

Cristal

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHO LOST THEIR CHILD TO
VERY-LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (VLCADD)

When our daughter, Cristal Marie Gara, 9 ½ months old, underwent surgery for cleft palate on Thursday, December 5, 2002, no one knew that a metabolic disorder was lurking. Cristal came through the surgery really well, impressing the medical staff. Unaware of the few hours left in our daughter's life, we took her home.

On the day after surgery, Friday, December 6, Cristal was running around in her walker. On Saturday, December 7, she began her day with juice and continued to accept more juice, but she refused baby food and formula. By that afternoon she began to look extremely sleepy, resembling a rag doll, no neck control, almost as if she were back in her infant state. Thinking fresh air might help, we dressed Cristal in her snow suit, and suddenly she stopped breathing. Her father immediately picked her up, and breathing resumed.



Hurriedly reaching the surgeon by phone and relating the symptoms, we were told that our daughter had recovered extremely well from the surgery and that she was tired due to the anesthesia. He advised us to let Cristal rest, but if we felt it necessary, we should not hesitate to take her to the hospital and to be sure to carry his card with us.

Through the night, at 1:00 A.M. on Sunday, December 8, Cristal passed away at home in my arms. Autopsy results revealed that Cristal had Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency, a condition in which the body cannot oxidize fatty acids because of a faulty or missing enzyme. This disease is treatable with diet and medication.

The regret is difficult: regret over not taking our child to the hospital, for trusting that the surgeon's observations were correct. I am not angry at any of the doctors involved in our daughter's care, but I am appalled that no blood work was performed prior to surgery. I know that the cleft palate had nothing to do with the VLCAD, but since she already had one genetic disorder, they should have tested her for additional disorders.

VLCAD Deficiency was added to New Jersey's newborn screening panel a few months after our little girl was born. Cristal Marie's first birthday was February 23, 2003. She never, ever, got to open a present, not her Christmas gifts, nor was she alive to celebrate her first birthday. I have decided to take it upon myself to do whatever it takes to make sure all infants/parents are entitled to newborn screening so other parents do not have to feel the pain or guilt that I feel. If this disease is easily treated, the death should not have occurred, unless God did, indeed, need another Angel.

Sincerely,

Debra Gara

Written February 2003 by Debra Gara

Mother of Cristal (VLCAD) February 23, 2002 - December 8, 2002