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Master Thesis Defense

Entitled

The association of methyltransferase genes with different methylation levels in fragile X Syndrome individuals

by

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Date & Venue

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Abstract

Fragile X Mental Retardation 1 (FMR1) gene produces a FMR protein (FMRP) which is known to regulate translation process in various organs. It has a significant role in neurons function, maturation and synaptic plasticity. FMR1 gene encompasses 5 - 30 CGG repeats in exon1 and the greater number of repeats has the potential to expand during gametogenesis. The expansion depending on the number of CGG repeat undergoes hyper-methylation and considered as a dynamic mutation in which the expansion increases through generation. Methylation of expanded CGG repeats result in inhibiting the transcription and silencing the gene. There are two types of affected individuals of Fragile X Syndrome, one has the methylated full mutation (no protein produced) and the other is mosaic (low amount of protein produced), which are a result of having different size expansion or both methylated and unmethylated alleles. It is not clear yet what causes the differential methylation in different individuals. Therefore we hypothesize that background gene, such as methyltransferase genes varies between individuals causing the observed epigenetic differences. The phenotypic severity depends on the number of repeats and the degree of methylation which corresponds to the concentration of FMRP. The study will focus on DNA and Histone methyltransferase genes which have an important role in genome imprinting, gene regulation, X chromosome inactivation, and embryonic development.

Keywords: Fragile X Syndrome – Mosaic – Full Mutation – DNA Methyltransferase – Histone Methyltransferase - Fragile X Mental Retardation 1