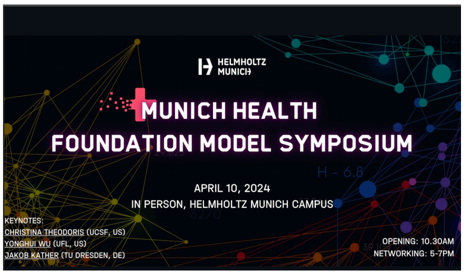

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A Machine learning based model for Multitrait Allelic Series Scoring

Allelic series describe how the range of mutations within a single gene impacts gene function, and their study can help identify new candidate targets[1]. Despite the advances in machine learning strategies to model the expected impact of variants aggregating various sources of variant annotation [2][3], methods describing effects across multiple phenotypes are missing. To fill this gap, we introduce a MultiTraitAlleleScore(MTAS) model, a machine learning based approach leveraging a comprehensive set of variant annotations to predict the effects of allelic series across multiple traits. In an application to whole-exome sequencing data (from the UK Biobank[4]), we show that the MTAS model can recover more genes compared to standard rare variant analysis methods and effectively describe allelic series effects across multiple traits.

1. McCaw et al.: https://www.sciencedirect.com/science/article/pii/S0002929723002410
2. Monti et al.: https://www.nature.com/articles/s41467-022-32864-2
3. Clarke, Holtkamp et al.: https://doi.org/10.1101/2023.07.12.548506
4. UKBB:https://doi.org/10.1038/s41586-018-0579-z

Primary authors: NAPPI, Antonio (Helmholtz Munich); CASALE, Francesco Paolo (HELMHOLTZ ZENTRUM MUENCHEN - GMBH)

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