Born with MCADD

Medium chain acyl-CoA dehydrogenase deficiency

Willem L. Middelkoop
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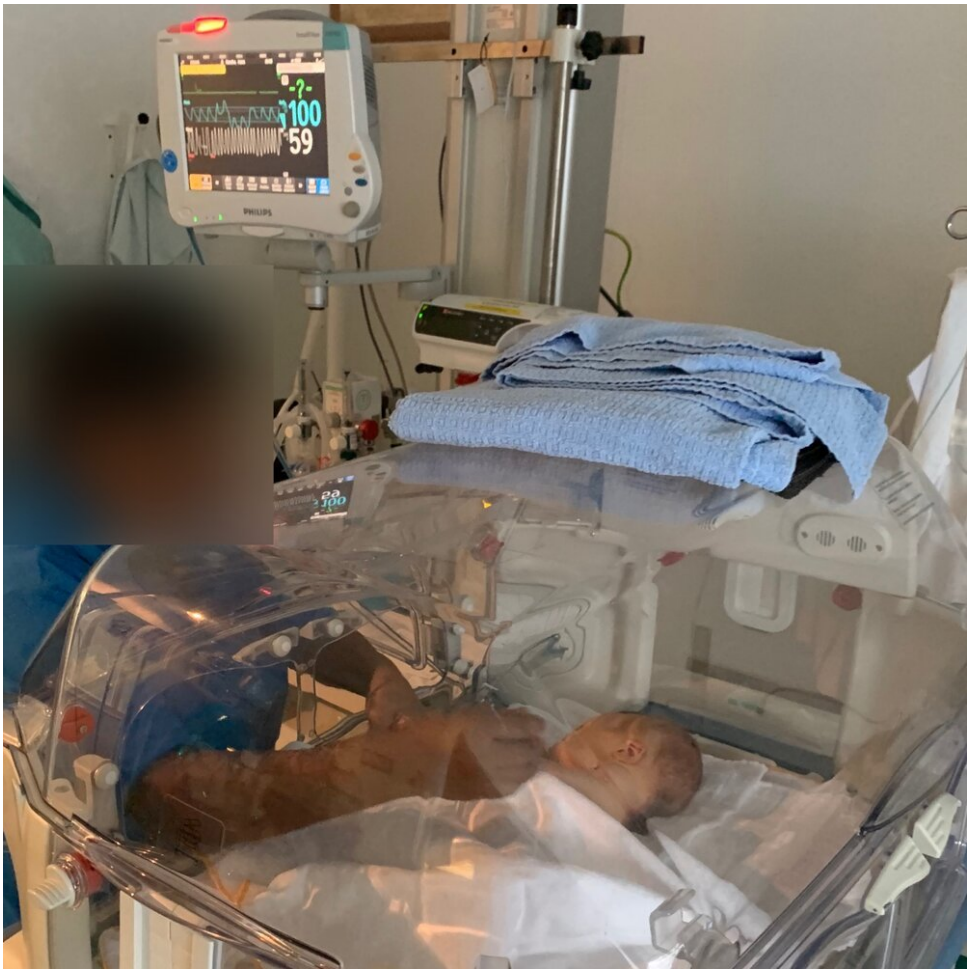
Just days after our son was born, we ran into trouble when our little man didn't want to eat anymore. Repeated attempts to feed him failed as he appeared to be too tired to drink from his mother. When we measured his body temperature we were shocked, only 33º Celsius! What followed was nothing less than an incredible emotional rollercoaster!

Hospitalised

We wasted no time and decided to take our son to the hospital, PRONTO! Because my wife was still recovering from giving birth (and therefore she was less mobile), we decided that I should go alone with our boy to be as quickly as possible. This was one very convenient time to have experience with driving a car as fast as possible!

Once in the hospital, the doctors knew of our arrival but couldn't believe the baby's temperature. They thought the measurement was an error, but their own reading confirmed the dangerously low body temperature. Before doing anything else, he was put in an incubator and was given some food via a syringe.



Saving the life of our son - note the heart rate monitor being unable to detect heart activity (the green question mark)...

Critical moments passed when the monitor had trouble detecting the kid's heart activity. He was still breathing, but his low temperature and weak appearance caused serious concern. It's a rather shocking experience to see your newborn boy like this.

Rapid recovery

The loving care of the nurses, the heath inside the incubator and the energy from the food did wonders for the little man. Within mere hours, his body temperature rose and he regained his appetite. I felt intensely happy to be able to feed him again!

It felt like victory to see him drinking again!

Although his vital signs returned to normal, nobody (including the doctors) had any idea what caused the boy's crisis. The doctors followed standard procedure, taking tests to look for an infection or any sign of (internal) trauma that might have gone unnoticed since his birth. They found none, by all their standard readings and measurements the boy was completely healthy.

Unsatisfied with the lack of a clear cause, the doctors advised to keep our son under close monitoring for some more time. We were welcome to join him spending some nights in the hospital. Three nights later, we were allowed to go home as he continued to appear completely healthy. We remained uncertain about what caused his critical condition, but not for long as the answer would present itself (rather literally) at our doorstep...

Newborn screening

While I was happily delivering birth announcement cards, I received a call from my wife: the doctor is here, please come home now!

Once home, the doctor started what seemed like a textbook procedure for a 'bad news delivery'. He told us that the newborn screening revealed an anomalous reading, indicating a rare genetic metabolic disorder called *"medium chain acyl-CoA dehydrogenase deficiency"* or MCADD.

We were asked to go to the hospital immediately and present ourselves at the emergency room where a specialised doctor would await us.

VU Amsterdam emergency room

MCADD diagnosis

The specialised doctor welcomed us in a friendly manner, she explained that they needed to confirm the MCADD indication before jumping to any conclusions. This was done by taking additional blood and urine samples from our little man.

As this was a different hospital (and doctor), we were asked how we experienced the days after our son's birth. Our story of his sudden hospitalisation did not seem to surprise the specialised doctor at all. Shedding new light on the crazy first days of his live, the doctor explained us what MCADD is.

What is MCADD?

MCADD is a rare genetic metabolic disorder affecting the body's ability to use fat as energy. The human body needs three primary nutrients in order to function:

- carbohydrates: are broken down into glucose
- proteins: are broken down into amino acids
- fats: are broken down into fatty acids

As we eat a meal, carbohydrates are broken down into glucose causing our blood sugar to rise. This causes insulin to be released into the bloodstream, enabling the glucose to enter our cells, distributing the meal's energy through our body.

If there is more glucose than the body needs, the excess is stored in a "glucose store" (glycogen) inside the liver and in fat cells. In between meals, the body can use the glucose store for energy. When the glycogen is depleted, the body turns to fat stores for energy.

Unfortunately, those individuals with the MCADD disorder lack (or have an insufficient amount of) an enzyme necessary to break down the fat to produce energy. The *medium chain acyl-CoA dehydrogenase* is an enzyme that is needed for the oxidation of medium chain fatty acids found in fats. The inability to oxidise these fatty acids, prevents the body from utilising fat as a source of energy. This can lead to an accumulation of unused fatty acids causing damage to liver and brain, leading to a metabolic crisis.

Simply put, with MCADD you run into trouble once all the energy from food is used. This leads to sleepiness, lethargy, coma and ultimately death.

At least he got a cool Garfield adhesive band aid

Living with MCADD

The additional blood and urine tests confirmed the MCADD diagnoses and marked the beginning of our son's life with MCADD, as there is no cure available (yet).

During additional visits to the hospital we learned that living with MCADD is perfectly possible by:

- **Eating often**: to prevent running out of energy, causing a metabolic crisis
- **Eating wisely**: to avoid the types of fats the body cannot break down, plan a high-carbohydrate, low-fat diet.
- **Taking medicine**: an additional supplement of L-carnitine might be prescribed to help the body remove harmful substances.
- **Being prepared**: for situations when appetite is low, such as illness or when one vomits, or when extra energy is needed (e.g. during intense and prolonged workouts)

Conclusion

Thanks to the newborn screening results we now know about our son's MCADD disorder. This helps us prevent future life threatening situations like the one we experienced just days after his birth.

This is one hell of a way to start our parenthood, but it could easily have been much worse. I'll use this blog to share our (future) lessons in dealing with MCADD as it may help others. With proper care, there's no reason why someone with MCADD cannot live a normal, healthy and active life!

Links

- http://minutesmatter-mcadd.org
- mcadd.be
- https://ghr.nlm.nih.gov/condition/medium-chain-acyl-coa-dehydrogenase-deficiency
- https://www.nhs.uk/conditions/mcadd/
- https://www.stofwisselingsziekten.nl (Dutch)

Feel free to contact me if you want to share a website or your experience with MCADD with me.