

OF SPECIAL CONCERN: BOWEL OBSTRUCTIONS IN THE KAT6 POPULATION

Parents of a child with a KAT6 variant are the first to recognize when the one they care for is in distress and needs medical care. People with KAT6 syndromes may quietly tolerate increasing pain until it has become severe and may show no signs of a bowel obstruction until it has progressed to a serious degree. Obstructions can happen again and again and can strike at any age. An obstruction can quickly go from serious to dangerous.



Low motility in the gut means weak contractions of the muscles that mix and propel contents in the gastrointestinal tract. When weak contractions fail to keep intestinal contents moving, this is ileus — not a blockage but a slow-down or stoppage.

If the bowel contents sit too long, they can begin to ferment and decay, with potentially serious results. If it does not eventually start moving on its own, it may respond to non-invasive treatments such as stimulants taken orally or a rectal enema. But if there is a physical barrier to continued movement, the problem can quickly become life-threatening.

During gestation, when a portion of the developing intestinal tract fails to move properly into place in the baby's abdomen, it is known as a malrotation. This is an anatomical defect and one that must be suspected if problems arise after birth. If it causes repeated interference in normal digestion or leads to an obstruction, then it can become a serious problem.

When a loop of intestine and the membrane that holds it in place twist around each other like sausage links, this causes an obstruction called a volvulus. Trapped intestinal material, already partially digested, continues to break down though, and some contents may be ejected as diarrhea or gas, while most of it will remain and swell the gut.

A person suffering a volvulus, who enters emergency surgery soon enough, may still lose part of the intestinal tract in surgery. Without emergency surgery a volvulus is almost certain to be fatal. A growing pocket of gas can be detected on successive x-rays, but not if the physician is treating the patient for a suspected food allergy or diagnosing tantrums due to anxiety.

A volvulus is a rare occurrence in the general population, but among the KAT6 population it seems common enough to be of serious concern.

It appears now that untreated bowel obstructions are the leading cause of death among children affected by KAT6 syndromes.

Communication problems are common with the KAT6 population as is a high tolerance for pain. Children and adults with KAT6 syndromes, especially those who can't tell us that something hurts or where it hurts, need to be monitored continually for lack of gut movement. Doctors need to trust what we are telling them, and so our information must be reliable. By educating ourselves, paying close attention to the signs our child gives us, and making sure we communicate consistently and accurately with medical providers, we can be our child's best advocates.

Many parents have observed GI benefits from a mitochondrial cocktail and supplements, such as Cytra-3. Learn more about these supplements by watching [Dr. Richard Kelley's presentation from our 2022 Conference](#). It is essential to consult your child's physician before starting anything new.