Dysfunctional breathing: a review of the literature and proposal for classification

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ABSTRACT Dysfunctional breathing is a term describing breathing disorders where chronic changes in breathing pattern result in dyspnoea and other symptoms in the absence or in excess of the magnitude of physiological respiratory or cardiac disease. We reviewed the literature and propose a classification system for the common dysfunctional breathing patterns described. The literature was searched using the terms: dysfunctional breathing, hyperventilation, Nijmegen questionnaire and thoraco-abdominal asynchrony. We have summarised the presentation, assessment and treatment of dysfunctional breathing, and propose that the following system be used for classification. 1) Hyperventilation syndrome: associated with symptoms both related to respiratory alkalosis and independent of hypocapnia. 2) Periodic deep sighing: frequent sighing with an irregular breathing pattern. 3) Thoracic dominant breathing: can often manifest in somatic disease, if occurring without disease it may be considered dysfunctional and results in dyspnoea. 4) Forced abdominal expiration: these patients utilise inappropriate and excessive abdominal muscle contraction to aid expiration. 5) Thoraco-abdominal asynchrony: where there is delay between rib cage and abdominal contraction resulting in ineffective breathing mechanics.

This review highlights the common abnormalities, current diagnostic methods and therapeutic implications in dysfunctional breathing. Future work should aim to further investigate the prevalence, clinical associations and treatment of these presentations.

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Introduction
Dysfunctional breathing is a term describing a group of breathing disorders in patients where chronic changes in breathing pattern result in dyspnoea and often nonrespiratory symptoms in the absence of, or in excess of, organic respiratory disease [1–3]. Many of these breathing patterns may occur as a physiological response to disease, but in the absence of organic abnormalities they can be considered pathological. Several different phrases have been used loosely and interchangeably in the literature, these include functional breathing disorder, breathing pattern disorder and behavioural or psychogenic breathlessness [4, 5]. The most widely recognised form of dysfunctional breathing is hyperventilation syndrome (HVS), which was first described over 70 years ago [6]. This term is often also used synonymously with dysfunctional breathing, whereas in fact it is just one type of breathing pattern disorder and hyperventilation is not necessarily seen in dysfunctional breathers. There is no formal definition of dysfunctional breathing and no gold standard diagnostic method.
The key symptom in dysfunctional breathing is breathlessness after organic causes have been ruled out, but associated symptoms may be attributable to hyperventilation (increased minute ventilation) and respiratory alkalosis (e.g. tingling, tetany and numbness), but these are not specific to dysfunctional breathing [7]. The patients’ overwhelming symptom is dyspnoea despite exclusion of, or treatment optimisation for, any organic disease. Breathlessness may manifest as hyperventilation, or symptoms that occur independently of hypocapnia and respiratory alkalosis such as deep sighing or the sensation of air hunger [2, 8, 9]. Dyspnoea is unlikely to be caused solely by inadequate or inefficient ventilation either due to hyperventilation or an abnormal breathing pattern and additional factors, such as poor subjective awareness of breathing by patients, are thought to play a role [10]. There is limited and largely uncontrolled evidence that when dyspnoea is related to an abnormal breathing pattern it may be improved by breathing retraining, but the sensation of dyspnoea may still remain [11].

We aimed to review the literature in order to summarise the epidemiology, presentation, assessment methods and treatment for dysfunctional breathing, with an additional aim of providing a suggested classification system for the different breathing patterns. Vocal cord dysfunction, a condition in which there is abnormal and excessive closure of the vocal cords and surrounding structures, typically during inspiration, has not been reviewed here. This is because it is a well-defined and distinct disorder with the principal source of breathlessness at the level of the larynx (i.e. extrathoracic) and is treated with speech and language therapy (rather than physiotherapy, as is typically the case for dysfunctional breathing).

Literature review
PubMed and Embase were searched using the terms dysfunctional breathing, hyperventilation, Nijmegen questionnaire and thoraco-abdominal asynchrony. A total of 115 papers were returned, of which 59 were considered relevant to this topic.

Epidemiology and comorbid conditions
It is not possible to accurately determine the prevalence of dysfunctional breathing in the absence of gold standard diagnostic criteria. Currently, diagnosis of dysfunctional breathing relies on exclusion of organic pathology before using several "methods of assessment" described later in this review. Many of the studies assessing epidemiology use the Nijmegen questionnaire as a method of diagnosis, which may not be valid in some circumstances. Most data are available for HVS, the prevalence of which is estimated to be in the region of 6–10% in the general population, rising to 29% in asthmatics [5, 12]. The Nijmegen questionnaire has recently been validated in asthmatics with a single study. The approximate prevalence of HVS in asthmatics was found to be 34% [13]. As many of the symptoms described in the Nijmegen questionnaire will occur in asthma the true prevalence of HVS in asthmatics is likely to be overestimated. One study assessing the prevalence of dysfunctional breathing in people with asthma found that most with high Nijmegen questionnaire scores had evidence of asthma at follow-up [14]. A further study found that up to 80% of patients diagnosed with HVS may in fact have an underlying diagnosis of asthma [15]. Asthmatics with HVS tend to be female, have poor asthma control, frequent exacerbations and comorbid anxiety states [16]. In asthmatic children the prevalence appears to be much lower in the region of 5% [17], although this study was performed in severe asthmatics and is therefore difficult to extrapolate to all patients. It must also be noted that the Nijmegen questionnaire has not been validated in children and that symptoms of dysfunctional breathing specific to children are not included in the Nijmegen questionnaire [17].

There are no data available to estimate the prevalence of other patterns of dysfunctional breathing either in the general population or in people with respiratory disease.

Dysfunctional breathing patterns and associated conditions
Dysfunctional breathing patterns may occur where there is coexistent respiratory disease, in particular asthma, and so difficulty arises when trying to untangle which of the two are contributing most to the reported symptoms. Due to the close link between asthma and dysfunctional breathing, it is important to identify any objective evidence of asthma and optimise asthma treatment by controlling features such as bronchial hyperresponsiveness and airway inflammation as far as possible prior to making a diagnosis of dysfunctional breathing. Recognising variations in breathing pattern in asthmatics may provide a way of identifying those patients who would benefit from breathing retraining to target abnormal breathing mechanics [18]. More recent studies of dysfunctional breathing in asthma have looked at combining use of the Nijmegen questionnaire with symptom limited exercise testing in attempt to improve specificity for dysfunctional breathing [16, 19]. This method has promise, but data from healthy controls are required before it can be recommended.

A clear link between dysfunctional breathing and other respiratory diseases has not been made in the literature, but in our experience dysfunctional breathing patterns do occur in some patients with diseases...
such as chronic obstructive pulmonary disease (COPD) and interstitial lung disease. Another condition that has been linked to dysfunctional breathing and in particular HVS is panic disorder, which comprises many of the symptoms listed in the Nijmegen questionnaire. Dysfunctional breathing, particularly HVS, is commonly seen in those with anxiety related disorders [20]. In these related conditions it is difficult to assess whether HVS is causative or simply a secondary effect of anxiety related disorders. There is an extensive body of research that implicates respiratory processes and particularly hyperventilation in panic disorder [21, 22]. Asthma itself is associated with an increased prevalence of reported panic disorder at 9.7% [5, 23]. Well-controlled asthma has a lower prevalence of significant anxiety than dysfunctional breathing when measured by the hospital anxiety and depression scale (HADS) (24% versus 56%) [24].

Methods of assessment

Prior to diagnosis of dysfunctional breathing, clinicians must first exclude, or adequately treat, organic disease and only then can dysfunctional breathing be considered. In cases where the usual investigations into a cause for breathlessness are normal or inconclusive cardiopulmonary exercise testing may be used to determine whether breathlessness can be explained (or not) by impaired cardiopulmonary reserve. If not, then further investigations are required to confirm the presence of a dysfunctional breathing pattern.

The most common method of diagnosing dysfunctional breathing relies on a positive Nijmegen questionnaire. Although this questionnaire was developed and validated only in people with exercise induced HVS its use has since been extrapolated, probably inappropriately, to aid the diagnosis of all forms of dysfunctional breathing in many settings. This questionnaire, developed by a group in the Netherlands, comprises 16 questions about symptoms. Of these, seven relate to respiratory symptoms, four to excessive dysfunctional breathing in many settings. This questionnaire, developed by a group in the Netherlands, comprises 16 questions about symptoms. Of these, seven relate to respiratory symptoms, four to excessive ventilation and five concern central nervous system symptoms relating to hypocapnia and "central tetany" [25]. During validation the Nijmegen questionnaire was shown to have a sensitivity of 91% and specificity of 95% [25]. A score of >23 is commonly used as a cut-off for HVS, although this was calculated using a positive hyperventilation provocation test as the gold standard, which itself is no longer considered a reliable way of diagnosing HVS [26]. This test was performed by asking the patient to voluntarily hyperventilate for several minutes and was considered positive if symptoms of HVS were reproducible. It was felt these symptoms were due to low end-tidal carbon dioxide, but a study in the Lancet found a high rate of false positives in patients where end-tidal carbon dioxide was controlled via a facemask [26].

Other suggested methods of diagnosis include other questionnaires such as the Self Evaluation of Breathing Questionnaire (SEBQ), end-tidal carbon dioxide measurement (measured using capnography with an expected low end-tidal carbon dioxide in hyperventilation), breath holding time (where a short breath holding time after normal expiration at functional residual capacity is considered an indicator of dysfunctional breathing) and manual assessment of respiratory motion (MARM) [27-30]. It is important to note that, other than MARM and the Nijmegen questionnaire (which correlate weakly), these methods have yet to been shown to correlate with one another [31].

The SEBQ describes two factors reported commonly by patients with dysfunctional breathing: the feelings of "lack of air" and "restriction of breathing" [32]. These factors were said to represent both biomechanical and chemoreceptive factors contributing to dysfunctional breathing, which have often been implicated in other studies [33].

MARM is a tool used for assessing rib cage and abdominal motion during breathing. It has been shown to have good inter-examiner reliability and is consistent with older and more complex methods such as respiratory induction plethysmography (RIP), which has limitations when posture is slumped [29]. In this method the user wears a series of bands across the chest to measure change in dimensions of the chest and abdomen; to distinguish between abdominal and thoracic breathing; and to determine a "phase angle" as a measurement of asynchrony [27]. Patients with both elevated Nijmegen questionnaire scores and abnormal breathing patterns as assessed by MARM may benefit from breathing retraining to produce more efficient breathing patterns [34].

Efficient breathing requires coordination between the diaphragm, abdominal muscles and muscles of the rib cage. Variations in coordination of muscle contraction are often implicated in dysfunctional breathing and sensations of dyspnoea [35]; particularly thoracic dominant breathing (also termed apical breathing). One study found a weak relationship between hyperventilation symptoms and breathing pattern in 74% of patients indicating that a large number of patients with elevated Nijmegen questionnaire score had abnormal breathing patterns [11].

In our experience tracking of respiratory flow, frequency and volumes during quiet tidal breathing, often performed before and after exercise, can give useful information to the clinician and be used for providing feedback to the patient. This may also help us to define and characterise different dysfunctional breathing patterns (figure 1).
Optoelectronic plethysmography is a novel method of assessing three-dimensional lung volumes across the pulmonary ribcage, abdominal rib cage and abdomen using markers on the chest wall and abdomen. Currently, research is limited to healthy subjects and some organic diseases such as COPD. Optoelectronic plethysmography may be a promising method of assessing breathing pattern and thoraco-abdominal asynchrony in future research [36].

Treatment

Treatment for dysfunctional breathing starts with an accurate diagnosis, which in itself can provide significant reassurance and relief of anxiety, and may be enough to reduce the severity and frequency of the symptoms. Physiotherapy directed breathing techniques may also relieve symptoms. In the acute setting rebreathing is no longer recommended and assurance and time are usually sufficient to allow patients to get over an acute attack. Patients can then be reviewed by a respiratory specialist physiotherapist to assess and train patients to gain quicker control over future attacks. Two common breathing techniques have been used: the Papworth and Buteyko methods.

The Papworth method (where patients are taught diaphragmatic breathing with an emphasis on controlled, slow nasal breathing) has resulted in some symptom relief and increasing carbon dioxide tension using small sample sizes of moderate asthmatics, but data on long-term follow-up are lacking [14, 37]. Another small study showed a similar improvement over 24 months in both the Nijmeegen questionnaire and HADS, but no correlation between the two scores [38]. A further study showed improvement in both the Nijmeegen questionnaire and dyspnoea-12 scores post-retraining [39].

The Buteyko technique describes the use of nasal breathing and increasing controlled pauses with the aim of reducing hyperventilation over time [34]. The Buteyko technique may have a role in those patients with thoracic dominant breathing patterns by way of increasing breath holding time, but it remains to be proven that it increases end-tidal carbon dioxide [34]. This method has been shown to reduce symptoms in asthmatics and reduce bronchodilator use without changing lung function, although the Buteyko method itself involves active encouragement to reduce use of bronchodilators. Another study comparing the Buteyko method with asthma education in asthmatics that scored over 23 on the Nijmegen questionnaire showed improvement post-training at 6 months in HADS, asthma-related quality of life and Nijmegen questionnaire scores with no change in respiratory physiology [40]. To extrapolate these data to all dysfunctional breathers is difficult and must be interpreted in the context of the studied sample population. The patients in this study had asthma and high Nijmegen questionnaire scores, but it was not...
clear that they had dysfunctional breathing. It is therefore not known whether asthma education may have 
been beneficial in people with dysfunctional breathing alone.

In recent years, yoga has been trialled in patients with COPD and asthma with limited effect and studies 
remain highly heterogeneous in design [33, 41, 42]. Future studies may provide further insight into the 
effects of yoga in dysfunctional breathing [43].

Pulmonary rehabilitation has clear benefits in COPD patients and has been trialled in other respiratory 
conditions. Although currently no literature exists regarding pulmonary rehabilitation for dysfunctional 
breathing further research would be beneficial to assess its efficacy in such patients [44].

Types of dysfunctional breathing: a proposal for classification

As we have demonstrated, there is a significant difficulty in reaching a consensus on the epidemiology, 
comorbidities and management of dysfunctional breathing while there is no clear agreement on the 
definition of the condition(s). In fact many patterns of dysfunctional breathing patterns are described in the 
literature (and evident in our clinical practice), and review of the symptoms and physiology described has 
enabled us to classify each into the following proposed categories (summarised in table 1 and figure 1). It is 
possible that several dysfunctional breathing patterns may coexist. The proposed categories include patterns 
described or observed in isolation as well as coexisting with one another.

We suggest differentiation between subtypes can be obtained via monitoring tidal spirometry with a full 
expiratory and inspiratory manoeuvre at the end. Further evaluation can be obtained by using MARM in 
the hands of an experienced practitioner to distinguish thoracic abdominal asynchrony and thoracic 
predominance of breathing pattern.

The quantity and quality of research varies considerably between subtypes, with HVS having the largest 
evidence base. Other categories exist to a lesser extent both in the literature and probably in the clinical 
setting, but may be equally distressing for patients and require further research.

Hyperventilation syndrome and its subcategories

This is the most commonly described and researched form of dysfunctional breathing (figure 1b). Diagnostic criteria initially required the demonstration of hypocapnia and respiratory alkalosis during 
attacks of hyperventilation. However, more recent research has questioned the importance of hypocapnia 
in inducing the associated symptoms of HVS as many of these symptoms can occur in normocapnia [26, 55, 56]. In fact many of the more recent methods used in diagnosis have a poor correlation with carbon 
dioxide levels [57]. The diagnosis is based on the Nijmegen questionnaire only after organic disease has 
been ruled out or optimally controlled, and is often difficult to distinguish from asthma and anxiety 
disorders [5, 16, 21].

Exercise-induced hyperventilation occurs in a subgroup of patients who develop chest discomfort and 
dyspnoea during cardiopulmonary exercise testing independent of bronchospasm and unresponsive to beta 
agonist therapy, thus distinguishing it from exercise-induced asthma [58, 59].

| Breathing pattern                      | Number of papers | Key references          | Linked conditions              |
|---------------------------------------|------------------|-------------------------|-------------------------------|
| Hyperventilation syndrome             | 43               | [1, 2, 5, 15]           | Asthma                        |
| Periodic deep sighing                 | 12               | [8, 9, 45–47]           | Panic disorder                 |
| Thoracic dominant breathing           | 4                | [11, 47–49]             | Asthma                        |
|                                      |                  |                         | COPD                           |
| Forced abdominal expiration           | 2                | [50, 51]                |                               |
| Thoraco-abdominal asynchrony          | 6                | [52–54]                 | Obstruction                    |

COPD: chronic obstructive pulmonary disease.
Postural hyperventilation occurs in a subgroup of patients where a change in posture from supine to standing is associated with hyperventilation. This may be due to the influences of the vestibular system on the autonomic and respiratory systems, which may be over stimulated in HVS [60].

**Periodic deep sighing**
This type of dysfunctional breathing is characterised by frequent sighing and irregular breathing patterns, sometimes overlapping with hyperventilation (figure 1c). Sighing may be defined as a tidal volume three times the normal volume and observed as a crescendo on a slow lung volume trace [6, 18]. Sighing occurs in healthy individuals and asthmatics, but is more frequent (up to 15 times in a 15 min period) and associated with dyspnoea in those with dysfunctional breathing [8, 45, 46, 61].

**Thoracic dominant breathing**
Thoracic dominant breathing or apical breathing occurs when there is predominant use of the upper thorax with lack of lateral costal expansion (figure 1d). Higher levels of dyspnoea are seen in thoracic dominant breathers who scored highly on the Nijmegen questionnaire. This type of breathing often occurs in patients with increasing ventilation needs such as those with cardiac or respiratory disease or those with decreased abdominal compliance such as the morbidly obese, but in their absence it can be termed dysfunctional [11, 48, 49]. Therapies already target thoracic dominant breathing in patients with COPD with improvements in quality of life scales [62].

**Forced abdominal expiration**
This is the least described breathing pattern in the literature, but can be observed in the clinical setting particularly in patients with COPD. These patients have an inappropriate and excessive abdominal muscle contraction in aid of expiration (figure 1e). This may be a normal physiological adaptation in patients with COPD and pulmonary hyperinflation, but is typically dysfunctional in the absence of these conditions [63]. Forced abdominal expiration can also be seen in morbidly obese patients with a prolonged expiratory phase, decreased functional residual capacity and chest wall compliance [64].

**Thoraco-abdominal asynchrony**
This is due to a delay between rib cage and abdominal contraction resulting in ineffective breathing (figure 1f). In extreme cases this is termed paradoxical breathing. Thoraco-abdominal asynchrony is sometimes seen as a normal physiological response in upper airway obstruction, neuromuscular disorders and in patients with acute respiratory failure, but may be seen in patients without evidence of these, and therefore considered dysfunctional [52].

**Discussion**
We have described the common abnormalities, current diagnostic methods and therapeutic options for people with dysfunctional breathing. The presentation of varying breathing patterns under the umbrella term of “dysfunctional breathing” appears to be relatively widely appreciated (anecdotally), but there is little recognition of this in the literature, and hence the topic presents a significant challenge for clinicians. HVS remains the most extensively studied dysfunctional breathing pattern, although other forms may coexist or appear in isolation. Advances in the management of dysfunctional breathing will first require relatively basic descriptive studies, to enable an estimate of prevalence, demographics and impact. We hope that we have facilitated this by providing a framework for classifying dysfunctional breathing that is useful to clinicians and researchers.

One issue that complicates the diagnosis and investigation of dysfunctional breathing is its overlap and interaction with associated respiratory diseases, in particular asthma. Dysfunctional breathing may present alone, as a somatic manifestation of psychological conditions or as a manifestation of underlying disease; and in any individual there may be more than one of these factors at play. We would advocate a rigorous approach in assessing for underlying or associated cardiorespiratory disease in patients’ suspected of having dysfunctional breathing, before this is confirmed as the dominant diagnosis contributing to breathlessness, in a similar way to the cough management guidelines that recommend a pathway to identify and treat comorbidities before the cough is labelled “idiopathic” or “treatment resistant” [65].

While we have proposed five type of breathing pattern based on the literature and our own experience, we would welcome debate and suggestions for improvement, to enable a consensus to be reached in order to drive better care and future research. We do not propose that the categories are mutually exclusive. For example, people with HVS may also have periodic deep sighing, and indeed the same patient may experience different forms of dysfunctional breathing at different times. Categorising the patterns in this way will also hopefully lead to treatment targeted at specific components of the breathing pattern. Currently most published treatment proposals are targeted at HVS, but these may not be appropriate (and
in our experience do not work) in patients with, for example, forced abdominal expiration or thoraco-abdominal asynchrony.

Without defined diagnostic criteria, current research is difficult to interpret. Many studies have used the Nijmegen questionnaire as a method of diagnosis and extrapolated its use to different dysfunctional breathing patterns in settings other than exercise-induced HVS. Without further diagnostic criteria and with an ongoing lack of awareness of dysfunctional breathing sample sizes were often low and included mixed breathing patterns. In addition, as for many conditions that lack clear criteria for diagnosis there is likely to be a significant publication bias. Many current breathing retraining methods have yet to be rigorously tested, and without clear definitions for cases and controls linking any improvements seen is difficult.

In conclusion dysfunctional breathing represents an important yet poorly understood topic and is probably associated with a disproportionately high cost to healthcare providers. A consensus needs to be reached on a definition for dysfunctional breathing and what the term encompasses. Further research is required to look at whether tailored breathing retraining methods can successfully target different breathing patterns.

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