

Obstructive Sleep Apnea (OSA)

Obstructive Sleep Apnea (OSA) occurs when the upper airway becomes blocked during sleep and the person cannot breathe properly. Both children and adults can have sleep apnea, but since not everyone is diagnosed, it is hard to know how common it is. If untreated, long term OSA can lead to multiple health problems. The treatment may include the use of **continuous positive airway pressure (CPAP)** therapy.

Causes of airway blockage

The upper airway can be blocked as a result of:

- Enlarged tonsils and adenoids
- Relaxation of the muscles of the throat during sleep
- Excess weight and fat around the airway
- Narrowing of the velopharynx due to a [surgery](#) that corrects palate problems
 - OSA can happen soon after surgery or [years later in adulthood](#)

What happens when the airway is blocked?

- When people cannot breathe normally, their oxygen level (in the blood) decreases and their carbon dioxide level increases.
- The brain sends signals to the muscles to reopen the airway. It also rouses them from deep sleep to light sleep, usually without fully waking them up. Their deep sleep is reduced but they do not realize it.
- In some people, this cycle happens repeatedly (up to hundreds of times) throughout the night, resulting in very broken sleep.

Symptoms of OSA

While sleeping

- Snoring
- Choking or gasping for air
- Breathing temporarily stops
- Restlessness

During the day

- Morning headaches
- Excessive daytime sleepiness
- Poor concentration
- Crankiness or irritability

Please talk to your doctor if you or your child may have OSA.

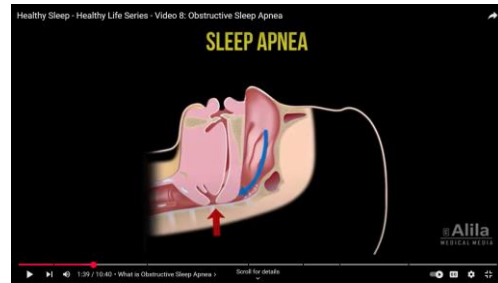
Diagnosis of OSA

A [study](#) showed that sleep symptoms reported by parents do not accurately predict the presence or severity of OSA in their children. The healthcare provider may ask the patient to do a sleep study ([Polysomnography](#)). During the sleep study (at home or in a sleep lab), the equipment monitors the heart, lungs, brain activity, movements, and oxygen levels to check for problems.

OSA and 22q Differences

Remember: Not everyone with OSA is diagnosed.

- A study on 60 [children](#) with 22q11.2 deletion syndrome (22q11.2DS) or 22q11.2 duplication syndrome (22q11.2DupS) showed that ~50% had OSA.
 - Among those who had sleep studies, ~75% were diagnosed with OSA.
- A study on 387 [adults](#) with 22q11.2DS showed that ~10% had OSA.
 - Among those who had sleep studies, ~50% were diagnosed with OSA.



Watch this [video on obstructive sleep apnea](#) from the American Academy of Sleep Medicine

Factors that increase the risk of OSA

Inherent factors:

- Older age
- Being male
- Menopause in females
- Family history

Anatomy:

- Changes in airway due to [palate surgery](#)
- Narrow airway
- Large tonsils or adenoids
- Large tongue
- Small jaw
- Lower jaw sitting farther back than the upper jaw
- Muscles at the throat not being strong enough

Medical conditions:

- **Obesity**
- Asthma
- Nasal congestion
- Others (e.g. 22q11.2DS and 22q11.2DupS)

Lifestyle factors:

- Use of alcohol (which relaxes muscles)
- Smoking

Note: Central sleep apnea is a different type of sleep apnea that happens when the brain fails to send proper signals to the muscles that control breathing. This info sheet is about obstructive sleep apnea, which is more common.

Obstructive Sleep Apnea (continued)

Impact of OSA on Health and Safety

OSA is a long-term disease. If OSA is not treated, it can:

- Affect overall functioning, school performance, and behavior
- Make it harder to learn, focus, think, remember, and plan things
- Increase the risks of mood disorders (e.g. depression)
- Increase the risks for high blood pressure, heart disease, irregular heart rhythms, and stroke
- Increase the risk of accidents when driving or using heavy equipment

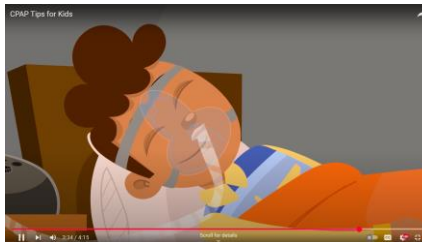
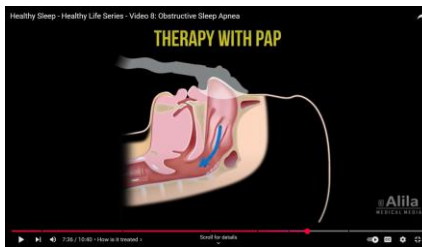
Management of OSA

People are rarely cured from OSA except through weight loss or successful corrective surgery. Medications are generally not very helpful. While mouth appliances can position the jaw for proper breathing, CPAP is the gold standard for treating OSA (see below).

Continuous Positive Air Pressure (CPAP) Therapy

CPAP is a machine that provides a continuous low pressure air flow through a mask, thereby keeping a person's airway open during sleep (see the [video about OSA](#) from the American Academy of Sleep Medicine (AASM)).

CPAP is the recommended treatment for OSA. With guidance, time, practice, and adjustments, most people can get used to the CPAP system and use it for long-term management of OSA. Check out this [CPAP Tips for Kids video](#) from the AASM.



Who are more likely to use CPAP in the long term?

- People who are white
- Older people
- People who used the CPAP machine consistently in the first week

What factors make it unlikely for people to use CPAP in the long term?

- Having sleep apnea even when using CPAP
- Not being able to be close with their bedpartner
- Having a mask that does not fit well or is uncomfortable
- Nasal dryness
- Noise of the machine

22q11.2DS and CPAP

In a [study](#) on adults with 22q11.2DS, 31/39 (79.5%) of the individuals with OSA were prescribed CPAP. 13 (44.8%) were able to use it regularly. These regular users include individuals with moderate to severe intellectual disabilities or schizophrenia.

Resources

- [Obstructive Sleep Apnea \(OSA\)](#) – Cleveland Clinic, USA
- [Obstructive Sleep Apnea](#) – Mayo Clinic, USA
- [Obstructive Sleep Apnea in Children](#) – Cleveland Clinic, USA
- [Pediatric Obstructive Sleep Apnea](#) – Mayo Clinic, USA
- [Pathophysiology of Sleep Apnea](#) – 2010
- [Obstructive Sleep Apnea \(video\)](#) – American Academy of Sleep Medicine
- [Treatment of Adult Obstructive Sleep Apnea with Positive Airway Pressure: An American Academy of Sleep Medicine Clinical Practice Guideline](#) – 2019
- Updated clinical practice recommendations for managing [\[children | adults\]](#) with 22q11.2 deletion syndrome – 2023
- [Adult-onset obstructive sleep apnea and pediatric pharyngoplasty in 22q11.2 deletion syndrome](#) – 2023
- [Polysomnographic findings in children with 22q deletion & duplication syndrome: relationship to genetic diagnosis, parent-reported symptoms, and calcium levels](#) – 2024
- [Velopharyngeal Dysfunction: Surgery](#) – International 22q11.2 Foundation – 2022