



INNOVATIVE APPROACHES IN THE DIFFERENTIAL DIAGNOSIS OF ALL SUBTYPES: INVESTIGATION OF *DE NOVO* VARIANTS BY RNA SEQUENCING

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THESIS ABSTRACT

Genetic alterations, particularly translocations, play a significant role in the development of acute leukemias and can be used diagnostically in certain subtypes. Translocations such as the Philadelphia chromosome (BCR-ABL fusion) activate protein kinases or transcription factors. Philadelphia-like ALL is a high-risk subgroup with poor prognosis and a gene expression profile similar to Ph+ ALL. Identifying and classifying this subtype is critical for appropriate treatment.

In this study, the aim was to identify genes specifically expressed in Ph-like ALL cases and investigate *de novo* variants. To achieve this, total RNA sequencing analysis was performed after isolating RNA from bone marrow samples of Ph+ and Ph- adult ALL patients. Bioinformatics analyses identified two Ph- patients with expression patterns most similar to the Ph+ group. DEG analysis conducted on the two patients with expression patterns resembling Ph+ ALL revealed 28 upregulated and 128 downregulated genes based on an adjusted p-value cutoff of 0.05.

APPLICATION AREAS OF THE THESIS RESULTS

In the thesis study, it is aimed to develop a gene panel suitable for clinical use in the differential diagnosis of Ph-like ALL after validating the data obtained from RNA sequencing analyses with RT-PCR analysis. This gene panel will enable targeted regulation of treatment protocols for individuals in the patient group and contribute to the development of personalized treatment strategies.

ACADEMIC ACTIVITIES

Gülşah Çeçener, Ebrucan Bulut, Rumeysa Fatma Balaban, **Nuseybe Huriyet**, Ufuk Ünal, Havva Tezcan Ünlü, Hülya Öztürk Nazlıoğlu, Ünal Egeli, Kazım Şenol, Seyit Ali Volkan Polatkan, Erdem Çubukçu, Mustafa Şehsuvar Gökgöz. Üçlü negatif meme kanseri moleküler alt tiplerinin belirlenmesine yönelik geliştirilen multi-gen paneli ile hedefe yönelik tedavi seçiminin değerlendirilmesi. 16th National Breast Diseases Congress. September 16-18, 2022, Cyprus.

Havva Tezcan Ünlü, Furkan Sarıdaş, Batuhan Topuz, Ufuk Ünal, Gülşah Çeçener, Özlem Taşkapılıoğlu, **Nuseybe Huriyet**, Rumeysa Fatma Balaban, Ebrucan Bulut, Ünal Egeli, Ömer Faruk Turan. Multipl Skleroz Tanılı Hastalarda, FokI (rs225870), TaqI, (rs7975232) ve ApaI (rs7975232) Vitamin D Reseptörü (VDR) Gen Polimorfizmlerinin İncelenmesi. 18th Uludağ Neurology Days. March 09-12, 2023. Bursa

E. Bulut, R.F. Balaban, **N. Huriyet**, M.M. Önal, U. Unal, H. Tezcan, G. Gokalp, U. Egeli, V. Polatkan, M.S. Gokgoz, G. Çeçener. Developing the AI program for TNBC subtyping. Esmo Breast Cancer 2023, 11.05.23-13.05.23, Berlin.

Eryılmaz, I. E., Çolakoğlu Bergel, C., **Huriyet N.**, Aröz, B., Çeçener, G., & Egeli, Ü. (2023). Luteolin disrupts cell health as a pro-oxidant agent against PC-3 metastatic castration-resistant prostate cancer cells, 9th BAU Drug Design Congress (pp.1). İstanbul, Turkey

Nuseybe Huriyet, Ecem Efendi Erdem, Gülşah Çeçener, Melike Sezgin Evim, Adalet Meral Güneş. Pediatrik Ph-Negatif ALL Vakalarında IKZF1Plus ile Relaps'ın Değerlendirilmesi. 18th Congress of Medical Biology and Genetics. October 26-29, 2023, Ankara, Turkey.

KEY WORDS

- ✓ Acute Lymphoblastic Leukemia
- ✓ RNA Sequencing
- ✓ Transcriptomic Analysis
- ✓ Gene Panel
- ✓ Philadelphia Like ALL

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