EUROPEAN RESPIRATORY journal
FLAGSHIP SCIENTIFIC JOURNAL OF ERS

COPD: can genetic Background inform about disease heterogeneity?

R F Santos Marçalo, G Rodrigues, T Córdova, V Neves, S Neto, M Pinheiro, M A S Santos, P Simão, A Mendes, L Andrade, A Marques, G R Moura
European Respiratory Journal 2022 60: 1811; **DOI:** 10.1183/13993003.congress-2022.1811

Article | Info & Metrics

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; FEV_1/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.

This work was funded by FEDER funds through COMPETE 2020, Operational Programme for Competitiveness and Internationalization (POCI-01-0145-FEDER-028806), CENTRO 2020 (CENTRO-01-0246-FEDER-000018; CENTRO-08-5864-FSE-000039) and by Fundação para a Ciência e a Tecnologia (UI/BD/151337/2021). The iBiMED is supported by FCT funds under UIDP/04501/2020.

COPD Genomics Genetics

Footnotes

Cite this article as *Eur Respir J* 2022; 60: Suppl. 66, 1811.

This article was presented at the 2022 ERS International Congress, in session "-".

Previous

Back to top

Vol 60 Issue suppl 66 Table of Contents

Table of Contents
Index by author

Email
Request Permissions
Citation Tools
Share

Jump To

Article
Info & Metrics

Tweet
Gosto 0

More in this TOC Section

Related Articles