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Imaging flow cytometry for identifying differences in SMN protein expression between spinal muscular atrophy patients and normal subjects

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Background: Childhood spinal muscular atrophy (SMA) is a common autosomal recessive disorder caused by mutations of the survival motor neuron 1 (SMN 1) gene, leading to progressive muscle weakness. A major goal of disease-modifying therapy is to increase SMN protein expression. There is a need for sensitive methods of quantifying SMN protein in accessible tissues. We tested the sensitivity and utility of imaging flow cytometry (IFC) for identifying differences in SMN protein expression between SMA patients and normal subject using cultured Epstein–Barr virus transformed B cells.

Materials & Methods: Following the immortalization of peripheral blood lymphocytes from a human healthy control subject and 2 SMA type 1 patients with 2 and 3 copies of SMN2, respectively, we utilized IFC analysis to identify significant differences in SMN expression. A bright detail intensity (BDI) analysis was used to investigate differences in the cellular localization of SMN protein.

Results: SMN expression was significantly decreased in SMA patient-derived cells relative to those derived from a healthy control subject. Moreover, SMN expression correlated with the clinical severity of SMA according to SMN2 copy number. The cellular accumulation of SMN protein was also significantly decreased in SMA patient-derived cells relative to those derived from a healthy control subject.

Conclusions: We developed an IFC method of SMN protein evaluation using human lymphoblasts. The benefits of IFC for lymphoblast analysis include its capacities for visualizing cell morphology, and evaluating the accumulation, localization, and expression of a target protein.

Biography

Reiko Arakawa is an Assistant Professor at the Institute of Medical Genetics, Tokyo Women's Medical University. She has also been participating in a clinical study of SMA supported by the Practical Research Project for Rare/Intractable Diseases from Japan Agency for Medical Research and Development (AMED) since 2014. Recently, her research has focused on new technology for SMN protein analysis in clinical SMA studies.

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