

NATIONAL INSTITUTE FOR HEALTH AND CARE EXCELLENCE

Interventional procedures

Patient Organisation Submission

IP1994 Phrenic nerve pacing for congenital central hypoventilation syndrome

Thank you for agreeing to give us your views on this procedure or operation and how it could be used in the NHS.

When we are developing interventional procedures guidance, we are looking at how well a procedure or operation works and how safe it is for patients to have.

Patient and carer organisations can provide a unique perspective on conditions and their treatment that is not typically available from other sources. We are interested in hearing about:

- the experience of having the condition or caring for someone with the condition
- the experience of having the procedure or operation
- the outcomes of the procedure or operation that are important to patients or carers (which might differ from those measured in clinical studies, and including health-related quality of life)
- the impact of the procedure or operation on patients and carers. (What are the benefits to patients and their families, how does it affect quality of life, and what are the side effects after the procedure or operation.)
- the expectations about the risks and benefits of the procedure or operation.

To help you give your views, we have provided this template, and ask if you would like to attend as a patient expert at the bottom of the form. You do not have to answer every question – they are there as prompts. The text boxes will expand as you type, the length of your response should not normally exceed 10 pages.

Please note, all submissions will be published on the NICE website alongside all evidence the committee reviewed. Identifiable information will be redacted.

About you	
1. Your name	Amanda Yardley
2. Name of organisation	CCHS UK
3. Job title or position	Charity Chair
4. Brief description of the organisation (e.g. who funds the organisation? How many members does the organisation have?)	<p>CCHS UK is a charity dedicated to supporting individuals and families affected by Congenital Central Hypoventilation Syndrome (CCHS). CCHS is a rare genetic disorder that affects the autonomic control of breathing, especially during sleep.</p> <p>The charity's primary mission is to provide information, resources, and a supportive community for those living with CCHS. They work towards raising awareness about the condition, fostering research initiatives, and advocating for improved medical care and understanding of CCHS within the healthcare system.</p> <p>Its funding comes from individuals through fundraising and legacies.</p> <p>There are 142 members</p>
<p>5. How did you gather the information about the experiences of patients and carers to help your submission?</p> <p>(For example, information may have been gathered from one to one discussions with colleagues, patients or carers, telephone helplines, focus groups, online forums, published or unpublished research or user-perspective literature.)</p> <p>We have gathered this information on Phrenic Nerve Pacing (PNP) through one-to-one discussions, online forums, a topic at a conference, views gathered on Facebook Support Groups, published and unpublished literature We have also consulted with families in the US, Poland and Sweden about the lived experience, where this treatment option is common.</p>	
Living with the condition	
<p>6. What is it like to live with the condition or what do carers experience when caring for someone with the condition?</p> <ul style="list-style-type: none"> • It is terrifying for both patient and carer as the condition is life-threatening • The patient is wholly reliant on mechanical ventilation (via tracheostomy or mask) to keep them alive 	

- They must be watched during sleep to stop them from dying, and they also have to be monitored while they are awake in case, they stop breathing or under-breathe
- Patients require ventilation when they are tired, concentrating, bored, exercising, eating or unwell
- It is demanding and exhausting for the carer to watch the patient 24/7, and it is intrusive for the patient to be watched and mechanically monitored, and embarrassing in public
- The patient has very little privacy due to carers watching them sleep, and as they transition to adulthood, it is difficult to become independent
- It is stressful if there isn't a reliable power source or access to power points
- There is a risk to life during stormy weather if there are power cuts
- It is also expensive for families, which is especially stressful with the rise in energy prices during a cost of living crisis
- Carers must pack bulky medical equipment everywhere the patient goes, which means travel can be limited
- The expertise of care required limits who can look after the patient, which in turn limits family relationships and freedom to be independent
- CCHS is a complex condition, which is also non-visible. This is frightening for parents and carers because others might assume there is nothing wrong when the patient is extremely high risk.
- Observational and mechanical monitoring can be impossible in certain situations (such as during exercise or in a PE lesson), making the risk of undetected hypoventilation even greater.
- Patients are unable to have sleep overs or go camping

Caring for a patient with a tracheostomy

- Tracheostomies have a huge impact on the carer and the patient; beside the daily cares that are needed to maintain the stoma and tracheostomy, they affect the patient's voice, as well as their swallow, smell, and taste
- Patients cannot shower, swim, play in sand or go to the beach.
- They require a hospital bed at home to safely receive cares and for easy access in an emergency.
- They easily catch infections due to the stoma, which means a patient stays in hospital for no less than 2 weeks per year
- Not all patients get on with the dry circuit which limits independences even further

Caring for a patient with a mask (non-invasive ventilation - NIV)

- A few patients transition to non-invasive mask ventilation when older
- Some patients struggle with this transition, which is often a lengthy process and distressing for both parent/ carer and patient

- Non-invasive ventilation can impact teeth enamel and hair, cause facial deformities, skin rashes and inflammation. It can also cause nasal problems and throat dryness
- While mask-ventilation is a credible option for some patients, many are still under-ventilated at points throughout the day. It can be more difficult to encourage young people to comply with wearing a mask in public settings due to feelings of self-consciousness and embarrassment
- Patients cannot talk, eat, drink or be ambulatory while wearing their mask, which means they face challenges in school and in the workplace

In summary, looking after someone who has CCHS is rewarding, stressful, tiring and guilt-ridden. The carer can't have an off day – the margins of error are too small. The patient can't doze on the sofa, sleep on a long car journey or stay over with friends, without having to be attached to a ventilator. It makes life physically and emotionally exhausting for all involved.

Advantages of the procedure or operation

7. What do patients (or carers) think the advantages of the procedure or operation are? Why do you consider it be to be innovative?

- You do not require a tracheostomy or NIV for ventilation which will immediately improve quality of life – improving communication, relationships, smell, taste and work opportunities
- Having the procedure reduces equipment requirements
- It reduces the need to be dependent on an electrical power source as it is AA battery operated
- It is discrete which means the patients will be more willing to receive ventilation and therefore ventilate more effectively and efficaciously, which will improve longer term outcomes for the patient in learning, health, growth, and confidence
- Potential secondary, or indirect, benefits might include academic and employment performance, travel opportunities, discrete treatment, and thus, reduced stigma, and withdrawal or exclusion from social activities
- It can be used easily when awake and asleep
- Having this procedure will help with independence as it is simple to use and lightweight, which also allows more people to care for patients
- The need for secondary specialist support will not be required

8. Does this procedure have the potential to change the current pathway or patient outcomes? Could it lead, for example, to improved outcomes, fewer hospital visits or less invasive treatment?

- Yes. It is proven in other countries that there are fewer hospital visits and the quality of life for the patient and carers is greatly improved.

Disadvantages of the procedure or operation

9. What do patients (or carers) think the disadvantages of the procedure or operation are?

- Keyhole Surgery is required
- A receiver could fail and need replacing

<ul style="list-style-type: none"> • Patient population
<p>10. Are there any groups of patients who might benefit either more or less from the procedure or operation than others? If so, please describe them and explain why.</p> <ul style="list-style-type: none"> • Some patients require 24-hour ventilation, and this will benefit them the most • Emerging evidence indicates that patients are under ventilated during the day, which impacts on concentration, performance and behaviour; so, this surgery would benefit all patients with CCHS
<p>Safety and efficacy</p>
<p>11. What are the uncertainties about how well this procedure works and how safe it is?</p> <ul style="list-style-type: none"> • There are very few uncertainties as this procedure and treatment pathway has been established in other countries for the last 30 years • There are two ways to surgically implant this device – thoracically or cervically. Thoracically will not have an impact on anyone who is required to have a heart pacemaker in the future. This is the UK preferred approach for this reason • The internal receivers are attached to the phrenic nerve. There was a myth about wearing out the phrenic nerve if you used the device too much – this has never happened and has been dispelled. • If a receiver does fail the other receiver carries on working
<p>Equality</p>
<p>10. Are there any potential equality issues that should be taken into account when considering this topic?</p> <ul style="list-style-type: none"> • None that we are aware of
<p>Other issues</p>
<p>11. Are there any other issues that you would like the Committee to consider?</p> <ul style="list-style-type: none"> • We are aware that there are other hypoventilation conditions, such as ROHHAD, that would also benefit from PNP • Spinal Cord Injury patients qualify for this device on the NHS; it is therefore a known and proved procedure and treatment method in use in the UK

Key messages

12. In no more than 5 bullet points, please summarise the key messages of your submission.

1. Patients are under-breathing when awake as the options to be mechanically ventilated are not practical. PNP is a discrete way to effectively ventilate a patient at all times
2. Independence and mobility are currently restricted, but PNP's would improve both
3. The caring load and cost are currently substantial, restrictive, invasive to patients, and complicated
4. This surgery has high safety and effectiveness; it has been successfully and efficaciously used in several countries (e.g., USA, Norway, Sweden, Poland, Germany etc) for CCHS, and is currently offered by NHS England for patients with Spinal Cord Injury
5. Offering the surgery would enable greater freedom, independence and quality of life, and opportunities for the patient, their families, and carers

Committee meeting

13. Would you be willing to attend the interventional procedures committee meeting to provide the view from your organisation in person?

yes

Thank you for your time.

Please return your completed submission to helen.crosbie@nice.org.uk and ip@nice.org.uk.