



Newborn Screening
Saves Babies
One Foot
at a Time

Damian

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS
3-METHYLCROTONYL-COA CARBOXYLASE DEFICIENCY (3MCC)

Damian was born on April 17, 2008 at 3:41 pm and weighed a healthy 8 lbs. 9 oz. He is our sixth child and all our children had been perfectly healthy so when the nurse took him to do his first newborn screen-ing test, we really didn't give it a second thought. And I didn't have to give it a second thought until Damian was five days old.

That's when the pediatrician called us personally at 7:00 pm. I instantly knew something was wrong. She told us that Damian's newborn screen-ing had come back abnormal for 3mcc. I was in such a state of shock that I didn't really ask her any questions.

She asked us a few questions about how he was doing (was he sleeping too much, difficult to wake, any vomiting, etc.) and we had to take him to our local children's hospital to get a second screening as well as some other lab work done.

After my initial shock wore off, I immediately got on the internet and began researching 3mcc. I was overwhelmed with devastating information and very sad stories. After getting the tests done, we went to see the pediatrician. She explained that the horrible things I found on the internet were what could happen to a child who had 3mcc and was not diagnosed early. Diet, supplements and special precautions can prevent all those things from happening.

She told me not to worry too much until we got the results from the second newborn screen and lab work. But of course, I worried about it every minute of every day!

After about a week, the results came in and again confirmed the 3mcc diagnosis. The doctor ordered some more follow up tests, which in another week came back with the same thing, Damian had 3mcc.

Admittedly, we had a very hard time accepting this. We had five other children who were completely healthy and it just felt unbelievable that we could carry such a genetic disorder. It was a lot to deal with initially. Time has helped us to realize that his early diagnosis was a blessing, as we now have the best chance at preventing the complications associated with his disorder.

We did have to have four of other children tested, because there was a chance that they also had the disorder. Only our fifth child had been newborn screened for 3mcc, as our State did not add 3mcc to their newborn screening panel until July of 2004. Although three of our children did have lab work results that suggest they are carriers, none of our other children have the disorder.

The journey we've been on since Damian's diagnosis has had its ups and downs, but has already taken us amazing places. We have met some of the most wonderful families and some of the kindest people we've ever come across in our lives.



Damian is an adorable, strong little guy that has truly changed our family. We have learned to see the blessings in everything, even in times when life is really difficult. When life feels overwhelming I learned to just take things day by day, hour by hour, or even minute by minute.

Sincerely,

Laura

Mom to Damian