



Newborn Screening  
Saves Babies  
One Foot  
at a Time

## Seth

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

Our son Seth was born on November 26, 2002, healthy, beautiful and responsive. We would never suspect anything to be wrong with him until we got the call from the Metabolic Genetic Clinic when he was five weeks old. At the time of the call Seth had a bad cough, which eventually developed into Respiratory Synthical Virus (RSV).

At the time of Seth's birth the California Expanded NBS research study was taking place. I was aware of this study, so I had signed the consent to have the extra testing done. The clinic contacted us with a sense of urgency to bring him in within 24 hours. I found this information disturbing, and my husband and I were skeptical. The following day after the call we brought Seth in and the same test was repeated. The results took five days to process, and in the meantime Seth was admitted to the hospital with RSV. Seth's doctor, Susan Winters, called us at the hospital (she was out of state at the time) with the lab results. It was confirmed that this was not a false positive, and the doctor ordered Carnitine and Biotin immediately. He was able to go home two days later, recovering well. The results of the two series of labs did not make total sense to the medical team, so they took a biopsy for further enzyme testing. Based on the biopsy results, they identified Seth's diagnosis as a partial deficiency of 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC). I have read so many unfortunate, sad stories of affected children. This is a happy one in that Seth is normal, healthy and has no disabilities due to his disorder. It was discovered only by the expanded testing which is now state mandated. My son has a delightful personality, and he is bright and beautiful. His life was saved, and his quality of life will be normal because of the expanded newborn screening. Thank you for your efforts to ensure that all children have the appropriate NBS testing.



Sincerely,  
Brenda, Seth's Mother  
Written by Brenda  
Mom to Seth - April, 2006