a wish for Evan

Evan is 11 years old and he has Sanfilippo Syndrome. He was diagnosed in August 2008, after 4 ½ years of searching for answers as to why he was falling behind, both physically and mentally. When Evan was a baby, he was on time in all his developmental milestones, such as sitting up, crawling, and walking. He was a sickly baby, having constant ear infections, respiratory infections, and seasonal allergies. Evan also had diarrhea frequently. When Evan was 2, we noticed that he did not talk like other 2 year olds talk, and that was the first delayment we noticed. We first thought he was deaf, but his hearing was normal. Also, during this time, potty training was very difficult due to the chronic diarrhea. Evan's gross motor skills as a toddler were fine. He was a very active little boy who loved sports, playing his guitar, and dancing and singing to his Barney or Wiggles songs. He was a very active little boy. As he got older, he babbled like a toddler learning how to talk, but never talked at the level he should have for his age. As the years went on and Evan got older, it became more evident that he was falling behind cognitively. He has difficulty with fine motor skills, but his gross motor skills are good. Evan does not talk anymore. He occasionally "babbles" a little bit, but does not say words anymore. He still has frequent diarrhea and is not potty trained. He occasionally has vomiting episodes. He still walks, but tires very easily and needs frequent rest periods. He is very active as far as constantly climbing on and off furniture, flicking lights switches, opening and shutting doors, clicking locks, etc. He has an attention span of less than 30 seconds and cannot sit still any longer than that. He takes medications daily to help control these symptoms. He also has difficulty sleeping at night and will wonder around the house getting into things if he is not supervised closely. He puts everything into his mouth, constantly chewing on whatever he can. He does not understand danger and therefore must be watched closely. Evan is like having a 4ft 5in, 80 pound toddler. He is very loving most of the time, but he does have aggression tendencies where he hits others frequently. Although raising Evan is challenging, he is such a blessing and teaches us how special life is every day. As Evan gets older, he will continue to regress to infant like quality of life, eventually losing his ability to walk and eat. He will continue to deteriorate mentally as well. Evan will always have to be cared for and will always be our special baby.

About Sanfilippo Syndrome: Sanfilippo syndrome is an inherited disease of metabolism that makes the body unable to properly break down long chains of sugar molecules called glycosaminoglycans (formerly called mucopolysaccharides). The syndrome belongs to a group of diseases called mucopolysaccharidoses (MPS). Specifically, it is known as MPS III.

The syndrome is inherited as an autosomal recessive trait. That means both your parents must pass you the defective gene in order for you to get this disease. Sanfilippo syndrome is possibly the most common forms of MPS. It is seen in about 1 in 70,000 births. A family history of Sanfilippo syndrome increases one's risk for this condition.

Unlike other forms of MPS, symptoms appear after the first year of life. A decline in learning ability typically occurs between ages 2 and 6. The child may have normal growth during the first few years, but final height is below average. Delayed development is followed by deteriorating mental status.

Other symptoms include:

- Behavioral problems
- Coarse facial features
- Diarrhea
- Full lips
- Heavy eyebrows that meet in the middle of the face above the nose
- Sleep difficulties
- Stiff joints that may not extend fully
- Walking problems

The syndrome causes significant neurological symptoms, including severe retardation. IQs may be below 50. Most persons with Sanfilippo syndrome live into their teenage years. Some patients live longer, while others with severe forms die at an earlier age. Symptoms appear most severe in persons with type A Sanfilippo syndrome.

We would really appreciate a donation from you and/or your company to help us with our fundraising efforts to raise money to assist families affected by Sanfilippo Syndrome and also for the research to find a cure. We are having a Spaghetti Supper October 20th. We will also be auctioning items and have a raffle drawing at the dinner. The money raised will go to the MPS Society. Last year we raised $10,000 (which kept the research going for 3 or 4 months .)Thank you so much for your thoughtful contribution.

National MPS Society
Support for Families. Research for a Cure.

For more information about MPS and related diseases, contact the National MPS Society at 919.806.0101. Also log on to its website at www.mpssociety.org.