

## FOD Concerns Need to be Addressed

[This was an Editorial posted in our July 2007 newsletter]

Over the course of the last 17 years, I've had the opportunity to talk with MANY new FOD Families. It's a great way to share what I've learned over 22 years of living with the death of an undiagnosed daughter and with a surviving son with MCAD, as well as try to answer the many questions they might have and also to listen to their many fears. From those conversations, I've noticed several themes that keep recurring that I feel all of us (Families and Professionals) need to be aware of. Many of these issues are addressed on our site and in past newsletters, as well as over our Email List (with 450 members), but I thought if they were posted here they might be a way to create a focused dialogue between ourselves and those that we come in contact with in regard to our child's/our disorders.

Even though EACH disorder has differences and EACH affected person responds differently to their disorder, the following points impact us all ~ affected and unaffected. This is not a complete list of concerns (others include being accused of munchausen and making your child sick, not being able to get coverage for formula or other supplements, school and therapy issues, etc), but it's a starting point for all of us to discuss with our Families and medical professionals. Feel free to cite any of these points in your discussions with others or when you might be presenting your own story in front of medical students ~ the more that are aware of these concerns the better ~ **COMMUNICATION and PROACTIVE BEHAVIOR are KEY when living with a rare disorder and ineffective communication and passive behavior can be fatal in some circumstances!** It would be great if you would create your own list of concerns and share it with us too ~ I can post more concerns/issues and your own positive or not-so-positive comments in future newsletters.

- With **expanded newborn screening (enbs)** spreading across the country (albeit too slowly for me!), there are several issues:

- 1) states/countries/hospitals that do NOT yet screen for the 'latest' full panel (that's another whole issue in itself – what they consider a full panel!) **NEED to tell parents that it's still possible to get the expanded nbs and where they can send the filtered paper!** [refer to our Med Info page, Diagnostic Labs]

- 2) be aware that the **enbs** is just that - **a screening tool – it is not the Diagnostic test** that will be done when the 'redflag' screen shows some abnormalities ~ even though both use the tandem mass spectrometry instrument, there are different ranges for the enbs and the diagnostic tests.

- 3) also be aware that there **may be false positives** in regard to the screening ~ this is where I hear the most disturbing comments from parents [note that this doesn't happen every time but even ONCE is too much!]

For example, and I've heard this several times, parents will receive a call on a Friday and the message is left on their answering machine that their infant "has a fatal disease and we need to see him/her immediately!" – but by the time they call the office back the staff has left for the weekend – so the parents are FRANTIC and DEVASTATED about this call and not sure what to do or whom to call!

First of all, stating that the infant HAS the disorder is not fully accurate UNTIL the follow-up DIAGNOSTIC test is performed (and sometimes even a further skin biopsy may be needed as well to confirm a diagnosis), and it's HOW the positive screen information is conveyed (by the staff/genetic counselor or other professional) – I do NOT feel it's appropriate to leave a frightening message on an answering machine and then not have a number the parents can call (even over a weekend) to get the FULL information. Some parents were told NOTHING about the FODs and what they might be able to proactively do while waiting for a follow-up DIAGNOSTIC test and the results (i.e. make sure to feed their infant often, avoid fasting, watch for vomiting, what might be done in a possible crisis, etc). Families have also had to endure weeks of waiting for results – only to find out that the staff knew the results 2 weeks before but failed to convey that to the Families! Of course they were relieved to find out their child DIDN'T have an FOD but the PROCESS to get to that point was HORRENDOUS for them!

Professionals --- if you see yourself in this scenario PLEASE be aware of HOW and WHEN you communicate a positive screen to a Family- it's important to convey the information without frightening them to death or putting them into panic mode by not giving them enough info. Again - **Effective Communication is KEY!**

4) Additionally, too often moms are made to feel guilty for supplementing with formula and/or pedialyte (versus just breastfeeding) after giving birth – we need to get the information out to medical professionals that **that supplementing with calories COULD be the difference between LIFE AND DEATH for some infants while waiting for the results of the enbs!** Encouraging breastfeeding is important BUT being so adamant about it and telling moms to ONLY breastfeed can be very dangerous to those yet-to-be diagnosed infants with an FOD.

- Another set of issues that many Families have discussed as I speak with them over the phone and also over our Email List, is the **use of the terms 'mild' and 'non-disorder' in regard to various FODs.**

Those infants being picked up via enbs most likely will NOT have the symptoms that many of our kids had (especially due to NOT being diagnosed at birth, some diagnosed after years of episodes or after their death) because they are being **DIAGNOSED and TREATED from birth BEFORE ANY SYMPTOMS CAN OCCUR!** And the parents KNOW what to do in an emergency BEFORE a serious problem can occur!

To be told their child has a 'mild' case ONLY based on the mutation they might carry makes new parents complacent – they think they don't have to follow any type of treatment protocol because the Dr told them it's a 'mild' case and the child shouldn't have any problems – they are being told it's not serious enough to be concerned about! That's a simplistic explanation (of the comments I have heard from many parents) on my part, but you get the point!

**How in the world does someone KNOW FOR SURE AT BIRTH how a child will respond to their disorder or to a future illness or stress???**  
**THAT – is what I am objecting to – NO ONE KNOWS FOR SURE!**

As for the '**your disorder is a NONdisorder and doesn't need to be treated' issue**: then why are the affected individuals so symptomatic?? We are hearing from SCAD Families that are being told SCAD is one of those NONdisorders – these comments really concern me. As Mary, our webmaster, commented to me, if someone was diagnosed with skin cancer, one that might be considered 'mild,' do you think they'd just let it go without treatment? Most likely NOT – they would treat it so it would not become more complicated and cause more medical problems and possibly a fatal outcome!

**In my opinion, if one has a metabolic disorder they either HAVE IT or they don't** (just like being pregnant – you either are or you aren't) **so TREAT TO PREVENT COMPLICATIONS**—why get stuck on whether it MIGHT be a so-called 'mild' case or a so-called NONdisorder??

If the professionals don't think it's a REAL disorder then why don't they RENAME it to reflect what it truly IS just like they did with LCAD years ago – renaming and re-diagnosing many initially diagnosed with LCAD with VLCAD. The same applies to some initially diagnosed with LCHAD – only to be re-diagnosed with Trifunctional Protein Deficiency (TFP) a few years ago. Yes, some of our Families have a dual diagnosis of an FOD and a mito disorder – so why don't they do further testing on these NONdisorder kids and find out if there's something else going on —instead of just calling the SCAD a NONdisorder and telling the parents no further treatment is needed!?

So what's next – if SCAD is a so-called NONdisorder, are we going to have NBS Labs making the decision to NOT tell Families that their baby screened positive for it because it's NOT a REAL or SERIOUS disorder?? I would think that would be pretty dangerous to NOT tell parents. Also what impact will being labeled a NONdisorder have on the entire enbs process – will states be the ones to just drop the disorder because SOME THINK it's a NONdisorder!?

Again, I think that would be setting a very dangerous precedent. As many have learned, these disorders can 'wax and wane' over time -- who's to say some trigger won't put an affected individual in a crisis? **Individuals have DIFFERING THRESHOLDS for when they may present with symptoms.**

Then the parents will happen to mention to the ER Drs that they were told their child/self had a 'mild' case or that their disorder was a NONdisorder and the staff may dismiss the seriousness of immediate treatment – **what are you going to say to that parent when that child/adult is medically damaged or died because it wasn't taken serious enough?!**

Lastly, one of those 'symptoms' that I hear a lot about is what many consider '**low blood sugar' or hypoglycemia** – I really think medical professionals **NEED to rethink that term in regard to FODs**. We've had many Families get to the ER only to be told to go home because their child's (or your) blood sugar was in the 60s or 70s and with fluids should be okay! As stated earlier – EACH affected person will respond DIFFERENTLY to DIFFERENT TRIGGERS and **blood sugar CANNOT BE TOTALLY RELIED ON to determine whether one is in crisis or not!**

If you read my Jan 2006 newsletter Editorial you would know what I am referring to – when my son, Kevin (19 at the time, MCAD) was in crisis after oral surgery where the anesthesiologist TOTALLY DISREGARDED the 10% dextrose IV protocol and only gave him 5% – even though his BS was 76 (which most consider close to normal range of 80-110) it was NOT at the NECESSARY 100-120 level to keep him from going into crisis from such a stress – and that ARROGANCE put Kevin in the ICU!!

**NEW Parents** – you will need to learn that waiting for the blood sugar to drop to 50 or below to go to the hospital (which is often what medical professionals call hypoglycemia) could be VERY DANGEROUS for an FOD individual. **We have to create an awareness about how 'normal' blood sugar levels don't always relate to FODs!**

I'll stop there for now – but it really bothers me that some are being so 'casual' about SCAD and some other FODs, as well as these other FOD concerns. Let's take ALL the FODs seriously so we can avoid major complications or another senseless death!

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