**Noninvasive ventilation as sole strategy in managing a child affected by hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD)**

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We describe the use of noninvasive ventilation (NIV) to avoid tracheotomy in a child affected by ROHHAD. Despite increased identification and advanced knowledge of the disease course, the variable onset and timing of phenotypic features in ROHHAD often result in delayed or missed diagnosis, potentially leading to fatal central hypoventilation, cardiorespiratory arrest, and impaired neurocognitive development (1, 2). The mortality and morbidity resulting from the high incidence of cardiorespiratory arrest may be prevented by an early tracheotomy and invasive ventilatory support (3, 4).

We describe a case of an old child affected by hyperphagic obesity, alveolar hypoventilation, adeno-tonsillar hypertrophy, daytime hypersomnolence, cephalea, central hypothyroidism, in treatment with thyroid hormons, sodium and water dysregulation, halitosis, strabismus. The patient came from an other hospital, discharged with diagnosis of idiopathic obesity with polyuria and polidipsia.He was referred to the dipartment of endocrinology of our referral hospital for a more specific diagnosis. In the year before his hospitalization, he also had presented 9 kilogram weight gain and suffered from abdominal pain, diarrhea, polydipsia. The parents said that he often fell asleep during the day, after night snoring with frequent awakenings and nycturia. On endocrinological examination, the child was in good general condition, happy, collaborative, he spoke and behaved as normal for his age, deambulation was regular. He presented abundant panniculus adiposus and normal muscular trophism with height below normal value. Head and neck were normally shaped. All main endocrinological screenings were prescribed (load glicemic curve, basal insulin level, ft3 ft4, Tsh, abtpo, prolattin, cortisolemia, ACTH, somatomedine, free urinary cortisol). The ENT examination also revealed tonsillar hypertrophy, and difficult nasal breathing with oral ventilation, The parents also reported frequent bronchitis treated with steroids and bronchodilators. Thorax was symmetrical, normothympanic, slightly hypoextensible with good thoracic-abdominal synchronization. Vescicolar murmur was clear. Heart sounds were normal. Heart rate was 120/min. Blood pressure Holter showed normal daytime systolic and diastolic values with few nocturnal values above the standard. Cardiac ultrasound examination showed no abnormality, nor pulmonary “p”, nor evidence of interatrial shunt. The onchologist suggested a total body TC scan, and hematologic evaluation of ferritin, LDH, NSE, omovanilic acid, vanilmandelic acid. After been sedated with a low dose of midazolam for Nuclear Magnetic Resonance (NMR) he presented an important oxygen arterial desaturation needing oxygen administration. NMR revealed incomplete areas of myelinization in temporalis white matter and an important airway narrowing because of the adenoid and tonsillar enlargement . NMR and a following CT scan did not show any neoplastic mass known to be associated with this syndrome. A nocturnal SpO2 monitoring showed a mean SpO2 of 83.5%. Nadir 69%. CT90 78.9%. Oxygen Desaturation Index (ODI) 34.1. The following day the child, referred to our pneumological ward, underwent a capillary arterial blood gases (ABG) (Table 1) and a full cardiorespiratory monitoring (CRM) showing a mean SpO2 85.4%, ODI 93.9/h with significant central and obstructive events of hypo-apnoea (Apnea Hyponea Index-ADI 24.5). Transcutaneous CO2 monitoring (TcPCO2) (Tina, Demori, Italia ) range was 59 – 64 mmHg. The patient was a bit drowsy but completely collaborative with no problems of feeding and of sputum retention as assessed by peak cough expiratory flow. After parental consent was obtained, daytime NIV was started in pressure support mode (PSV) with a back-up rate (BUP/RR) (Elisée 350, ResMed, Australia) by using a total full face mask (TFFM) (Philips, Netherland) (5). A TFFM was used because of the adenotonsillar hypertrophy and the possible night time air leak (table 1). Nocturnal SpO2 monitoring on NIV showed a mean SaO2 =98% Nadir SaO2= 84% C T90=0.6% ODI=1.7. Night time TcPCO2 range was about 40 mmHg. As a respiratory alkalosis was found in ABG of the following morning , PSV and the BUP/RR were reduced (Table1). Time on NIV was gradually increased until reaching about 8 hours during the day due to a significant TcPCO2 increase during spontaneous breathing (SB) period . One week after admission with normalized ABG (Table 1) the patient underwent adenotonsillar surgery. After extubation there was a need 24 hour/NIV for 2 days due to the patient’s incapacity to sustain even short period of SB (ABG are shown in table 1). Nocturnal mean SpO2 was 97.6%, SaO2 nadir 91%, CT90 0%, ODI 3.7. The last CRM before discharge showed a mean SpO2 of 96.5% and an AHI of 1.1. Patient was discharged on day 33 after admission with a diagnosis of ROHHAD (ABG are shown in table 1). NIV with the TFFM was continued at home during sleep and 8 hours/daytime (1 hour on NIV and 4 hours off). Hormone therapy with Levothyroxine 25 mcg once a day, diet therapy, Fluticasone spray 50 mcg two puffs twice a day. In case of exacerbation, salmetherol plus fluticasone and salbuthamol. The child it still on NIV after two years, in good health, with good quality of life and human relationship.

Table 1 Patient ‘s clinical variables, NIV timing and settings

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Timing | pH§ | PO2  mmHg | PaCO2  mmHg | HCO3-  mmol | SaO2 | Respiratory Rate#  breath/m | Heart Rate#  beats/m |
| On admission SB | 7.35 | 62 | 56.6 | 30 | 91.2 | 24 | 120 |
| After 2 hrs on NIV PSV 12 / PEEP 8 cmH2O BUP/RR 22/breaths/m, Ti 1.2 sec Inspiratory Trigger 1 Expiratory trigger 10% | 7.48 | 62.3 | 45.2 | 33.3 | 95.1 | 28 | 100 |
| After a night time on NIV\* (unchanged settings) | 7.52 | 66.8 | 33.4 | 28.3 | 97.9 | 22 | 100 |
| Before surgery after a night time on NIV\* (PSV 10 / PEEP 8 cmH2O BUP/RR 17/breaths/m. Other ventilatory settings unchanged ) | 7.44 | 72 | 38 | 26 | 97.1 |  | 90 |
| Following surgery in SB after extubation | 7.13 | 72 | 84.5 | 29 | 94.8 | 18 | 140 |
| Following surgery after a night time on NIV\* (PSV 15 / PEEP 8 cmH2O BUP/RR 17/breaths/m Other settings unchanged) | 7.54 | 67.1 | 30 | 27 | 96.8 | 22 | 98 |
| Before discharge after a night time on NIV\* (PSV 13 / PEEP 4 cmH2O BUP/RR 18/breaths/m | 7.41 | 66 | 44.3 | 27.3 | 95 | 18 | 98 |
| After 2 hrs in SB before discharge | 7.39 | 60 | 48.3 | 27.7 | 94 | 24 | 100 |

# Patient’s respiratory and heart rate were the average of 3 consecutive minutes. § All capillary ABG obtained in room air.

\* Other ventilatory settings unchanged

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