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THE EXPRESSION OF *SMN1*, *MART3*, GLE1 AND FUS GENES IN SPINAL MUSCULAR ATROPHY

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Introduction: Spinal muscular atrophy (SMA) is one of the most common genetic causes of death in infants due to a mutation of the motor neuron 1 (*SMN1*) gene. The *SMN1* gene encodes for the multifunctional SMN protein. SMN has been shown to be implicated in pre- mRNA splicing, mRNA transport and translational control. Also other mRNA processing proteins, such as GLE1, Marten (*MART3*) and Fused in Sarcoma (FUS), have been linked to neurodegenerative diseases. The aim of the study was to determine the expression of SMN, GLE1, *MART3* and FUS genes in cell lines of the fibroblasts derived from SMA patients and normal controls.

Material & Methods: Total RNA was extracted from purchased fibroblasts acquired from three SMA type I patients and fibroblasts of three age-matched healthy controls. The RNA was then subjected to qPCR analysis using primers specific for the GLE1, *MART3*, FUS and *SMN1* genes vs. GAPDH as internal control gene.

Results: *SMN1* mRNA levels were at least x10 lower in fibroblasts of SMA patients compared to controls. Gle1 and *MART3* gene expression was x2 downregulated whereas FUS mRNA levels appeared to be x3 upregulated in SMA cells when compared to controls. We found a high correlation between FUS gene expression level to the *SMN1* at gene expression level of fibroblast cell lines of SMA type I patients ($r=0.994$, $p<0.0001$).

Conclusions: Our preliminary data show an intriguing expression profile of Gle1, *MART3* and FUS genes in SMA, and suggest a critical role of FUS protein in the SMA pathogenesis.

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