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Characteristic of the spinal muscular atrophy cell model

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Motivation and Aim: After the induced pluripotent stem cells were obtained by the Yamanako group, experiments on the development of cellular models of disease have become widespread in medical biology. Here we performed a comparative description of a previously obtained cellular model of spinal muscular atrophy (SMA) at different stages of differentiation. We compared the expression of well-known and newly founded by genome-wide linkage analysis genes, involved in molecular pathogenesis of SMA.
Methods and Algorithms: We utilized induced pluripotent stem cells, obtained from conditionally healthy patient and patients with SMA I and II types [1]. Using the open Internet resource BLAST we selected primer pairs to estimate by Real-time PCR the number of *SMN1, SMN2, PLS3, SLC23A2, NCALD, RPL6, CDKA2P1* genes [2–4], which products are involved in the development of SMA. Statistical processing of the results were carried out using the non-parametric Kruskal-Wallis criterion for independent groups using the Statistica 64. Differences were considered significant when the probability parameter value was $p<0.05$.
Results: We shown a decrease in the expression level of the full-sized *SMN* transcript at all stages of neural differentiation in cell models with the SMA phenotype of types I and II in comparison with control. An increase in the level of the incomplete *SMN* transcript was also detected. We shown a change in the transcription of modifier genes: in case of SMA type I, the number of *RPL6* transcripts increases in mature motor neurons, and the expression level of *PLS3, NCLD, CDK2AP1* decreases.
Conclusion: The earlier obtained cell model of SMA complies well-known literary characteristics of mature motor neurons with SMA phenotype and can be used for further molecular applications. Newly founded by genome-wide linkage analysis genes (*RPL6* and *CDK2AP1*) can be also involved in pathogenesis of SMA.
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