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A novel approach to SMN protein-based evaluation as biomarker in spinal muscular atrophy

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Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease caused by deletion or mutation within the survival motor neuron 1 (SMN1) gene, leading to progressive limb and trunk muscle weakness associated with muscle atrophy. SMA results in the reduction of survival motor neuron (SMN) protein expression and SMN complex in the anterior horn of the spinal cord and other tissues. Recently, SMN protein has been used as a therapeutic biomarker in recent SMA clinical trials using enzyme-linked immunosorbent assay (ELISA). In our study, we investigated whether imaging flow cytometry (IFC) can be a viable source of quantification and cellular localization on the SMN protein. Using IFC (Merck-Millipore, Germany), we first demonstrated that IFC can successfully identify different expression patterns and subcellular localization of SMN protein in healthy human and SMA patient-derived fibroblasts. In addition, we could also significantly evaluate the therapeutic effects of SMN protein expression by valpronate-treated SMA patient-derived cells relative to those from non-treatment SMA cells in vitro ($p<0.05$). Therefore, our study provides a strong evidence to develop a new evaluation method of SMN protein using IFC in SMA clinical trials.

Biography

Masayuki Arakawa is a member of Research Committee of Spinal Muscular Atrophy (SMA), the Ministry of Health, Labour and Welfare of Japan in 2010. He has also done the clinical study of SMA supported by the Practical Research Project for Rare/Intractable Diseases from Japan Agency for Medical Research and Development (AMED) in 2014. Recently, his work is focused on a new technology for the SMN protein analysis in SMA clinical study.

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