Health & Translational Medicine Research Cluster (HTM)

POSTER NO. : B001-HTM
TOPIC : METHYLATION STATUS IN MALAYSIAN CHILDREN WITH BECKWITH WIEDEMANN SYNDROME AND RUSSELL- SILVER SYNDROME

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ABSTRACT

Introduction: Beckwith-Wiedemann (BWS) and Russell-Silver syndromes (RSS) cause gigantism and growth retardation respectively. Imprinting centre (IC) 1 controls the imprinting of IGF2 and H19 whereas IC2 controls the imprinting of COKN1C and KCNQ1OT1.

Methodology: Multiplex ligation-dependent probe amplification (MLPA) was used to detect the methylation status in imprinted genes of chromosome 11 for both BWS and RSS. Genomic DNA was extracted from peripheral blood. Twenty-three samples derived from BWS/RSS subjects, as well as two designated controls were blind-tested by MS-MLPA. The resulting PCR products were sent for fragment analyses.

Results and discussion: With the BWS samples, 1 had paternal UPD11 (hypermethylation in IC1 and hypomethylation in IC2), 1 had maternal UPD11 (hypomethylation in IC2) and 6 had hypomethylation in IC2. All BWS patients showed normal methylation for IGF2. IGF2 is a paternally expressed fetal growth factor gene with an important role in cancer development, while H19 is a maternally expressed non-coding RNA with possible tumour suppressor functions. With the RSS patients, all 15 showed hypomethylation at IC2 specifically at COKN1C which is a growth inhibitor. In addition, 4 patients also showed hypomethylation at IC1 and/or IC2.

Conclusion: The findings showed that the molecular basis for BWS and RSS are heterogeneous. The findings will be further confirmed by using sequencing and microsatellite analysis.

POSTER NO. : B002-HTM
TOPIC : THREE-DIMENSIONAL QUANTITATIVE EVALUATION OF FACIAL MORPHOLOGY BETWEEN ADULTS WITH UNILATERAL CLEFT LIP AND PALATE AND NORMAL PATIENTS

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ABSTRACT

Introduction: The aim of this study was to assess the quantitative values of measurements using proportion indices in the several craniofacial regions in repaired, non-syndromic complete unilateral cleft lip and palate (UCLP) patients and compared them with a normal control group using the three-dimensional (3D) image capturing system.

Methods: Three-dimensional (3D) facial images of 15 repaired UCLP and 100 non-cleft subjects aged 18 – 25 years of Malay ethnicity were recruited to the analysis. 3D images of the subjects were captured using VECTRA-3D Facial System (Canfield Scientific, Inc. USA). Eleven craniofacial proportions assessed through combination of 18 linear measurements obtained from 21 anthropometric soft tissue landmarks. These measurements were used to produce proportion indices to find the differences of morphological features between the groups, and assessed using independent sample t-test and z-scores.

Result: Significant differences were observed between both groups in 7 out of 11 craniofacial proportion indices (P < 0.05). Z-scores of 2 indices showed disproportionate; they were nasal index, which was severely supernormal and upper lip index, which was moderately supranormal. Patients with UCLP had more positive mean z-score values.

Conclusions: Clinically significant differences were found mainly in the nasolabial area where the nose and the upper lip were wider, larger or flatter in UCLP patients. Classifying the level of these disproportions is crucial in order to identify the respective elements to be surgically repaired and analyzing the morphological changes in the soft tissue of cleft in order to determine the quality of surgical treatment outcome.