Genetic Analysis of CD16+ Monocyte, CD16- Monocyte and CD4+ T Lymphocyte Cells to Identify Novel Gene Expression Signatures and Develop a Diagnostic Tool for Systemic Lupus Erythematosus

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Systemic lupus erythematosus (SLE) is an incurable chronic autoimmune disease in which the immune system attacks its tissues, causing widespread inflammation and organ damage. Due to the lack of a single test for SLE diagnosis, multiple generalized methods are currently employed by doctors to detect the presence of the disease. The purpose of this study was to evaluate gene expressions in CD16+ monocyte, CD16- monocyte and CD4+ T lymphocyte cells to identify gene expression signatures unique to SLE to improve diagnostic processes and outcomes for patients with SLE, or at risk of acquiring SLE. Raw gene probe data was obtained from the Gene Expression Omnibus. Statistical significance was determined by calculating p-values for gene probe expressions between healthy patients and those with SLE, covering all 54,675 gene probes and three cell types. The gene probes with the most significant statistical difference were identified and evaluated. Excluding transcription genes, the top three gene probes identified in the study were associated with the ATP6V0C, UBA1, and TGFB1 genes. Quantile-quantile plots confirmed statistical appropriateness for genetic analysis. Further evaluation determined that the ATP6V0C, UBA1 and TGFB1 genes associated with the CD16- monocyte cell type represents a unique and novel gene expression signature for the identification of SLE in patients. Gene probe expression ranges were established for these gene probes which serve as a diagnostic tool for the presence, or genetic predisposition, of SLE. This diagnostic tool can detect SLE in a single blood sample, improving diagnostic outcomes for patients and reducing healthcare costs.

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