



COPD: can genetic Background inform about disease heterogeneity?

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Abstract

People with COPD vary substantially on their pulmonary (e.g., airway obstruction) and extra-pulmonary (e.g., symptoms, functional status) manifestations. The aim of this study was to relate this high heterogeneity to the patient's genetic Background, namely focusing on polymorphisms associated with COPD and COPD-associated phenotypes and features.

Summary statistics for COPD and COPD-associated phenotypes and features (emphysema, FEV₁, FEV₁/FVC, smoking, BMI, asthma, airway responsiveness, coronary heart disease, blood pressure, pulmonary artery enlargement, resting heart rate and resting oxygen saturation) were obtained from GWAS Catalog (<https://www.ebi.ac.uk/gwas/> accessed in August 2021). A local COPD cohort was genotyped using Global Screening Arrays (GSA-Illumina) and polygenic risk scores were calculated per phenotype/feature. A cluster analysis was then carried out to determine how patients would group according to their assessed genetic risks.

The study currently includes 255 participants with COPD (68 [61, 74] years old; 79.61% male; FEV₁/FVC 53.02 [41.24, 61.94]). Our preliminary results show that people cluster into 3 main groups based on their genetic risk for emphysema, followed by COPD, whilst their clinical characteristics remained similar among groups. Future work is currently being conducted to further explore these clusters and perform their validation.

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Footnotes

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