Giulia, CCHS

Female

Age at first observation 8 years, 4 months

Diagnosis of CCHS (Mutation (poly Ala repetition 20-26) on day 15 of life

Hirschsprung disease

Tracheostomy placed at the age of 2 months

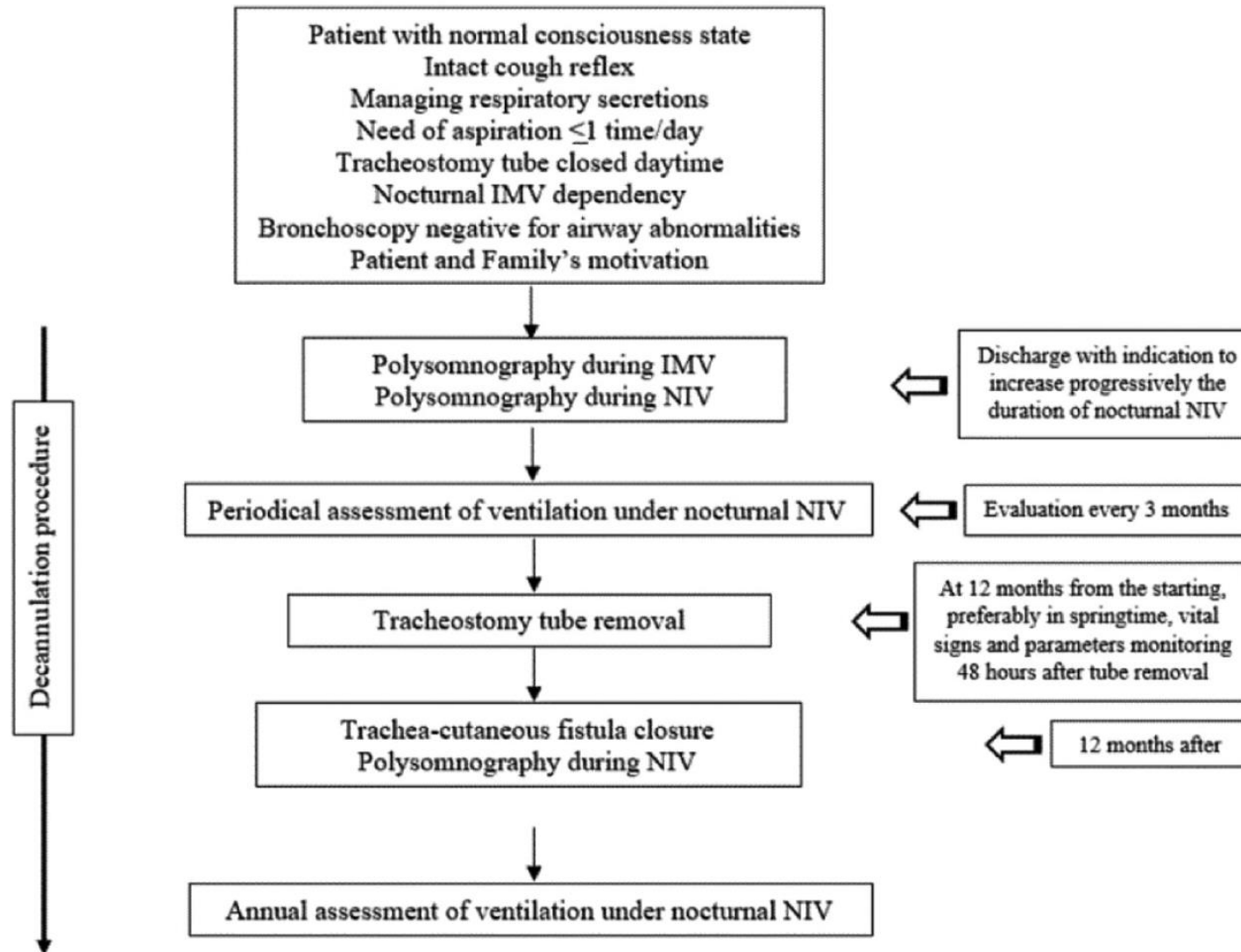Invasive Mechanical Ventilation (IMV) only at night

Pressure control mode

Bronchoscopy: normal

Tracheostomy removal protocol started at request of patient and her parents.

Tracheostomy decannulation protocol adopted in our patients affected by congenital central hypoventilation syndrome

Legend: IMV, invasive mechanical ventilation; NIV, non-invasive ventilation.

Sleep Study results on IMV at first observation

Ventilator Setting: **PCV**: IPAP 14 cmH$_2$O, EPAP 4 cmH$_2$O, FR 18 breaths/min.

Parameters	Values
Total Sleep Time (h e min.)	07.54
mean SpO$_2$ (%)	99.7
lowest SpO$_2$ (%)	97
SpO$_2$ < 90 % (% TST)	0.0
mean Heart rate (bpm)	75
AHI (N°/h)	0.0
Central Apneas (N°/h)	0.0
peak tcpCO$_2$ (mmHg)	36
mean tcpCO$_2$ (mmHg)	33.2
tcPCO$_2$ >50 mmHg (% TST)	0.0

Recommendations at discharge

- use of a speaking valve

- later on, routine tracheostomy capping

- progressive acclimatization to nasal mask while awake

Start approaching Non Invasive Ventilation (NIV)

NIV initially tolerated 2 hours/night

NIV Setting: **PCV**: IPAP 16 cmH$_2$O, EPAP 4 cmH$_2$O, FR 18 breaths/min.

Parameters	Values
Total Sleep Time (h e min.)	1.49
mean SpO$_2$ (%)	98.8
lowest SpO$_2$ (%)	88
SpO$_2$ < 90 % (% TST)	0.3
mean Heart rate (bpm)	79
AHI (N°/h)	0.0
Central Apneas (N°/h)	1.6
peak tcpCO$_2$ (mmHg)	42
mean tcpCO$_2$ (mmHg)	30
tcPCO$_2$ >50 mmHg (% TST)	0.0

The goals of NIV support

- mean CO_2 less than or equal 45 mm Hg,

- $PtcCO_2$ greater than 50 mm Hg less than or equal to 10% of total recording time,

- mean O_2 saturation 96%,

- time spent on NIV at least 6 hours per night.

Berry RB, Chediak A, Brown LK, et al. Best clinical practices for the sleep center adjustment of noninvasive positive pressure ventilation (NPPV) in stable chronic alveolar hypoventilation syndromes. J Clin Sleep Med. 2010;6(5):497-509.

- Double prescription (IMV; NIV)

- Progressive increase of NIV use at night

NIV tolerated all night long

Age 9; cm 138; Kg 26

Sleep Study results on NIV

NIV Setting: **PCV**: IPAP 16 cmH$_2$O, EPAP 4 cmH$_2$O, FR 18 breaths/min.

Parameters	Values
Total Sleep Time (h.min.)	6.50
mean SpO$_2$ (%)	97
lowest SpO$_2$ (%)	88
SpO$_2$ < 90 % (% TST)	0.2
mean Heart rate (bpm)	76
AHI (N°/h)	0.0
Central Apneas (N°/h)	0.0
peak tcpCO$_2$ (mmHg)	54
mean tcpCO$_2$ (mmHg)	41.6
tcPCO$_2$ >50 mmHg (% TST)	26.2

What would you do?

Increased IPAP to 18 cmH$_2$O, added volume 250 ml

PCV: IPAP 18 cmH$_2$O, EPAP 4 cmH$_2$O, FR 18 breaths/min, Vt 250ml

Parameters	Values
Total Sleep Time (h e min.)	06:13
mean SpO$_2$ (%)	98
lowest SpO$_2$ (%)	91
SpO$_2$ < 90 % (% TST)	0.0
mean Heart rate (bpm)	74
AHI (N°/h)	0.0
Central Apneas (N°/h)	0.0
peak tcpCO$_2$ (mmHg)	51
mean tcpCO$_2$ (mmHg)	40.2
tcPCO$_2$ > 50 mmHg (% TST)	0.2

Age 11

Capped tube during daytime
NIV at night

Decannulation *

Sleep Study results on Non Invasive Ventilation
PCV: IPAP 18 cmH_2O, EPAP 4 cmH_2O, FR 18 breaths/min, Vt 250ml

Ventilation mode	APCV
Mean SaO_2, %	97
Minimum SaO_2, %	84
SaO_2 <90%, %	0.8
Mean CO_2, mmHg	37.8
Apnea/hypopnea index, *n*/hour	0.3

* Heffner JE. The technique of weaning from tracheostomy: criteria for weaning; practical measures to prevent failure. J Crit Illn. 1995;10(10):729-733.