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# <u>Title: How is dysfunctional breathing characterised in the clinical</u> literature? A scoping review.

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# **Abstract**

#### Introduction

Dysfunctional breathing (DB) is a complex condition that affects health and quality of life. It can occur alone or with other conditions and presents with various symptoms. Despite its prevalence, there is no consensus among healthcare professionals on its definition, diagnosis, or classification.

### **Aims & Methods**

This scoping review aimed to explore how dysfunctional breathing is characterised across the literature. To achieve this, a comprehensive search strategy was employed, covering both peer-reviewed and grey literature from relevant academic databases and websites.

#### Results

Seventy one relevant sources published between 2001 to 2024 were identified resulting in 6 key themes. These consisted of 'Defining DB' (69% of studies), 'Diagnosing and Assessing DB' (86%), 'Symptoms and Dimensions' (87%), 'Comorbidities' (64%),' Psychophysiological Links' (43%), and 'Classification' (50%).

#### Conclusion

There are inconsistencies in the terminology, assessment methods, and classification systems used to describe (DB). This highlights a lack of standardisation across the literature. The use of multiple, and at times overlapping, definitions for the same condition poses a significant challenge for clinical decision-making and treatment planning. To address these issues, future research should prioritise achieving consensus across these domains acknowledging the multidimensional nature of the condition.

# Keywords

- Dysfunctional breathing
- Dyspnoea
- Classification
- Assessment
- Comorbidity
- Diagnosis
- Phenotypes

# **Highlights**

- Six recurrent themes identified in 71 studies on dysfunctional breathing literature.
- No consensus exists on DB definitions, assessment, or classification
- Multidimensional models of DB are underused in current clinical practice
- Common comorbidities include asthma, anxiety, and COPD
- Standardised diagnostic tools for DB remain unvalidated and inconsistent

## **Educational Aims**

- To provide a comprehensive overview of how dysfunctional breathing (DB) is characterised in clinical literature
- To explore inconsistencies in DB definitions, assessment methods, and classification systems

- To highlight the multidimensional nature of DB, including biomechanical, biochemical, and psychophysiological domains
- To identify diagnostic challenges and the impact of comorbid conditions such as asthma and anxiety
- To inform clinicians and researchers of current gaps that affect diagnostic accuracy and patient care

## **Future Research Directions**

Future research should prioritise the development of a consensus definition of dysfunctional breathing that reflects its multidimensional nature. Additionally, there is a pressing need to validate diagnostic tools that capture the full spectrum of DB phenotypes. Establishing standardised classification systems and assessment frameworks will improve diagnostic reliability, guide targeted interventions, and enhance comparability across studies. Collaborative, multidisciplinary approaches—such as Delphi consensus studies—may be instrumental in achieving these goals.

# Introduction

Breathing is a fundamental physiological process essential for life, performing key functions such as air transport and gas exchange, which are central to the respiratory pump (1). Beyond these physiological roles, breathing also has a biomechanical function, supporting posture and movement through the expansion of the thoracic cavity (2). Moreover, the subjective experience of breathing significantly influences psychological well-being, interacting with the autonomic nervous system to regulate emotional and stress responses (3).

Dysfunctional breathing (DB), a condition where abnormal breathing patterns are present without an identifiable organic cause, has become a subject of increasing clinical interest. It is associated with a high symptom burden, high healthcare-related costs and has a negative impact on health-related quality of life (HR-QoL) (4,5). Various studies report its prevalence in the general population ranging from 6-10% but with some estimates reaching up to 60-80%, particularly in patients with asthma (6,7). However, it has been highlighted that due to a lack of a diagnostic criterion, prevalence data for the condition could be over or underestimated (8). This has also led to DB being often ill-defined, with considerable ambiguity surrounding its definition, diagnosis and optimal management (9,10).

Multiple factors are reported to contribute to DB, with it being described in the literature as a multi-dimensional construct consisting of at least 3 dimensions; biomechanical, psychophysiological and biochemical (3,9). While the influence of these domains on patient experience is thought to vary between individuals, the degree to which these three dimensions have been integrated into clinical practice remains uncertain (3,11). The predominant symptom associated with DB is dyspnoea, either at rest or during exercise (12) which may occur with or without other commonly reported respiratory symptoms such as breath-holding, air hunger, and rapid mouth breathing (4,13). Non-respiratory symptoms often considered to be related with DB include palpitations, dizziness, paresthesia and chest tightness (8,14). DB is also commonly observed in patients with anxiety; the presence of the aforementioned symptoms may exacerbate anxiety and, in turn, provoke further breathing irregularities, creating a self-

perpetuating vicious cycle (6,15). This complex interplay between psychological and physiological symptoms often encountered in other common cardiopulmonary conditions such as asthma and COPD has been suggested to contribute to under recognition of DB's and high rates of misdiagnosis (8). This lack of clarity has profound implications. It has been suggested that this may lead to inappropriate management and reduced quality of care, including the overprescription of corticosteroids when symptoms experienced during DB exacerbations are misattributed to an underlying organic respiratory condition (16). The prevalence of misdiagnosis in cases of DB indicates a limited understanding of the condition among healthcare professionals, which may hinder the delivery of appropriate treatment to affected patients (8).

With this in mind, it is no surprise that a lack of understanding amongst Health Care Professionals (HCP's) has been reported within the literature (17,18). In a qualitative evaluation, Grillo et al (19) reported the use of inconsistent nomenclature associated with DB which has impacted assessment and management of the condition. Physiotherapists used different terms interchangeably with DB including breathing pattern disorder, however, Barker et al reported breathing pattern disorder to be a form of DB (16). A potential contributing factor may lie in the inconsistent use of terminology when discussing DB. A literature review highlighted the ambiguity surrounding the term DB, with "hyperventilation syndrome (HVS)", "anxiety-related breathlessness" and "sighing dyspnoea" among others all being used to describe the same condition (16). This variability in terminology has compounded confusion among healthcare professionals (HCPs) and contributed to the inconsistent assessment and management of DB (19). In addition to the variable terminology, there have been different proposed classifications of DB. Some authors argue that the condition should viewed multifactorial phenomenon, biomechanical, be as involving

psychophysiological, and biochemical domains, which can independently or collectively contribute to its manifestation (20,21). Despite these insights, a consensus on the precise characterization of DB remains elusive.

A further consequence of this lack of consensus is the absence of a universally accepted diagnostic instrument for assessment (9,10). Although various tools designed to evaluate different aspects of the condition have been described in the literature, their diagnostic application remains limited due to insufficient rigorous validation (5). For example, the Nijmegen questionnaire (NQ) is often used as a diagnostic tool for DB, despite this not being its intended purpose (22), as well as the Self-Evaluation of Breathing Questionnaire (SEBQ) (12), Manual Assessment of Respiratory Motion (MARM) (23) and capnography (7).

Despite its potentially significant prevalence, there is little consensus in the literature and in clinical practice on how DB is defined, assessed and managed (8,16). Given these challenges, it is essential for healthcare professionals and researchers to collaborate towards developing a unified understanding of DB. As a first step to achieving this, our scoping review aimed to provide an in-depth exploration on how DB is characterized within the clinical literature. The review examines how DB is defined, highlights key concepts surrounding assessment and diagnosis, and identifies research gaps. These insights may inform future research and clinical practice, contributing to improved diagnostic accuracy and treatment strategies for those affected by DB.

# **Methods**

## Study design

This scoping review was conducted using the Joanna Briggs Institute (JBI) framework (24) following advancement and refinement of the original framework for developing scoping reviews (25). The Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) checklist was used to structure and ensure quality of the review (26).

#### **Search strategy**

The following databases were screened due to their subject specificity relating to the current research topic: Cumulative Index to Nursing and Allied Health Literature (CINAHL), Medical Literature Analysis and Retrieval System Online (MEDLINE), Scopus, Science Direct, Web of Science, Pubmed, google scholar and nursing databases to ensure a comprehensive and multi-disciplinary approach to the literature search. Grey literature was also searched as it has been found to be increasingly recognised as valuable sources of literature when conducting scoping reviews in recent years (27). Subject librarian in Sheffield Hallam University was consulted.

Databases were searched from inception to September 2024. Search results were exported to Refworks before importing to Covidence<sup>TM</sup> (28).

## **Keywords**

A preliminary search was conducted in CINAHL & MEDLINE to assess the terminology and keywords most commonly used. From December 2023 to January 2024, a thorough search of PubMed, Web of Science and Scopus was conducted. The search query included keywords related to "Dysfunctional breathing", "respiratory",

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"respiration" "paradoxical", "definition", "classification", "Nijmegen questionnaire", "breathing pattern disorder".

Several search terms were used in combination with Boolean operators 'AND', 'OR', alongside truncation and wildcard functions to aid in broadening the search potential across databases.

### Inclusion/exclusion criteria

Inclusion and exclusion criteria (Table 1) were defined using the population, concept, context (PCC) framework, as recommended for scoping reviews (24).

Table 1.

PCC criteria	Inclusion	Exclusion
Population	Literature characterising	Non-human (animal)
	dysfunctional breathing	participants
	for paediatric and adult	
	populations of any	
	biological sex, gender	
	identity and race	
Concept	Characterisation of DB as	Characterisation of
	a term in the clinical	related terms or
	literature	respiratory condition focus

		without specific
		characterisation of DB
Context	Literature published on	Literature published in
	any date in any	non-English language
	healthcare setting or	
	country written in the	
	English language	

## **Study Selection**

Screening involved a three stage process using Covidence<sup>TM</sup> software and required consensus from two members of the research team (28). Conflicts encountered at all stages of the screening process required a third independent researcher for consensus decision. After duplicate removal, stage one involved screening titles and abstracts using predefined inclusion and exclusion criteria (Table 1) and keywords. Stage two involved full text screening of the remaining articles.

#### **Charting the data**

Data extraction was performed using a data charting table which included study characteristics such as title, author, year of publication, study design, key findings and overall themes, the table was created with collaboration of all researchers and based on JBIs standardized extraction form for scoping reviews (24). Data from each included article was summarised in supplementary materials 1.

#### Summarising and reporting of results

The results were synthesised using a narrative approach aligned with the JBI framework recommendations for scoping reviews (29). Key Themes and gaps within literature were identified and agreed upon within the research team.

# Results

A total of 6026 records were identified through the database search published between 2001 and 2024. After removing 1803 duplicates, 4223 unique records remained. These records underwent title and abstract screening, from which 4057 articles were excluded, leaving 166 full text articles to be assessed for eligibility. Out of these 166, 95 were excluded as a result of not meeting the eligibility criteria. Seventy - one studies (3-5,7-18,20-23,30-80) were included into the scoping review for data extraction. The process of evidence selection is illustrated in the PRISMA flow chart on figure 1.

Themes assigned to relevant articles were: definition and classification of DB, symptoms and phenotypes of DB, assessment and diagnosis, comorbidities and diagnostic challenges. What studies contributed to the development of these themes can be found in supplementary materials 1.

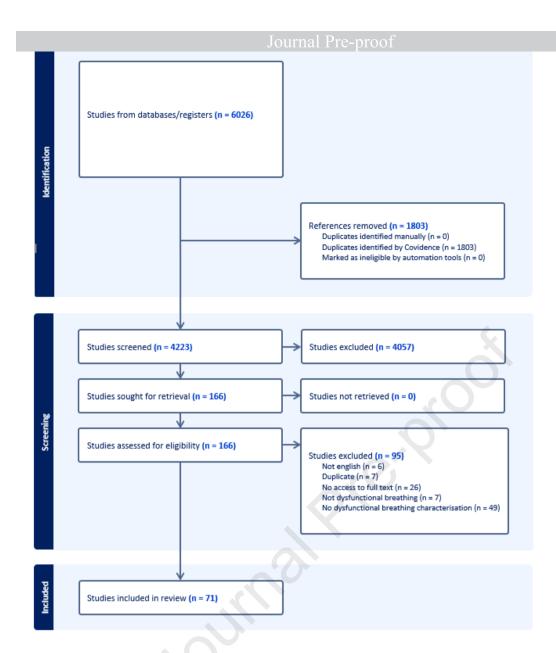


Figure 1: PRISMA flow chart

#### **Definitions and classifications of DB**

Forty nine publications (69%) discussed the concept of defining DB, all of which state that there is no consensus. Sixteen publications (23%) defined DB or cited another author's definition. The most cited definition of DB was that proposed by Barker and Everard (25%): "an alteration in the normal biomechanical patterns of breathing that result in intermittent or chronic symptoms which may be respiratory and/or non-respiratory"(16) . All definitions of DB are presented within supplementary materials 2. The remaining 33 (47%) reported DB as being difficult to define, poorly defined, that there is a lack of agreement on a definition, or that DB is not definable. Ten publications (14%) referred to DB as an "umbrella" or "overarching" term. DB was differentially defined according to a change in breathing pattern (n= 12), the presence of respiratory and non-respiratory symptoms (n= 3), symptoms being intermittent or chronic (n = 3), occurring in excess of or without organic disease (n = 4), being multidimensional (n= 1).

Four classification systems for DB have been proposed, as referenced in sources (4,7,8,10), with further details provided in Supplementary Materials 2. In total thirty-six publications (50%) discussed DB classification and/or the use of overlapping terms. The use of different terms to describe the same phenomenon was consistently linked to a lack of consensus for DB classification and confusion around the condition. The use of terms such as "breathing pattern disorder", "functional breathing disorder", "hyperventilation syndrome" and "behavioural/ psychogenic breathlessness" were sometimes used interchangeably with DB, however some of these terms were used

to describe DB. Nine studies have identified HVS as an outdated term and 19 studies classify HVS as a subtype of DB and 10 using it synonymously.

### Symptoms and phenotypes of DB

Sixty two of publications (87%) discussed the various symptoms and phenotypes associated with DB. Ten articles (14%) reported DB symptoms can be intermittent or chronic. Both respiratory and non-respiratory symptoms were commonly reported to be associated with DB, and these symptoms can be linked to different phenotypes (biomechanical, psychophysiological, biochemical) of DB. Dyspnoea was the most commonly reported symptom of DB (n=32). Other symptoms were variably reported to be associated with DB including: chest tightness/pain (n=30), thoracic/upper chest/apical dominant breathing (n=25), (deep) sighing (n=30), frequent yawning (n=14), fatigue (n=4), anxiety (n=11), dizziness (n=13), hyperventilation (n=17) and paraesthesia (n=3).

#### **Assessment and Diagnosis**

Sixty one publications (86%) discussed diagnosis and assessment of DB, commonly reporting the lack of gold-standard diagnostic method or criteria.

The Nijmegen questionnaire was the most frequently reported tool used to assess DB (n = 50). Other tools reported for DB assessment included: spirometry (n = 5), breathing pattern analysis (n = 2), respiratory inductance plethysmography (n = 2), cardiopulmonary exercise testing (CPET) (n = 8), Hi-Lo breathing assessment (n = 4), manual assessment of respiratory motion (MARM) (n = 9), Self-Evaluation of Breathing Questionnaire (SEBQ) (n = 14), breathing pattern assessment tool (BPAT) (n = 5),

capnography (n= 2). There was a common agreement that individual diagnostic tools in isolation may neglect certain DB phenotypes. Twelve articles (16%) commented on the need for a multidimensional approach to assessment and diagnosis, and highlighted how combinations of tools are required for a comprehensive assessment of DB.

## Comorbidities and diagnostic challenges

Forty six publications (64%) discussed comorbidities and diagnostic challenges associated with DB. The most commonly reported comorbidity was asthma (n = 25). Other comorbidities associated with DB included: chronic obstructive pulmonary disorder (COPD) (n= 4), long COVID (n= 2), musculoskeletal (MSK) complaints (n= 3), postural orthostatic tachycardia syndrome (POTS) (n=2) and bronchiectasis (n=1). The coexistence and symptom overlap of DB with other conditions was commonly reported to lead to diagnostic confusion, particularly with asthma patients. Hancox et al. reported wheeze as a cardinal feature of asthma which is not regarded as a DB symptom (42). Eleven publications (15%) acknowledged DB can exist in the presence or absence of organic disease.

Thirty-one (43%) of publications discussed a link between psychophysiological factors and DB. Stress (n = 11), anxiety (n = 20), PTSD (n = 2) and panic disorder (n = 4) were reported to be linked to DB.

# **Discussion**

The aim of this scoping review was to explore how DB is characterised in the clinical literature and represents the most comprehensive exploration of this topic to date. Our

review identified a diverse range of sources consisting of 36 quantitative, 28 qualitative, and 7 mixed-methods approaches, collectively encompassing 23 different study designs. There was a significant disparity in how DB was characterised across the majority of sources identified, reflecting a lack of consensus in relation to different elements of the condition.

Multiple, heterogeneous definitions of DB were reported, with no universally accepted definition of DB being agreed upon. Nair et al. highlights how this can pose significant challenges to healthcare professionals in relation to clinical reasoning and decision making (22,43,75,81). It has also been noted by Karkouli et al how the absence of a consensus definition in DB potentially limits its identification in clinical practice (43). This lack of clarity also has the potential to negatively influence the patient experience, with DB patients expressing frustration with a lack of recognition and delays in diagnosis (19,43).

The definition proposed by Barker and Everard was the most cited in the literature (16). Of the articles that defined DB, the majority included terms relating to an alteration in breathing mechanics, suggesting a physical/ biomechanical component is seen as an important definitive feature of DB. Interestingly, only one article included "multidimensional" ("biomechanical, biochemical and psychophysiological") in their DB definition explicitly (21). This is despite contemporary perspectives acknowledging DB as a multidimensional clinical phenomenon (15). This appears to reflect a transition from the more historical term of HVS. While contemporary perspectives recognize HVS as a subtype of DB (15), the findings of this study indicate that HVS, along with several other terms, is still used interchangeably to describe what seems to be the

same condition, albeit with a different and incomplete definitional approach. Courtney (82) previously discussed this issue, suggesting that it may contribute to the ongoing conceptual confusion surrounding the condition.

The reason for this lack of consensus is likely multifactorial and driven by a number of factors. Firstly is the complex, multidimensional nature of the condition itself, as highlighted by Morgan et al. (67) who describe DB as "not a precise or definable entity". This notion is supported by the findings of our review which identified over 20 distinct symptoms associated with DB, encompassing respiratory, non-respiratory, and psychological features, thus highlighting its complexity. A recent editorial by Vlemincx explores how this, along with its apparent existence in the absence of organic respiratory disease places DB as a potential 'transdiagnostic dimension' associated with a range of disease states (83).

Our findings showed frequent associations of DB with conditions such as asthma, long COVID, neck pain, musculoskeletal (MSK) complaints, chronic obstructive pulmonary disease (COPD), postural orthostatic tachycardia syndrome (POTS), bronchiectasis, and vocal cord dysfunction. Of these, asthma was the most frequently reported comorbidity (35% of results), and this high prevalence of DB among individuals with asthma underscores a significant diagnostic challenge. This has also been considered in regards to comorbid anxiety, it being described as both a cause and consequence of DB (83). The presence of comorbidities in these studies affects the quality and reliability of the results when answering the research question on DB's characterization. When comorbid conditions are present in study populations, they introduce additional variables that can mask or mimic DB symptoms, creating diagnostic ambiguity. This overlap complicates efforts to identify DB's unique features,

as symptoms like breathlessness, chest tightness, and dyspnea are often shared with these comorbid conditions (75). To advance understanding of DB as a distinct entity, future research should aim to clarify its relationship with these overlapping conditions.

This breadth of symptomatology observed within the literature is in keeping with the multidimensional model proposed by Courtney who recognised DB as a multidimensional construct, consisting of psychophysiological, biomechanical and biochemical domains (21). This model highlights the complexity of DB and supports the view that it cannot be understood through a unidimensional framework. Several authors have attempted to classify DB based on phenotypes and symptoms with no single system gaining widespread acceptance (see supplementary materials 3). However, these efforts have predominantly focused solely on biological and structural factors, overlooking the broader multidimensional nature of DB. This trend is also consistent with existing definitions of the condition previously discussed, which tend to prioritize biological aspects over a more integrative perspective.

The lack of agreement surrounding the underlying nature of the DB and symptom overlap with associated conditions poses further barriers to achieving definitional and phenotypic consensus. A further consequence of this is a lack of agreement in how DB should be assessed and diagnosed. The absence of a universally accepted definition, gold-standard diagnostic criteria, or standardized assessment tools creates a significant challenge in establishing a consensus on the optimal assessment method (9,10). It has also been suggested that this may be a contributing factor to misdiagnosis rates in DB patients (6). While various tools for assessing different aspects of DB are discussed in the literature, their use in diagnosis is limited due to a lack of validation

within this population (5). For instance, our findings showed the Nijmegen Questionnaire (NQ) to be the most frequently cited tool used to assess DB. This is despite it not being designed for this purpose, having been initially developed to screen for 'hyperventilation complaints' (22,46). Some authors have confirmed that the NQ was validated against the hyperventilation provocation test, however it may lack sensitivity to detect non-hyperventilation DB patterns and lacks rigorous validation in these contexts (5,84). This observation becomes yet more problematic in light of our earlier discussion surrounding the persistent interchangeable use of DB with other diagnostic labels such as HVS.

Twelve of the reviewed articles (16%) commented on the need for a multidimensional approach to assessment and diagnosis, highlighting that combinations of tools are required for a comprehensive assessment of DB. Kopitović and Mirić suggest that combining different assessment tools could enhance diagnostic accuracy (6). A notion supported in a recent literature review highlighted the multidimensional nature of DB renders the use of a single assessment criterion as inadequate (43). Despite this, our findings show there to be no consensus as to what tools, or combination thereof, are required (19). This creates challenges for researchers in comparing and validating findings, and for professionals in providing accurate diagnoses and effective treatments, ultimately impacting patient care.

# **Strengths**

This scoping review offers a comprehensive and methodologically robust exploration of how dysfunctional breathing (DB) is characterised in the clinical literature. Guided

by the Joanna Briggs Institute (JBI) framework and adhering to the PRISMA-ScR guidelines, the review systematically synthesised evidence from 71 studies, selected from over 6,000 screened records. The inclusion of a wide range of study designs—quantitative, qualitative, and mixed-methods—enabled a multidimensional understanding of DB across diverse healthcare settings and populations. The application of broad inclusion criteria ensured a multidisciplinary perspective, which is essential given the complex nature of DB. The thematic synthesis—encompassing definitions, diagnostic approaches, symptoms and phenotypes, comorbidities, psychophysiological associations, and classification systems—offers a structured and comprehensive overview that fills a critical gap in current literature.

#### Limitations

Several limitations must be acknowledged. The primary challenge stemmed from the marked heterogeneity in terminology, definitions, and conceptual frameworks used across studies, which limited the ability to synthesise findings cohesively and draw generalisable conclusions. The absence of critical appraisal—typical in scoping review methodology—means the quality of included studies was not formally assessed, necessitating cautious interpretation of reported findings, especially concerning diagnostic tools and classification systems. The restriction to English-language literature introduces a potential language bias, which may have excluded pertinent international research. Moreover, the frequent co-occurrence of comorbid conditions within the study populations introduces confounding variables, complicating efforts to characterise DB as an independent clinical entity.

### Recommendations

To advance both clinical practice and research, the development of a consensus definition of DB is imperative. Such a definition should encompass its multidimensional nature—biomechanical, psychophysiological, and biochemical—moving beyond historical, reductionist labels such as hyperventilation syndrome. The Delphi technique may offer a structured method for achieving expert consensus on terminology, classification, and diagnostic criteria. Additionally, future research should prioritise the validation of a multidimensional assessment tool that accurately captures the breadth of DB phenotypes. This tool should reflect the complexity of the condition and allow for differentiation from overlapping disorders, such as asthma or long COVID. Consistent use of terminology, standardised outcome measures, and refined inclusion criteria are essential to improving the comparability and clinical applicability of future research findings.

### Conclusion

This review reveals significant inconsistency and conceptual ambiguity in the characterisation of dysfunctional breathing within clinical literature. Despite increasing scholarly and clinical interest, the absence of a unified definition, standardised classification, and validated diagnostic criteria continues to hinder effective identification, management, and research progression. Recognising DB as a multidimensional construct is a necessary step toward improving diagnostic accuracy and enabling more targeted, patient-centred interventions. Establishing conceptual clarity through consensus-building efforts will enhance the reliability of future studies

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and improve clinical outcomes for individuals affected by this complex and often underrecognised condition.

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