

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

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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 – defined as a set of health outcomes directly reported by patients). 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. Databases were searched until May 2017. 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, subdivided in 7 options from 1 poor-7 excellent. 13 studies reporting 5 PROs in association with genes were included. Studies were rated as “good quality” (n=8) and “moderate” (n=5). The most reported PRO was frequency of exacerbations (n=7/13), which was mainly associated with MBL2 gene variants. Other PRO's were health-related quality of life (HRQOL) (n=4/13), depressive symptoms (n=1/13), exacerbation severity (n=1/13) and breathlessness, cough and sputum (n=1/13), however, commonly associated with other genetic variants. 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/severity of exacerbations, depressive symptoms and HRQOL. 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.

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