Children with motor neuron disorder treated with gene therapy for first time

Eleven-year-old Hannah Sames's curls were the first thing her parents, Lori and Matt, noticed when she was born. "Their other daughters, Madison, five, and Reagan, two, had stick-straight hair, as do Lori and Matt. When the birthing goop had dried, Hannah's curls were odder still, weirdly dull, like the 'before' photograph in an ad for a hair conditioner," I wrote in my gene therapy book. A more recent story about a little girl with curly hair but straight-haired siblings and parents in the Times of India is remarkably similar.

Hannah has giant axonal neuropathy – GAN. It's like amyotrophic lateral sclerosis (Lou Gehrig's disease) in a child, a gradual failure of motor neurons to stimulate muscles and eventually failure of sensory neurons too. Swollen intermediate filaments (IFs) stuff the axons in what one researcher terms a "logjam." Whole-body effects are slow yet profound, and ultimately overwhelming. I've covered Hannah's story here, beginning with "A Little Girl With Giant Axons, A Deranged Cytoskeleton, and Gene Therapy," and most recently the wonderful news that a gene therapy clinical trial is finally underway.

Thanks largely to the herculean efforts of the Sames family, the first child received an infusion of working gigaxonin genes into her spinal cord at the NIH Clinical Center on May 27. But it isn't Hannah. She can't participate until the researchers figure out a way to dampen a potential immune response. Hannah's two mutations are full deletions, and so if the missing protein suddenly shows up, her immune system could go into overdrive.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis. Read full, original post: Gene therapy clinical trial underway for children's motor neuron disease