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Vents, Stomas, and Masks, Oh My!

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No Conflicts of Interest to Disclose

Learning objectives:

- Discuss modalities of ventilatory support in CCHS
- Discuss the advantages and disadvantages of different modalities of assisted ventilation
- Describe pathways for tracheostomy decannulation to mask ventilation and diaphragm pacing
- Review the timing and technique of closure of the tracheostomy stoma

Respiratory abnormalities in CCHS

- **Hypoventilation is the hallmark of CCHS**
- **Hypoventilation is worse during sleep but may also be present during wakefulness**
- **Absent subjective or objective responses to hypoxia or hypercapnia**
- **Abnormal or absent arousal responses to hypoxemia**
- **Signs of respiratory distress such as tachypnea, retractions & nasal flaring are absent in CCHS**

Weese-Mayer, D.E., et al. *Amer. J. Resp. Crit. Care Med., 181:* 626-644, 2010
Kasi, A.S., et al. *Pediatric Health Med Ther* 2016:7 99-107

PHOX2B gene and hypoventilation

- **PHOX2B genotype-phenotype correlation for severity of hypoventilation**
- **The duration of assisted ventilation per day generally depends on the PHOX2B genotype**
- **Ventilator requirement may range between sleep-only to full-time dependence**

Weese-Mayer, D.E., et al. *Amer. J. Resp. Crit. Care Med., 181:* 626-644, 2010
Trang H et al (2020). *Orphanet J Rare Dis* 15:252.

Treatment of CCHS

- **Assisted ventilation whenever CCHS patients hypoventilate**
  - **PHOX2B 20/24 and 20/25 PARM usually require ventilatory support only during sleep**
  - **PHOX2B 20/27 or higher & NPARM usually require full-time ventilatory support awake and asleep**
- **Require assisted ventilation throughout life**

Ventilatory Support in CCHS

- **Positive pressure ventilation via tracheostomy**
- **Noninvasive positive pressure (mask) ventilation**
- **Diaphragm pacing (DP)**
- **Negative pressure ventilation**

- **CCHS patients are relatively unstable in the first 2-4 years of life, so ventilation via trach recommended initially.**
- **Consider early DP awake for full-time ventilator dependent patients.**
- **If ventilator dependent only during sleep and stable, consider mask ventilation ~5-6 years of age.**

Weese-Mayer, D.E., et al. *Amer. J. Resp. Crit. Care Med., 181:* 626-644, 2010

Home Ventilation in CCHS

- **Ventilators adjusted to fully meet the ventilatory demands**
- **Goal P_{ETCO2} 35-40 mm Hg, SpO2 ≥ 95%**
- **Settings ideally titrated during sleep study**
- **Monitor with continuous pulse oximetry and P_{ETCO2} during sleep**
- **The modality and changes to alternative modes of ventilation are individualized to each patient based on age, duration of ventilatory requirement, and risk-benefits of different forms of ventilation**

Weese-Mayer, D.E., et al. *Amer. J. Resp. Crit. Care Med., 181:* 626-644, 2010
Trang H et al (2020). *Orphanet J Rare Dis* 15:252

Positive Pressure Ventilation via Tracheostomy

- **Recommended during infancy to optimize oxygenation and neurodevelopment**
- **Pressure control mode preferred**
- **Should include age-appropriate respiratory rate setting**
- **Visual and audible ventilator alarms recommended**
- **2nd ventilator setting could be programmed for use during illness**
- **Relatively smaller and uncuffed trach tubes preferred**
- **Periodic evaluation of airway and tracheostomy size**

Ventilator Sprint Weaning

- During early infancy, full time ventilation is provided
- Later in infancy or childhood, children may develop the ability to breathe spontaneously while awake
- Ventilator sprint weaning: brief disconnection (30-60 min) of ventilator while awake with pulse oximetry and CO_2 monitoring in-hospital
- If tolerated, a sprint weaning progression plan is provided to gradually increase time off the ventilator while awake with pulse oximetry and CO_2 monitoring
- Close medical follow up

Positive Pressure Ventilation via Tracheostomy

Advantages	Disadvantages
• Effective during baseline health and illness • Provides continuous prolonged ventilation • Airway is secured, prevents obstructive apneas • Facilitates suctioning of secretions • Can be used in all age groups • Newer, smaller ventilators with longer battery life	• Requires tracheostomy • Increased burden of care (trach care, suctioning) • Increased cost • Risk of speech delay and swallowing problems • Trach-related complications: infections, bleeding, granuloma

Noninvasive (Mask) Ventilation

Indications:

- **For patients with late-onset CCHS**
- **For patients with tracheostomy requiring sleep-only ventilation for tracheostomy decannulation**

- **Not generally recommended in infants and young children**
- **BPAP most often used**
- **BPAP ST or T modes should be used (has respiratory rate)**
- **BPAP S mode and CPAP modes should not be used**
- **Use home ventilator instead of BPAP device**

Noninvasive (Mask) Ventilation

Careful mask selection, a well-fitting headgear and mask acclimation are important for adequate mask adaptation in children.

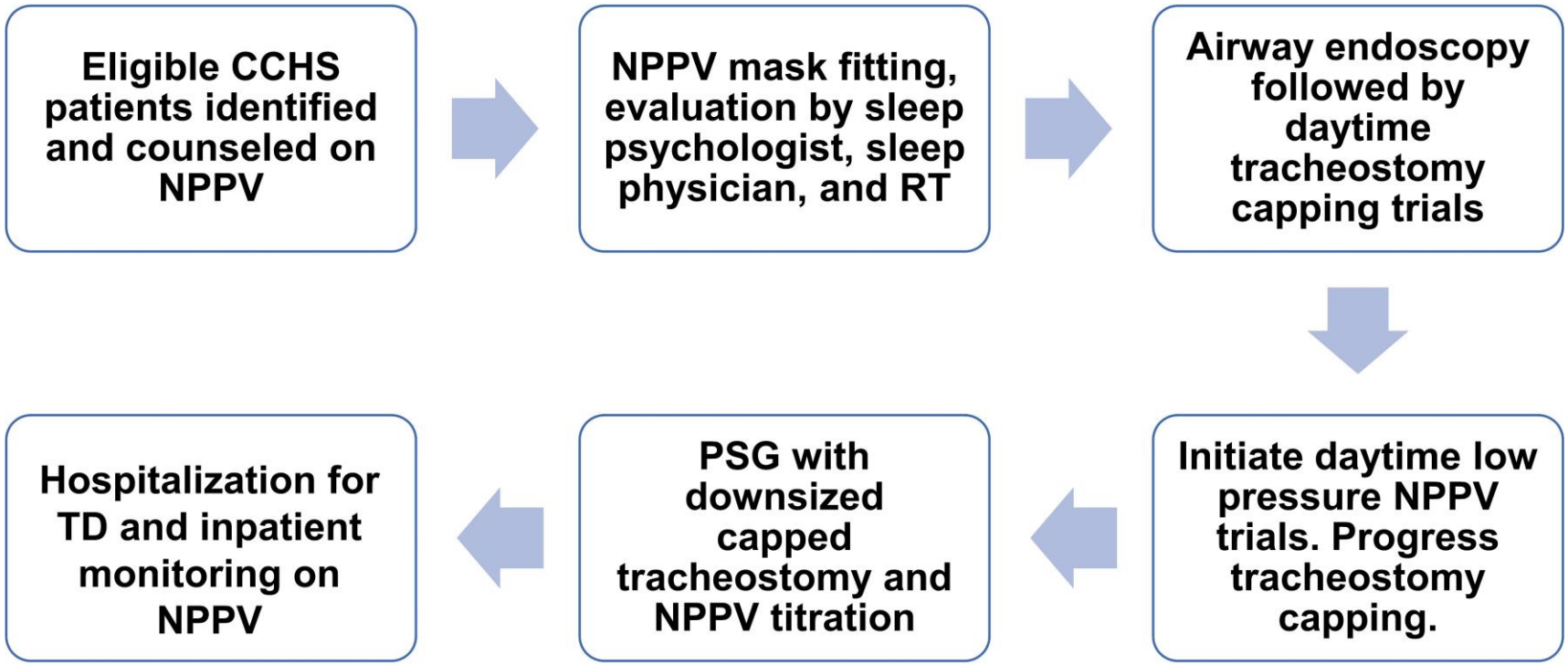
M.L. Castro-Codesal. Paediatr Respir Rev 32 (2019) 66–72
Amin R. Pediatr Pulmonol 2016 Apr;51(4):335-48

Noninvasive ventilation

- **Eligible patients for mask ventilation seeking tracheostomy decannulation:**
  - **Age ≥6 years, cooperative**
  - **Requires sleep-only ventilation**
  - **Does not require full-time ventilation with illness**
  - **Stable without frequent admissions**
- **BPAP settings should be titrated during sleep study**
  - **Goal PtcCO2 35-40 mm Hg; SpO2≥ 95%**

NPPV Titration Task Force of the American Academy of Sleep Medicine.
J Clin Sleep Med 2010;6(5):491-509

Transition from tracheostomy to NPPV



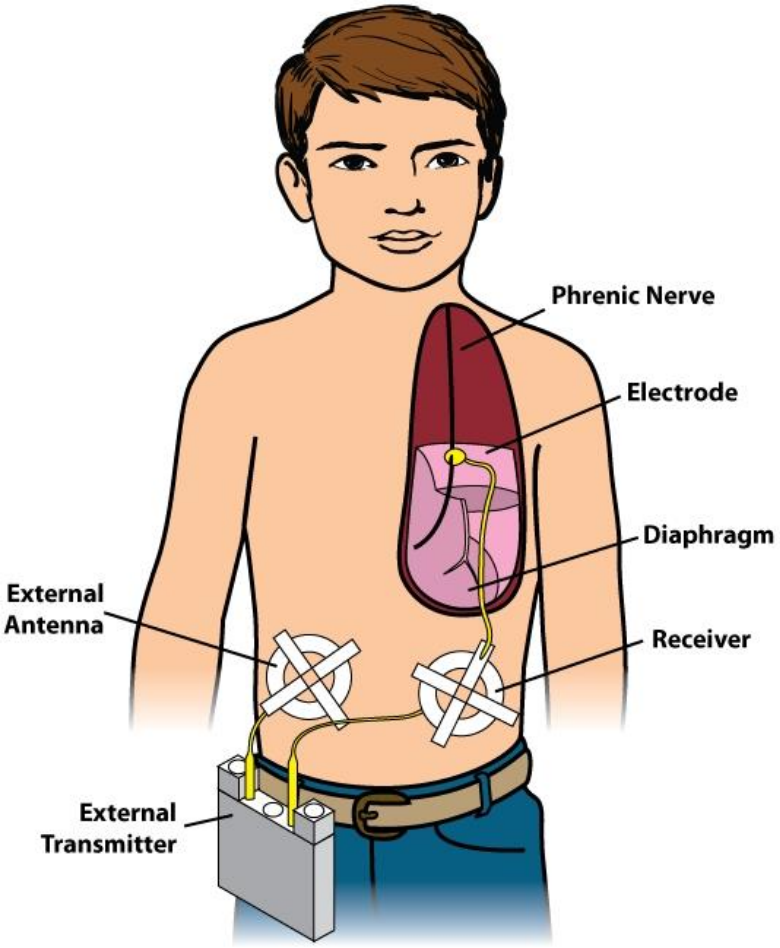
Kasi et al. Journal of Multidisciplinary Healthcare 2022:15 455–469

Noninvasive (Mask) Ventilation

Advantages	Disadvantages
• No tracheostomy • Relatively shorter training for family compared to tracheostomy • Possibility of trach decannulation	• Airway is not secured • Limited mask options in children; periodic re-fitting • Difficult to use for > 14-16 hours per day • May require temporary intubation during illness • Risk of facial deformity with prolonged use • Potential discomfort, pressure sores, aspiration

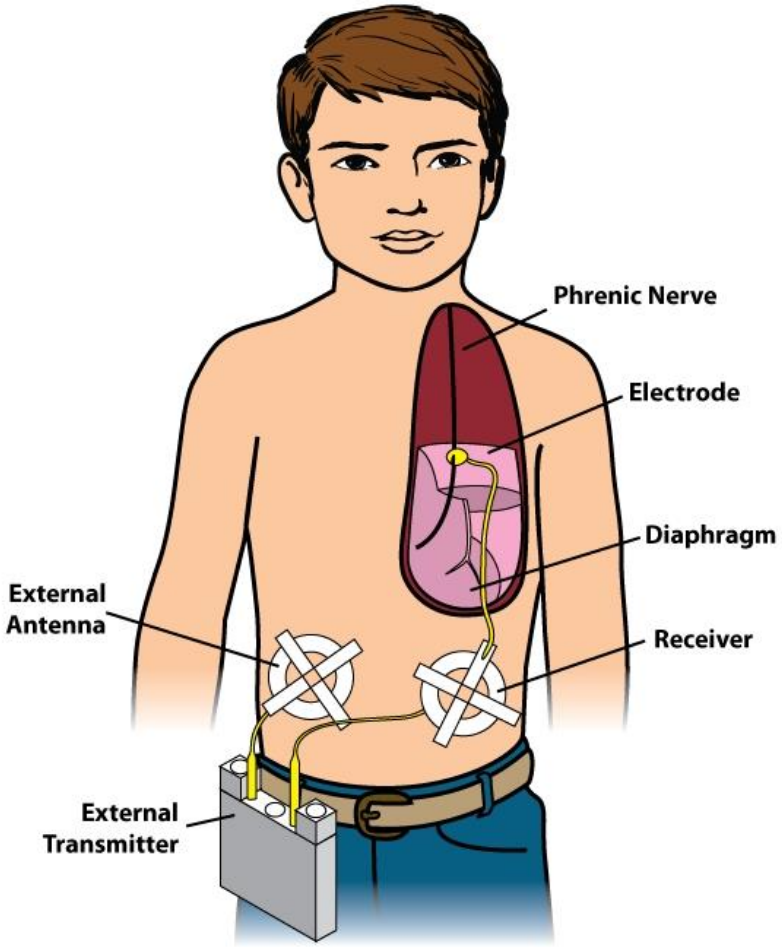
Trang H et al (2020). *Orphanet J Rare Dis* 15:252.

Diaphragm Pacing



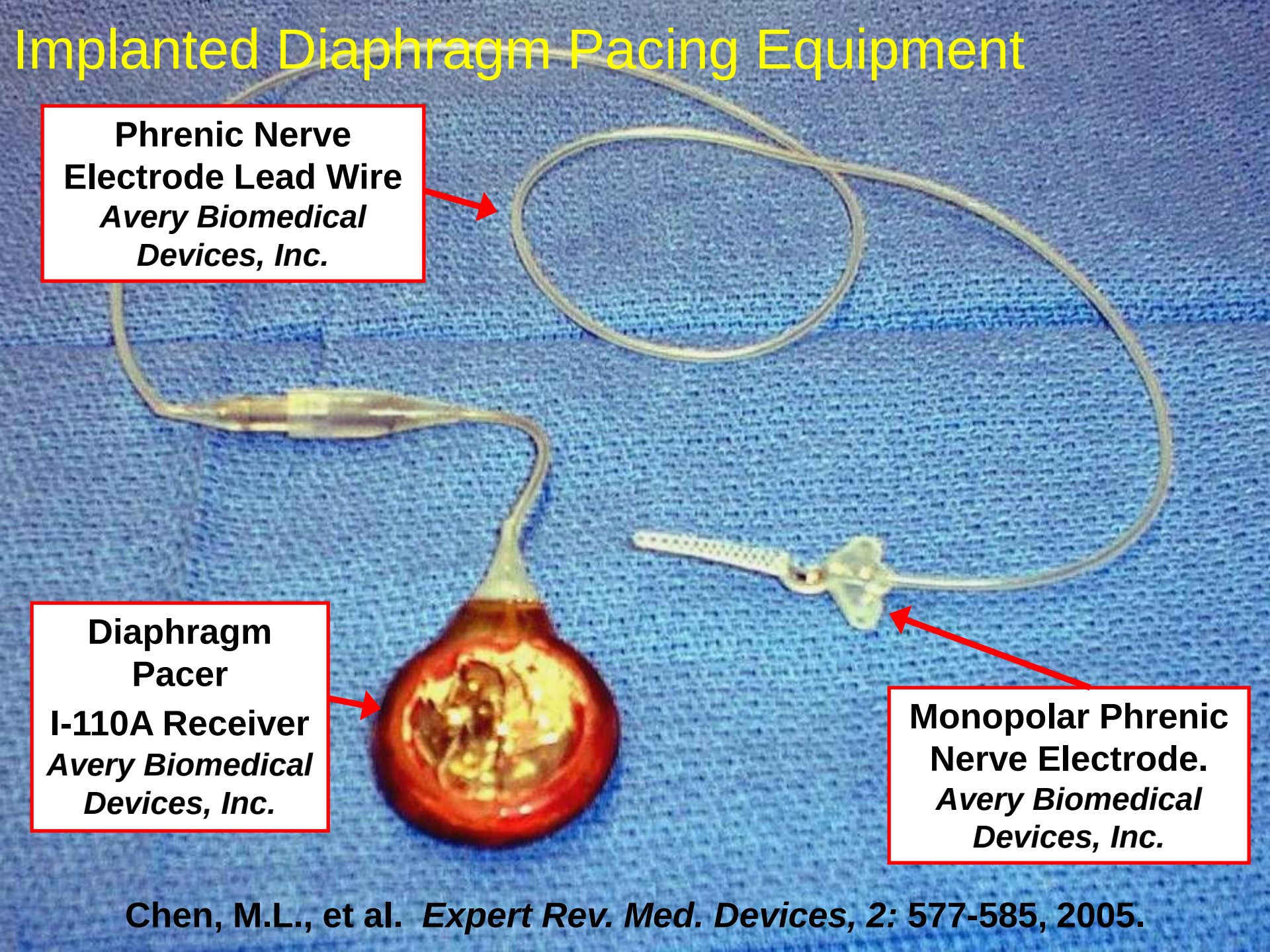
- **Diaphragm pacing is a type of ventilator, which uses the patient's own diaphragm as the ventilator pump.**
- **Tracheostomy may be decannulated in some patients requiring ventilation only during sleep.**

Diaphragm Pacing



- **Electrical stimulation of the phrenic nerve causes diaphragm contraction.**
- **Normal intact phrenic nerve is required.**
- **Normal diaphragm muscle is required.**

Implanted Diaphragm Pacing Equipment



Chen, M.L., et al. *Expert Rev. Med. Devices, 2:* 577-585, 2005.

Diaphragm Pacer

Image from: Avery Biomedical Devices

Diaphragm Pacer Surgery

- **Deflation of each lung**
- **Placement of the electrode on the phrenic nerve**
- **Securing the electrode to the tissue**
- **Testing the DP equipment intraoperatively**
- **Implantation of the receiver and connecting it to the electrode lead wire**
- **Repeating the same steps on the other side**

Shaul, D.B., et al. *J. Pediatr. Surg., 37:* 974-978, 2002

Pacing Timeline

Time (weeks)	Procedure
0	Surgical implantation
6-8	Initiate Pacing (1-2 hours/day)
12-16	Increase Pacing Time (muscle training)

Chen, M.L., et al. *Expert Rev. Med. Devices, 2:* 577-585, 2005.

Initiating Pacing

- **Usually can start pacing at 1 - 2 hours per day**
- **Monitor ventilation using pulse oximeter and $P_{ET}co_2$**
- **Begin with lowest setting on each side and increase until contraction palpable**
- **Increase tidal volume setting until a reasonable contraction is observed**

Chen, M.L., et al. *Expert Rev. Med. Devices, 2:* 577-585, 2005.
Keens, T.G., et al. *J. Appl. Physiol., 44:* 905-908, 1978.

Initiating Pacing

- **Shoulder pain means tidal volume setting is too high**
- **Increase pacing time by 30 - 60 minutes per week**
- **Usually takes 3-months to train for full pacing**
- **Continuous pacing is not usually possible**
- **Limited to 12 - 14 hours/day**

Chen, M.L., et al. *Expert Rev. Med. Devices, 2:* 577-585, 2005.
Keens, T.G., et al. *J. Appl. Physiol., 44:* 905-908, 1978.

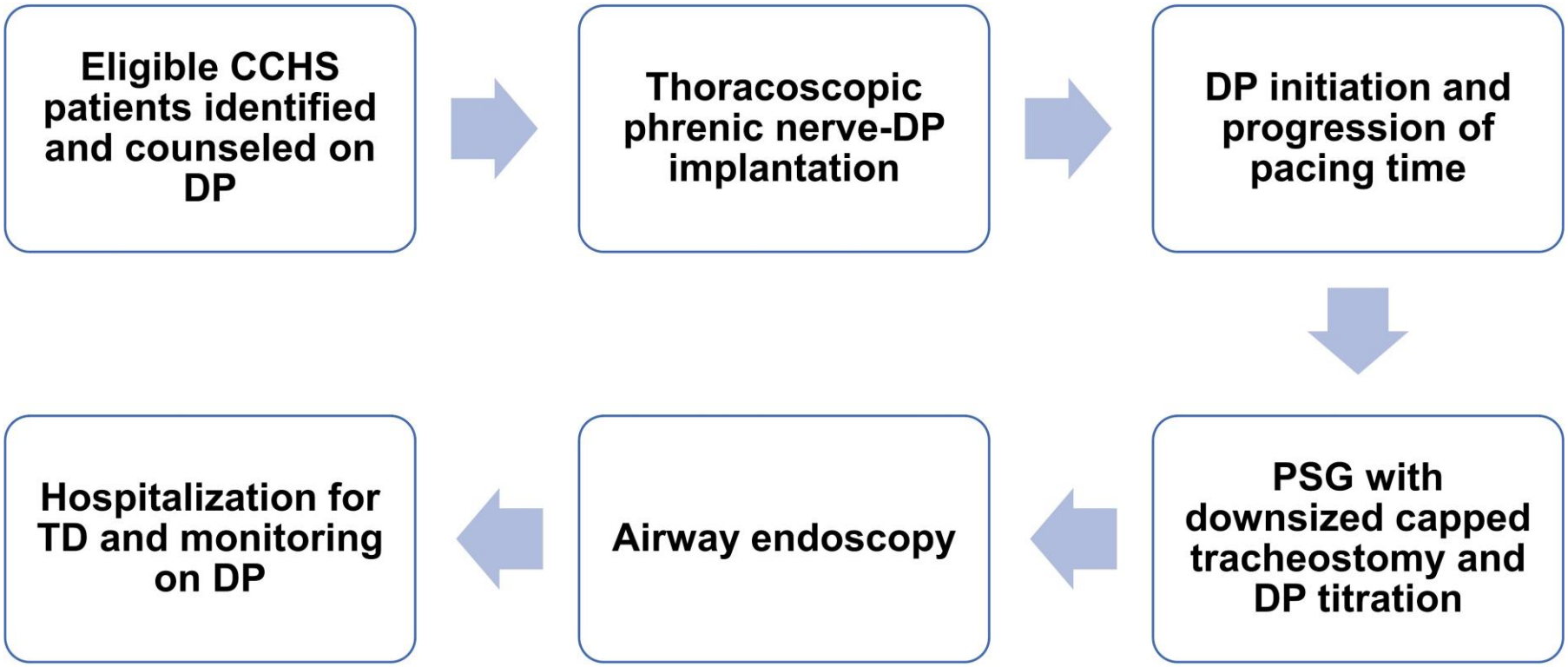
Diaphragm Pacing: Eligible Patients

- **In patients requiring full-time ventilation**
  - **DP during the day**
  - **Ventilator via trach during sleep**

- **In patients requiring ventilator only during sleep**
  - **Age ≥ 4 years**
  - **Minimal or no lung disease**
  - **Not requiring full-time ventilation with illness**
  - **No obesity**

Chen, M.L., et al. *Expert Rev. Med. Devices, 2:* 577-585, 2005
Diep B et al. *Respiration* 2015;89(6):534–538

Transition from Trach to Pacing



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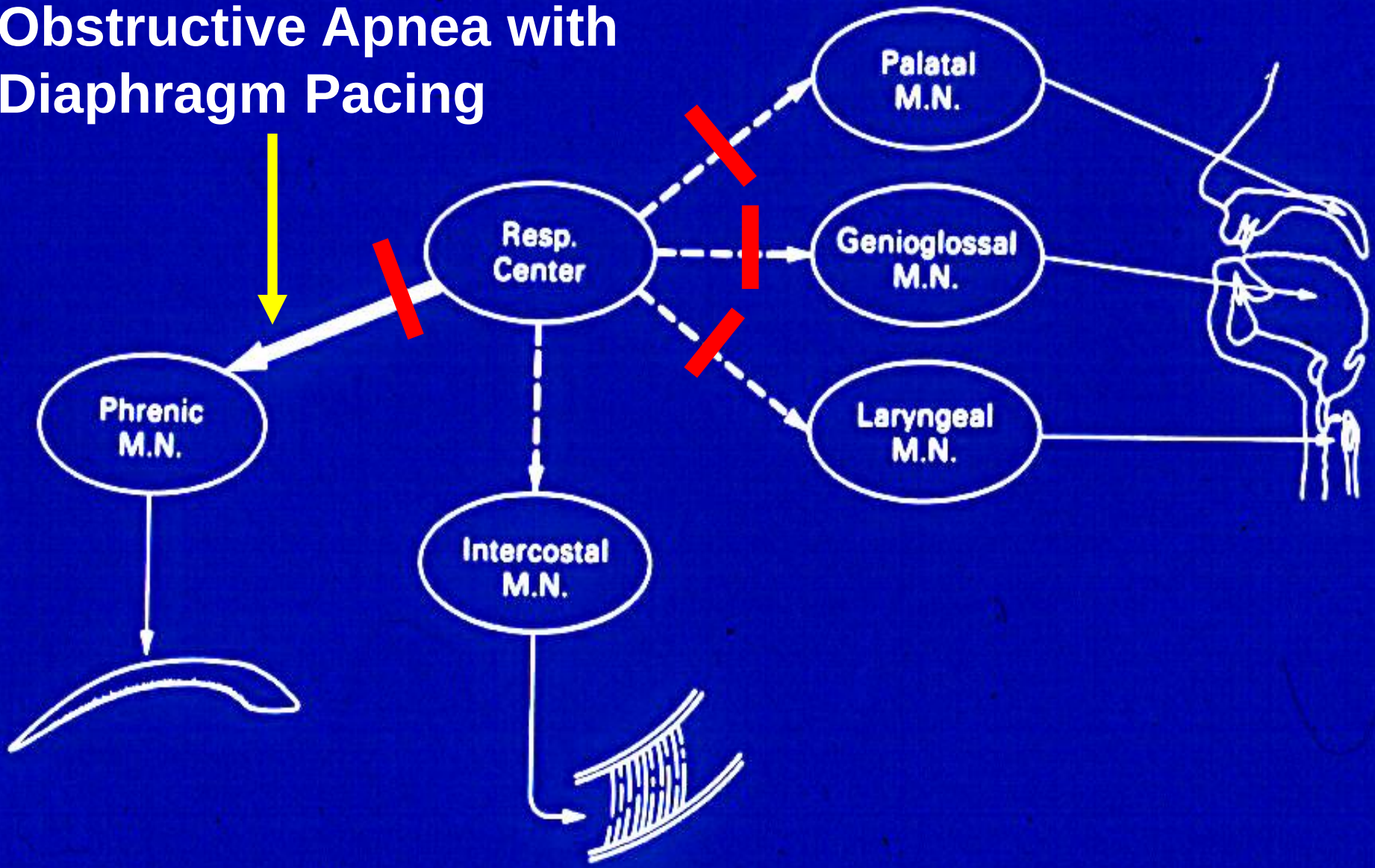
Transition from Trach to Pacing

- **If oxygen saturation < 94% and/or $P_{ET}co_2$ > 45 on overnight sleep study with DP and capped trach:**
  - **Obstructive apnea is usually the problem**
  - **Adjust pacer settings**
  - **Consider adenotonsillectomy, sleep endoscopy**

Obstructive Apnea with Diaphragm Pacing



Remmers, J.E., et al. *Clin. Chest Med., 1:* 57-71, 1980.

Goals of Diaphragm Pacing

- **Ventilator dependent only during sleep:**
  - **To potentially remove the tracheostomy**

- **Full-time ventilator dependent:**
  - **To improve mobility while awake**
  - **Improves quality of life, primarily because of its portability**

Diaphragm Pacing

Advantages	Disadvantages
• Improves mobility • Possibility of trach decannulation • No negative impact on speech or swallowing or facial growth	• Requires surgery • Can cause obstructive sleep apnea • May require additional form of ventilation during illness • Management in specialized CCHS centers • Limited to 12-14 hours per day

Trang H et al (2020). *Orphanet J Rare Dis* 15:252

Outcomes: Single-center study, n=30

- At diagnosis, most patients (80%) required full-time ventilation
- Median age of transition from full-time to sleep-only AV was 9 months (IQR 6-14 months)
- During respiratory infections, several young children who were weaned off ventilator during wakefulness required continuous ventilation
- Median age at trach decannulation (n=9/14) was 11.2 years (IQR 5.9-15.7 yrs)

Table 3. Comparison of assisted ventilation modalities.

AV MODALITY	AT DIAGNOSIS, N (%)	CURRENT, N (%)
PPV-T	28 (93%)	19 (63%)
NPPV	0 (0%)	9 (30%)
Diaphragm pacing[a]	0 (0%)	3 (10%)
Oxygen	2 (6.7%)	0 (0%)

Fain et al. Clinical Med Insights Pedr 2023

Tracheostomy stomas

- **When trach stoma does not close after trach tube removal - Tracheocutaneous fistula – connects trachea to skin**
- **Less likely to spontaneously close based on longer duration of tracheostomy (>12 mo)**
- **Risks: secretions, skin irritation, infection, may affect speech**
- **Ideal timing: 3-6 months after trach decannulation**

Tracheocutaneous fistula

- **2 surgical techniques for closure**
  - **Excision of fistula with primary closure in layers**
  - **Excision of fistula with healing**

- **Both techniques had similar success rate >90%; no difference in post-op complications**

- **If using mask ventilation, stoma may need to be covered with gauze and tape until the stoma is closed**

Stern Y. Ann Otol Rhinol Laryngol. 1999 Sep;108(9):880-3
Lewis S. et al. Laryngoscope, 127:241–246, 2017

THANK YOU!

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