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**EVOLIUCINĖ MEDICINA:
ŽMOGAUS RAIDOS PERSPEKTYVOS IR SVEIKATA
BESIKEIČIANČIOJE APLINKOJE**

**EVOLUTIONARY MEDICINE:
PERSPECTIVES ON HUMAN DEVELOPMENT AND HEALTH
CONSEQUENCES IN A CHANGING ENVIRONMENT**

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Vilniaus universiteto Medicinos ir sveikatos mokslų
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PHENOTYPING PRETERM BIRTH HETEROGENEITY: COMPOSITE NEONATAL ADAPTATION IN A LITHUANIAN COHORT

Svetlana DAUENGAUER-KIRLIENE¹, Alina URNIKYTE², Ingrida DOMARKIENE², Ausra MATULEVICIENE¹

¹Department of Human and Medical Genetics, Institute of Biomedical Sciences, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

²Populational Genomics Laboratory, Translational Health Research Institute, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

Contact e-mail: svetlana.dauengauer-kirliene@mf.vu.lt

Background and Aim: Preterm birth (PTB) remains difficult to predict and biologically heterogeneous, limiting the reproducibility of genotype–phenotype studies based on binary clinical outcomes. We aimed to develop and apply a quantitative composite neonatal phenotype — the Major Neonatal Outcome (MNO) score — to improve characterisation of preterm neonatal adaptation and support integrated genotype–phenotype analyses.

Materials and Methods: The MNO score was developed in a retrospective cohort of 208 preterm neonates using weighted multivariable regression of clinically relevant neonatal outcomes and applied to a study cohort comprising 71 preterm neonates and 25 term-born controls. Parent–offspring trios included 23 preterm neonates in the final *de novo* variant (DNV) analysis. Genomic data were obtained using SNP arrays and short-read whole-genome sequencing. Rare variants and DNVs were identified and evaluated in relation to MNO score variability.

Results: The MNO score integrated gestational age, birth weight < 5th percentile for gestational age, cord blood IL-6 levels, premature rupture of membranes, prolonged latency, and induction of birth into a continuous clinical severity phenotype. The model demonstrated good discrimination for MNO (AUC = 0.894, sensitivity = 0.90, specificity = 0.76). No significant association was observed between total DNV burden and MNO score ($p = -0.23$, $p = 0.28$). However, clinically severe cases with high MNO scores harbored rare coding and regulatory DNVs in developmental signaling, structural integrity, adhesion, and cellular process pathways, including *ROR2*, *MUC17*, *DNAH3*, and *ZAN*. Multiple regulatory variants frequently co-occurred within individuals rather than acting independently.

Conclusions: These findings suggest that quantitative composite phenotyping may provide a more informative framework for studying genomic heterogeneity in PTB than categorical outcome definitions alone. The MNO score may enable more refined genotype–phenotype analyses in future large-scale genomic studies of PTB.

Keywords: composite outcome score, *de novo* variants, genotype–phenotype integration, neonatal phenotype, preterm birth