

Ray Fantel's Story



Ray was born a happy, healthy baby on October 17, 2008. When Ray was 4 months of age, we went to our pediatrician for his well checkup. We knew Ray seemed floppy, especially compared to our older son, Ethan. We assumed he would catch up. At that visit, our pediatrician expressed concern with Ray's low muscle tone and recommended we see a neurologist.

Shortly thereafter, we went to see Dr. Richard Finkel, a neurologist (and long-time Spinal Muscular Atrophy (SMA) researcher and public speaker) at Children's Hospital of Philadelphia (CHOP). We will never forget March 30, 2009 when we received a tentative diagnosis of SMA Type 1. Dr. Finkel said, "I hope it is not SMA, but it seems highly likely." Ray's Dad Googled it while we were in the office and he found that SMA is an incurable, terminal disease. Moreover, SMA is the #1 genetic killer of infants. The average life expectancy of an infant diagnosed with SMA is 2 years of age.

When we heard the diagnosis, we were crushed. I remember Matt holding me while I cried hysterically in the parking garage. We love our boys more than anything. As we digested Ray's diagnosis over the next few months, we vowed to do everything possible for him – to give him the best chance and to make sure he enjoyed life no matter what length of time he has.

Since March 2009, life will never be the same. We have learned to take life one day at a time and try to enjoy every day as best we can. In June 2009, we learned all too well how medically fragile Ray is and how serious respiratory illnesses are for Ray. He was in the ICU at CHOP for 22 days and intubated twice with a cold then pneumonia. It was so scary to see someone so young and innocent breathing via a machine and wondering what would happen next. Since that hospitalization, Ray has had 8 hospitalizations...6 from respiratory illnesses and 2 for surgeries (most recently was major back surgery in August in which hardware was implanted along his spine and his rib cage to treat scoliosis and help him breathe easier).

SMA is a degenerative, neuromuscular disease. As a result, Ray has never crawled or walked; nor can he sit up without assistance. Ray has a feeding tube to

help him get the nutrition he needs. He has lifesaving medical equipment in the house and we are approved for 16 hours of nursing a day. SMA has not affected his spirit nor his happiness.

Ray never says that he cannot stand or walk. In fact, he says he WILL stand and walk someday. We love his attitude and do not tell him he will not be able to walk. No one knows what the future holds. In fact, there are several drugs in clinical trials and one was approved by the FDA in December 2016. The drug, SPINRAZA, is injected into the spine. Ray would need six injections the first year and three injections per year moving forward. We are submitting for insurance approval and are anxious to see if this drug can stop the disease's progression and help Ray.

We are just like other close families.....we love to spend time together! When it is not cold and flu season, we go bowling, swimming and to the movies. Ray goes to Ethan's baseball games and cheers for his big brother. Ethan likes going to Ray's Miracle League baseball games and helps Ray bat. During cold and flu season, we are home all the time and play board games, iPad games and watch DVDs. In many respects we are very blessed; we have learned to be thankful for what we have.

When Ray was diagnosed, I stopped working and became Ray's primary caregiver and advocate. I research any and all new therapies, treatments, and equipment to see if any of them can help Ray. I train every nurse who cares for Ray and am Ray's nurse when a nurse gets sick or we have an open nursing shift.. Now, I am his advocate ensuring he receives the education he is entitled to as well as trying to get insurance approvals for SPINRAZA and additional medical equipment.

Over the years, we have realized that Ray has many "needs" that, although not covered by insurance, would greatly add to the quality of his life (e.g. overhead lift system, handicapped van, repairs to the van's lift, travel to out of state doctor's appointments, iPad, wheelchair lift for access to the house). We are so touched that the Shannon Daley Memorial Fund has selected Ray and our family as a beneficiary. Thank you for your support and assistance.

