

# Andrew's 10 year Journey with a UCD

Andrew is a very active and spirited ten year old boy—his passions are very diverse. From church, school, and reading to playing the drums, singing, and playing baseball, Andrew is always on the go. He reads at a second grade level and loves to ride his bike, go to the water park, and hauls logs to burn in his family's wood stove during the winter.

He was diagnosed with Neonatal Onset Citrullinemia, one of several UCD's. Andrew's genetic code does not have the "recipe" to make a specific enzyme which is needed to digest protein. Without special Ammonia scavenging medications and specialized metabolic formula, the level of ammonia in Andrew's blood could escalate out of control, causing symptoms from tiredness and vomiting, to coma and even death from swelling of the brain. Below is a timeline which documents important milestones in Andrew's medical and personal life.

- 3/23/2000- Andrew born (10 days late)
- 3/25/2000- Andrew was discharged at 48 hours old (had been vomiting)
- 3/26/2000- Andrew was readmitted to the ER at 60 hours old due to poor feeding, hypertonia, lethargy & unresponsiveness.  
-At 68 hours old Andrew had a plasma ammonia level of 1300 and was diagnosed with a Urea Cycle Disorder. He was admitted to Children's Memorial Hospital where he was given the specific diagnosis of Citrullinemia.
- 4/2000- After being treated with Buphenyl (Sodium Phenylbuterate) and Arginine and put on a low protein diet including essential Amino Acid formula, Andrew was released to home utilizing these treatments.
- 1<sup>st</sup> year- Andrew grew slowly (hugging the 5<sup>th</sup> % for height and weight) but his physical development was fairly normal...crawling at 9months and walking by 16months.
- 2<sup>nd</sup> year- Andrew hung on to the low end of the growth curve but taking his formula and meds by mouth was a constant struggle.  
  
-Andrew's oral development progressed so that he learned to eat normal table foods. Although progress was a bit slower than a normal child, he learned to bite, chew, and swallow normally.  
-Andrew showed developmental delays in expressive speech and language (receptive language skills were much more normal). Any activity requiring the use of executive function skills was difficult. Andrew made many sounds but spoke no words (not even "mama").
- 3<sup>rd</sup> year- Getting Andrew to consume all of his metabolic formula and meds became so difficult that we finally decided to have a G-tube placed. We were glad we waited until Andrew learned to eat table foods but it was definitely time! What a huge relief. Andrew could finally get 100% of his meds and formula every day. Also, when Andrew got sick, we could give him additional hydration and nutrition very easily. The G-tube also allowed us to treat mild hyperammonemic episodes at home.
- 4-7<sup>th</sup> year- Andrew was fairly healthy without any major hospitalizations until he turned 7 years old. During this time, however, he struggled developmentally. With hard work and persistence, his skills improved steadily. Behaviorally, Andrew also struggled, but a diagnosis of ADHD shed much

light on these problems. It helped us to discipline him more effectively. To this day, he still struggles with listening and following through with directions.

- 8-10<sup>th</sup> year- Andrew was hospitalized over a dozen times in 2 years—requiring dialysis five times since his initial presentation as an infant. The hyperammonemic episodes were gradually becoming MUCH more frequent and they were occurring with no other intercurrent illnesses (as they had in the past). It was only by the grace of God that we were getting him treatment in time.
- 4/2010- Andrew had a severe hyperammonemic episode and afterwards we sadly realized that he lost some of the math skills that he had worked so hard to acquire...that was it! We realized very vividly that a liver transplant was our only option to save Andrew's life and his quality of life.
- 5/10/10- After a liver transplant evaluation, Andrew was approved by the Transplant Team at Children's Memorial Hospital and officially put on the National Liver Transplant List. Because of his genetic disorder, he was given a PELD score of 30 out of 40, with 40 being the most in need of a transplant. Also, if he wasn't transplanted within 30 days of listing, his status would move up to a level 1B (Above 40 and just below emergency level-1A).
- 6/10/10- Andrew's status moved to a level 1B.
- 6/11/10- Andrew received an offer for a liver.
- 6/12/10- Andrew's new birthday- he received his new liver!
- 7/31/2010- Andrew is 7 weeks from his transplant, is home and is back to his normal physical self! He is eating foods he never tried in his entire life (eggs, hotdogs, yogurt, peanut butter...), he is riding his bike and he is even swinging across the monkey bars! THANKS BE TO GOD!

Along our ten year journey, we have had so much professional support...the NUCDF was a major factor in helping us make well informed decisions along the way...We attended the national conferences almost every year.

From the beginning of its inception, we enrolled in the longitudinal rare disease study for UCDs funded by the US Federal Government. Preliminary feedback from this UCDC study was very helpful. Many thanks to the other UCD patients who participated, to the private donors who gave additional financial support to the study and also to the professionals at Cleveland's Rainbow Babies Children's Hospital who followed Andrew over the past five years. We strongly recommend participation in research studies like these to ALL metabolic patients and their families...this is the KEY to understanding these disorders and in making progress for treatments of these disorders!

Last but not least was the **OUTSTANDING** care and love provided to our family by Dr. Joel Charrow, Dr. Barbara Burton, Kathryn Kim (genetic counselor), Heather (dietician), and the entire genetics team at Children's Memorial Hospital in Chicago. They are the blessings given to us by God who helped Andrew not only survive ten years, but helped him to have a good and happy life from his infancy until now without a liver transplant!

Many thanks, also, to the donor and the donor's family for giving Andrew a chance at life with a renewed sense of health and the opportunity for a newfound freedom and independence!

