

MCADD- Medium-chain acyl-CoA dehydrogenase deficiency

Newborn screening had a very big impact on my family. Both of our children have been diagnosed with the same deficiency. In the beginning, we felt like this was the worse thing in the world. But with the proper education, they are able to live normal lives.

To be honest, I really don't want to think about how our family would have been affected if our daughters had not been diagnosed when they were. Both of our babies were very happy and healthy.

There was a situation that happened with my youngest child. She had just gotten over a chest cold and one morning had to go and physically wake her up from her night's sleep, normally she was up by 7am, but it was 8:30am. I brought her downstairs, made her some pancakes and she was just kind of like sitting there slouching in her chair staring off into nowhere. Picked her up and she was very lethargic. Her eyes were barely opening. I called my mother-in-law. She came right over. We knew we had to get her to the hospital.

When look back probably should have just called 9-1-1 but my oldest daughter never had any episodes like this before.

The entire ride to the hospital she was staring off and not really responding to me talking to her. It was a very scary moment. I remember it as if it happened yesterday.

We got to the hospital and they immediately started IV fluids and she perked up a little bit.

While she was sick with her chest cold, her appetite went way down and it eventually caught up to her, which is why she had the episode.

Now, if we had never known about her deficiency I would have probably just chalked it up to her still being sick and maybe put her back to bed. That could have definitely made things worse.

My husband and I reaction to the newborn screening results were both shocked and overwhelmed for our first child. It was expected for our second and definitely easier to handle.

We were both thinking we have this perfectly normal baby and when our pediatrics doctor called to tell us the news and HE couldn't even give us any information about it. It was very scary moment for us.

Both of our children go to Children's Hospital Metabolic Clinic in Southfield.

They have been wonderful from the start and have made all the difference in both of our children's lives.

I will never forget my first phone call from the Nurse, Tammy. She was so nice, informative and calming.

She assured me this was something we could get definitely handle, that we could get through it together and the kids would live normal lives.

I would tell a parent that is considering refusing to screen to their baby to definitely reconsider it. There was really no history of this in our family. The deficiency that my children have could be life-threatening if not handled correctly.

It was very overwhelming being a first time parent, especially when they are diagnosed with something that is not so common, but we received the proper knowledge and were put into contact with parents with the same situation and it just helped us that much more.

You only want what is best for your children and I think this is something that would be very beneficial to them for their future. (read my story above)

All it takes is a few drops of blood!