TRANSCRIPTOME PROFILING AND EXAMINATION OF INDIVIDUALS DIAGNOSED WITH ESOPHAGEAL SQUAMOUS CELL CARCINOMA ORIGINATING FROM KAZAKHSTAN

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Esophageal cancer stands as the eighth most prevalent cancer globally and the sixth most prevalent in Kazakhstan, with esophageal squamous cell carcinoma (ESCC) as its primary histological subtype, often diagnosed in advanced stages. This project aimed to unveil the genetic underpinnings of ESCC by examining differentially expressed genes (DEGs) via whole-transcriptome sequencing of Kazakhstani patients. Tissue samples from 22 individuals undergoing Ivor-Lewis esophagectomy at the Oncology Center in Astana were collected. Utilizing STAR software and DESeq2 package, DEGs were mapped and defined, followed by functional analysis using various R packages.

The study encompassed 13 males and 9 females, averaging 65.72 ± 8.26 years old, with 86% diagnosed at advanced stages T3-T4. Analysis of tumor and normal esophageal tissues unveiled 6689 DEGs, including 2056 upregulated and 4633 downregulated genes (adjusted p-value <0.05). Additionally, 42 up-regulated and 2 down-regulated KEGG pathways were identified (p-value <0.05). The top 300 DEGs were mapped to a protein-protein interaction (PPI) network, and functional enrichment analysis revealed three modules comprising closely connected genes. Within our patient cohort, ESCC with moderate dysplasia emerged as the most prevalent histologic subtype, associated with poor prognosis. High-throughput sequencing techniques facilitate the identification of molecular pathways implicated in esophageal carcinogenesis, potentially revolutionizing diagnosis and treatment strategies.

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