



Family Helping Others Learn More About SMA (Spinal Muscular Atrophy)

(New York-WABC, September 19, 2002) — For parents, few things are more difficult than watching their child struggle with a serious illness. When a muscle disease threatened their daughter, a frustrated family looked for a treatment and then found a way to help others facing the same crisis. Now, ABC7's On Call with Dr. Jay Adlersberg.

It is a beautiful family portrait that is two-and-a half year old Arya and her parents standing in a park. But Arya has trouble standing. She can't walk without help. On her own, she can't even roll over in bed. Arya has Spinal Muscular Atrophy, a genetic muscle disease that slowly cast a cloud over the first year of her life.

Loren Eng, Arya's Mother: "She was late in hitting her milestones and when we saw that we became alarmed."

But with the opening of the Spinal Muscular Atrophy Center at Columbia Presbyterian, Arya's family is helping to turn the alarm into action. The center will give patients and families access to experts in the disease and its management, and will stimulate research and treatment.

Dr. Daryl De Vivo, Columbia-Presbyterian: "It is a problem that is as promising as any other genetic disease as far as a breakthrough in gene therapy or some variation of gene therapy."

Both patients have to be carriers for the SMA gene to transmit it to a child, and an expectant mother can have a prenatal test for it.

A gene that is defective in SMA fails to make a substance that protects nerve cells in the spinal cord. When the cells die, the muscles in the limbs and chest start to fail.

Some children born with SMA don't live until age two. For others like Arya, the future is uncertain.

Eng: "You envision what it is like on her first day of school and as this disease is unfolding, you see all of those things melting away. Most families with this illness have kids who die before they are two. Our child is at least walking around, and we have to focus on the good things we have."