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Clinical case: Spinal Muscular Atrophy type 1 - Vivo 45 LS is a valid option for long-term ventilation.

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Case description

A five-day old newborn with a normal pregnancy but, with decreased foetal movements in the last week of pregnancy was admitted for assessment. Apgar test results were 9/10/10 at birth, and no resuscitation was required. The parents reported hypotonia since birth. Key features on examination were, soft cry, severe hypotonia and poor spontaneous movement, poor feeding with a low weight. Paradoxical breathing with a respiratory rate 70-78 breaths per minute.

At Day14, genetic results confirmed the clinical suspicion of spinal muscular atrophy (SMA) presenting during the neonatal period and the child was diagnosed with SMA type 1.

SMA is a genetically determined, congenital neuromuscular disorder, which presents with the progressive deterioration of the motor neurons in the anterior horn cells of the spinal cord. This leads to progressive muscle wasting including the respiratory muscles. SMA is classified on a functional scale. Type 1 are unable to sit unaided, type 2 are unable to walk unaided and type 3 who can walk unaided. Within the types there is a wide range of weakness. SMA type 1 is the severest form and therefore has the poorest prognosis due to the severe involvement of the respiratory muscles. Those patients that present with respiratory symptoms at birth or close to birth are classified further as SMA type 1a. In recent years, there are some treatments that can change the natural course of the disease but the effect on the bulbar and respiratory symptoms are not fully known. It is for this reason that, despite new medications, ventilation, preferably non-invasive, remains an essential support for these children with SMA type 1.

Due to the severe hypotonia and the bulbar involvement. Non-invasive ventilation (NIV) was indicated to alleviate the symptoms of breathing difficulties, maintain a more stable airway, prevent pectus excavatum, facilitate the drainage of secretions and slow down the progress of the disease to terminal respiratory failure in addition to other therapies previously highlighted.

This patient with SMA type 1a, was offered NIV. However, there were several different challenges:
1) The patient's weight means limited licenced ventilators
2) The patient's high respiratory rate, which results in very short inspiration times
3) The need for very high inspiration and expiration sensitivity to enable in spontaneous triggering
4) High daily usage due to sleep wake pattern of infants.
5) Finally, it would be desirable if the equipment was small, light and resistant.

Ventilation mode	PCV(A+TgV)
Target Volume	50 ml
Breath Rate	40 bpm
Maximum inspiratory pressure	14 cmH_2O
Minimum inspiratory pressure	10 cmH_2O
PEEP	6 cmH_2O
Inspiratory trigger	1
Rise time	2

Table 1: Initiation of NIV with Vivo 45 LS:

During sleep the Vivo 45 LS was used with a nasal mask and a 15 mm passive circuit with active humidification. At that time the total RR was around 38-40 and each breath had an estimated Vt of around 40-45 ml. When awake spontaneous triggering was around 30-60% and asynchrony was only present during periods of tachypnoea (RR 90bpm). The patient was generally comfortable when sleeping and with SpO_2 100% without supplementary oxygen and normal transcutaneous carbon dioxide levels ($TcCO_2$ 42 mmHg).

"the sensitivity of the triggers, and the precise measuring systems facilitate the adaptation of the ventilator to the child"

	Chest circumfer-ence (cm)	Head circumfer-ence (cm)	Chest/head circumfer-ence	SpO_2 (%)	$PVCO_2$	$EtCO_2$	Median HR (beats per min-ute)	Median RR (breaths per minute)
Day 1 of NIV	30.5	34	0.897	98	50	48	140	70
Day 10 of NIV	32	34.5	0.927	99	47	44	130	60
Day 40 of NIV	37	37	1	100	42	42	120	40

Table 2: The course of some of the clinical parameters

At two months, the patient was discharged home with the Vivo 45 LS, using it for 12 hours a day. However, during the first month at home, the patient was readmitted due to increased bulbar symptoms such as difficulties in swallowing and laryngospasm leading to a respiratory tract infection. On admission, NIV continued with oxygen entrained. The Vivo 45 LS settings were changed, RR increased to 60 bpm and maximum inspiratory pressure to 30cmH_2O to achieve the same Vt. However, due to the progressive hypoxaemia and the instability of the airway, the patient required intubation and ventilation. After 21 days of IMV and two failed attempts of extubation, the decision was made to perform a tracheotomy and continue with invasive home mechanical ventilation.
At four months old, the Vivo 45 LS settings were further modified. Current average usage was 18 hours per day. Taking advantage of the programable profiles, three profiles were set: Profile 1, aimed at supportive awake ventilation was pressure support ventilation with target volume (PSV(TgV)). Profile 2, aimed for ventilatory support whilst sleeping and was set to pressure control with target volume (PCV(TgV)). Profile 3, aimed to be used when nebulising was assisted pressure control ventilation PCV(A). All profiles had disconnection and rebreathing alarms set. The family was instructed on how to change from one profile to the other and were given instructions on how and when to use each profile.

The patient remained stable during the following months. When the patient reached the age of one, they continued with invasive respiratory support on average of 10-12 hours a day. Settings were adjusted, in accordance with the patient's change in weight and respiratory pattern being his spontaneous respiratory rate much lower and his respiratory effort more effective. Further adjustments were made but, by 1 year old the child used pressure support mode and only required assist control mode when unwell or extremely fatigued.

Discussion.
In infants, the available technology is limited and adapting a ventilator to a small child, both in invasive and non-invasive ventilation, can be challenging. The most significant problem is asynchrony: the device needs to have a high sensitivity of the inspiratory and expiratory triggers to allow synchronisation at high respiratory rates. It is technically challenging to support breathing in a child whose respiratory rate is greater than 40 bpm. Short inspiratory times, in very young children, are often insufficient to activate the inspiratory trigger and do not cycle into expiration appropriately. Also if the trigger is very sensitive it can be activated by any small movement in the circuit (e.g. water), causing auto-triggering and ineffective breathing. To eliminate asynchrony, we used a 15mm active circuit, with a higher back-up respiratory rate and an appropriate Target Volume for the patient. In our case, during an initial period of non-invasive respiratory support in a patient with estimated tidal volume less than 50 ml, the use of a PCV(A+TgV) mode allowed two inspiratory pressure levels to be programmed, which, at times of increased resistance in the airway, enabled ventilation to remain more or less uniform at the expense of small variations in inspiratory pressure.
Secondly, due to the progression of the natural history of the illness, it was necessary to initiate prolonged invasive ventilation, by tracheostomy. Ventilators need to have appropriate modes and alarms for life support. In this case, the presence of fewer leaks, the sensitivity of the triggers, and the precise measuring systems facilitate the adaptation of the ventilator to the child, thus allowing the use of a support mode, PSV(TgV), which is more suited to the spontaneous breathing pattern of the child. All of this possible with the same ventilator the Vivo 45 LS.

Conclusions:
In very young children it is essential that a ventilator has the ability to use different circuits, very sensitive inspiratory and expiratory triggers, the possibility to cope with a high respiratory rate (>60 bpm) and the possibility of ensuring a tidal volume of 50 ml. If the progress of the disease results in the need for invasive ventilation, as in the case presented, the possibility of using the same equipment, with multiple programable profiles and with different ventilation modes facilitates the process of adaptation to prolonged respiratory support at home.

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