

Editorial

Disease definitions in respiratory and sleep medicine: changes in diagnostic criteria and categories over time and clinical implications

We often think of disease definitions as absolute and unchangeable, however, diagnostic criteria and categories for diseases are not static [1]. They change over time, and different definitions can coexist at the same time.

Have you ever wondered how diseases get defined? Often definitions are based on several diagnostic criteria, which might include symptoms and/or results of diagnostic tests. They are created and revised by expert panels, for example guideline committees or professional society groups. Determining the presence of disease is not always as straightforward as one might think. How “high” does the pulmonary arterial pressure have to be before it is pulmonary hypertension? How many apnoeas and hypopnoeas are considered “normal” during sleep and when is it labelled obstructive sleep apnoea?

Changes in disease definitions/diagnostic criteria can significantly impact the conceptualisation of a condition. For example, the inclusion of symptoms as a diagnostic requirement means that asymptomatic patients do not receive a disease label. The change of thresholds to distinguish normal variation from disease can change the incidence and prevalence of a disease, as well as the perceived effectiveness of treatment. For example, as the forced expiratory volume in 1 s (FEV₁)/forced vital capacity (FVC) ratio is known to decline with age, the prevalence of COPD in a population could

be higher when using the fixed FEV₁/FVC ratio compared with using an age-adjusted definition of airflow obstruction or the lower limit of normal [2]. Including earlier and/or milder disease cases by lowering disease thresholds might bias results to suggest better treatment efficacy.

Widening disease definitions to include early disease stages (*e.g.* pre-diabetes, pre-hypertension, pre-COPD) has the potential to allow earlier detection and treatment of a disease, but potential harms from overdiagnosis also need to be considered [3]. An evidence-based systematic evaluation of the benefits and harms before widening disease definitions is therefore indicated.

Disease labels help to define groups of patients who will benefit from specific treatments. Inconsistencies in definitions of diseases/clinical conditions in clinical trials may therefore have consequences for the applicability of the results. The article “Towards precision in defining COPD exacerbations” in this issue of *Breathe* outlines the challenges of finding a standardised definition for what constitutes an exacerbation of COPD [4], an important disease state and outcome measure in clinical trials because of its impact on patients and health systems.

Other articles in this issue explore the definitions of pulmonary hypertension, obstructive sleep apnoea, and latent tuberculosis infection [5–7],

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outlining how these disease labels and categories evolved over time, and how changes in diagnostic criteria impact on clinical practice. The article “Challenging the paradigm: moving from umbrella labels to treatable traits in airway disease” puts forward the idea that umbrella terms such as “asthma” or “COPD” should be replaced by the concept of treatable traits [8]. The lung function corner article in this issue on the “Pathophysiology of pulmonary function anomalies in COVID-19 survivors” proposes COVID-19 phenotypes based on different types of lung function abnormalities found in survivors [9].

With this issue, my tenure as Chief editor of *Breathe*, the leading educational journal for respiratory professionals, is coming to an end. It has been a privilege to serve in this role for the past 3 years. I wish the incoming Chief editor Brian Kent all the best for this challenging but fulfilling role.

Based on my experience of working with Brian on the Editorial board of *Breathe*, I have no doubt that he will do an outstanding job taking the journal into the future. I would like to express my sincere gratitude to the hard-working publications team at the Sheffield office for their invaluable support over the past 3 years, especially during the difficult time of the COVID-19 pandemic. I would also like to thank all Editorial board members; in particular, Andrea Aliverti and Pierantonio Laveneziana, the Section editors for the Lung function corner, Jayesh Bhatt, who has been doing a fantastic job commissioning and managing paediatric contributions, and Frits Franssen and Alexander Mathioudakis, the former and current Section editor for the journal club papers. Thanks to all the reviewers, contributors, and readers of *Breathe*. It is my hope that you, our reader, find the journal content useful in your everyday clinical practice.

Affiliations

Claudia C. Dobler^{1,2}

¹The George Institute for Global Health, University of New South Wales, Sydney, NSW, Australia. ²Dept of Respiratory and Sleep Medicine, Liverpool Hospital, Sydney, NSW, Australia.

Conflict of interest

None declared.

References

1. Tresker S. A typology of clinical conditions. *Stud Hist Philos Biol Biomed Sci* 2020; 83: 101291.
2. Thomas ET, Glasziou P, Dobler CC. Use of the terms “overdiagnosis” and “misdiagnosis” in the COPD literature: a rapid review. *Breathe* 2019; 15: e8–e19.
3. Dobler CC, Glasziou PP. Overdiagnosis in respiratory medicine. *Respirology* 2019; 24: 939–941.
4. Jenkins CR. Towards precision in defining COPD exacerbations. *Breathe* 2021; 17: 210081.
5. Kovacs G, Olschewski H. The definition of pulmonary hypertension: history, practical implications and current controversies. *Breathe* 2021; 17: 210076.
6. Riha RL. Defining obstructive sleep apnoea syndrome: a failure of semantic rules. *Breathe* 2021; 17: 210082.
7. Migliori GB, Ong CWM, Petrone L, *et al*. The definition of tuberculosis infection based on the spectrum of tuberculosis disease. *Breathe* 2021; 17: 210079.
8. Bush A, Pavord ID. Challenging the paradigm: moving from umbrella labels to treatable traits in airway disease. *Breathe* 2021; 17: 210053.
9. Laveneziana P, Sesé L, Gille T. Pathophysiology of pulmonary function anomalies in COVID-19 survivors. *Breathe* 2021; 17: 210065.