

Statement of Elizabeth Florian and Lee Florian RN  
Senate Bill 465, An Act Requiring Newborn Screening For Adrenoleukodystrophy  
February 27, 2013  
Senator Gerratana, Representative Johnson, and Public Health Committee Members

The youngest of our four children, Joshua Seth Florian, was a funny, charming and dashing handsome blonde boy with big blue eyes. My husband Lee and I always said he would be a politician someday because he loved to smile and wave at everyone he passed like he was riding on a float in a small town parade. He had a distinctive laugh, for someone so tiny; hearty and deep like a pirate. On June 26, 2008 Joshua came down with a fever, like any normal toddler. He woke up at dawn and asked for a drink, as he did on many mornings. My husband sat him on his hip, and handed him a cup. Joshua handed the cup back and then suddenly fell back in his father's arms and stopped breathing. The trauma of that day is hard to talk about, but that morning we and our three other children, Emma, Ben, and Adam, witnessed the heroic efforts to save their brother's life. Just hours after Joshua took his last drink of water, our children kissed their brother goodbye and Joshua was taken off life support, he died in our arms. He was 2 years 2 months and 2 days old.

Joshua's life could not be saved that day because none of the doctors knew what was wrong with him. Had Joshua been diagnosed, he could have been given a simple, inexpensive medication to treat his symptoms. When we left the hospital that day, we were told that because there was no cause of death, this was now a police investigation and could take up to 10 months. But that investigation never happened because the next morning, the medical examiner told us it was Addison's Adrenal insufficiency of an unknown origin.

Our grief took us on a marathon search for the origin of this disease. We wanted reassurance that our other children would not suddenly die one morning. After 4 years of intense, personal scientific research, countless dollars in medical tests, and meeting with many empathetic doctors, we found Doctor Raymond who kindly agreed to test the last remaining drops of our son's blood as one last reassurance to us. Last Christmas, we found out our son Joshua had a very rare presentation of Addison's adrenal insufficiency caused by Adrenoleukodystrophy, or ALD. According to Dr. Raymond the world expert on ALD, Newborn Screening would have given the early diagnosis needed to save Joshua's life. Had this testing been implemented when our son was born 6 years ago, Joshua could have been here in this meeting himself. With Joshua's natural confidence, he would have looked you in the eye and asked you directly if you would please allow the State of CT to do Newborn screening for ALD. But Joshua will never get that chance and his short life will have to serve as his testimony to the importance of testing for this disease. And we, his family, now must live with the painful fact that our son could have been protected by the knowledge that comes from a simple newborn screening test that costs \$1.50. Young as he was, Joshua had the personality of a leader. Now our only hope is that you can feel his story leading you to do all you can to pass Bill 465 and protect Connecticut's children. Our family is urging you to act so that what happened to our son will not happen to another child in the state of CT.