

# The 2017 Update of the Standard of Care Recommendations for SMA



## Nutrition

**Dr Finkel:** Nutrition has taken on an increased focus. There is still a lot we don't know about proper nutrition of both infants and children with SMA. We don't have growth charts for SMA.

**Dr Crawford:** It's a most difficult one to study.

**Dr Finkel:** We have a general sense of what a well-nourished infant or child with SMA looks like, and we have some general guidelines about when you look at the growth charts. We want to see a child who continues to gain weight, not plateau, not lose weight. But what is being undernourished and what is being overnourished is still I think a bit of an art form.

I use my dietician a lot to help with that. Not all places have a dietician who has expertise in SMA, but I think it's still an important area.

We are again focusing on bone health topics, making sure there's adequate calcium in the diet or a need to add supplemental calcium, monitoring vitamin D levels, looking at DEXA scans in older patients. How do we put that all together along with weight-bearing to promote optimal bone health? To me that is still an area where there's a lot to be learned.

**Dr Crawford:** One of the difficult things about nutrition is that muscle mass will comprise 40% to 45% of the weight of an adolescent male, and this disease is going to compromise a substantial amount of that. What would be the normal value? Even the idea of what is an appropriate growth chart may be difficult given that there are different levels of severity for the SMA. These are the challenges.

I'm not even certain it's possible to imagine a growth chart, so we have to defer to other metrics of nutritional health. A large part of it is good sense, and an awareness of the challenges, and watching children as they grow over time to have a sense of "Where are they now and is this right for this child at this time?"

**Dr Finkel:** We don't know if some popular special diets, such as elemental amino acid diets, are effective, or if they are even healthy for a growing child. There is certainly some support for that in the animal models, but is it appropriate to have that as a major component of a diet in a growing child? I don't think we know that answer yet.

**Dr Crawford:** I think most of the expert opinion was a little bit on the negative side for at least the amino acid diet.

What about micronutrients? Overall calories are going to be diminished. The children are not exercising that much, they don't have to supply as much other organs. A new focus

# The 2017 Update of the Standard of Care Recommendations for SMA



in the guidelines was that micronutrient deficiencies are more probable than not in a lot of the children, and a change in dietary composition addressing that need was a big focus of the standard of care guidelines, and I think it was appropriate.

## Pulmonary Care

### Pulmonary Care

**Dr Crawford:** Pulmonary care is a difficult topic, another area that is difficult to do science on and yet, over the years, we have learned a lot about care. Certainly, the number one concern that patients with SMA have, the major difficulty they report, is airway clearance – over and above everything else. Children with SMA have a deficient cough. The muscles in the chest wall required to generate the Valsalva movement that produces cough are deficient. They are selectively impaired early.

The second common concern is sleep hypoventilation. Everybody's breathing slows during sleep, but if you have normal muscle strength, slower respiration is easily accommodated. With diminished muscle strength in children with SMA, it requires extra effort to breathe, and it is common that children with SMA will underventilate during sleep. This is something you may not see in any other dimension than the sleep study.

There is also emphasis on prevention of aspiration. Everybody aspirates a little bit, but children with SMA, because of problems around the hypopharynx, are prone to more aspiration, including aspiration of gastric contents owing to gastric reflux. These are all complicated areas, which are key elements of the guidelines.

We have some new therapies. In 2007, we started to use airway clearance devices, to assist in stimulating the cough reflex, effective in producing a cough in children who can't cough otherwise. These devices should be used early on in treatment.

**Dr Finkel:** It is interesting that the care guidelines also took a look at other modalities, such as the mechanical vest, and basically said there was no evidence for support. There was certainly an emphasis on cough assist and suctioning. What about noninvasive ventilation?

# The 2017 Update of the Standard of Care Recommendations for SMA



**Dr Crawford:** The other big advancement – BiPAP machines are better, bilevel support in which a child is given an additional burst of pressure via a mask or nasal pillows. When they are breathing in, it allows for a deeper breath, and especially when applied at night, these children who are otherwise inclined to hypoventilate can be brought into normal respiration for a long time. We have seen a good number of children who were tailing off in a number of dimensions all of a sudden brighten up, do well, become healthier, when they are able to get a good night's sleep.

I used to worry that somehow I was making them weaker by supporting their breath; now I realize a good night's sleep is essential to being healthy, and it makes a big difference. It is part of the care guidelines.

The problem, of course, is the BiPAP isn't available everywhere. It is an expensive technology, it requires a lot of expertise at getting these interfaces to work right, and that becomes one of the challenges. Insurance guidelines, requiring that nocturnal hypoventilation be demonstrated, may be a little bit too strict. That may be a little bit late. We can help people with these therapies at an earlier stage.

This goes to the issue of what is the best way to evaluate children? It is interesting that one of the best ways to evaluate children is with our own good judgement and vision and awareness of SMA. A pulmonologist, a pediatrician, or a neurologist, can tell when children are having difficulty with breathing, when breathing is a struggle. They may be able to do it, but it is work, and working means that they might benefit from some assistance.

**Dr Finkel:** You emphasized an important point: the physical exam is still pretty important here. To look at the child breathing, and sometimes have a parent take a video of what the child's breathing pattern looks like in sleep so you can get an idea of the chest wall and abdominal motion – is there a paradoxical pattern or a hypoventilation-type picture?

It looks like the pulmonologists still largely rely on spirometry, pulmonary function tests, and, occasionally, sleep studies.

## **Pulmonary Care Algorithm**

# The 2017 Update of the Standard of Care Recommendations for SMA



**Dr Crawford:** These are the tools they have, and they are obviously good. That is what we have available to evaluate in standards of care, but I think the standard of care didn't emphasize the need for opinion, and opinion actually still matters.

**Dr Finkel:** Clinical judgment.

## Pulmonary Care

**Dr Crawford:** There is one other dimension that was brought up – paradoxical breathing arises when there is a strong diaphragm but a weak abdominal wall and a weak chest wall. When the diaphragm presses down, there is no abdominal tension to resist its expansion and there is no intercostal activation of the internal intercostals to suppress chest wall depression from atmospheric pressure, so the appearance of paradoxical breathing is both the abdomen coming up, which is normal in babies, and the chest wall going down, which is not. The earliest expression of paradoxical breathing is that the belly seems to expand more than you would have thought, because the abdominal wall is so weak.

There was interest in whether the early appearance of paradoxical breathing is going to lead to the classic thoracic insufficiency, these tiny little chest walls with the diaphragm flattened and the ribs suppressed, and whether or not we wouldn't think about using BiPAP prior to that time to prevent that deformity? There have been several cases in which people demonstrated either reversal if it is applied early, or enthusiasm of starting early. The problem is that is a place where it is hard to accumulate evidence.

**Dr Finkel:** That is a good example where those guidelines are identifying an area that certainly needs more work.

## Acute Care

### The Acute Care Setting

**Dr Finkel:** Let us segue into the patient with SMA in the acute care setting. This is a new area for the standard of care guidelines and it's largely pulmonary management but also nutritional support. It would include patients who have an acute illness, usually acute

# The 2017 Update of the Standard of Care Recommendations for SMA



respiratory decompensation, and need inpatient support, and those who need perioperative care.

We've learned we can't make these patients NPO for hours in advance of surgery. We have to make sure they have good nutrition before and after the procedure. It may mean placing a feeding tube to provide caloric support and fluids throughout the perioperative period.

If a patient is coming in for an expected procedure, we mobilize our teams in advance. Make sure the pulmonologist sees patients before they come in for surgery, and is available to help work with the intensive care team on that transition from extubation to BiPAP, and to wean patients from BiPAP in preparation for release to go home.

Coming in for spinal surgery is a big operation still. Having a team with expertise in managing the hospital stay both for an expected admission and for an unanticipated, urgent one is quite important. There are some good suggestions in the current care guidelines that were not present in the initial ones.

**Dr Crawford:** Patients with SMA can have difficult airways; often their jaws don't open up all the way. Knowing about that ahead of time and being certain about the optimal way of intubating a patient before big surgery is not trivial and is worth advance thought.

## The Acute Care Setting

We've mentioned two things; one of course is the management of the expected difficulties with surgery, but there are also new guidelines about how to anticipate complications as we go on.

**Dr Finkel:** Part of the education of parents of children diagnosed with SMA is to help them develop an acute or urgent care plan: what can they do at home, such as cough assist, nebulizers, suctioning. If they have to come to the hospital, it's important that they remember to bring their equipment, to bring the interface that they are using for their BiPAP, and not start from scratch. They should be prepared to work with the pulmonary care team in the hospital so that there is a good transition there.

**Dr Crawford:** Parents can expect that pulmonary or primary care at the hospital will help them develop a care plan for emergencies. Setting it up in advance makes a big difference.

**Dr Finkel:** Also to know which hospital to go to. Emergency service personnel may opt to go to the closest hospital. We often have to write a reminder note that says, "Please go to this facility." It might be an extra 5 miles away, but this is where the patient can get optimal care.

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