



Newborn Screening
Saves Babies
One Foot
at a Time

James and Samuel

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHO HAS TWO CHILDREN
WITH TRIFUNCTIONAL PROTEIN DEFICIENCY (TFP)

It is hard to believe that almost five years have gone by. James was born January 12, 1999 and is a miracle and a survivor. I can only highlight our experience because the story can go on for hours.

James was born not only with liver disease (biliary atresia) but also Trifunctional Protein Deficiency. We always start our story with "If James hadn't had liver disease, both my boys would probably not be here today." James (almost 5 yrs old now), and Samuel (almost 4 yrs old) both have Trifunctional Protein Deficiency.



Unless you have lived through similar traumas, it is difficult to really understand what life is like with two volatile chronically ill children. Yet in the same breath, we thank God for all the research, the support sites and the efforts that keep our sons on a "healthy" path. We thank God for our doctors, our family & friends and other FOD families.

I have to admit that life without James and Samuel would be horrifying. And we certainly have had extreme "episodes." Two liver transplants, cardiac arrest, respiratory failure, seizures and numerous intubations. And that is just for James. Although he has been affected mentally and physically from all of his trauma, he is interactive and tries really hard on a daily basis to move forward. He is wheelchair bound and totally dependent. He says two phrases now: "Dao" for Daddy and "Ah Oh Ew" for I love you. Mommy currently is "Olm." We keep working! He is a very happy and vocal child and we do all that we can to allow him to be his best. There were days, and still are days, which we think he may not make another year. But he does.

Sam (Born Feb 2000) is a spit fire, yet he can end up in the ER with a CPK of 120,000 simply from playing. He has been protected by the knowledge gained from James' diagnosis. Sam was tested at birth and treated with IV glucose as a precaution even though he appeared to be a perfectly healthy newborn, weighing 8lbs, 12ozs. Sam has a wide gait and low muscle tone in his lower extremities. He also has feeding and sleeping issues. Cognitively, he is sharp -- -and exhausting --- in a good way! He tries very hard to keep up with his older sister and peers, but it is difficult for him. He loves firetrucks and all sports balls. Our thoughts about Sam are that he is the "healthier" of the two, yet really he is very fragile. Doctors even think that James may be benefiting on a TFP level, due to his different liver. Only time will tell.

And then there is our Gabrielle. Our completely healthy 8 year old. I always mention her in the boys' story since she too is affected by our daily routine, yet is simply a joy and a big help. She is a big part of the boys'

lives. (She would love to hear from other siblings.)

I want to share what has protected my boys and our family.

1. Constant attention to their baselines. 99% of the time we catch an illness at the onset. I believe this has saved them.

2. Ask questions and learn what works for your child. Not all treatments work the same. Not all medications interact properly with one another. Don't hesitate to call the oncall doctor.

3. Bring the protocol letter to the Emergency Room. At least bring the knowledge in your head of what MUST be done. Help educate the staff about the necessity of immediate treatment. ie: IV fluids, lab work, urine analysis. And about the seriousness of the illness!! Even though your child may not appear ill to them, always say something pertinent. I say: "Yes, he seems well, but he's had CPK's of 120,000 within hours." "He's a metabolic kid." Or "He's a transplant kid." They'll listen!

4. The #1 In-patient Rule Is: Be Alert. Constantly check things and ask what is going on with your child. Mistakes can happen unintentionally.

5. Constant Nutrition. Both of my boys have a gastro tube and an infusaport. These pieces of medical apparatus have saved them on numerous occasions. My boys have learned to eat, but the gtube feedings of special formula are what gives them the nutrition they need to grow safely. Don't be afraid of something which can save your child's life, like the ventilator.

6. Communication with your Doctor. Learn to work with your physicians. Listen, learn and also give them feedback. With these rare disorders, parents often see more.

7. Be Grateful and Humble. To God, family, friends, teachers, staff, and the facilities that keep your child alive and well. Reach out and make calls.

8. Advocate Newborn Screening. Tell everyone you know about the websites: fodsupport.org and savebabies.org.

9. Be A Witness. Tell your story when you can. Write it down to get your feelings on paper. Call the media.

10. Pray. Pray for protection and stability for your whole family unit.

Our daily life sometimes seems impossible. But I truly believe that my boys are survivors to help explore and develop solutions. And to give other families hope.

With love,
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James, 4.5yrs Trifunctionl Protein Deficiency;
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