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Asthma/COPD Differentiation Classification (AC/DC): Machine learning to aid physicians in diagnosing asthma, COPD and asthma-COPD overlap (ACO)

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Implementation Science/Service Development

Aim: To assess machine learning (ML) approaches for differential diagnosis of COPD, asthma and asthma-COPD overlap (ACO).

Brief outline of context: Diagnosis of patients presenting with chronic respiratory symptoms is difficult, because the symptoms of COPD and asthma may be similar, and their diagnostic criteria overlap. However, treatment recommendations for COPD and asthma differ, and inappropriate treatment, as a result of misdiagnoses, has the potential to increase the risk of exacerbations, morbidity and mortality, and reduce quality of life.

Brief description of the change/intervention and why you thought it would work: ML offers an innovative approach to mining data from large electronic health records (EHR) to develop diagnostic algorithms for disease differentiation.

Strategy for change: From a database of US electronic health records covering primary care, specialist care and hospital medical records, cohorts of patients aged ≥35 years who had specialist diagnoses of asthma, COPD or ACO on ≥2 occasions, were created. A total of 240,378 COPD, 143,748 asthma and 27,437 ACO cases were identified. The specialist diagnosis was used as the case label. Over 60 clinical features, including spirometry results, blood test results, comorbidities and symptoms, were extracted from patients' EHR data within 12 months before and 12 months after patients' incident diagnosis. Eleven supervised ML methods were investigated to perform disease classification on 85% of the labeled cases, and the remaining 15% were used as a hold out data set for model validation.

Effects of changes: The Extreme Gradient Boosting model with Bayesian hyper-parameter optimisation had the best performance. The model with 12 clinical features, including spirometry results, pack-years, body mass index, symptoms, and allergic and chronic rhinitis, achieved a sensitivity of 0.98, 0.98 and 0.78, and an F1-score (accuracy measure) of 0.98, 0.98 and 0.84, in diagnosing COPD, asthma and ACO, respectively.

Lessons learnt: The findings from the study support the potential for ML-based approaches as a tool for differential diagnosis in clinical settings.

Message for others: Machine learning is a powerful tool that may assist family physicians in the differential diagnosis of asthma, COPD and ACO. Additional studies are needed to evaluate the model in other settings and countries, and to assess its safety for guiding treatment decisions.

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