



# Genetic profile and patient-reported outcomes (PROs) in COPD: a systematic review

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## Abstract

Chronic Obstructive Pulmonary Disease (COPD) may impact differently on patients at similar grades, suggesting that factors other than lung function may influence patients' experience of the disease. Recent studies have found associations between genetic variations and patient-reported outcomes (PROs). Identifying these associations might be fundamental to predict the disease progression and develop tailored interventions. This systematic review aimed to identify the genetic variations associated with PROs in COPD.

Pubmed, Scopus and Web of Science were searched until May 2016 (PROSPERO Reg. CRD42016041639). Additional searches were conducted scanning the reference list of the articles. 2 independent reviewers assessed the quality of studies using the Q-Genie checklist. This instrument is composed of 11 questions, each subdivided in 7 answer options from "1-poor" to "7-excelent".

10 studies reporting 3 PROs in association with genes were included. Studies were rated as "good quality" (n=3) and "moderate" (n=7). The most reported PRO was frequency of exacerbations (n=8/10), which was more often associated with MBL2 gene variants. Other PRO's were depressive symptoms (n=1/10) and respiratory health status (n=1/10) which were associated with SLC6A4 and GATA-4 gene variants, respectively.

Although a limited number of PRO's have been related to genetic variations, findings suggest that there is significant association between specific gene variants and the number of exacerbations, depressive symptoms and health status. Further research is needed to confirm these findings and assess the genetic influence on other dimensions of patients' lives, since it may enhance our understanding and management of COPD.

## Footnotes

This abstract was amended on 27 December 2017 to correct an error in the author list.

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## We recommend

Exacerbation of respiratory symptoms in COPD patients may not be exacerbations of COPD.  
Bianca Beghé et al., European Respiratory Journal

Polymorphic variation in surfactant protein B is associated with COPD exacerbations.  
M G Foreman et al., European Respiratory Journal

Relationship between depression and exacerbations in COPD.  
J K Quint et al., European Respiratory Journal

Exacerbations of chronic obstructive pulmonary disease.  
B R Celli et al., European Respiratory Journal

Global Strategy for the Diagnosis, Management, and Prevention of Chronic Obstructive Lung Disease 2017 Report: GOLD Executive Summary  
David M.G. Halpin et al., European Respiratory Journal

Association of chronic obstructive pulmonary disease and postresection lung cancer survival: a systematic review and meta-analysis  
Lan-Eng Tan et al., J Investig Med

Genetic mannose binding lectin deficiency is associated with airway microbiota diversity and reduced exacerbation frequency in COPD  
Gisli G Einarsson et al., Thorax

Exome-wide analysis of rare coding variation identifies novel associations with COPD and airflow limitation in MOCS3, IFIT3 and SERPINA12  
Victoria E Jackson et al., Thorax

Anaemia secondary to erythropoietin resistance: important predictor of adverse outcomes in chronic obstructive pulmonary disease  
Rahul Kumar Sharma et al., Postgrad Med J

Depression in Patients With Chronic Obstructive Pulmonary Disease  
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