Managing OSA in Children and Adolescent Down Syndrome Patients

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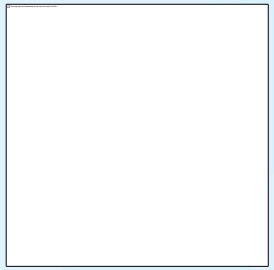
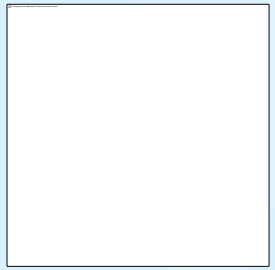
Objectives:

- Define risk factors for obstructive sleep apnea in Down syndrome
- Review guidelines for sleep studies in Down syndrome
- Outline treatment options for OSA and how to monitor usage/effectiveness
- Review/discuss options for hypoglossal nerve stimulation in pediatric Down syndrome

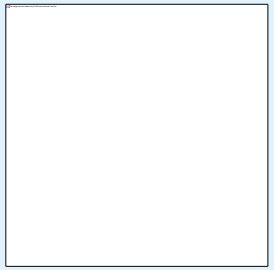
What is Obstructive Sleep Apnea (OSA)



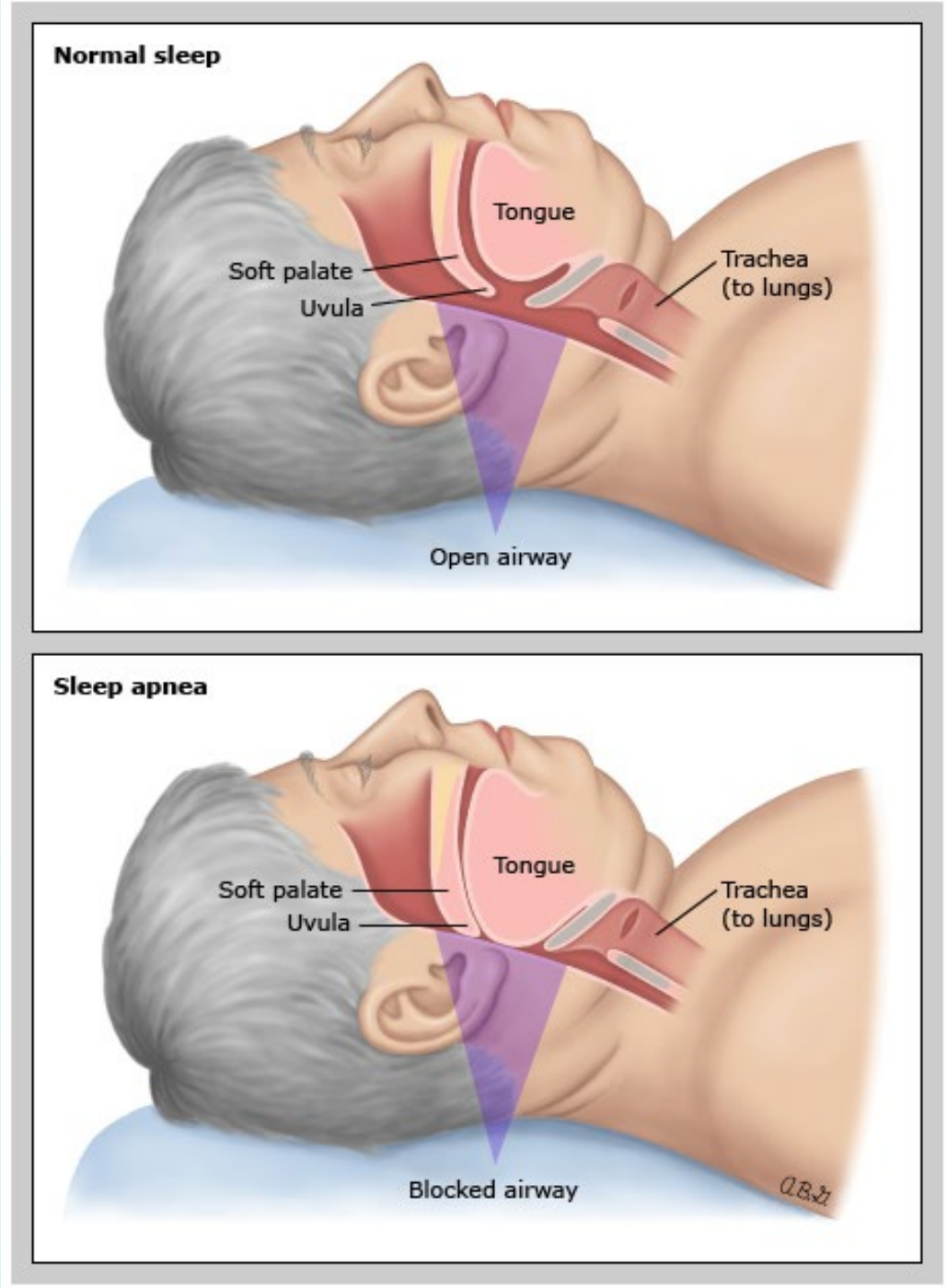
Apnea – absence of breathing



Obstructive apnea – absent breathing that is the result of something getting in the way of normal airflow



Obstructive sleep apnea – when intermittent obstructive apneas occur during sleep



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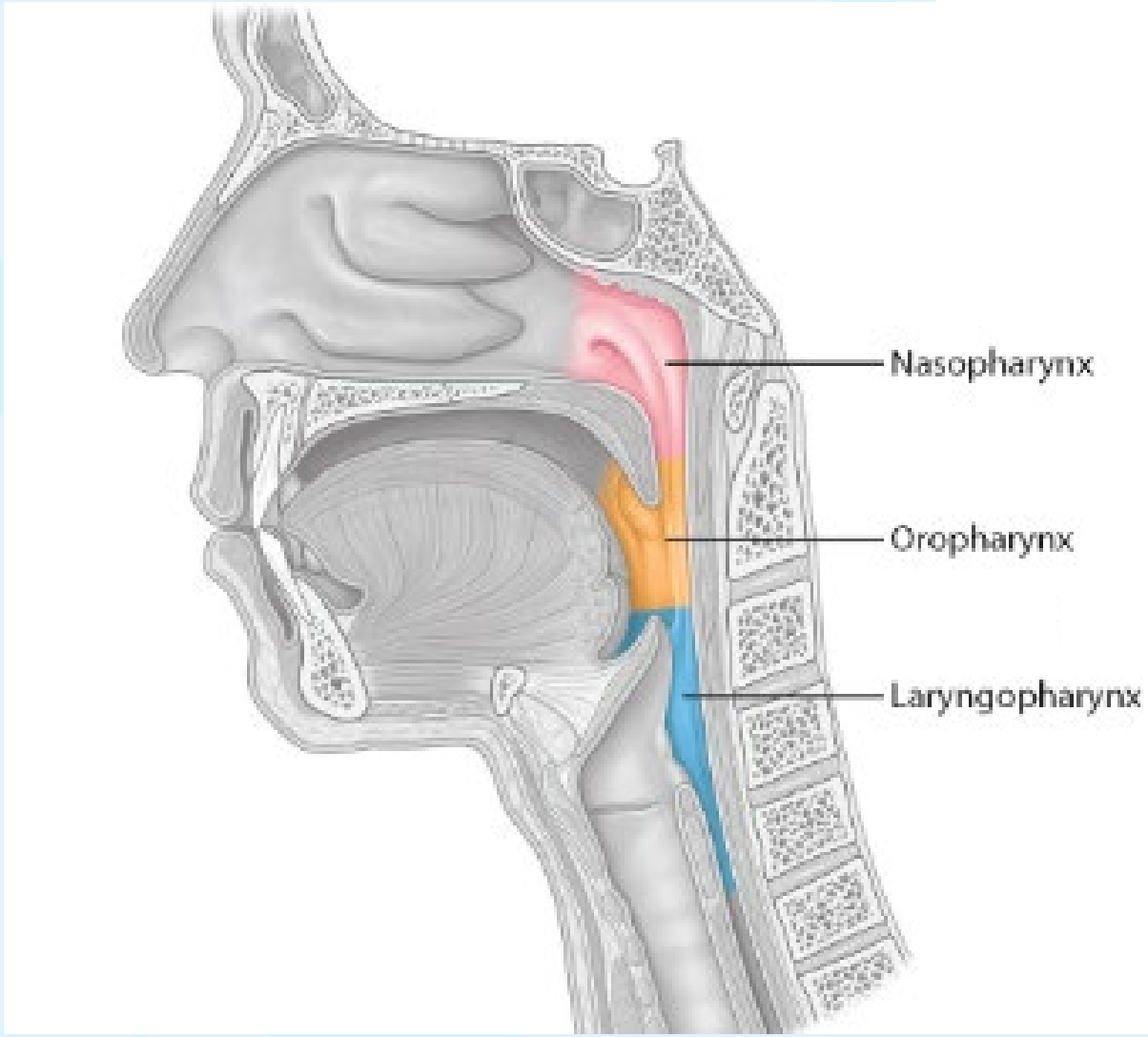
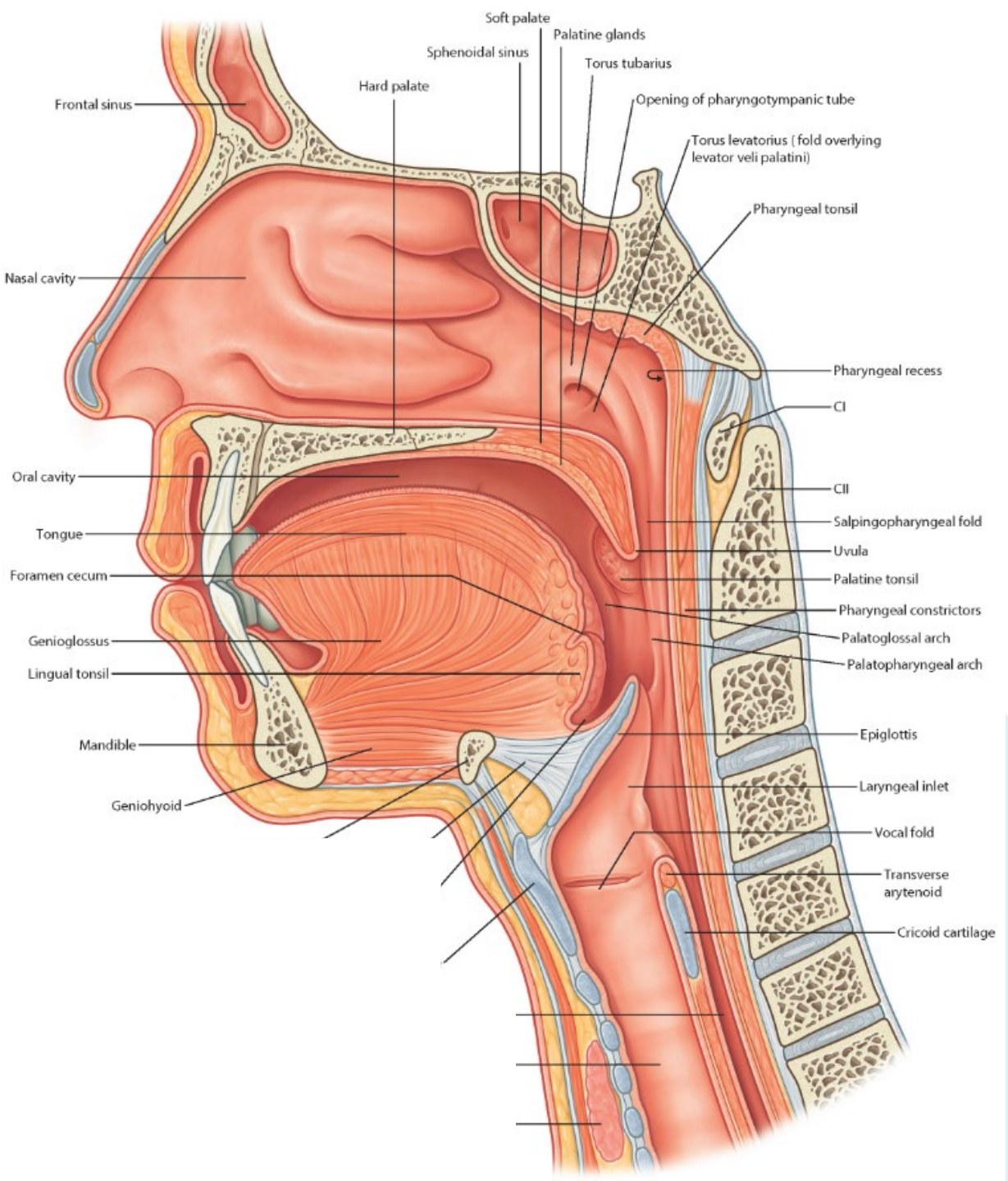
UpToDate





Gray's Atlas of Anatomy

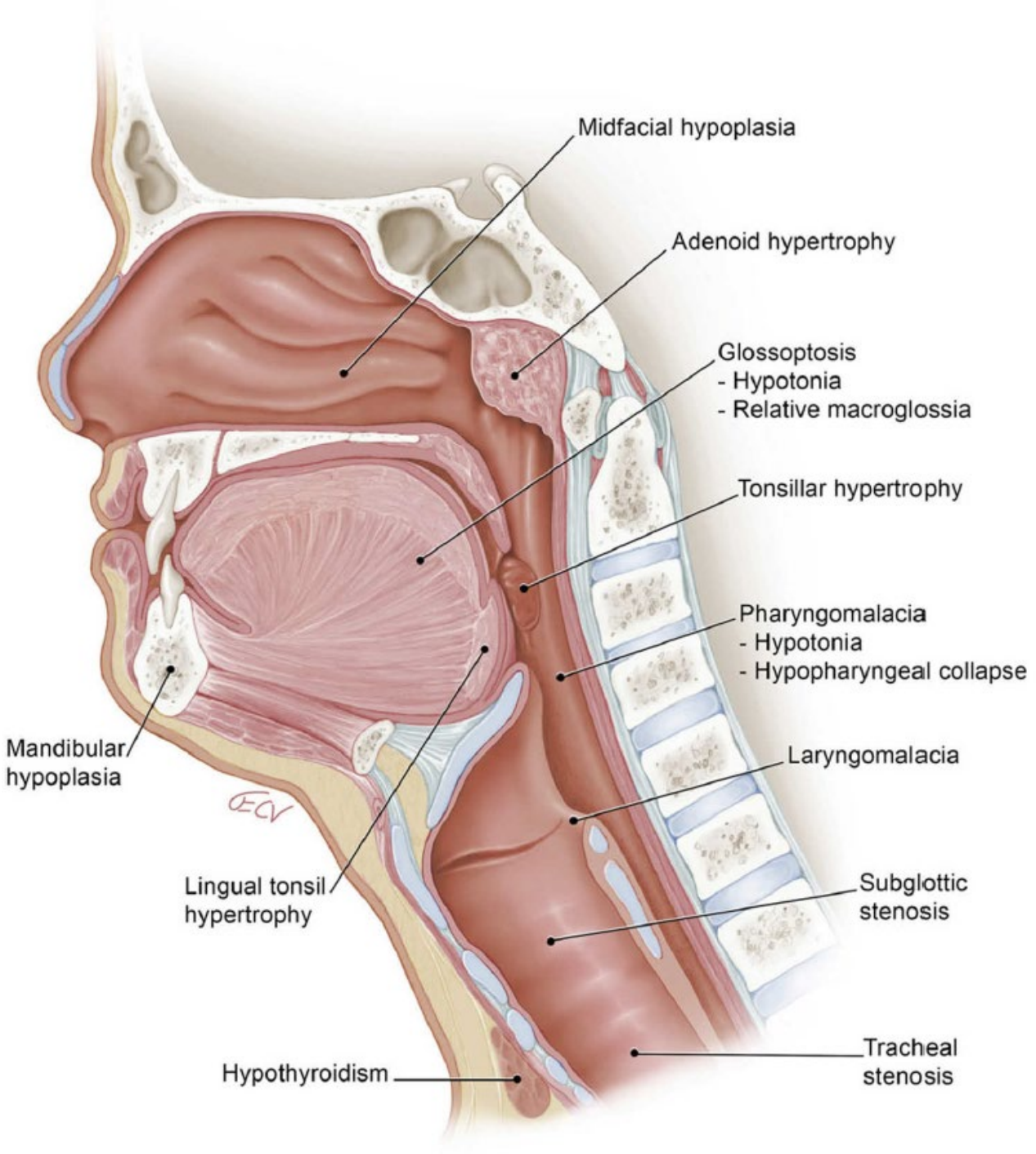
How common is OSA in Down Syndrome?

- Very
- Prevalence of OSA in people with Down syndrome is:
  - 50-80% for children
  - >90% for adults
- For context, the prevalence of OSA in the general population is:
  - 1-5% for children
  - 15% for adult males
  - 5% for adult females

AAP 2022 Guidelines, Ingram 2018 Nat Sci Sleep

Why is OSA common in people with Down Syndrome?

- Enlarged (aka hypertrophied) tonsillar tissues: adenoids, palatine tonsils, lingual tonsils
- Large tongue (aka macroglossia) relative to other upper airway dimensions
- Smaller anatomy of the middle face structures (aka midface hypoplasia)
- Low muscle tone
- Low thyroid levels (hypothyroidis)
- Increased prevalence of obesity
- Gastroesophageal reflux



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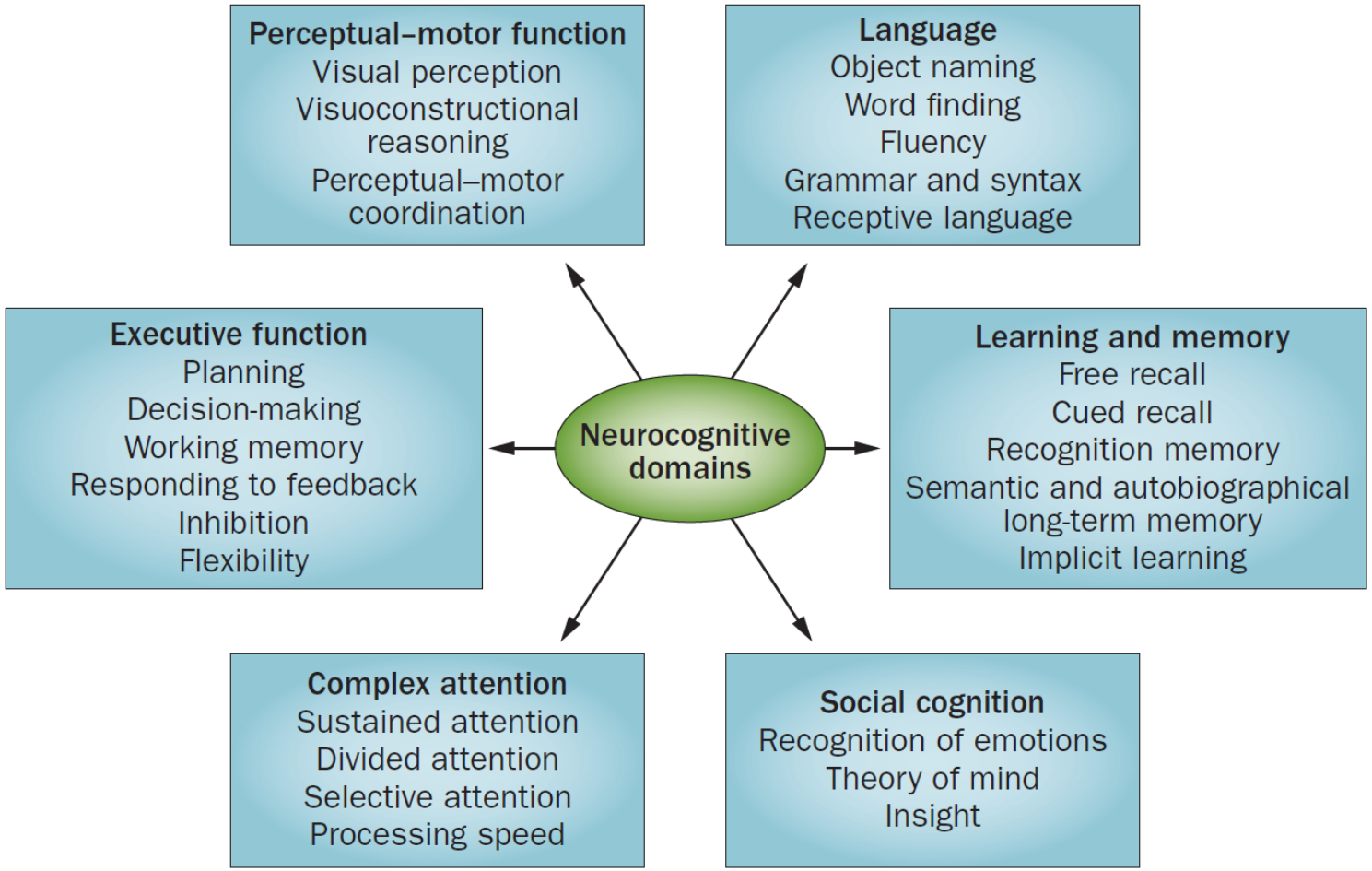
Lal 2015 Chest

Complications of Untreated OSA

Neurocognitive Complications

- Cognition – the mental action or process of acquiring knowledge and understanding; all forms of knowing and awareness
- Neurocognition – cognitive processes in relation to the specific pathways by which they occur in the brain



Sachdev 2014
Nat Rev Neurol

Cardiovascular Complications

- Pulmonary hypertension – high blood pressure specific to the blood vessels of the lungs
- High blood pressure
- Heart failure – can be result of impaired squeeze of heart (termed systolic dysfunction) or impaired filling of heart (termed diastolic dysfunction)
- Abnormal heart rhythms*
- Coronary artery disease*

**More specific to adults*

Other Complications

- ADHD – attention deficit/hyperactivity disorder
- Fatigue
- Bedwetting
- Disrupted sleep / insomnia
- Behavioral difficulties

Signs and Symptoms of OSA

- Aforementioned complications can be the presenting symptom(s)
- Snoring
- Abnormal breathing patterns during sleep
- Observed apnea during sleep
- Restless sleep
- Atypical sleeping positions

❖But…symptoms are not enough

- Studies have shown that 50% of asymptomatic children with Down syndrome will have OSA on an overnight sleep study
- Studies have also shown that symptoms alone do not predict OSA
- Therefore sleep study testing is needed

Diagnosing OSA

- In-lab sleep study (aka polysomnography [PSG]) is the gold standard for diagnosing OSA in children
- Home sleep testing (HST) is not recommended for the diagnosis of OSA in Children (per American Academy of Sleep Medicine)
  - In-lab testing utilizes more sensors, more sensitive sensors, and sleep techs who can adjust sensors
- Down syndrome is an exclusionary reason for HST based on “potential respiratory muscle weakness due to neuromuscular condition – hypotonia”
- Some studies show feasibility for moderate-severe OSA
  - Sensitivity 0.81, Specificity 0.75 (Cielo, J Clin Sleep Med, 2023)

What's Involved with a Sleep study

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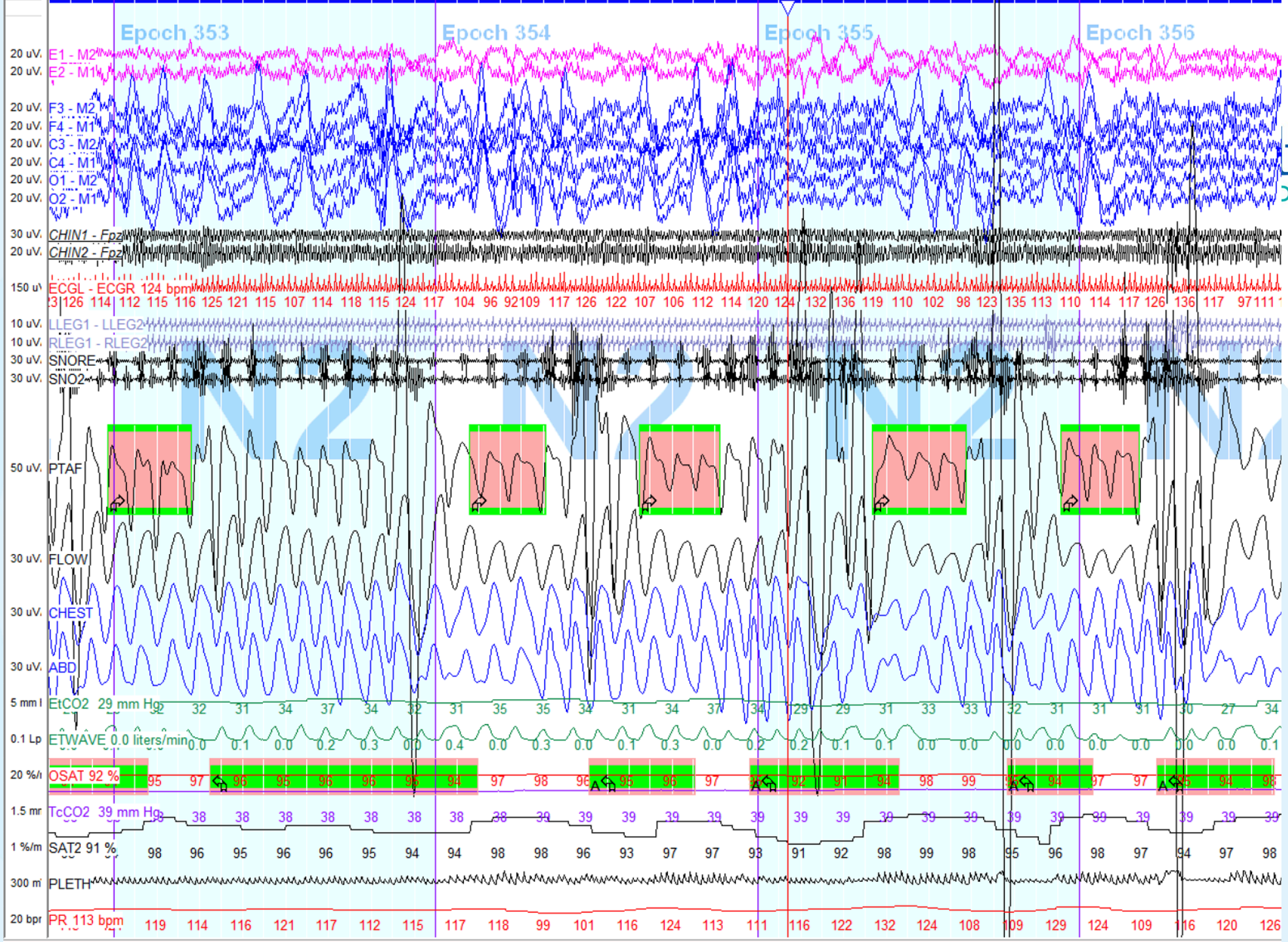
Preparing for your Sleep Study at Children's – YouTube

1:00-2:17

Getting Ready for the Sleep Study Equipment

- Desensitization Kits
- Sleep psychology

Nasal Cannula Placement - YouTube


Epoch 353
Epoch 354
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Epoch 356
E1 - M2
E2 - M1
F3 - M2
F4 - M1
C3 - M2
C4 - M1
O1 - M2
O2 - M1
CHIN1 - Fpz
CHIN2 - Fpz
ECGL - ECGR 124 bpm
LLEG1 - LLEG2
RLEG1 - RLEG2
SNORE
SNO2
PTAF
FLOW
CHEST
ABD
EtCO2 29 mm Hg
ETWAVE 0.0 liters/min
OSAT 92 %
TcCO2 39 mm Hg
SAT2 91 %
PLETH
PR 113 bpm
N2

Sleep study result terminology

- Apnea: ≥90% reduction in airflow for 2 breaths
  - Obstructive apnea: apnea associated with obstructed airflow (trying to breathe, but can't get the air in)
  - Central apnea: apnea associated with absent or reduced of respiratory effort (not trying to breathe)
- Hypopnea: ≥30% reduction in airflow for 2 breaths that is associated with an arousal or oxygen desaturation
  - Obstructive hypopnea: hypopnea associated with obstructed airflow (trying to breathe, but can't get the air in)
  - Central hypopnea: hypopnea associated with absent or reduced of respiratory effort (not trying to breathe as deep)
- Apnea hypopnea index (AHI): number of obstructive apneas and obstructive hypopneas averaged out over the total sleep time; reported as average number of events per hour

Screening for OSA

- As previously mentioned, there is poor correlation between observed symptoms and sleep study results
- Therefore, the American Academy of Pediatrics (AAP) recommends an in-lab sleep study for all children with Down syndrome by 4 years of age.
- Inquire about symptoms of sleep-disordered breathing at all Health Maintenance Visits into adulthood

Action	Pre-natal	Birth up to 1 mo	1 mo up to 1 yr	1 yr up to 5 yr	5 yr up to 12 yr	12 yr up to 21 yr
21. Assess for sleep-disordered breathing; if present, refer to physician with expertise in pediatric sleep disorders.			At least once by 6 mo, then all subsequent HMV thereafter			
27. Obtain polysomnogram.				Between 3-5 yr		

AAP 2022 Guidelines

Guidelines for Sleep Study Prior to Tonsillectomy

5. Indications for polysomnography	Before performing tonsillectomy, the clinician should refer children with obstructive sleep-disordered breathing (oSDB) for polysomnography (PSG) if they are <2 years of age or if they exhibit any of the following: obesity, Down syndrome, craniofacial abnormalities, neuromuscular disorders, sickle cell disease, or mucopolysaccharidoses.	Recommendation

Table 6. Role of PSG in Assessing High-Risk Populations before Tonsillectomy for oSDB.

Role of PSG	Rationale
Avoid unnecessary or ineffective surgery in children with primarily nonobstructive events	Identify primarily nonobstructive events or central apnea that may not have been suspected prior to the study and may not benefit from surgery
Confirm the presence of obstructive events that would benefit from surgery	The increased morbidity of surgery in high-risk children requires diagnostic certainty before proceeding
Define the severity of oSDB to assist in preoperative planning	Children with severe OSA may require preoperative cardiac assessment, pulmonary consultation, anesthesia evaluation, or postoperative inpatient monitoring in an intensive care setting
Provide a baseline PSG for comparison after surgery	Persistent OSA despite surgery is more common in high-risk patients than in otherwise healthy children
Document the baseline severity of oSDB	High-risk patients are more prone to complications of surgery or anesthesia

Abbreviations: OSA, obstructive sleep apnea; oSDB, obstructive sleep-disordered breathing; PSG, polysomnography.

Otolaryngology Guideline 2019

Treatment

- Mild Disease
  - Nasal steroid spray
  - Control of nasal allergies, reflux, low-thyroid levels
  - Healthy weight management
- Moderate & Severe Disease
  - Surgery
    - T&A (tonsillectomy & adenoidectomy)
    - Lingual tonsillectomy
    - Tongue base reduction surgery
    - Supraglottoplasty surgery for laryngomalacia
  - CPAP (continuous positive airway pressure)
  - Inspire – hypoglossal nerve stimulator
  - Healthy weight management

❖Dental appliances not typically used because of sub-optimal efficacy in setting of multilevel obstruction (only 50-60% reduction in AHI)

Sleep Endoscopy can be helpful to determine sites of obstruction and possible further surgical treatment

(multilevel collapse is common)

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Efficacy of T&A for OSA in down syndrome

- In general population T&A is curative in up to 80% of cases
- Studies suggest that 50 – 75% of children with Down syndrome and OSA will have residual OSA after T&A due to multi-level nature of obstruction
  - **However**…most children with Down syndrome and OSA will have substantial improvement in objective sleep study parameters after T&A
  - So, even though a large number of children with Down syndrome continue to have OSA after T&A, their OSA is still improved
  - On average children with Down syndrome have a 50% reduction in their AHI after T&A
  - Much easier to treat with CPAP if lower AHI present (lower cmH2O)

Ingram 2018 Nat Sci Sleep

CPAP Therapy

- Can be highly effective, downside is it only has an effect when it is being used
- “Playing the long game” – focus at initiation is on gradual desensitization
- Cellular modem allows for frequent evaluation and adjustments

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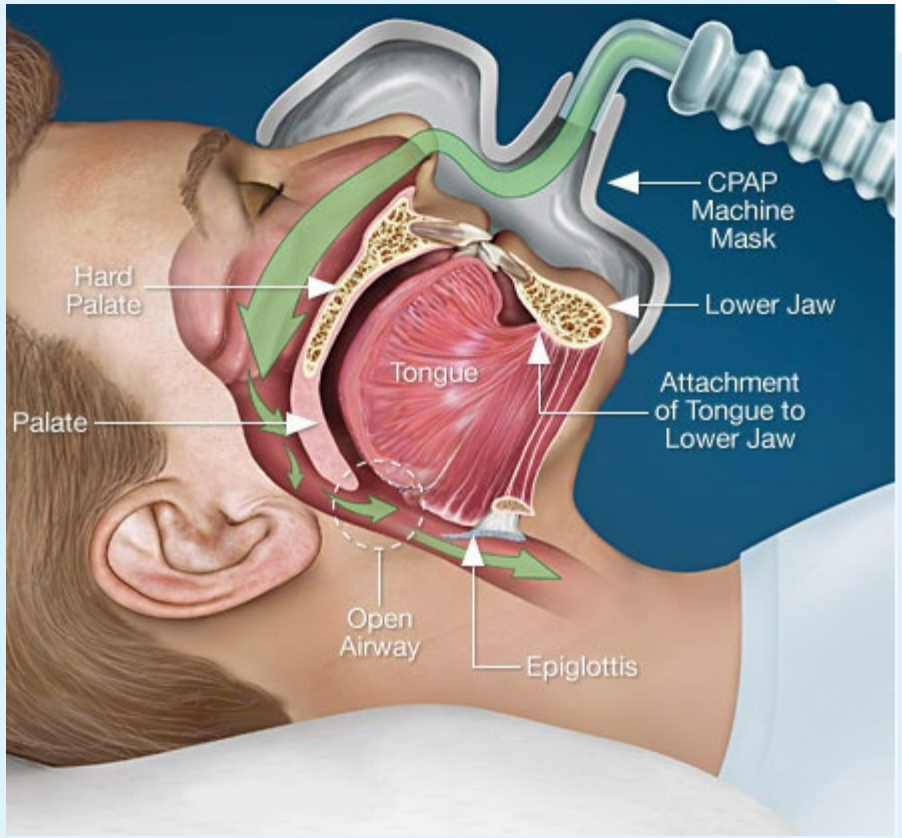
Wisp

Devices Used

- Most companies have a download capability
- Integrated modems are ideal
- Patient/parent portal also helps with compliance
  - Kids like reviewing data on iPhone/iPad
  - Parents (and myself) like to point out children that are lying about their CPAP use
- I prefer a report that allows waveform views
  - Algorithm for adults sometimes identifies CSA as OSA in children
  - Likely due to small airways and pressure pulse “bouncing” off the carina (my explanation anyway)

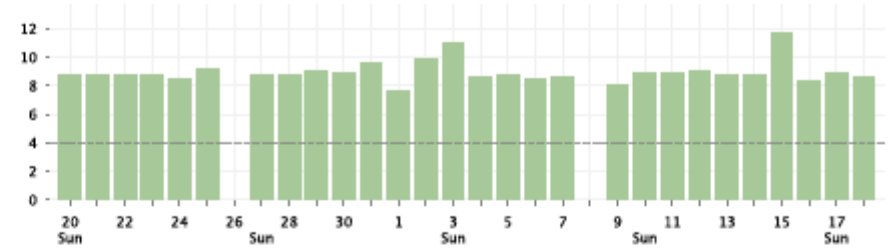
Pressure Pulses

- Machine delivers short burst of pressure when it detects cessation of breathing
- If pressure “bounces back” = obstruction
- If pressure dissipates = central
- Small airways in kids can give some signs of “bouncing back” even though event was a central pause



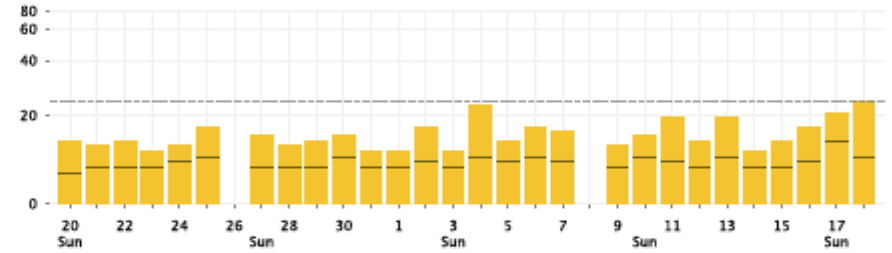
Usage (hours)

Usage days	28/30 (93%)
>= 4 hour days	28 (93%)
< 4 hour days	0 (0%)
Days not used	2 (7%)
Days no data	0 (0%)
Used/day (avg.)	9.0 hrs.



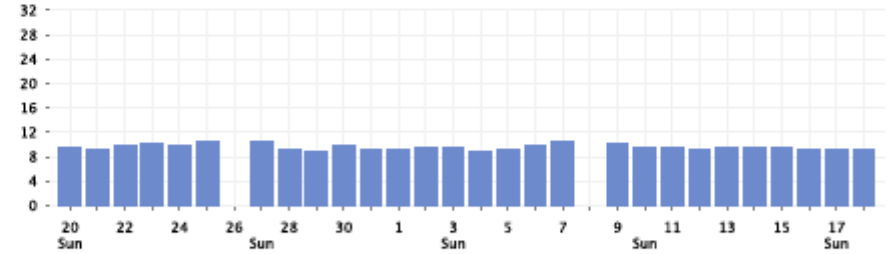
Leak (L/min)

Set threshold	24.0 L/min
Maximum (avg)	22.3
95th % (avg)	14.7
Median (avg)	8.2



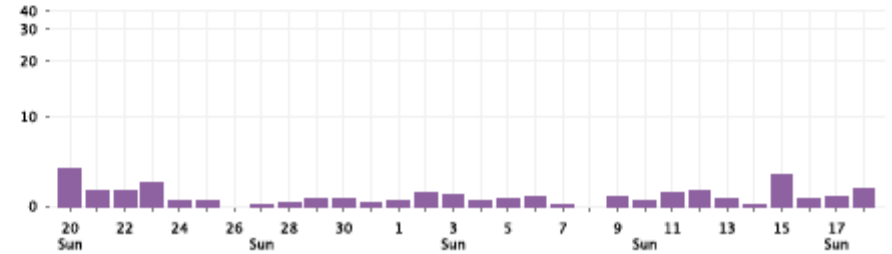
Pressure (cmH2O)

Mode	AutoSet
Set EPR	Fulltime, 3.0
Set Max Pressure	15.0
Set Min Pressure	8.0
Maximum (avg)	10.4
95th % (avg)	9.5
Median (avg)	8.2



AHI (events/hour)

AHI	1.0
HI	0.0
AI	1.0
CAI	0.7
OAI	0.2
UAI	0.0



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Clinic vs Telemedicine

IMPRESSION:

1. Apnea-hypopnea index has improved since changing pressure range from 5- 10 cmH2O to 5- 7 cmH2O, however, remains elevated.
2. Compliance is good at 93%
3. Mask fit is good.
4. CPAP setting appears inadequate

RECOMMENDATIONS:

1. Keep titration sleep study in sleep lab as scheduled on 8/23/2017. Start CPAP at a pressure of 5 cmH2O, titrate to 7, 9 & 11 cmH2O, then switch to BiPAP 13/7, then 14/8 cmH2O.
2. Further recommendations to be made after the study.

DATA:

PAP/Vent Download Report: 8/21/2017	**PAP/Vent Download Report: 6/27/2017** - personally reviewed & interpreted by myself
Respiratory DME: Children's Home Health	Respiratory DME: Children's Home Health
Modem: Active	Device Name: Home Auto CPAP
Mask Type & Size: AG-small child	Mask Type & Size: AG-small child
Home BiPAP/CPAP: Auto CPAP	EPAP Max (cm H2O): (S) 10 cm H2O
EPAP Max (cm H2O): 7 cm H2O	EPAP Min (cm H2O): 5 cm H2O
EPAP Min (cm H2O): 5 cm H2O	90th %tile Pressure: 9.1
90th %tile Pressure: 6.8	30d Use > 4hrs %: 96.7
30d Use > 4hrs %: 93.3	30d Any Use %: 100
30d Any Use %: 96.7	Avg % Large Leak: 0.4
Avg % Large Leak: 0.3	**AHI: 13.7** (CI 2.9, OI 10.1, HI 0.7)
AHI: 6.8 (CI 0.8, OI 5.4, HI 0.6)	

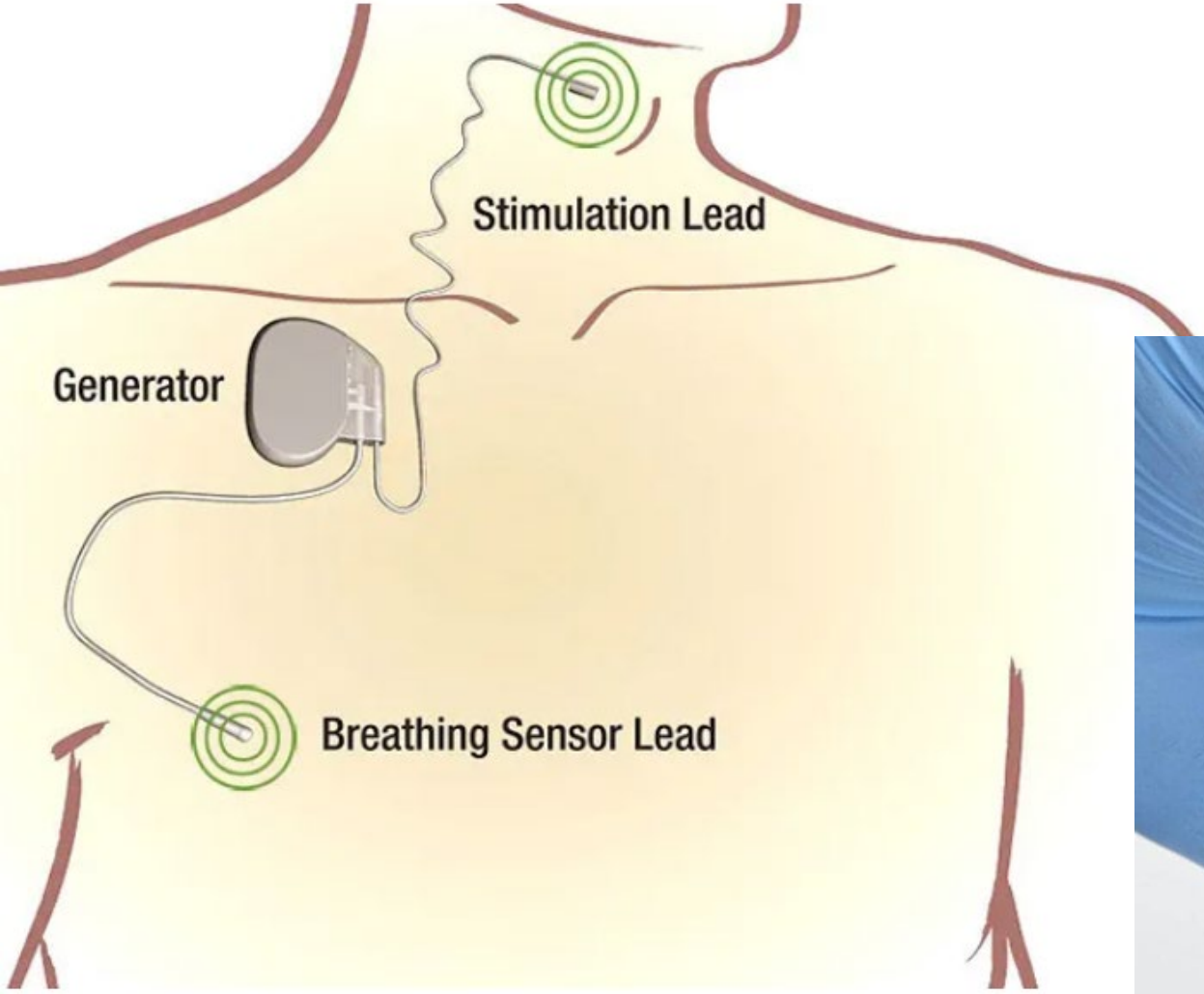
- Use data to generate reports for RPM (remote patient management) visit to avoid frequent travel and to guide therapy
- Helps limit the need for surveillance sleep studies

Trend Oximetry

- Not a replacement for a sleep study
- Can help decide if referral straight to ENT vs obtain a PSG
- Many times use it to monitor for resolution of desaturations following an intervention
  - Post T&A vs medical treatment with nasal steroids +/- Singulair
  - More helpful when sleep study verifies that ODI was main determinant of events scored (i.e. events not scored due to arousals)
- Look more at the desaturation index than I do at the overall oxygen saturations
- Internal QI study compare the sleep study derived ODI with a home trend oximetry derived ODI
  - Good correlation noted (controlled averaging time and desat determination)
  - Average of the ODI from the combined 3% and 4% desaturation report matched the sleep study ODI

Inspire

- System consists of implanted pulse generator (in chest), stimulation lead (in base of tongue), and breathing sensing lead



University Hospitals – 1:28

Metrohealth – 1:10

Prior FDA indications for inspire

- Age >/=18yo
- BMI <35kg/m2
- Moderate to severe OSA (AHI 15-65)
- Predominantly obstructive events (central events <25% of total AHI)
- CPAP failure or intolerance
- Do NOT have complete obstruction of the upper airway
- Are not, or would not be, effectively treated by T&A

Updated pediatric indications for Inspire

- On 3/21/2023 the FDA approved use of the Inspire device for children with Down syndrome who:
  - are at least 13 years old
  - have an AHI of 10-50
  - have had CPAP intolerance/failure

- However, CPAP tolerance is typically pretty good for Down Syndrome
  - 81% compliance for > 4 hrs (Dudoignon, Am J Med Genet, 2017)
  - OCD tendencies tend to help with compliance (regular & consistent schedule)

Literature supporting new pediatric indication for Inspire

- Phase I clinical trial published April 2022. *Yu PK, Stenerson M, Ishman SL, et al. Evaluation of Upper Airway Stimulation for Adolescents With Down Syndrome and Obstructive Sleep Apnea. JAMA Otolaryngol Head Neck Surg. 2022;148(6):522–528.*
- 42 people between the ages of 10 and 22yo underwent implantation of the Inspire system
- At one year follow-up:
  - On average, patients' AHI was reduced by 12.9
  - 30 patients (73.2%) had an AHI under 10; 14 patients (34.1%) had an AHI under; three patients (7.3%) had an AHI under 2
  - Quality-of-life surveys filled out by parents reported significant improvements in daily functioning, behavior, and language.

Thank You !!

Questions??

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