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Review Article

Disparities and genetic risk factors in obstructive sleep apnea

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ABSTRACT

Obstructive sleep apnea (OSA) is an increasingly prevalent condition. A growing body of literature supports substantial racial disparities in the prevalence, risk factors, presentation, diagnosis, and treatment of this disease. Craniofacial structure among Asians appears to confer an elevated risk of OSA despite lower rates of obesity. Among African Americans, Native Americans, and Hispanics, OSA prevalence is increased, likely due in part to obesity. The burden of symptoms, particularly excessive daytime sleepiness, is higher among African Americans, although Hispanics more often report snoring. Limited data suggest that African Americans may be more susceptible to hypertension in the setting of OSA. While differences in genetic risk factors may explain disparities in OSA burden, no definitive genetic differences have yet been identified. In addition to disparities in OSA development, disparities in OSA diagnosis and treatment have also been identified. Increased severity of disease at diagnosis among African Americans suggests a delay in diagnosis. Treatment outcomes are also suboptimal among African Americans. In children, tonsillectomy is less likely to cure OSA and more commonly associated with complications in this group. Among adults, adherence to continuous positive airway pressure (CPAP) is substantially lower in African Americans. The reasons for these disparities, particularly in outcomes, are not well understood and should be a research priority.

1. Background

Obstructive sleep apnea (OSA) is one of the most prevalent sleep disorders with moderate to severe disease affecting up to 17% of middle-aged men and 9% of middle-aged women [1]. OSA is associated with numerous adverse consequences including excessive daytime sleepiness, motor vehicle accidents, hypertension, and cardiovascular disease (CVD) [2]. A large body of literature has identified risk factors for OSA, consequences of the disease, and treatment options. However, studies evaluating the extent to which the development, presentation, consequences, and management of OSA vary by race have not been as extensively considered. This article will review known differences in OSA by racial background as well as point out areas where further research is needed.

2. Disparities in OSA prevalence

Few studies have directly compared the prevalence of OSA across racial groups. In addition, the lack of consistent criteria to define OSA limits comparisons of OSA prevalence across studies. Nevertheless, available data indicate an elevated prevalence of OSA among African Americans, Hispanics, and Native Americans as compared to US whites, while the prevalence of OSA in Asians appears comparable to whites.

The strongest evidence for a racial disparity in OSA exists with regard to African Americans. Several studies have found a higher rate of OSA in African Americans, particularly African American children [3]. Among pediatric patients evaluated in a sleep clinic, African American race is associated with a 20% increase in OSA severity [4] and greater oxygen desaturation [5]. African American children are four to six times more likely to have OSA compared to white children [6,7]. Even among young adults <26 years of age, African Americans are 88% more likely to have OSA as compared to whites [8]. Among middle-aged populations, the evidence for a disparity in OSA prevalence is weaker as differences in OSA prevalence from community-based studies are evident in some but not all studies [9–12]. By contrast, data from older populations suggest that a disparity may reemerge in this age group. While African Americans had similar prevalence of OSA to whites (32% and 30%, respectively) in a community-based survey of individuals 65 years of age and older, this group was 2.1 times more likely to have severe OSA [13].

Data are somewhat more limited regarding OSA prevalence in US Hispanics. The Hispanic Community Health Study (HCHS) used portable sleep monitoring to evaluate the prevalence of OSA in a diverse US Hispanic cohort of over 14,000 adults. The prevalence of mild, moderate, and severe OSA in this cohort was 25.8%, 9.8%, and 3.9%, but OSA risk was found to vary substantially by Hispanic background, being greatest among Cuban men. Consistent with other

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racial groups, older age, male gender, and obesity were independent risk factors for OSA in this cohort [14]. Although the prevalence is somewhat greater than estimates of community-based white populations, the monitoring system used is very different making direct comparisons difficult.

A cross-racial survey utilizing overnight oximetry, however, did find a higher rate of OSA in Hispanics compared to whites [11]. By contrast, an analysis of data from one site of the Multi-Ethnic Study of Atherosclerosis (MESA) found that the rate of OSA in Hispanics was similar to whites [15]. However, a more recent analysis evaluating subjects at all MESA sites has reported a higher prevalence in Hispanics [16].

Information about OSA in Native Americans is sparse. The best evidence comes from the Sleep Heart Health Study (SHHS) where the odds of moderate to severe OSA was 1.7 times greater than that found in whites [10].

Unlike African Americans, Hispanics, and Native Americans, the prevalence of OSA in Asians and Asian Americans appears similar or lower than that of whites. In a cross-study analysis comparing Japanese participants in the Circulatory Risk in Communities Study (CRICS) to whites in MESA, the prevalence of OSA among Japanese was roughly half that of whites (18.4% vs. 36.5%) [15]. However, in other studies, Asians have been found to have similar OSA severity to whites [17]. In the Male Study of Osteoporosis (MrOS) cohort of older men, Asian American background was an independent risk factor for OSA [18]. This is consistent with population-based studies from Asia where high rates of OSA have been found in China, Japan, Korea, and India, despite low rates of obesity [15,19–21].

In summary, current data from population-based studies suggest that the prevalence of OSA is greater among African Americans, Hispanics, and Native Americans, although direct comparisons particularly for Hispanics and Native Americans compared to other groups are limited. The greater prevalence in African Americans is particularly notable in younger and older age groups. Asians and Asian Americans appear to have comparable rates of OSA to whites despite markedly lower levels of obesity.

3. OSA risk factors

Understanding the basis of disparities in OSA prevalence requires an evaluation of disparities in the risk factors for OSA as well as an assessment of racial heterogeneity in how risk factors contribute to OSA pathogenesis. Craniofacial shape and obesity are among the most studied OSA risk factors.

3.1. Craniofacial shape

Craniofacial shape has been recognized as an important contributor to OSA risk. Both skeletal features, such as maxillary–mandibular shape, inferior hyoid position, and small cranial base, and soft tissue features, such as size of the tongue, soft palate, tonsils, pharyngeal walls, and parapharyngeal fat pads, have been identified as OSA risk factors. In general, studies suggest that soft tissue factors may be more relevant to predicting risk in African Americans while skeletal features are more predictive in Asians [22–24].

Studies comparing African Americans to whites have found tongue area to be significantly larger in African Americans with OSA. By contrast, skeletal features such as brachycephaly (a skull shape with a greater lateral compared to anteroposterior dimension) were predictors of OSA severity in whites but not African Americans [25]. By contrast, Asians with OSA have more skeletal restriction than their white counterparts as measured by a shorter cranial base as well as difference in length and positioning of the maxilla and mandible [26–30]. In addition, both an inferiorly positioned hyoid and an extended craniocervical angle have been demonstrated to predict OSA risk in Asians [27,28]. However, it is important to note that heterogeneity does exist across Asian backgrounds in the relationship between craniofacial risk factors and OSA [31].

Table 1
Soft tissue and skeletal risk features associated with OSA, with racial/ethnic differences noted.

Craniofacial risk factors for OSA
Soft Tissue
Tongue
• Enlarged in African Americans with OSA
Skeletal
Brachycephaly
• Predictor of OSA among Caucasians, not among African Americans
Midface length
• Shorter in Asians with OSA
Cranial base
• Shorter and extended angle in Asians with OSA
Maxilla
• Shorter length predicts OSA in Asians
• Retroposition may be associated with OSA in Hispanics and Asians
Mandible
• Length and position predict OSA in Asians
Hyoid
• Inferiorly positioned in Asians and Caucasians with OSA
Anatomical imbalance (tongue area relative to intermaxillary length)
• Large tongue area relative to intermaxillary length associated with OSA in Caucasians but not African Americans or Asians

As compared to African Americans and Asians, there are much sparser data on the relationship between craniofacial shape and OSA risk in Hispanics and Native American groups. Only a few studies have evaluated differences in craniofacial shape between Hispanics and whites that could contribute to differences in OSA risk and these have been inconclusive. One study found bimaxillary retroposition to be more common among Hispanics with OSA than apneics of other races [32]; however, another study did not find any differences between Hispanics and whites [33]. Table 1 summarizes the contributing soft tissue and skeletal contributors to OSA, with racial differences noted where literature is available.

3.2. Obesity

Obesity is one of the strongest risk factors for OSA, with >50% of OSA diagnoses attributable to being overweight [34]. Obesity likely contributes to OSA through increased fat deposition in neck subcutaneous fat and other soft tissue structures as well as a reduction in lung volumes.

Obesity risk varies widely by race. The prevalence of obesity was 37.1% in African American men and 56.6% in African American women in the US in 2012 [35]. As compared to other groups, African Americans have a 51% increased likelihood of obesity, even after age, sex, comorbidities, and socioeconomic factors are considered [36]. The reasons behind the higher obesity rates among African Americans are not fully understood. One potential explanation is decreased physical activity – particularly among African American women [37–39]. This may reflect competing time interests that leave no time for exercise or other physical activities. Indeed, data from the National Health and Nutrition Survey (NHANES) suggest that African Americans are more likely to have no leisure time activity as compared to whites; nearly three-quarters of middle-aged African Americans reported no physical activity during leisure time in 2010, as opposed to 43% of whites [38].

Another potential explanation is a diminished basal metabolic rate and energy expenditure among African Americans as compared to whites [40–42]. By contrast, no consistent differences in caloric intake by race have been identified [38]. Socioeconomic factors may also contribute through effects on access (in terms of proximity and cost) to fresh fruits and vegetables versus processed foods, dietary choices, and walkability of neighborhoods.