

PP 29 - Identifying *PROZ G79A* gene variant in a cohort of patients affected with young stroke in Sri Lanka

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Introduction: Ischemic stroke in younger adults is far less common than among older adults. The underlying risk factors are more diverse. Approximately 10%-15% of all strokes occur in adults ages 18-50. The diagnosis of stroke in younger adults can be challenging to differentiate from stroke mimics and to identify the cause or underlying pathogenesis. There are more than 50 monogenic diseases that can cause stroke. Given the increasing incidence of early-onset stroke, there is an objective need for early detection. The aim of this study was to identify genetic variants associated in a cohort of Sri Lankan patients affected with young stroke in Sri Lanka.

Methods: A literature review was conducted to determine the genetic variants associated with young stroke. A novel tetra-primer amplification refractory mutation system-polymerase chain reaction (T-ARMS-PCR) was performed to determine the presence of *PROZ G79A* gene variant/s known to be prevalent in the Asian region. The optimized assay was Sanger validated. The genotyping was done using 90 samples from patients who had given prior consent for such studies.

Results: Out of the ninety patients 57 were homozygous for the normal variant, 22 were heterozygotes for the pathogenic variant while 11 patients were homozygous for the pathogenic variant. The minor allele frequency for the g.79G>A genotype was 0.244 (chi squared = 11.79) according to the Hardy Weinberg equilibrium.

Conclusion: *PROZ G79A* variant is a genetic marker for symptomatic young stroke. This genotypic assay can be implemented as a sensitive, specific and simple diagnostic technique for screening associated genetic variants for young stroke.

Keywords: Key words: stroke, early-onset ischemic stroke, gene variants, ARMS-PCR, *PROZG79A*

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