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Identification of C1QTNF2 and Its Combination with AASS As a Novel Biomarker of Uterine Cancer: RNA-Sequencing and Machine Learning Analysis

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ABSTRACT

Introduction: Uterine Cancer is known as the sixth most common cancer in the world. In 2020, the number of individuals with uterine cancer worldwide was 417,367. It is predicted that in 2023, almost 66,200 individuals in the United States will be diagnosed with Uterine cancer. Further, it is anticipated that the number of people who have this cancer will increase by 52.7% by 2040, and the death rate will reach 70.6%. Hence, finding new methods for early detection and screening of this type of cancer has gained much attention. Identification of diagnostic genes associated with uterine cancer can be productive. Advances in bioinformatics and machine learning techniques provide powerful tools for assessing numerous genomic data and discovering potential biomarkers indicative of disease states. In this study, we explored the application of this prospective procedure to identify key genes involved in uterine cancer. This approach may present a promising strategy to enhance the treatment of patients.

Methods and Materials: RNA expression profiling of uterine cancer patients was performed using data obtained from the Genomic Data Commons database. Survival curve evaluation was performed using COMBIO-ROC analysis to identify diagnostic biomarkers. Compared with these, ROC curves, as other potential biomarkers predictive of patient outcomes, were assessed. A vast analysis was conducted using advanced machine-learning algorithms to detect significant differentially expressed genes associated with the progression of uterine cancer.

Results: Our research showed that differential expression of the *C1QTNF2* gene significantly contributed to cancer cell expansion. Also, mutual evaluation of both *C1QTNF2* and *AASS* gene expression was found to be relevant to uterine cancer pathogenesis. The evaluation metrics demonstrated the efficacy of the machine learning model, with a minimal mean squared error of 5.1096067E-5 and a root mean squared error of 0.007, indicative of accurate predictions. The R-squared value of 0.99 underscores the ability of the model to explain a substantial portion of the variance in the data. *C1QTNF2* and a combination of *C1QTNF2* and *AASS* can be considered diagnostic markers with sensitivity, specificity, and area under curve values of 0.97, 1.00, and 0.99, respectively.

Conclusion and Discussion: Through combined machine learning and bioinformatics procedures, we identified the *C1QTNF2* and the combination of *AASS* and *C1QTNF2* genes as diagnostic tools for uterine malignancies. This finding improves our perception of uterine cancer molecular pathogenesis. Identifying new uterine cancer biomarkers has promise for adequate care, improved prognosis, and early diagnosis. Further studies are required to confirm the clinical ability of this identified gene as screening, diagnostic, and monitoring tools for uterine-related conditions.

Keywords: Biomarkers, Machine learning, Uterine neoplasms

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