

The 2017 Update of the Standard of Care Recommendations for SMA



Multidisciplinary Care

The Importance of Multidisciplinary Care

Dr Finkel: Let's change the topic for a moment to the multidisciplinary care clinic, because the standard of care guidelines really promote this as a central theme. Tell me your thoughts, Tom, on why this might be important.

Dr Crawford: Probably the single most important thing about multidisciplinary care is it involves a model in which the whole and the parts are reflective of each other. It enables parents to have a role. It is critically important that parents know what's going on and can contribute. There are many times where parents say, "In my situation this won't work" or "This is something I am attracted to" and that matters. That matters in the life of children.

I have become more and more enthusiastic about the idea that when people don't follow my recommendation, it's because I somehow didn't value everything in the same way that I should have valued things. The result was what may be called noncompliance, but it was actually parental compliance with the larger concerns.

Dr Finkel: While these guidelines are directed toward the physician and the healthcare provider, it sounds as if there is also an important role for the parents. It empowers them in a way. It gives them some understanding of how these different type of care fit together.

Dr Crawford: I am going to take it further. It puts them in the center of things. It means that we have to care for children in the most holistic fashion, and that word is not a fluffy word. It means that there are elements of importance in all dimensions that multidisciplinary care will accomplish.

Dr Finkel: It really, then, enables parents to be an effective advocate for their child?

Dr Crawford: It is required that they become an effective advocate for their child. But the other issue, of course, is that the experience of SMA, when you ask children and adults who have SMA, "What does it feel like to be in that body?" – they very rarely talk about weakness. They talk about the pulmonary problems and caregiving. They talk about the GI problems or the orthopedic problems. The experience of the disease is very much in these other domains. And while our specialists in one or another of these dimensions know a lot about that area, they may not know about the interaction of their specialty with the other specialties. So multidisciplinary care has to involve a cross-talk: "Yes, I think that there is something

The 2017 Update of the Standard of Care Recommendations for SMA



important in the pulmonary domain, but that might interact with how we deal with orthopedics or nutritional issues.” So there is a back and forth that goes toward the center and out from the center. Everybody is talking to everybody, with the parents at the center, and that’s critical to the efficacy, to doing a good job.

Dr Finkel: How do you suggest that this care be coordinated? It sounds like it is not a one-size-fits-all process.

Dr Crawford: It is so complicated because every jurisdiction has different resources, and we have places where the center of the universe is a child neurologist. Of course as a child neurologist I think that’s the standard, it should always be! But, obviously, that’s not always possible and, in fact, it is probably not optimal always.

We need to have a place at which the centers of interest are able to convene and discuss things with one another. It’s also mindful to the standard of care, but not every jurisdiction has every specialist. Certainly these highly advanced centers that we have the opportunity to work at, where we have everything in abundance – that’s not the standard everywhere. There are lots of places where they have maybe a pulmonary specialist in pediatrics, maybe not. SMA is a common disease of low distribution everywhere, so many of our patients are at places that don’t have every specialty all the time.

A care center has to work in the real world, and figure out how best to manage and leverage the resources and interests that are available to them, and that includes all the family resources too.

Dr Finkel: And to provide this locally, in a way that is accessible to them.

Physical Therapy

The Changing Role of Physical Therapy and Rehabilitation

Dr Finkel: Let’s talk now about the role of physical therapy and rehab, this is an important topic and has undergone some evolution. Now that we have a treatment that has been approved for SMA, we are expecting to see a changing phenotype. We see that these children in many cases are actually getting stronger, much to our surprise. They are gaining new skills. So the role of the physical therapist is changing. It is to promote optimal motor function as we are seeing these changes evolve in these babies and young children.

The 2017 Update of the Standard of Care Recommendations for SMA



Also, we need to keep in mind adults with SMA. That is a group that, in some cases, has been neglected, they were not included in the clinical trials. Now they are eligible for treatment and we need to think about how are they going to respond to the drug as well, and what is the role for physical therapy for them?

Dr Crawford: A lot of care for SMA has been in children's hospitals, and that is one of the reasons why we found with this new therapy that adults are coming back to our therapies saying "We didn't know that existed". It is really quite extraordinary.

Dr Finkel: The broad role of physical therapy and rehabilitation specialists is to promote independent function, independent living as patients grow up, obviously to reduce the burden of the disease on them, to try to limit any pain or discomfort with activities.

The Changing Role of Physical Therapy and Rehabilitation

For babies, the role of physical therapy is to provide proper positioning, whether it is at home or in the car. What is the role of exercise in these children? What about orthoses, bracing? Another important area is bone health, and the importance of weight bearing, so we are using more standing frames and opportunities for children who are not able to walk, maybe not yet, but hopefully someday. But at least to have some weight bearing, and keep those bones strong.

We know that contractures are a significant issue in a lot of these infants and children with SMA. As we see improvement in strength, as they are treated with nusinersen for example, we are hoping to avoid the contractures that might limit their motor function, so we are seeing that there is still a very important role for physical therapy, even in the treated patient. Perhaps even more so.

Dr Crawford: I might have a minority opinion: I think we are better able to prevent contractures than to make a fully developed contracture whole again by stretching, so I am on a mission to make certain we "right-size" our therapy to the potential gains.

Dr Finkel: That would again go back to that focus on pre-emptive or proactive care, not waiting for the contractures to develop so that you need to try to reverse them.

The 2017 Update of the Standard of Care Recommendations for SMA



Dr Crawford: If you wait, it is too late.

Dr Finkel: Okay, but we are finding also that there is a shortage of physical therapists with expertise in neuromuscular disorders, so sometimes patients have to go to a center of excellence, see a physical therapist who knows SMA, and then go back to where they live to see if they can get that care locally, whether it is with the school or in the community. It remains a challenge there.

Goals of Physiotherapy

It is also important to keep in mind that the role of the physical therapist is different in the non-sitter, the sitter, and the walker. Again, the general theme is trying to promote well-being and independent function. For example, for a child who is in a wheelchair, sitting but non-ambulant, the focus will be on upper limb function and how to function independently through the use of different adaptive devices, for example.

Dr Crawford: This is probably a good time to bring up another issue. If SMA is a relentlessly progressive disease in which people are only going to decline over many years, there is a kind of a nihilism about “Why do we want to burden somebody with this or that therapy because they are only going to lose?” Now, in this period of hope where we can at least stabilize, or hope to be able to stabilize, things I think it is really important that sights be lifted as to the possibility. I am very interested in making certain that children don’t underestimate what they can do, especially with not only improved SMA therapy but also with the improved assisted devices and electronics. We have children, the two of us, who are doing extraordinary things in school and professionally that was never expected. Even at the time of the 2007 guidelines, it would have been unusual for some of these children to make it through college. Now they regularly do, and they get advanced jobs.

One of the things I think the therapists can provide, as well as the entire care center, is expectation of improvement. We can actually pull off some extraordinarily meaningful things.

The 2017 Update of the Standard of Care Recommendations for SMA



Orthopedic Care

Orthopedic Care, Growth and Bone Health Care

Dr Finkel: Why don't you share with us your thoughts on what is going on with orthopedic care, because that is a rapidly changing area?

Dr Crawford: Yes, bones are important, and bones are pulled around by muscles. We just talked about the contractures, and contractures have a role in bone health, but deformity has always been a confounder, accumulating complications of weakness, that then creates a life of its own and burdens things.

Orthopedic Care, Growth and Bone Health Care

Scoliosis is a regular complication of SMA type 2 in the way it used to be. We are not certain how much of it is going to be a problem now if we treat early, or how much more of a problem it is going to be, but it is quite clear that once scoliosis starts to advance, it can further encumber children from doing what they need to do, both from a pulmonary standpoint and from a biomechanics standpoint, and certainly sitting difficulties.

Dr Finkel: There are really two populations here: there is the child who is developing progressive spinal curvature and there is the child that has already had spinal fixation. They are two different groups, but I think the standard of care guidelines are trying to address both of them.

Dr Crawford: Yes, the new orthopedic opportunities for growth-enabling surgeries are a big deal. We used to put off surgery as long as possible, have those bones grow as much as possible, because the only thing we could do would be a spinal fusion surgery. Now, in centers that have this expertise, it is possible to give various different kinds of growth-enabling therapies. We don't have to get into the specifics because each institution has its favorite way of doing it, and I don't know that there is an argument for one being better than the other yet, but we can do things to enable maintenance of emerging scoliosis and keep the bones straight for longer until we finally do the definitive spinal fusion surgery, if that becomes necessary.

The 2017 Update of the Standard of Care Recommendations for SMA



One of the big changes between 2007 and 2017 is these new growth-enabling surgeries. It really should be managed by people who have experience in that area and have done it many times before, because experience seems to matter.

Dr Finkel: Instead of waiting until maybe 8 or 10 years of age, close to skeletal maturity for the spine, we are now able to do this type of surgery much earlier.

Orthopedic Care Algorithm

Dr Crawford: The standard of care has a nice algorithm about what would be the thresholds for being able to refer. A key issue is that if patients are under 15 to 20 degrees of scoliosis by the Cobb angle, which is something orthopedics and radiologists know how to do, then we basically just watch them. If they are between that degree and perhaps about 50 degrees, then we can manage with bracing, or maybe, towards the advanced end, start talking about the new growth-enabling surgeries.

Certainly, in children that are greater than 50 degrees they should be managed because the sense is that they are going to advance relatively quickly after that time, and proactively it is better to prevent it than trying to repair it.

There has also been advancement in the management of the hips, and the other deformities that were brought together. We know the prospective management of orthopedic problems, again, makes a difference.

Orthopedic Surgery and Intrathecal Access

Dr Finkel: Can you comment on the particular challenge of being able to administer nusinersen in the patient who needs spinal surgery?

Dr Crawford: One of the real complications of nusinersen is how difficult it is to give. It has to be given by spinal tap. In a nice limber back in a young infant, that is not too difficult. You and I have been doing it quite a bit now, but the further along children get, and certainly the more deformed their back might get, it is going to get more and more difficult to get a needle into that space.

There was enthusiasm amongst the orthopedic surgeons, and certainly I share that, that at the time of scoliosis surgery it is possible to cut a window, a small

The 2017 Update of the Standard of Care Recommendations for SMA



laminectomy that makes it possible to approach from posterior approaches. There may be a requirement for other approaches, whether it is transforaminal or by pumps. There is a lot of work going on now, and I suspect there is going to be some advancement in that area. Working out the best way to get a needle into the intrathecal space on a repetitive basis with no complications and minimal morbidity is going to be one of the new challenges, but at the time of scoliosis surgery we would obviously encourage thinking about it and figure out what the best way is going to be in the future.

Dr Finkel: And to at least try to leave a space in the lumbar. Move your rods around and do what you can.

Dr Crawford: Leave a hole, yes. Leave a window.