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Session 3 – Poster presentation *S3-12*

IMPROVING THE DIAGNOSTICS OF RARE LUNG DISORDERS USING A UNIQUELY DESIGNED PIPELINE FOR ANALYSIS OF NGS DATA

Andjelkovic Marina, Skakic Anita, Stevanovic Nina, Parezanovic Marina, Komazec Jovana, Klaassen Kristel, Spasovski Vesna, Stojiljkovic Maja, Pavlovic Sonja

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Rare lung diseases (RLDs) are a group of diseases that individually affect one in 2,000 people, with an estimate that about 80% of RLDs have a genetic origin. Despite the variations among RLDs in clinical characteristics and manifestations, most of these diseases similarly damage the lungs, making diagnosis difficult. The utility of NGS technology in RLDs for diagnostic purposes allows a better understanding of the genetic background, however, the identification and classification of disease-causing variants are challenging. Further, numerous VUS (variants of uncertain significance) that cannot be precisely defined and classified are produced. The main goal of this study was to create a unique guideline that will enable the standardization of the assessment of novel genetic variants in RLDs causative genes. The designed pipeline consists of three main steps: (1) sequencing, detection, and identification of genes/variants, (2) classification of variants, and (3) characterization of variants using in silico structural and functional analysis. The pipeline validation was performed through the analysis of variants detected in a disease-causing and candidate genes of one of the RLDSs, and detected VUS variants have gained diagnostic significance. The application of this pipeline resulted in the identification and classification of novel variants, through analysis at the transcriptional, translational, and posttranslational levels, and led to accurate diagnosis.

Keywords: pipeline, rare lung diseases (RLDs), classification of variants, characterization of variants, sequencing, detection, identification of genes/variants

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