

Families of Spinal Muscular Atrophy

Transcript of SMA Questions Chat, September 29, 2004

This transcript has been somewhat edited and revised to match questions and answers, correct typos, and to remove extra remarks. Please note there may still be errors and omissions.

Our expert this month is Dr. John Kissel of the Ohio State University.

<i>mikki :</i>	Is it usual for adults to have low creatine readings on blood tests?
<i>drkissel :</i>	mikki, I am not sure what you mean...did you have your creatine kinase enzyme level checked? If so, the creatine kinase level or CK CAN be low, especially if your muscle mass is very reduced. I should also add that that (ie, a low CK) is not bad in any way for the body.
<i>bryce's_dad :</i>	Hi Doc Kissel: What type of expectations could adults with SMA have for drugs being trialed now and in the near future? Will the possible benefits of increased SMN protein possibly improve SMN/Muscle communication - ie additional "nodding" of nerve cells to muscles?
<i>angiesma2 :</i>	is the debate over sma being an aggressive disorder that never stops versus being switched on then off?
<i>drkissel :</i>	Those are great questions, and I think the answer to both of them is yes. That is, I think that increasing SMN protein levels may indeed prove to be useful in improving strength. Also, there is a very real possibility that increasing SMN could act in ways not directly related to the motor nerve itself, either at the level of muscle or of nerve-muscle communication.
<i>drkissel :</i>	angiesma2 - that is a tough question, and I do not think the debate is over. I think most of us would agree that SMA is not aggressive in the sense that it continues to progress rapidly from month to month. What seems clear is that function declines, although very slowly, in SMA 2 and 3 patients. Whether this is all due to progression of weakness or other factors is still a little unclear. We and others are trying to examine this issue in more detail now.
<i>angiesma2 :</i>	for me, my abilities remained reliable from birth to about 35 yrs. knowing basic info on human bodies, can my weakening be attributed to the "aging process"
<i>Cuco :</i>	Hello Dr. Kissel. What is your advice on diet? Should a person with SMA eat more carbohydrates due to less glycogen stored in the muscles, or is a normal, healthy diet more appropriate?
<i>nekrosys :</i>	I have been reading about the different nutritional requirements in children with SMA, and I was wondering if adults have different nutritional needs too. It seems like I need a lot of extra protein to function properly. I also take "predigested whey protein" and vitamin B complex. If any of these are missing from my daily diet I lose energy quicker and have more intestinal track problems.

<i>drkissel :</i>	Cuco, Nutrition is certainly one of the other factors I referred to in my last answer, and we are still appalling ignorant of the nutritional needs of SMA patients. My own view is that in adults at this time, a generally normal, healthy diet is best. Most patients I know have various issues and food-stuffs that they do better on in the sense that they feel better and seem to function better. I obviously tell me patients that have found something like this to "go with it" and stay on that diet. There is simply not enough good information at this time to make hard and fast rules for everyone.
<i>earl :</i>	Is SMA as progressively degenerative in adults as it is in children? I'm 41 years old, diagnosed at around age 4. I've been (relatively) healthy except for pneumonia every 5 years or so, and of course very weak muscles.
<i>drkissel :</i>	Earl, based on the best data we have, you may get some functional deterioration in he coming years or decades.
<i>angiesma2 :</i>	I am now taking mestinon to give me back my "mojo" and desire to keep moving yet the strength I had isn't returning.
<i>andy :</i>	I'm from Argentina, my mother has SMA, she is 56 years old and she has SMA since 4 years ago. Her doctor gives her this medicine – Rilutek / riluzol drug.
<i>drkissel :</i>	AngieSMA2, that is the \$64,000 question; that is, is the loss of function you may be experiencing all due to SMA, to SMA and aging, just aging, or other factors. The data in the mouse model would suggest that there is some motor nerve loss late, but we haven't been able to confirm that in adults with SMA.
<i>drkissel :</i>	Earl, I sort of answered that question above in my prior 2 answers, but I would be interested to hear whether angiesma2 thinks the Mestinon has helped.
<i>mikki :</i>	Speaking of pneumonia, is there a recommendation how often to get the vaccine?
<i>drkissel :</i>	Did that drug help your mother Andy? And does she have genetically proven SMA, or a form of ALS?
<i>andy :</i>	She has (I don't know how to say it in English) affected the "asta anterior medular". She is just beginning the medicine. Do you think that she may take Mestinon? Can one take both?
<i>drkissel :</i>	Andy, mestinon might be worth a trial in your mother. One can take riluzole and mestinon at the same time.
<i>drkissel :</i>	Mikki, I recommend vaccination in SMA patients like in other patients, depending on their age and their associated medical problems. Most of my patients get yearly flu shots, and the pneumomvax vaccine every year.
<i>angiesma2 :</i>	I am on 30mg (Mestinon) in the morning and another 30mg around 8-10 hrs later depending on how active I get. Are there better kinds? This is ideal for Myasthenia Gravis (MG) patients, so if there's others better suited for the skeletal muscle's acytecoline, let me know.
<i>drkissel :</i>	AngieSMA2, that is a fairly common dose, although in myasthenia

	gravis patients, we often go up to 60 mg 4 times a day or more. Have you noticed any effect?
<i>angiesma2 :</i>	Yes, I "feel like moving" and I don't feel "heavy", yet my strength has dwindled from 5lb lifting to 1lbs. I recoup faster when tired from intense activity too.
<i>questioner:</i>	Ok, I'm glad we are not face-to-face because I have an embarrassing question. I'm 37 with type III SMA, and I'm wondering if it can cause you to lose your sexual desire?
<i>drkissel :</i>	There is no need to be embarrassed, and that is a good question. Sexual desire is obviously a complicated matter. Neurologically, there is nothing that we know about that should cause SMA to automatically result in reduced sexual desire. Patients with all sorts of chronic illness frequently wrestle with this issue, and as you know, fatigue, depression, medications, and social factors can all be operative. I do know that there has not been much work done in this area!
<i>bryce's_dad :</i>	Have you heard about any of the De Vivo & Columbia U research and upcoming trials? Similar drugs? Is there any coordination with AmSMARt and/or Project Cure? Are any of the trials going to include adults - seems like adults would have much less toxicity issues with some of the drugs being trialed...?
<i>drkissel :</i>	Bryces dad, I agree about trying the meds in adults, and that is why we are trying to study the adults especially here at OSU (Ohio State University.) Most of the large groups are trying to look at the same drugs, and the information is shared freely.
<i>female :</i>	My question is a little peculiar: have you heard about any contraindications for IVF treatment in SMA women? How can I be sure that the drugs are safe for me? Do you think the dosage should be different regarding of the weight and the weakness? I'm afraid of pulmonary consequences.
<i>drkissel :</i>	I am no expert in that area, but I do not think there would be any contraindications except for the respiratory issues, and I assume those could be watched closely and dealt with. Good luck if you decide to proceed.
<i>drkissel :</i>	AngieSMA2, glad to hear you are doing well with that med. I hope it continues.
<i>angiesma2 :</i>	Side effects aren't nice. To get optimum benefit causes more intense side effect. Hard to find the ideal balance. I am on Mestinon 30mg to 60mg/day - it's my ideal dosage that gives me more of the benefit and none of the side effects like diarrhea.
<i>nekrosys :</i>	How does one volunteer for the OSU studies?
<i>drkissel :</i>	If you send us your name and number, our coordinator can get in touch with you, or you can call our office directly. FSMA can give you my contact information.
<i>nekrosys :</i>	Thank you!
<i>mawlis :</i>	I have two boys, I understand that they will now be carriers of SMA - do they need to have their future wives tested to see if they too are

	carriers before they decide to have children of their own?
<i>drkissel :</i>	Mawlis, that is correct. Most SMA carriers want their mates to be tested IF it will help them make decisions about pregnancy, children, etc.
<i>KELLY :</i>	Hi, I just joined so I'm not sure if you have had this question. I'm 31 and have SMA 3 and still walking unassisted, my question is since I have had this disease all of my life are my muscles damaged? So if they are would a treatment work on people like me?
<i>drkissel :</i>	Kelly, we did deal a little with that and you can scroll up to check the answers. The bottom line, though, is that ambulatory SMA patients should be particularly responsive to the treatments that are being proposed.
<i>arobit2 :</i>	I would like to know if fatigue is a common symptom of SMA (type 3) like it is for multiple sclerosis. And if so, is there any treatment or recommendations. Thank you very much.
<i>drkissel :</i>	arobit2, fatigue is very common in SMA patients, and is often a multifactorial problem of weakness, poor sleep, medication effects, depression, so-called "over-use" phenomena, and a number of other problems. Most of the time these can be addressed individually or as a group, and your doctor can help with that. Meds like those used in MS can also be helpful.
<i>angiesma2 :</i>	is there any way to create more acetylcholine (MN nerve spit)? or would that just make our muscles keep firing?
<i>drkissel :</i>	Angiesma2, there is no way we know of to make more Ach, and you are right, it might be counterproductive since it might be pumping a low well (so to speak)!
<i>KELLY :</i>	Thank you, also my husband and I have thought of having a baby and I know the weight is a big issue. Is it known that if I had a baby would I be able to walk like I do now after the baby is born. Is there a for sure way of knowing this?
<i>mawlis :</i>	Kelly, I have had two over 9 lb. babies and I walk just the same as I did before. I actually take better care of myself now and lost more weight than what I started out at and I feel better.
<i>Cuco :</i>	Kelly, my sister, age 25, type III, has had 2 children and she is walking pretty much like she used to before she had her first child (3 years ago), but I guess it's an individual question
<i>angiesma2 :</i>	my friend, sma 2, had 2 boys too.
<i>drkissel :</i>	Kelly, there is no way to know for sure what effect a baby would have on your disease. I recently reviewed all of the information I could find on this, and was not impressed at all that patients necessarily got worse with pregnancy. In several reports, the women (and babies!) did fine!
<i>KELLY :</i>	how old were they when they had the babies?
<i>angiesma2 :</i>	my friend was 27
<i>mawlis :</i>	I had my first at age 22.
<i>nekrosys :</i>	I have SMA 2 and have two daughters I gave birth to via c-section at ages 23 and 27. I was basically a quadriplegic. I had some use of my right arm for typing and such, but I need help with all activities of daily

	living. I did not use and still do not use any ventilation aids though. My website is http://www.isoc.net/brokeninside/nekrosys
<i>KELLY :</i>	If I wouldn't be able to walk after I wouldn't do it because it would put a burden on my husband to take care of me and a new baby.
<i>drkissel :</i>	I am very interested in the testimonials about pregnancy and childbirth, and would be interested in hearing from you or your loved ones about your experiences. FSMA can give you my contact information.
<i>angiesma2 :</i>	is mestinon the only inhibitor that comes in a liquid form or crushable pill?
<i>drkissel :</i>	Angiesma2, there are generics for mestinon, but they are the same basic chemical.
<i>Cuco :</i>	Do VPA and PBA inhibit the same HDAC?
	<i>(this question was missed in the discussion and not answered)</i>
<i>rall38 :</i>	Over the last few years, I have noted that I am uncomfortable with environmental temperatures below about 75 degrees. There is no evidence of cardiac problems, hypothyroidism, or anemia. Is this common in SMA?
<i>drkissel :</i>	Rall38, the cold intolerance you describe is a complaint of some of my patients, but many do not have this problem. I suspect it is not directly related to your SMA.
<i>mawlis :</i>	Rall38, I too have an intolerance to cold. I have Raynaud's which causes my finger tips to turn white when I get cold. I take Ginkgo during the winter and it helps them from turning white.
<i>bryce's_dad :</i>	Is Stem Cell Therapy for SMA realistic anytime soon in your opinion - regarding Dr. Kerr recent studies etc.? Seems to me that such therapy would be fairly straight forward theoretically compared to other diseases - since the cells to be replaced aren't as many (many as in varied)? When would you see any such therapy being realistic? 10 Years? 20? Who knows?
<i>drkissel :</i>	That is a good, and very difficult question. I guess my best, most honest answer would be 'no', I do not think stem cells are going to be a viable treatment option anytime soon. There are just too many issues that have to be worked out for it to be "viable". The other question, concerning how long until it IS viable, is even more difficult to answer, and I would not even want to hazard a guess. I do think that within the next decade, we will gather enough information to tell us whether stem cells are going to be a reasonable approach to this disease. <i>(this answer was added after the chat)</i>
<i>KELLY :</i>	Dr. Kissel, how long do you think it will be before they have some type of treatment for adults? It seems like and I understand they are only having trials for children.
<i>drkissel :</i>	Kelly, one of the things I am trying to get moving is clinical trials in adults with SMA. I think they are a great population to study, have been sort of neglected in regards to this disease, and could provide a wealth of information that could be applicable for children. You and I think alike!

<i>angiesma2 :</i>	What is riluzole? What the difference between riluzol & mestinon?
<i>drkissel :</i>	Riluzole is a drug that is used in ALS that we think is neuroprotective. Mestinon acts on a chemical that communicates the signal between nerve and muscle. It usually is used in a disease called myasthenia gravis.
<i>mawlis :</i>	I have heard of some adults with sma 3 have breathing machines. Do I have to expect this in the future?
<i>drkissel :</i>	It is obviously hard to predict without seeing you, but there are MANY SMA 3 patients who never go on breathing machines. There is actually good data on that issue.
<i>mawlis :</i>	That's a relief...but how do you know if you are having trouble with breathing. I seem to have a hard time at times and have been tested for asthma but do not have it.
<i>KELLY :</i>	I have been in one trial, but nothing to do with receiving any treatment, just giving blood to test the smn levels, Dr. Sumner at Ninh is doing it. Are you familiar with that study?
<i>drkissel :</i>	Kelly, yes I familiar with Dr. Sumner's study. As most of you probably know, there was a study of a drug done in adults with SMA. The drug was gabapentin, and although it did not show any benefit, we did get useful information about the disease in adults that we can apply to future studies.
<i>angiesma2 :</i>	does mestinon focus more on MG symptoms rather than sma symptoms?
<i>drkissel :</i>	angiesma2, there is no question that mestinon is a good treatment in MG. In SMA, it is, quite honestly, a "shot in the dark."
<i>angiesma2 :</i>	it has proven to be consistent because I keep having to increase or decrease as another of its side effect is weakness/fatigue, sigh.
<i>mikki :</i>	Are SMA 2 adults more often over or underweight. My son is 23 yrs and although in generally good health (no pneumonia) he is very frail. Seemingly no muscle mass at all.
<i>drkissel :</i>	mikki, SMA patients, especially type 2, can be quite frail, as muscle, which makes up a significant proportion of our weight, is decreased in these patients. Too much weight can obviously be a problem as well.
<i>administrator :</i>	We have about 5 minutes left - if your question hasn't been answered please post it again.
<i>Cuco :</i>	I was recently in a 4 month treatment with VPA (500mg/day I weigh ~55kg) but without any results (my muscular strength did not improve). However, I experienced a loss of stamina. I was not taking any carnitine supplements during this treatment, do you think this could explain the outcome? I have type III SMA.
<i>drkissel :</i>	People are now looking at the role of carnitine in treating SMA patients who are also taking VPA. I am curious, if you don't mind my asking, as to how old you are, what type of SMA you have, and were you in a study, or did your own doctor give you the VPA (valproic acid)?
<i>Cuco :</i>	I'm 23, I have type III and my treatment was in co-operation with my neurologist.

<i>drkissel :</i>	Cuco, yes, I would agree with you on that.
<i>Cuco :</i>	I know now that VPA depletes carnitine levels
<i>KELLY :</i>	In the U.S. we can't take that, can we Dr. Kissel?
<i>drkissel :</i>	Cuco, did your function improve at all, and was your strength measured by computer. Also, have you had the genetic test for SMA?
<i>drkissel :</i>	Kelly, you can take it if you can find a doctor willing to prescribe it and willing to follow you with it.
<i>Cuco :</i>	Yes, I had a genetic test in 1997. My function did not improve at all (measured by isokinetic dynamometri/computer). My knee and elbow muscles were measured.
<i>angiesma2 :</i>	is VPA okay for me to try considering I am on Mestimon?
<i>drkissel :</i>	Cuco, thanks for the information. Don't get discouraged, I think there will be better drugs coming along.
<i>rall38 :</i>	Due to the muscular atrophy in SMA, is it common to develop pelvic floor weakness and problems like rectal prolapse?
<i>drkissel :</i>	rall38, I think pelvic floor weakness and even rectal prolopse would not be unexpected in a disease like SMA
<i>bryce's_dad :</i>	Do you think the Gabepentin study might have shown benefit if studied over a longer period of time? Rome wasn't built in a day...
<i>drkissel :</i>	bryce's dad, quite possibly. We were looking for a marked increase in strength, so there is no question that we could have missed a small effect.
<i>bryce's_dad :</i>	Was MUNE (Motor Unit Number Estimate - accomplished through a nerve conduction) used in the Gabapentin study?
<i>drkissel :</i>	bryce's dad, I think you are a "ringer" because your questions are too good! We did not use MUNE in the initial gabapentin trial. We are trying to look at that now in adults. Motor unit number estimation. It is an electric way to look at the number of motor nerves present in people with SMA and other diseases.
<i>Various:</i>	Thanks very much to Dr Kissel!! also to FSMA! It would be nice to have the chats more frequently!
<i>drkissel:</i>	Have a great day all, and thanks for signing on!!!