

November 2016

This is the time of year we surround ourselves with the love of family and friends and give thanks for our many blessings. At the Clinic for Special Children, we give thanks for you; your trust and support allow us to alleviate suffering and change lives.

We are inspired to share children's stories, like that of Sara, a compassionate and vibrant teenager who first started to suffer from motor disability as an infant. Sara's disease progressed rapidly, until she was finally confined to a wheelchair by the age of 6. Thereafter, she suffered daily from severe dystonia, involuntary and forceful muscle contractions that impose painful contortions on the body and interfere with the smooth, intentional movements we otherwise take for granted. Through the help of the Clinic, Sara was diagnosed with a rare genetic condition called tyrosine hydroxylase deficiency, more commonly known as Segawa Syndrome, which is caused by mutations of the *TH* gene. Our doctors used this crucial information to offer Sara a precise, safe, and life changing therapy in the form of L-DOPA, a chemical indispensable to normal motor function of the brain. Slowly her muscles loosened, she regained purposeful movement of her hands, her voice gained power and fluidity and she was able to stand on her own two feet. Now is able to function as a typical teenager, Sara texts, talks and snapchats with her friends. She is gaining strength daily and has set a goal of walking in May!


Segawa Syndrome is one of more than 220 genetic disorders we treat and diagnose at the Clinic, and for most of these conditions, we are able to reduce the burden of disease, prevent disability, or prolong life in ways that transform the lives of children and their loved ones. Our personalized approach to genomic medicine often illuminates understanding about complex diseases found throughout the world, and advances science and patient care one child at a time.

Working together, we can enrich the lives of children like Sara, and your support is vital to this mission! Last year more than 60% of our financial support was raised from charitable sources. Whether you donate your talent, buy a quilt from a benefit auction, send a generous holiday gift, or donate each month, we are grateful for you!

Warmly,

A stylized, handwritten signature in black ink, appearing to read 'Kevin Strauss', enclosed in a thin black rectangular border.

Kevin Strauss, MD  
Medical Director

A handwritten signature in black ink, appearing to read 'Christine Stickler', written in a cursive style.

Christine Stickler, JD  
Development Director