Sleep-related breathing disorders, particularly obstructive sleep apnoea syndrome (OSAS), are highly prevalent and represent an increasing part of clinical respiratory practice. OSAS represents something of a paradox in clinical medicine. On the one hand, the clinical disorder has been widely recognised only in recent decades, although the sleeping characteristics of OSAS have been recognised in the medical and classical literature for centuries. On the other hand, OSAS is now recognised to be highly prevalent with recent prevalence figures approaching those of asthma and chronic obstructive pulmonary disease (1,2), and there is increasing evidence that OSAS is associated with many adverse sequelae, both behavioural and physical. Behavioural features include daytime sleepiness, impaired memory and concentration, whereas physical consequences include cardiovascular and metabolic disorders (3). 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 (4) 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”.

In 1965, Gastaut and co-authors (5) provided the first comprehensive account of OSAS, describing polysomnography in obese hypersomnolent patients with frequent nocturnal apnoeas. During the following decade, the clinical (6) and pathophysiological (7) features of OSAS were described and it became clear that the pathophysiological basis of obstructive apnoea fundamentally relates to a narrowed upper airway. This narrowing is partly genetic in origin, but acquired factors such as obesity also contribute (8). Upper airway narrowing compromises the balance between collapsing forces affecting the upper airway during inspiration and the counteracting forces of upper airway dilating muscles (9). Although early research focused on clinical and pathophysiological aspects, more recent research has increasingly focused on genetic and molecular factors (10,11), particularly in the development of comorbid conditions, such as cardiovascular and metabolic disease. 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 so highly prevalent, affecting up to 10% of adult males and 3% of adult females in the developed world (2), and this prevalence is growing in parallel with the growing prevalence of obesity. However, many 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 (12).

The failure to recognize 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 (13), in addition to a significant risk of automobile accidents and injury in the workplace as a consequence of excessive sleepiness (14). 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 sleep apnoea without sleepiness and the differing clinical presentations of sleep apnoea in the elderly (15), in children (16), and in females (17). 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 (17). Sleep apnoea is very prevalent in the elderly population, but affected patients appear to be relatively less symptomatic (15), and the disorder may have less severe clinical consequences in this age group.

The physical morbidity of OSAS relates principally to the cardiovascular system (13) although there is increasing evidence of independent associations with metabolic disease also, particularly diabetes mellitus (18). Systemic hypertension occurs in
up to 50% of OSAS patients and studies of the prevalence of sleep apnoea in patients with systemic hypertension demonstrate the disorder to be the most prevalent cause of resistant secondary hypertension in hypertension clinics (19). However, this patient population of patients has a high incidence of other co-existing cardiovascular risk factors such as obesity, hyperlipidaemia, increased age, male sex, smoking history and excessive alcohol intake, which makes the identification of a clear independent association of OSAS with cardiovascular disease more difficult. Nonetheless, there is now convincing evidence from many studies of an independent association of OSAS with hypertension (13) and this association has been reinforced by studies demonstrating a reduction in blood pressure levels with nasal CPAP therapy (20). A growing body of evidence points to an independent link between OSAS and ischaemic heart disease, cardiac dysrhythmias and stroke.

The relationship of OSAS to road traffic accidents has been recognised for many years and various studies have demonstrated an increase in accident rate between 3 and 7 times that of the general population among untreated OSAS patients, which falls to normal levels after successful therapy (14,21). There is also evidence that occupations such as long-haul truck driving are particularly associated with a risk of sleepiness while driving, and an increased risk of accident, particularly where there is evidence of associated OSAS. These findings may not be surprising given the relative sedentary and monotonous nature of this occupation, and the fact that long-haul truck drivers frequently drive for many hours at a time. The risk of accident also calls into question the suitability of untreated OSAS patients to hold a driving licence and many countries have introduced regulations in this regard. However, there is evidence that patients with OSAS who are successfully treated no longer pose an increased accident risk (22), which emphasises the importance of encouraging affected patients to seek medical help and effective treatment.

Treatment approaches to OSAS have evolved in parallel with the increased pathophysiological understanding. The development in 1981 by Sullivan and co-workers 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 revolutionized the whole field of sleep medicine (23). CPAP is now the mainstay of therapy, particularly in moderate and severe cases of OSAS. Surgical approaches to enlargement and/or stiffening of the upper airway were also developed; the most widely used being uvulopalatopharyngoplasty (24) but the efficacy of pharyngeal surgery in the management of OSAS is debated (25). Newer approaches are evolving in the management of OSAS, most notably pacing of the hypoglossal nerve (26). As yet, there is no effective pharmacological management for the disorder but an increasing understanding of the complex pathophysiological nature of the disorder may open new possibilities in the future (27).

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 (28). 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.

References

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