

A Closer Look: GI and Volvulus

Nirel's Story

We want to thank Nirel's mother, Gayle, for sharing Nirel's story and experiences with us.

Since Nirel was born, she has had problems with reflux and constipation. Her pediatrician prescribed medications to help with the reflux, but Nirel was not a happy baby for the most part because she was so often in pain or discomfort. Taking what we learned from information provided by the CdLS Foundation, we decided to start seeing a pediatric gastroenterologist when Nirel was a few months old. We thought perhaps a specialist would be able to monitor Nirel's GI issues and help us find the right solutions.

When Nirel was three years old, we read about the risk of malrotation in *Reaching Out*. We expressed our concerns to the GI specialist about doing diagnostic testing. However, his reaction was not supportive. He was quite dismissive about the need to rule out malrotation because he felt it was so rare. In addition, he felt that we could go her whole life without knowing she had malrotation, and no problems would arise.

Coincidentally, the Foundation sent families an information letter written by Dr. Tonie Kline about the typical GI issues in individuals with CdLS. The letter was intended to provide information for health care providers (particularly primary care and GI specialists). The letter explained about the increased incidence of malrotation in CdLS and the dangers of volvulus if left untreated. We showed the letter to Nirel's GI specialist and we had to push hard to get him to comply. He finally agreed to a barium swallow test, a simple non-invasive test that only took a few minutes to do. But he made it clear that he was not expecting any abnormal results. He was patronizing, and made us feel like we were just being pushy parents.

The day after the test, we got a call from the GI specialist. The tone of his voice was very different. He gave us the news that Nirel did indeed have malrotation. It was bad news, but also good news that we had an early diagnosis and could do surgery to prevent volvulus. The surgery was called the "Ladd procedure." The GI specialist told us that surgery was not urgent, but we pushed again and told him that we wanted surgery as soon as possible.

The first surgeon we met with was an experienced general surgeon who had recently moved from a high-profile institution to the children's hospital where we were taking Nirel. He had done many Ladd procedures in the past, and he explained what was involved with the Ladd procedure as an



open surgery.

A few days later, the Foundation put us in touch with another family whose son had a Ladd procedure. The father said that the surgery went fine, but he wished he had opted to do it by laparoscopy because the recovery of open surgery was rough on his son.

So, we called the surgeon the next day to ask if we could do the Ladd procedure via laparoscopy. He admitted that he did not know how to do the laparoscopic surgery, and that he only could perform it by open surgery. We told him we wanted to speak with another surgeon in the department who could do the laparoscopic surgery.

The second surgeon was no-nonsense about the benefits of laparoscopic versus open surgery. He made us feel confident in him and his surgical ability, but most importantly, he listened to our concerns.

The surgery went well. Nirel only spent one night in the hospital, and her recovery was smooth. It is reassuring to know that having Nirel undergo the Ladd procedure prevents the complications associated with malrotation. In addition, it is important to us because Nirel still has abdominal discomfort and pain often, and we are able rule out that it is not due to a complication of the malrotation.

