



FOD Family Support Group

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I am writing to you as a concerned parent, as well as the Director of an international Family Support Group, that was 'born' out of the death of our daughter, Kristen, who was born on October 6, 1983 ~ then died suddenly 21 months later on July 21, 1985, all because she was **not diagnosed AT BIRTH with a DIAGNOSABLE and TREATABLE genetic metabolic disorder called MCAD. If she was diagnosed and treated AT BIRTH, today she would be an active 18-year-old young woman finishing her first year of college.**

MCAD is one of several Fatty Oxidation Disorders (FODs) whereby fatty acids cannot be broken down in the body to be used as energy, especially in times of fasting or illness and are treated through dietary changes and medications. These disorders are currently part of several states' expanded Newborn Screening panels either mandated, as here in North Carolina, or on a pilot study basis. Obviously I would **ideally like to see ALL states universally screen for several disorders**, but that will take time and a lot of perseverance. However, until then, **educating families and professionals about the availability of this expanded screening is IMPERATIVE to preventing more tragic deaths.** Therefore, I applaud Senator Dodd and others for bringing this issue to the forefront and **strongly encourage your support for making AWARENESS and EDUCATION OF EXPANDED NEWBORN SCREENING in the United States a priority.**

I thank you for this opportunity to share with you **why** I feel it's so **very important** for families and professionals in **every state to be aware that these tests are presently available**, especially if their state or hospitals are not yet utilizing them.

The pictures enclosed in this testimony are my family's 'WHY!' Our 21-month-old daughter, Kristen, was joyfully running through the sprinkler on the night of July 20, 1985 and **by 6:05 pm on Sunday the 21st, she was DEAD!**

She woke up that morning vomiting and by 11am was in a coma (although we thought she was sleeping since I had just conferred with the on-call nurse and she said 'The flu is going around, let her sleep'). At 4 pm I called the nurse again because I was concerned about dehydration and Kristen sleeping so much. While on the phone, Kristen stopped breathing. We immediately started CPR until the paramedics arrived and they took over. Although the ER physicians did their very best trying to help Kristen, she was not responding. They had no idea what was causing her total system crisis. After talking with the Doctors and with great sorrow, my husband, Dan, and I had them turn off the machines. **That horrendous moment in time changed our lives forever.**

My family lived for 1 year with the nebulous 'diagnosis' of Reye's Syndrome as the cause of our daughter's sudden death before we finally discovered it was MCAD, but not until after our 2nd child, Kevin, was born and tested positive for MCAD. **Fortunately, the Medical Examiner in Illinois saved some of Kristen's liver tissue** and the frozen specimen was tested by Dr. Charles Roe at the Duke University Medical Center (presently at Baylor University Medical Center in Dallas). She was the first documented post-mortem MCAD diagnosis in this country. **It was definitely not an 'honor' that we wanted, but at least we finally knew why she died.**

If we hadn't read about MCAD while I was pregnant with Kevin (my sister's pediatrician sent us a January 1986 *Journal of Pediatrics* article by Dr. Roe and colleagues) and insisted on testing him at birth (June 12, 1986) and discovered his metabolic disorder, he too most likely would have died or experienced severe complications at 6 months of age when he had his first illness. **Because we KNEW he had MCAD, we KNEW what to do in an emergency. We weren't given that chance with our daughter!**

As you can see from Kevin's picture, he is now a healthy and strong 16-year-old young man enjoying life as an active teenager, along with his younger brother, Brian, who is an MCAD carrier and not directly affected. Kevin is at this point in his life **ONLY** because he was **TESTED AT BIRTH instead of AFTER a CRISIS or DEATH!**

Unfortunately my family's experience is not unique. As shown in the **attached FOD Cost Benefit Analysis Survey Results***, you can **SEE how being AWARE and EDUCATED about this expanded testing would definitely SAVE CHILDRENS' LIVES and much heartache.** This information is specific to our Group's disorders, but I am sure there are MANY other metabolic Support Groups across this country that unfortunately could replicate our results.

In 1999, I surveyed our Families to determine the **Costs of having an infant diagnosed at birth versus those not diagnosed at all or diagnosed months to years after one or several critical metabolic episodes** resulting in severe medical, cognitive, physical and emotional complications or death.

Specifically note the 2nd MCAD Family (case 41) listed on the Survey's Complication page ~ **it took 5 years before a diagnosis was made, but not until after accumulating over \$1 million in medical costs and SEVERAL MAJOR MEDICAL COMPLICATIONS!** The same is true for the LCAD Family (case 5) that waited over 2 years for a diagnosis and had **\$800,000 in medical costs and residual complications.** Note also **ALL the DEATHS on that page due to a LATE or MISDIAGNOSIS** (MCAD case 29, 30, 25, 46, and 31; other FODs case 63, 8,7, 60, and 58).

Compare those cases with MCAD case 34 or the GA2 case 1 for example ~ **BOTH infants were SCREENED and DIAGNOSED AT BIRTH with NO complications!** Today they are thriving because of that early diagnosis.

Our Survey Results definitely showed that **EARLY DIAGNOSIS and TREATMENT at BIRTH saved lives, as well as prevented debilitating COSTLY complications and death.**

My family, as well as over 100 others presently in our Group, are living the ramifications of NOT having their detectable metabolic disorder diagnosed at birth, **experiencing 1, 2 and even 3 deaths BEFORE a diagnosis was ever made.** Several of those deaths were MISdiagnosed as SIDS or Reye's Syndrome leaving families to grieve the unknown and **literally putting their other children and future children at great risk.**

My point is that **these deaths** (and major residual medical complications) **and deaths that are presently occurring** because many states have not implemented this VITAL testing and/or because families and professionals are unaware of the expanded screening tests, **could have been and CAN BE PREVENTED!** **No Family should have to endure such a tragic loss.** The technology is already in place and continues to be further developed (i.e. at Baylor, NeoGen Screening, and Mayo), so **30+ metabolic disorders do not have to go UNDETECTED and UNTREATED!**

I am **URGING** you to **support EDUCATING Families and Professionals** across this country to make them **AWARE of the availability of these expanded newborn screening tests** so that **ALL** infants in the United States will have that chance to live. **EVERY family DESERVES that!**

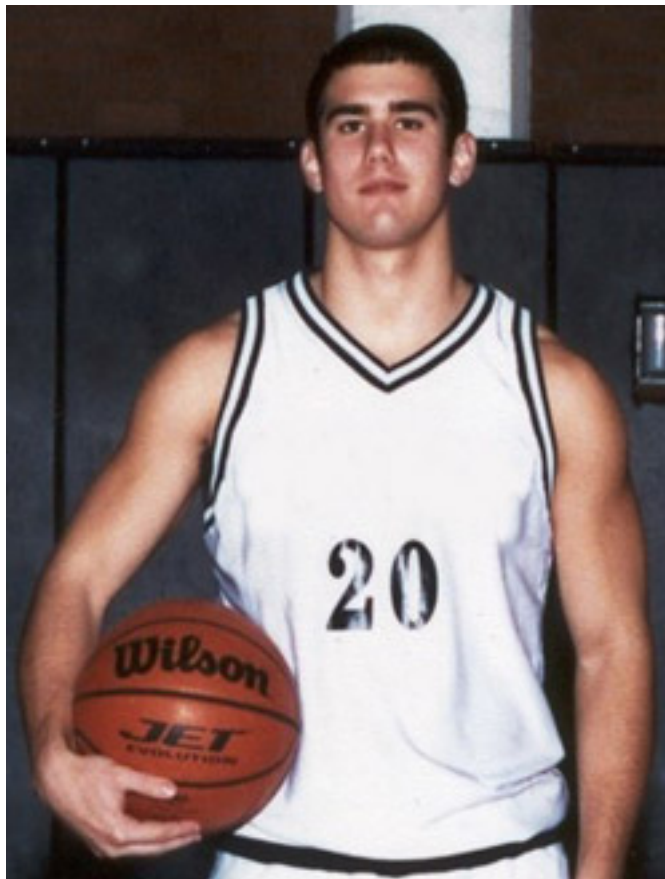
Respectfully,

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* Detailed Survey Results ~ on our site's NBS page



Kristen Marie Gould
•••
Born October 6, 1983 ~ Died July 21, 1985
Undiagnosed MCAD
(originally misdiagnosed as Reye's Syndrome)



Kevin Robert Gould
•••
Born June 12, 1986
MCAD ~ Diagnosed at Birth