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# Identification of microRNAs associated with Human Fragile X Syndrome using Next-Generation Sequencing

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Fragile X Syndrome (FXS) is caused by a mutation in the FMR1 gene which can lead to a loss or shortage of the FMR1 protein. This protein interacts with specific miRNAs and can cause a range of neurological disorders. Therefore, miRNAs could act as a novel class of biomarkers for common CNS diseases. This study aimed to test this theory by exploring the expression profiles of various miRNAs in Iranian using deep sequencing-based technologies and validating the miRNAs affecting the expression of the FMR1 gene. Blood samples were taken from 15 patients with FXS (9 males, 6 females) and 12 controls. 25 miRNAs were differentially expressed in individuals with FXS compared to controls. Levels of 9 miRNAs were found to be significantly changed (3 upregulated and 6 downregulated). In Patients, the levels of hsa-miR-532-5p, hsa- miR-652-3p and hsa-miR-4797-3p were significantly upregulated while levels of hsa-miR-191- 5p, hsa-miR-181-5p, hsa-miR-26a-5p, hsa-miR-30e-5p, hsa-miR-186-5p, and hsa-miR-4797-5p exhibited significant downregulation; and these dysregulations were confirmed by RT- qPCR. This study presents among the first evidence of altered miRNA expression in blood samples from Patients with FXS, which could be used for diagnostic, prognostic, and treatment purposes. Larger studies are required to confirm these preliminary results.

**References**

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