Abstract No: BO-03 **Biological Sciences**

Molecular diagnosis of Williams syndrome using quantitative polymerase chain reaction (qPCR) in a cohort of Sri Lankan patients

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Williams Beuren Syndrome (WBS) is a genetic cause of congenital heart defects associated with developmental delay, hypercalcaemia and characteristic facial features. Its cause is a 1.5 to 1.8 Mb hemizygous deletion of chromosome 7q11.23 involving the loss of around 23 genes including the elastin (ELN) gene. The deletion results in copy number alterations. The aim of this study was to identify whether a group of Sri Lankan children with a clinical diagnosis of WBS could have their diagnosis confirmed or refuted by the use of genetic testing using a validated low cost method. A quantitative PCR method was evaluated for use in deletion screening. Twenty four suspected WBS cases were recruited following ethical clearance and informed consent. DNA was extracted, spectrophotometrically quantified and qPCR performed. The target used for deletion screening was the ELN gene and TES was used as the reference gene for normalization. In all assays, a 10 fold dilution series of standards, a no template control (NTC) and a negative control (NC) were included. The fold copy number change (ΔKC_t) was determined and the mean for normals (n=6) was -0.087 ± 0.11 representing no loss while the mean for previously clinically diagnosed WS patients and confirmed by either Fluorescent *in situ* hybridization (FISH) or microarray analysis (n=6) was -1.39 ± 0.086 representing the loss of one copy (deletion). Among twenty four suspected cases, 19 (79%) had an ELN gene deletion while 5 cases did not and the findings correlated strongly with the clinical suspicions. This qPCR method was able to distinguish ELN deleted cases from the non-deleted ones. The preliminary data supports this as a useful diagnostic test for WBS. Validation of this test using FISH has been performed for five patient's samples and the microarray confirmed positive which correlated with the qPCR results.

Keywords: Elastin (ELN) gene, qPCR, Quantitative polymerase chain reaction, Williams Beuren Syndrome

Acknowledgment: *Financial assistance by Research Grant AP/3/2/2014/RG/02 of University of Colombo.*