Finding hope for children with multiple sulfatase deficiency

Willow [Olson] was born on August 21, 2013. At first she seemed fine, except for difficulty nursing. “Nothing in itself was significant, but a combination of things over time got worse. She crawled a little late. She did walk, but late, at 18 months. She had frequent ear infections, and dry skin that was very bad on her back and her scalp. And she never talked. No words, not even dada. She just made a few sounds,” recalled her mother, Amber Olsen.

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Willow has a gap in both copies of a gene, SUMF1, that normally encodes a protein called sulfatase modifying factor 1, making her one of about 50 people in the world with multiple sulfatase deficiency(MSD).

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In the MPSs, stuff (“storage material”) builds up, drowning and eventually destroying cells in many body parts. In the most severe form, MSD, the corneas cloud, hearing vanishes, ears clog, tonsils and adenoids swell, and the windpipe narrows.

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Eventually, Amber’s networking led to Steve Gray, a gene therapy guru at the University of North Carolina. […] And Dr. Gray has helped. The first mice with MSD were born on September 2 at UNC, and the first gene transfer experiments are slated for November.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: A Genetic Disease With a Domino Effect: Multiple Sulfatase Deficiency