Validation of C8ORF4 mRNA Expression in Deep Infiltrating Endometriosis via Bioinformatics Screening and Correlation Analysis with Systemic Immune-Inflammation Index

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Abstract

Obiective To identify the key genes of deep infiltrating endometriosis (DIE) using bioinformatics and machine learning algorithms, and to investigate their correlations with systemic immune-inflammatory indices (SII) in patients with DIE. *Methods* The GSE141549 dataset from the Gene Expression Omnibus (GEO) was analyzed to identify Differentially Expressed Genes (DEGs) between the DIE group and the non-DIE group. Using bioinformatics methods and three machine learning algorithms (LASSO, Random Forest, and Support Vector Machine), the key feature genes were identified.Subsequently, the expression levels of these key genes in normal endometrial tissues (NE), DIE, superficial peritoneal endometriosis (SPE), and ovarian endometrioma (OMA) were evaluated. Moreover, the infiltration profiles of 22 immune cell types in DIE patients and their correlations with key genes were analyzed. Clinical data were collected from patients diagnosed with OMA alone (control group) and those with OMA accompanied by DIE (observation group). Statistical analyses were performed to determine the independent risk factors for DIE. Additionally, the relative mRNA expression levels of key genes in OMA, DIE, and NE tissues were measured using RT-qPCR, and their correlations with the corresponding independent risk factors in patients were assessed. *Results* Through bioinformatics analysis and machine learning methods, 133 differentially expressed genes were screened out, among which C8ORF4 was identified as the key regulatory gene of DIE. In the dataset, the expression level of C8ORF4 in DIE was significantly higher than that in OMA, SPE, and NE groups ($P < 0.05$). Immune infiltration analysis revealed significant differences in various immune cell populations between the DIE group and non-DIE groups, with C8ORF4 showing a close association with multiple immune cells. From January 2021 to December 2024, data were collected from 193 patients in the control group and 76 patients in the observation group.Statistical analyses indicated that fibrinogen and SII were independent risk factors for DIE. RT-qPCR results demonstrated that the level of C8ORF4 mRNA in DIE tissues was significantly higher than that in NE tissues ($P < 0.05$). Correlation analysis further showed that SII was positively correlated with the relative expression of C8ORF4 mRNA ($P < 0.05$). *Conclusion* C8ORF4 has been identified as a potential key gene associated with DIE. The relative expression of C8ORF4 mRNA is positively correlated with SII levels, which may provide a new potential target for the diagnosis and treatment of DIE.

Keywords

Deep Infiltrating Endometriosis, C8orf4, Systemic Immune-Inflammatory Indices, Machine Learning Algorithms