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RNA sequencing revealed potential biomarkers in Iranian children with B-cell acute lymphoblastic leukemia

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Abstract

Backgrounds: B-ALL (B-cell acute lymphoblastic leukemia) ranks amongst the most common malignancies in pediatric patients, causing increased proliferation of immature lymphoid cells and resulting in reduction of normal bone marrow cells. As the search for approaches resulting in earlier diagnosis is still ongoing, novel biomarkers present an attractive target for studies as they may help in early detection and improve clinical outcomes. Thus, RNA sequencing (RNA-seq), a powerful technique for transcriptome profiling, presents an ideal tool for biomarker discovery. Here, we utilize the RNA-seq method to obtain a transcriptome profile of pediatric B-ALL patients to identify potential biomarkers.

Materials and Methods: Bone marrow aspiration samples were obtained from 10 newly diagnosed B-ALL patients and 2 Immune thrombocytopenic purpura (ITP) as non-malignant controls. Then, using Ficoll density gradient centrifugation, mononuclear cells were isolated, followed by total RNA extraction. Paired-end RNA sequencing (~100 million reads per sample) was performed on a NovaSeq6000 instrument. Raw RNA-seq data was processed and analyzed using bioinformatics tools. Our raw RNA-seq data are publically available at BioProject under PRJNA589314 accession.

Results: 1216 genes were upregulated and 920 downregulated as compared to the control group ($|\log2FC| \geq 2$, padj < 0.05). Functional analysis and protein-protein interaction networks revealed *ESR1*, *NRIP1*, *MYSM1*, *BCL7A*, *UCKL1*, *SPRING1* and *UBASH3B* may act as suitable biomarkers in pediatric B-ALL patients.

Conclusion: *ESR1*, *NRIP1*, *MYSM1*, *BCL7A*, *UCKL1*, *SPRING1* and *UBASH3B* may act as potential biomarkers in pediatric B-ALL patients. Further studies on larger datasets are necessary for validating results presented in this study.

Keywords: RNA sequencing, Transcriptome profiling, B-ALL