

# Introduction

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Sleep-related breathing disorders, particularly obstructive sleep apnoea (OSA) syndrome (OSAS), are highly prevalent and represent an increasing part of clinical respiratory practice. OSAS is now recognised to be one of the most common chronic respiratory disorders, with only asthma and possibly chronic obstructive pulmonary disease having a higher prevalence. Thus sleep apnoea represents an important topic for the *European Respiratory Monograph*, and the present volume provides a comprehensive overview, with chapters ranging from basic mechanisms to clinical practice. This issue provides an update of a previous Monograph on *Respiratory Disorders during Sleep* published in 1998 [1].

Humans spend up to a third of life asleep, and yet surprisingly little is known about the biological role of sleep. There is general agreement that sleep is beneficial. William Shakespeare's Macbeth described sleep as the "chief nourisher in life's feast", and the age-old remedy for the sick person is to get "plenty of sleep". Some classical writers, however, have viewed sleep with deep suspicion. Sir Thomas Browne wrote "Sleep, in fine, is so like death, I dare not trust it with my prayers", and Alfred, Lord Tennyson described sleep as "death's twin brother".

There have been many references to presumed sleep apnoea in the classical literature and throughout history, including several kings of ancient Greece, and a detailed review of the historical aspects of OSAS is provided in the previous Monograph on *Respiratory Disorders during Sleep* [2]. Furthermore, the giant in the tale of Jack and the beanstalk very probably suffered from OSAS, and Charles Dickens's description of Joe, "a wonderfully fat boy", in *The Posthumous Papers of the Pickwick Club*, clearly demonstrates features of OSAS. Dickens noted that Joe snored heavily, "as if the roaring of cannon were his ordinary lullaby". He was plethoric, "red-faced", had dropsy (peripheral oedema), cognitive dysfunction ("the fat boy's perception being slow") and "divided his time into small alternate allotments of eating and sleeping". These descriptions strongly indicate that Joe suffered from the sequelae of untreated OSAS, with excessive daytime sleepiness, intellectual impairment and cor pulmonale. The term Pickwickian was first used by William Osler, in 1905, to describe similar patients with hypersomnolence and obesity, and was used to describe many early cases of sleep apnoea reported in the medical literature [2]. However, this term is rarely used today, as there may be different causes of obesity and hypersomnolence.

Although sleep apnoea was first described as a specific clinical entity in the late 1950s, there are several descriptions in earlier clinical journals that clearly refer to the disorder. The sleeping characteristics of obstructive apnoea were clearly described by BROADBENT [3] in the late nineteenth century: "there will be perfect silence through two, three, or four respiratory periods, in which there are ineffectual chest movements; finally air enters with a loud snort, after which there are several compensatory deep inspirations".

The practice of modern sleep medicine and the investigation and diagnosis of sleep-related breathing disorders owes much to the development of appropriate diagnostic tools and techniques. The pivotal discovery of electroencephalography by BERGER [4], in 1930, demonstrated clear differences between wakefulness and sleep. The use of the electro-oculogram by

ASERINSKY and KLEITMAN [5], in the early 1950s, revealed the presence of slow and rapid eye movement (REM) during sleep, and they observed that REM is associated with a lightening of sleep, dreaming and a number of cardiovascular and respiratory phenomena. The association of REM sleep with muscle relaxation was later made by JOUVET *et al.* [6].

During the late 1950s and early 1960s, several different clinical reports described the causative associations between obesity, chronic hypoventilation, pulmonary hypertension and cor pulmonale, but the excessive sleepiness was typically attributed to hypercarbia and depression of the respiratory centre. Cor pulmonale was associated with tonsillar and adenoidal hypertrophy and micrognathia in other reports, but the underlying aetiology was not clarified. In 1965, GASTAUT *et al.* [7] provided the first comprehensive account of OSAS, describing polysomnography in obese hypersomnolent patients with frequent nocturnal apnoeas. During the following decade, the clinical and pathophysiological features of OSAS were described, and it became clear that the pathophysiological basis of obstructive apnoea fundamentally relates to a narrowing of the upper airway, which is partly genetic in origin, but to which acquired factors, such as obesity, also contribute. This narrowing compromises the balance between collapsing forces affecting the upper airway during inspiration and the counteracting forces of upper airway dilating muscles. Although early research focused on clinical and pathophysiological aspects, recent research has increasingly focussed on genetic and molecular aspects, particularly in the development of comorbid conditions, such as cardiovascular and metabolic disease. OSAS is now recognised to be highly prevalent in disorders such as congestive heart failure, systemic hypertension and renal failure. Although much has been learnt, substantial knowledge deficits remain, and the basic mechanisms and consequences of OSAS represent an exciting area for future research.

The relatively recent clinical and pathophysiological descriptions of OSAS are surprising given that the disorder is now widely recognised and highly prevalent, affecting  $\geq 4\%$  of adult males and  $\geq 2\%$  of adult females in the developed world, and with this prevalence growing in parallel with the growing prevalence of obesity. However, most of these cases are clinically unsuspected, since the two most common symptoms of loud snoring and a tendency to fall asleep during the daytime are often considered normal variants, and patients frequently do not seek medical attention. Unfortunately, many patients who do seek medical attention are dismissed as having no significant illness, without formal assessment, and it is very common for patients who have been symptomatic for many years to present to sleep clinics.

The failure to recognise clinically significant OSAS is particularly unfortunate for many reasons. First, the condition carries significant morbidity and mortality, and is associated with an increased risk of heart attack and stroke [8], in addition to a significant risk of automobile accidents and injury in the workplace as a consequence of excessive sleepiness [9]. Secondly, the condition is very treatable, and severe forms of OSAS can respond dramatically well to the continued home use of nocturnal nasal continuous positive airway pressure (CPAP) therapy. Additional clinical challenges in the assessment and management of OSAS include the presentation of OSA without sleepiness and the clinical spectrum of OSA in the elderly and in females. Although OSAS has traditionally been regarded as a disease affecting males, there is increasing recognition that the disease is also prevalent in females, particularly after the menopause, and that the clinical manifestations may differ from those in males [10]. OSA is prevalent in the elderly population, but affected patients appear to be relatively less symptomatic, and the disorder may have less severe clinical consequences in this age group [11].

Treatment approaches to OSAS have evolved in parallel with the increased pathophysiological understanding. Most treatment modalities have been mechanistic, with nasopharyngeal airways and tracheostomy being used as a means of bypassing the site of obstruction in the upper airway in the 1970s [12]. However, these treatments were neither indicated nor acceptable to patients without very severe disease. Surgical approaches to enlargement and/or stiffening of the upper airway were also developed, the most widely used being uvulopalatopharyngoplasty, which was described by FUJITA *et al.* [13] in 1981. The development by SULLIVAN *et al.* [14], in 1981, of CPAP

therapy delivered *via* a nasal mask was a crucial development in the history of sleep-related breathing disorders, providing a highly effective but noninvasive modality of treatment for OSAS that revolutionised the whole field of sleep medicine. CPAP is now the mainstay of therapy, particularly in moderate and severe cases of OSAS.

The high prevalence of OSAS has focused attention on simplified approaches to diagnosis, and there is an increasing trend towards diagnosis and therapy in the ambulatory setting. Although polysomnography remains the gold standard for diagnosis, such studies are resource-intensive since they generally require the facilities of a full sleep laboratory and a trained technician. Thus polysomnography is impractical in many sleep centres for routine assessment in the majority of patients with typical clinical presentations of OSAS. An increasing number of limited diagnostic systems are available to meet the high clinical demand, and ongoing research is directed at identifying novel signals in order to simplify and improve the diagnosis of OSAS in the home setting.

## Statement of Interest

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None declared.

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