

## A Closer Look at Immunodeficiency

### **Matthew's story**

Matthew's trouble began in February 2006, when the then three-year-old started running a fever of 100 degrees or higher. Initially, he was ill with a bacterial infection, but after several weeks, the fever remained even though the other symptoms subsided. The fevers continued into May, according to Matthew's mom, Lori.

The cause of the fever remained a mystery. Lori contacted a pediatric infectious disease doctor, who ran a new set of blood work. The results were normal except for Matthew's IgG levels (IgG are antibodies that are very important in fighting bacterial and viral infections.), which were low.

The doctor referred Matthew to an immunologist, who diagnosed him with hypogammaglobulinemia, an immune disorder caused by a lack of B-cells, resulting in low level of immunoglobulins (antibodies) in the blood.

There were two treatment options: once monthly immunoglobulin replacement therapy done via an intravenous line at the hospital or a subcutaneous immunoglobulin treatment done weekly at home. Both deliver antibodies into the body.

The family chose the home option. Matthew's home nursing staff and Lori were trained in how to give the treatments, which involve a small needle, the size of a pin, inserted into fatty tissue.

The benefits of the treatment take anywhere from three to six months to notice, Lori says. Over time, Matthew's temperature gauge was back to normal. He is monitored with blood tests and an annual visit to the immunologist.

"I believe if we hadn't figured out that Matthew had this immune disorder, he would have continued to get sick with pneumonia, MRSA or something else, and probably would not be alive today," says Lori.

### **Olivia's story**

Olivia, born in 2004, spent most of the first six months of her life in the hospital with complications, including upper respiratory infections and pneumonia. During the next five years, she suffered occasional ear, sinus and upper respiratory infections, and while the number of infections were no more than those of her siblings, they were more severe, says her mom, Adrienne.



In 2009, Olivia became ill with pseudomonas pneumonia and septic shock. “The intensive care doctors were scratching their heads as to why she would contract this type of bacterial infection,” says Adrienne. “We were told that it was probably a fluke and that it should not happen again.”

Adrienne’s parents were not satisfied with the idea that such a life-threatening experience was a fluke. Olivia’s pulmonologist wasn’t satisfied either and referred the family to an immunologist. Tests showed that, like Matthew, Olivia had hypogammaglobulinemia.

Olivia started immunoglobulin replacement therapy intravenously. Unfortunately, her IgG levels remained too low, even when the dosage of the medication (Privigen) was increased many times.

The family reached out to Dr. Soma Jyonouchi at the Children’s Hospital of Philadelphia, who is researching immune issues in children with CdLS. He recommended trying weekly subcutaneous treatments of Hizentra (the same as Matthew). This past spring, the family was trained how to give the treatment, which lasts about 45 minutes.

“It’s wonderful to treat her at home,” says Adrienne. “She is comfortable in her own setting and is familiar with the process. Last week, Olivia saw me getting out the supplies, gave a resigned sigh, and then climbed up on her bed to get ready for treatment.”

Recently, the family learned that Olivia’s IgG levels are right where they should be.

“While the diagnosis was initially frightening and a bit overwhelming, we are in a place now of confidence and gratitude,” Adrienne says.