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Chairman Ritter
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Members of the Public Health Committee
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RE: Testimony in Support of HB6580
An Act Creating an Advisory Council on Rare Diseases
Public Hearing Public Health Committee, February 20, 2015

Ladies and Gentlemen:

I am Heather Harwood's mother and she asked me to talk about how our grandchildren's medical conditions have affected my life.

When my kids were young, I worried about the usual things--would they grow up to be happy, independent, successful? With my grandchildren, I worry--will they grow up?

When Maren was born, it was one of the most exciting days of my life. I was with Heather for her birth, and I fell in love with her immediately. Three days later, I got a call from my hysterical daughter. Something was wrong with the baby. The newborn testing had shown she had something called MCAD. After the initial panic, I called my sister who was a nurse at the University of North Carolina. She had her PhD in nursing administration and was on many public health committees. When anyone in the family had a medical question, it was "call Jan." She will know the answer. She was astounded. She had never even heard of the condition and couldn't believe that we lived in a state that had newborn testing for a disease she had never even heard of. I thank God every day that we live in Connecticut that actually tests for this condition. If Heather hadn't gotten that call when she did and rushed to the doctor, the lab and the hospital, Maren wouldn't have lived through the night. Days, months, years of trying to feed her every couple of hours, countless trips to the hospital. It really did take a village to keep the child alive.

Then Jack was born. We were assured he was healthy except he wasn't. He cried constantly--that cry that a mother recognizes as a child in pain. Heather took him to endless

doctors—they couldn't find anything wrong. Except he was not gaining weight, was not hitting all the milestones he should have. We were told he might "outgrow" it.

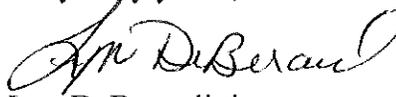
Lastly, Caitlyn came along. Again, she seemed perfectly normal. Until she wasn't. Heather found her lying on the floor next to her toys seemingly asleep, but she couldn't wake her up. Because of her experience with MCAD, Heather took her blood sugar and realized it was dangerously low. The ambulance ride to the hospital. Testing. No one knew what was wrong with her. Home she went. She wasn't eating, wasn't talking, wasn't gaining weight. One of the tests showed that her blood sugar was incredibly low over night so it was decided to insert a feeding tube so she could get continuous overnight nutrition. What a difference. They decided that it was pretty likely that whatever was wrong with Caitlyn might be wrong with Jack, so he got his own "tubie."

They are all doing well now, but their diagnosis is mitochondrial disease—which basically means that they know something is wrong but don't really know how to treat it or how it will progress. I ride around with medical protocols in my glove compartment in case I'm the one who gets the call from the school nurse. If they get the flu or a stomach bug, it is pretty likely that they are in for a long stay at the hospital. My daughter has spent endless hours there trying to comfort a sick child while worrying about the children still at home.

When someone has a chronic disease, we hold out hope that someday someone will "find a pill" to cure it. It happens. It is not going to happen with these rare diseases unless someone is actually looking for a cure, and why would they? How can they make money when such a small percentage of the population is affected? Obviously, we need research. Just as obviously, we need better support for these children and their parents who struggle to have a "normal" life but face a community that doesn't know what to do with them. The symptoms make no sense. They don't fit into any known category. Maybe the parents are over reacting. Maybe the child isn't really sick. Maybe if we give them a couple of stickers and send them home, they will just "outgrow" it.

My three grandchildren are alive because of my daughter's relentless pursuit for answers, but we need your help. Connecticut is a leader in the newborn testing that saved Maren's life. Let it be a leader in passing the legislation to help these families with what most of us take for granted—enjoying watching their children grow up.

Very truly yours,



Lyn DeBerardinis