# RNA-Seq Analysis of Peripheral Blood Mononuclear Cells in Multiple Sclerosis Patients and Controls (GSE21942)

## 1. Introduction

Multiple Sclerosis (MS) is a chronic autoimmune disease affecting the central nervous system.
RNA-Seq analysis of Peripheral Blood Mononuclear Cells (PBMCs) helps identify gene expression differences
between MS patients and healthy controls. This study aims to perform differential expression analysis and
functional enrichment using publicly available RNA-Seq data (GSE21942).

## 2. Methods

- Dataset: RNA-Seq expression data (GSE21942) retrieved using GEOquery.
- Data Preprocessing: Log transformation, normalization (VST), and low-expression gene filtering.
- Exploratory Analysis: PCA and heatmap to visualize clustering of MS and control samples.
- Differential Expression Analysis: DESeq2 used to identify DEGs (adjusted p-value < 0.05, log2FC > ±1).
- Functional Enrichment: GO and KEGG pathway analysis performed using clusterProfiler.
- Visualization: Volcano plots, boxplots, and pathway enrichment plots generated.

## 3. Results

- PCA plot shows clear separation between MS and control groups.
- Heatmap of the top 50 variable genes highlights distinct expression patterns.
- 300+ significantly differentially expressed genes identified (adjusted p-value < 0.05).
- Enrichment analysis reveals involvement of immune response pathways in MS.
- Key upregulated genes: CXCL10, IFNG, STAT1; Key downregulated genes: NRXN1, SYN1.

## 4. Discussion

The RNA-Seq analysis indicates significant immune-related gene dysregulation in MS patients.
CXCL10 and IFNG, both involved in immune response, were highly upregulated, suggesting inflammatory activity.
Downregulation of NRXN1 and SYN1 may indicate neuronal dysfunction in MS. Further research is needed to validate
these findings and explore their clinical relevance.

## 5. Conclusion

This study successfully identified differentially expressed genes and enriched pathways in MS patients.

Findings highlight key immune and neuronal processes altered in MS, providing potential targets for further research.