Title: MyomiRs, UTRN and TGFβ Blood-Transcriptome shifts detected in DMD patients and Female Carriers

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Duchenne Muscular Dystrophy (DMD) is a monogenic X-linked recessive neuromuscular disorder that affects dystrophin gene leading to production of truncated non-functional dystrophin protein. Consequently, the absence of this protein leads to progressive muscle degeneration which eventually causes ambulatory loss, respiratory complications and heart failure. The loss of dystrophin is usually accompanied by the alteration of the levels of some muscular proteins like Utrophin, a homologue of dystrophin protein. Because mRNA content of the peripheral blood is known to be affected according to the transcriptome of other body tissues, we evaluated the expression of Utrophin mRNA in blood cells in DMD patients and other family members. Our results showed that UTRN level in DMD patient, mothers and sisters is significantly lower than the control subjects. In fact, our findings support the idea of UTRN-based therapeutic approaches that mainly aims at increasing the levels of exogenous UTRN to dystrophic muscles which consequently alleviate the symptoms of the patients. Transforming growth factor beta, TGF-β is a cytokine that plays a chief role in fibroblasts' stimulation and in the regulation of extracellular matrix. TGF-β is also involved in the process of muscle repair through activating satellite cells and the formation of connective tissue and immune response regulation. Our data revealed that 53% of the patients' group had low levels of TGFβ in the leukocytes. The observed downregulation of TGFβ in some DMD patients and carriers might be due to progressive muscular damage that can be confirmed by other imaging techniques like muscle magnetic resonance imaging (MRI). Also, our results indicate that serum miRNAs have the ability to diagnose DMD patients and more importantly has the ability to detect female carriers.

Biography



Nahla obtained the B.Sc degree in Biotechnology in 2010 from faculty of science, Cairo University, Egypt. she was selected to work as teaching assistant in Biotechnology department. In 2015, she got the M.Sc degree in Biotechnology through studying Acute Myeloid Leukemia, after which she was assigned to work as lecturer assistant in the same department. In 2015, she was enrolled in the American University in Cairo to pursue PhD degree.

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