

## Usher Syndrome Conference Call, January 11, 2010

Overview:

The main topic of discussion will be "Families and Treatments". Some background:

The good news is that a number of treatments for various aspects of Usher syndrome are nearing clinical trials. For these treatments to reach the masses someone is going to have to participate in the clinical trials. There are also treatments for Usher syndrome currently available, some that are widely discussed and supported to varying degrees (like Vitamin A and sunglasses) and others that offer hope to families but which are supported by less science. What should families be considering when they are approached to participate in a clinical trial? How do families know what science to trust? What should we be doing to ensure that families feel confident when participating but don't put themselves at undue risk?

Present:

Amy Lovelette  
Melissa Chaikov  
Mark Dunning  
Nausheen Fatima Khan  
Irene Goodman  
Irene Kamaroff  
Marly Kenna  
Bill Kimberling  
Heidi Rehm  
Steve Rose  
Krista Scheall  
Karmen Trzupsek  
Tracy Evans-Luisilli

Mark: Nausheen has volunteered to help with Usher Cookbook (kid-friendly recipes). She is contacting local restaurants to see if they will contribute.

Karmen: I'm meeting with Lisa at Children's to discuss the upcoming family conference. There will be an all-day conference Saturday, with stuff for the kids. Saturday evening, we're finding a local Seattle park to have a BBQ get-together. I'll have more updates after this meeting.

Mark: Here's what I'm hoping to discuss today – I'll give you some context from what I've been seeing from families here. I've been contacted by families who have been contacted by researchers who are doing clinical trials and asked whether they should be involved or not. I don't know what to tell them and I'm not sure families have the right amount of information to make decisions. I think families feel they have no other choice because there are limited options. I'd like to give advice on when they should be involved, how, etc. ...what is your advice?

Steve: I would say, talk to your ophthalmologist. We sit and talk, I tell them what you need to be thinking about, asking about, what you should know about participating in a clinical trial – I talk people through it, but I always say, you have to talk to your own personal physician and ophthalmologist – also, any trial they're hearing about should be listed on [clinicaltrials.gov](http://clinicaltrials.gov).

Marly: Mark, you said you've heard that families of Usher patients have been contacted out of nowhere?

Mark: It's not that – families are scrambling, as you know. There is a particular trial running out of the Mayo Clinic – not geared directly to Usher patients, but Usher patients could be candidates. That's one example. I've also been contacted by families who have heard of things going on internationally – I know nothing about what's going on internationally. Does [clinicaltrials.gov](http://clinicaltrials.gov) have anything to do with international trials?

Marly: Anything being run in the US should be registered on [clinicaltrials.gov](http://clinicaltrials.gov).

Steve: And in fact if you look at the website, you'll see trials that are European registered on there. Most are using that as a way of letting people know. While we're talking, I've gone on [clinicaltrials.gov](http://clinicaltrials.gov) and I'm trying to find - the only thing in Minnesota on vision is for patients with Acute Optic Neuritis (AON) that I can find. I just searched vision. I'm not sure what else to say...

Bill: Maybe Mayo hasn't registered yet.

Steve: Mayo has to register before they actually start registering patients.

What about the registry with the FFB?

Steve: We'll certainly publicize if asked - we do send e-blasts for registration - but to check with your ophthalmologist...  
Bill is this what I think this is -

Bill: Anti-inflammatory?

Steve: Yes, I don't know who at Mayo is doing that.

Marly: Literature and website info would help patients better understand involvement in a clinical trial.

Steve: That's what [clinicaltrial.gov](http://clinicaltrial.gov) has - inclusion/exclusion, but it might not be completely clear to the patient.

Marly: I think you need a point person, someone to call after they've reviewed [clinicaltrial.gov](http://clinicaltrial.gov).

Steve: It's something that is very important for people to understand what the realities are - you have to have a realistic understanding of what the potential is. And that doesn't always come across.

Mark: So that's kind of the issue we keep running into - not solely with clinical trials but I've also been contacted by people about a number of treatments. They don't know whose advice to take about these things. These treatments can range as commonplace as sunglasses and vitamin A to more off the wall such as acupuncture and other things I've heard. I'm not a physician and when people contact me about vitamin A I tell them to contact their physician and try to get them in touch with Dr. Eliot Berson (studying effects of Vitamin A on retinitis pigmentosa) who is very good about that. I'm not sure what the best approach is for families - I want them to get Eliot's side of vitamin A, but they've often gotten other opinions.

Steve: It's definitely not a unified opinion. The patient education is huge, it's not only in this field, whether I mean retinal disease or Usher, it's the same thing. It's not a wholesale business, it's a retail business of people having to ask questions and make decisions. The thing that bothers me the most is the stem cell transplants in China. I get questions about that all the time and I'm really scratching my head on that one.

Marly: We had a patient who went to the Dominican Republic to get a "stem cell transplant" and it's kind of spooky.

Steve: Also with acupuncture - we're visiting with a guy whose doing micro-acupuncture with electrical stimulation and we're visiting with a physician to review medical records for which we have received permission from the patients, as required.

Marly: There are so many things, especially if we're looking at acupuncture or nutrients or whatever, hard to have follow up to see if it's working - it's great that Eliot is doing the vitamin A study.

Irene G.: There's this doctor, he's a sort of maverick - blood sugar can have an effect on the inner ear - my son has Ushers, he's hypoglycemic and turns out blood sugar has a lot to do with the inner ear - the doctor put him on something that strengthens bones and a sugar free diet. We figured it can't hurt because if he's already hypoglycemic, he needs it anyways. When we went back, he actually experienced a slight improvement in hearing. The improvement was in speech recognition, not volume, because the ringing in his ears goes away when he's on the sugar free diet. Has anyone heard of this happening?

Steve: Do I believe it's going on? I never tell someone that they're not experiencing what they say they are.

Irene G.: There's no question that it's happening.

Marly: If you manage the inner ear tinnitus (ringing in the ear) – salt, sugar, those kinds of things balance, so - who knows? Short answer? It's possible, yes. I'm an ear doctor so most efforts have been focused on vision because fortunately we have some things to offer people with hearing loss but very little to offer people with vision loss.

Irene G.: The goal was to maintain it, not to improve it, but there was improvement in the speech recognition.

Marly: Tinnitus management (ringing in the ears) - a lot of that seems to be diet based – hearing may not be improved, but may be managing the tinnitus better.

Mark: You get a lot of “well, it can't hurt” types of responses. Often it's left to the families to figure out whether it could or couldn't hurt.

Marly: If you have eight different people saying they're all on a hypoglycemic diet, then we should do the study. If it's random things, you want to make sure before you study it that it's safe.

Mark: From a larger perspective - getting from here to a cure – we need families to be involved in the clinical trials. We need them to feel fully educated and not feel like guinea pigs. “There's no real treatment for this so how about you try this.” On the other end of the spectrum, I have families that are willing to do anything, they'd be willing to try it without necessarily having a scientific support of what it is they're doing.

Marly: It seems you're asking for a [clinicaltrials.gov](http://clinicaltrials.gov) clearing house that isn't associated with [clinicaltrials.gov](http://clinicaltrials.gov). to claim whether it's safe. I think we could have a clearing house with links, with a caveat that this is available, ongoing but they have to talk to the study people.

Irene G.: You talk to a very good doctor and ask, is this safe? The kind of clinical trial you really want is something very cutting edge and medical that will really help.

Marly: I think what Mark is getting at – after you look into a trial – who do you speak with next?

Mark: It sounds like [clinicaltrials.gov](http://clinicaltrials.gov) is a good link to point people to. If it's not on the list, you should be suspicious.

Marly: Before you get approval for the trial, you have to prove it's registered on [clinicaltrials.gov](http://clinicaltrials.gov). It's a website that they're maintaining for ALL clinical trials. A lot of trials were only publishing positive data – this is supposed to be one of the ways that get the people doing the trials to be more forthcoming about the outcomes, rather than just the positive.

Mark: As for hoaxes, things that have been debunked in some way, Steve you mentioned the stem cell replacement in China. First of all, I don't know, is this a complete hoax or just ahead of its time? Like the acupuncture, it seems it may be legitimate but it might not be? How do I address that with families? If we suspect there is something that might be worth pursuing, how do we let the families know?

Steve: You're getting into sticky territory because that gets into liability, what you're saying about others, etc. I had someone who went to China to look at the study groups, came back with the data, and said no. Mark, I don't think it's anything you can just throw up on the website, because it always comes back to a discussion. For example, we're now looking at micro-acupuncture with electrical stimulation – I don't know what I don't know. That's why we're sending physicians to visit.

Mark: For me personally, as a parent, those things didn't personally interest me – am I doing my family a disservice by not pursuing something? To be able to say there's no discernible proof that acupuncture ... however, the FFB has doctors going to look into this - my suggestion to the family would be why don't you wait until the FFB comes back with results. Same thing with China.

Irene G.: I like what I'm hearing; maybe they know something we don't, who knows? When I hear the FFB or an organization I trust is out there checking it out, that's enough for me.

Mark: I'm not sure how exactly to go about this and Steve, I get what you're saying. I think from the family's perspective, you think I don't just have to take this one person's perspective. I think that's what Eliot runs into with the vitamin A stuff; he's so enthusiastic about it, has a lot of data, but everyone knows he has a vested interest in its success. If you go based solely on one person, this guy says it's going to help me, but it's going to help him if it helps me. - Eliot Berson is a doctor studying the effects of vitamin A on retinitis pigmentosa. There's been quite a bit of debate on whether it's applicable to Ushers.

Marly: A lot of the diagnoses were made before genotyping was done, but a lot of the follow-up he's doing now is terrific.

Mark: A lot of people say I've heard about the vitamin A treatment but my ophthalmologist says don't do it. I recommend talking to Eliot, but then they're in the spot of my ophthalmologist says don't do it, he says do.

Irene G.: I would say what can it hurt?

Bill: Well, the answer to that is that in large doses, it can be harmful. Anything taken in large amounts can cause issues. There are people that take standard ibuprofen, that can cause problems. It's rare, but it happens.

Irene G.: For people in that position that's an issue, for my son, it's not. He'll try anything he can.

Mark: When you get into stem cell or acupuncture, things that families find on their own – it's probably good to tell families here's where you should go for info and if it's not there, not on [clinicaltrials.gov](http://clinicaltrials.gov), FFB hasn't heard of it, it's probably something you don't want to be involved in.

Well thank you everyone. I think that [clinicaltrials.gov](http://clinicaltrials.gov) will be a useful resource and Steve, thank you for sharing about FFB.

Steve: My answer to you, if you all wind up with someone that wants to talk, I'm here. I'm the Chief Research Officer of Foundation Fighting Blindness. Someone came to me about something we knew nothing about, in a country in Asia; we got on it right away. I should be plugged into all those things, but sometimes things slip by.

Irene G.: I'm a literary agent in New York, I recently held a charity auction where 26 people bid to get their manuscripts read by me – see more information at [www.IreneGoodman.com](http://www.IreneGoodman.com) – all proceeds go to the FFB and Deafness Research Foundation. I'll keep doing it.