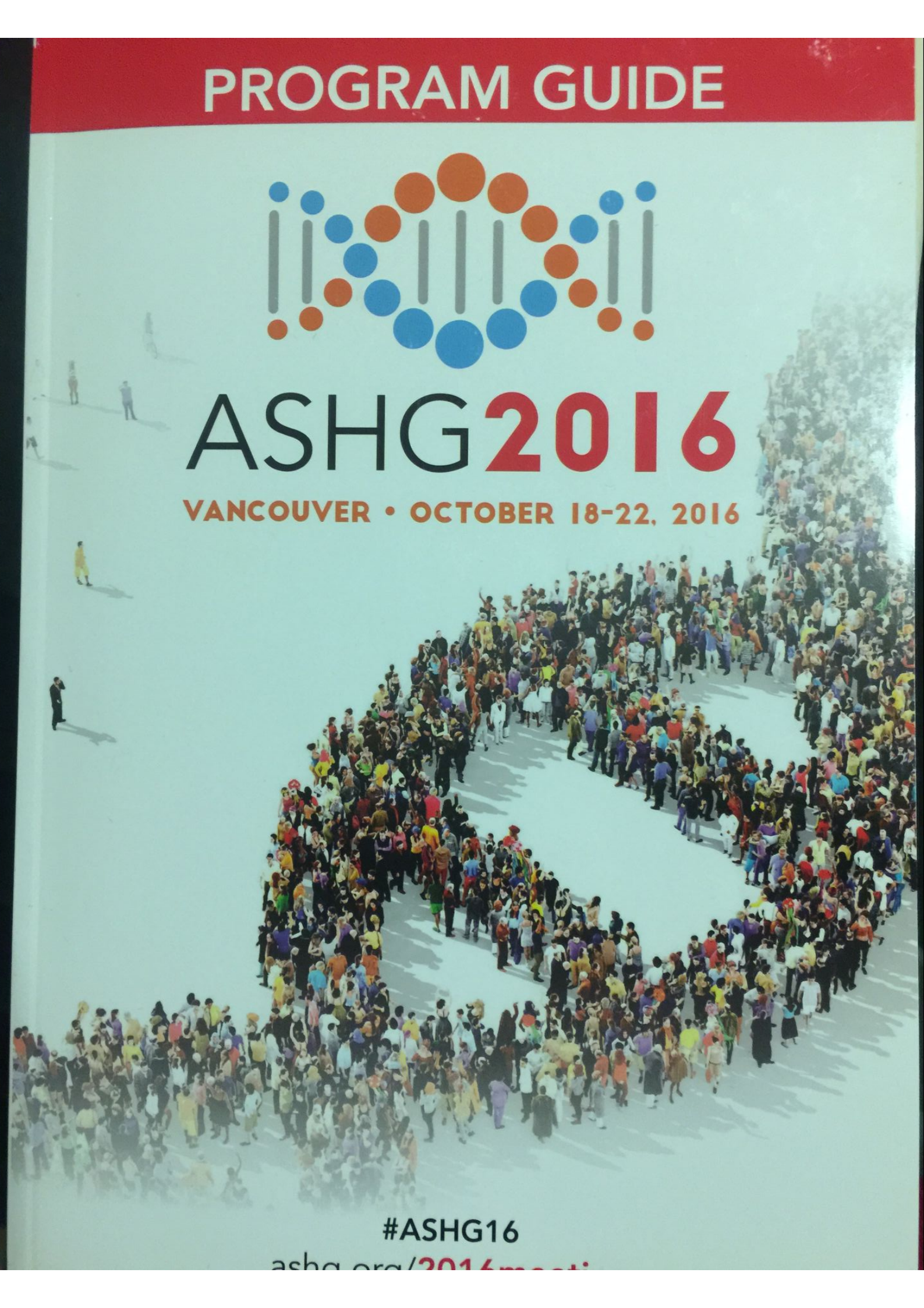
PROGRAM GUIDE

ASHG2016

VANCOUVER • OCTOBER 18-22, 2016



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3248T Development of a novel targeted assay for non-invasive prenatal testing of fetal trisomies and sub-chromosomal abnormalities exhibits diagnostic level accuracy. P.C. Patsalis.

3249F Non-invasive prenatal testing for *de novo* dominant monogenic diseases using maternal cell-free DNA. J. Zhang.

3250W Microarrays in prenatal diagnosis – some interesting cases during validation using the Affymetrix CytoScan 750K SNP chip. L. Gole.

3251T Frequency of prenatal diagnosis of most common chromosomal abnormalities in 289,076 births in Bogota and Cali, Colombia: 2011-2015. P. Hurtado.

3252F Diagnostic utility of chromosomal microarray analysis in neonates: A comprehensive multi-year study. T. Sahoo.

3253W Pregnancy and neonatal outcome associated with genome-wide paternal uniparental disomy. L. Wallace.

3254T Genomic abnormalities in products of conception in the CMA era. H. Yonath.

3255F Case Report: Prenatal diagnosis of X-linked Optiz G/BBB Syndrome by chromosomal microarray in a male fetus presenting with fetal hydrops, increased nuchal fold and absent nasal bone. N. Martin.

3256W Prenatal CMA findings in urinary system malformations fetuses with normal karyotype. H. Wang.

3257T Differentially expressed miRNAs in trisomy-21 placenta. I. Svobodova.

3258F Molecular and functional characterization of a novel mouse mutant of the Planar Cell Polarity gene, *Vangl2*. A.R. El-Hassan.

3259W Implementation of non-invasive prenatal testing by semiconductor sequencing in a genetic laboratory. A. Dheedene.

3260T Assessing the mutational spectrum of 7-dehydrocholesterol reductase and the toxicological effects of pharmacological inhibition during the prenatal period. M.R. Boland.

3261F Dynamics of mitochondrial DNA heteroplasmy in human multi-generation pedigrees. M. Su.

3262W Interphase Chromosome Profling (ICP) as a rapid, sensitive and cost-effective diagnostic tool for Amniotic fluid (AF) and Products of Conception (POC) testing. S.K. Bhattacharya.

3263T‡ Accurate detection of segmental aneuploidy in preimplantation genetic screening using targeted next-generation DNA sequencing. M. Umbarg...

3264F Study the high incidence and mechanism of chromosome 22 aneuploidy in the embryos of male Robertsonian translocation carriers. W. Wei.

3265W Novel autosomal genes linked with male infertility. D.V.S. Sudhakar.

3266T Single cell testing versus amniocentesis and NIPT: Ultra-high resolution preimplantation genetic screening for microduplication-microdeletion syndromes. G. Altarescu.

3267F Prospective analysis of maternal duplications improves positive predictive value in prenatal cell-free DNA screening. M.D. Maxwell.

3268W‡ Clinical utilization of NGS in Preimplantation Genetic Diagnosis (PGD)/Screening (PGS) for chromosomal rearrangements. S. Madjunkova.

3269T A change in fetal risk after prenatal diagnosis: A case of HbS/City of Hope. B. Clark.

3270F Identification and characterization of rare hemoglobin variants: Experience at good start genetics. J. Wilkinson.

3271W Evaluating early pregnancy loss by whole exome sequencing. H.M. Byers.

3272T A systematic evaluation of RNA quality of human conceptual tissue and its relationship to the post-mortem interval. D. O'Day.

3273F Preconception carrier testing using genome sequencing: What categories of results do patients want to receive? P. Himes.

3274W Investigating *HOXA4 and HOXA11* expression-affected implantation under the influence of N-acetyl cysteine. P. Afsharian.

3275T *Nelf* knockdown alters gene expression in GnRH neuron migration. E. Ko.

3276F Finding genes for infertility: A balanced translocation in an oligospermic male uncovers *SYCP2* overexpression. S.L.P. Schilit.

3277W First day urine of discordant monozygotic twin reflected fetal metabolic programming in metabolomics study. M.K. Thong.

3278T‡ Effects of paternal age on child's telomere length and number of *de novo* mutations. S.W.W. Wong.

3279F Performance evaluation and clinical implementation of a new paired-end MPSS approach for cfDNA based prenatal screening of common chromosome aneuploidies. V. Cirigliano.

3280W‡ Computing confidence intervals on positive ...al screening.

3256W

Prenatal CMA Findings in Urinary System Malformations Fetuses with Normal Karyotype. *H. Wang 1,2 , Z. Dong 1,2 , M. Chen 3 , L. Xiong 4,5 , Y. Song 3 , J. Lou 6 , Y. Kwok 1 , K. Choy 1,2 .* 1) Department of Obstetrics and Gynaecology, The Chinese University of Hong Kong , Hong Kong, Hong Kong; 2) Shenzhen Research Institute, The Chinese University of Hong Kong; 3) Key Laboratory of Reproductive Medicine of Guangdong Province, Key Laboratory for Major Obstetric Diseases of Guangdong Province, Key Laboratory of Reproduction and Genetics of Guangdong Higher Education Institutes. The Third Affi liated Hospital of Guangz; 4) Bao An Maternal and Child health hospital, Shenzhen, Guangdong, China; 5) Shenzhen Key Laboratory of Birth Defects (Number: ZDSYS201504301707152); 6) Dong Guang Maternal and Child health hospital, DongGuan, Guangdong, China.

Background: Fetal urinary system malformation, accounts for one third of all congenital malformations detected by routine fetal ultrasonography. Also known as congenital anomalies of kidney and urinary tract (CAKUT) are observed in 0.3 to 0.6% of live births and accounts for 40 to 50% of the etiology of chronic kidney disease in children. Chromosomal microarray analysis (CMA) has been proven to detect submicroscropic genomic imbalances prenatally because of its high-resolution and time-saving. However, prenatal studies on isolated or syndromic CAKUT and its advantage over traditional cytogenetics are limited. In this study, we aimed to investigate the diagnostic performance of CAKUT among prenatal cases with normal karyotype. **Methods:** This study recruited 108 prenatal samples, with detailed documented ultrasound fi ndings and CMA results from four prenatal diagnosis centers. Pathogenic copy-number variants (CNVs) were identifi ed based on the American College of Medical Genetics guideline. **Results:** The overall detection rate was 9.2% (10/108) for pathogenic CNVs by CMA. The extra diagnostic yield compared with karyotype was 1.7% (1/60) for the isolated CAKUT, and 18.7% (9/48) for the syndromic group. The incidence of pathogenic CNV is signifi cantly diff erent ($p<0.05$) between the isolated group and syndromic group. **Conclusion:** This study shows the improved diagnostic yield by CMA in prenatal CAKUT, compared with conventional karyotyping, with or without syndromic anomalies. Our data supports that CMA should be provided in prenatal diagnosis as a fi rst-tier test for fetus with urinary system malformations. The pathogenic CNVs identifi ed help to provide further understanding of the pathogenesis of the urinary system malformations and CNV spectrum.