Focusing on Chiari
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The Sosenheimer family (clockwise from front): Hunter, Rick, Markee Rase, Heather, Drake and Kedrick. Photo by Dan Dry
Little-known genetic disorder causes debilitating headaches and other neurological symptoms

Heather Sosenheimer stands at the side of her 7-year-old son’s hospital bed. Kedrick’s long eyelashes are at rest. His left leg is fully extended, his right slightly crooked. A picture of a dog rests at the foot of the bed for when he awakens. For the time being, he’s at peace.

Kedrick is still sedated after an MRI. His mom hopes the test will reveal the cause of his continuing headaches, pain so awful he has to lie down every few hours. She has lived through this scene so often that she can’t recall how many times. She’s seen her four children through at least 11 MRIs during the past two years, as well as 11 surgeries.

Kedrick, his brothers Drake and Hunter, older sister Markee, mom and dad all have a genetic condition called Chiari malformation, a brain protrusion at the base of the skull and the top of the neck that restricts the flow of cerebrospinal fluid.

First documented in the late 19th century, the condition remains largely a mystery. There’s no known cure, just means of relieving symptoms. For the time being, surgeries and shunts are the only methods for giving Chiari sufferers relief.
Markee, whose biological father does not have Chiari, has only an occasional headache. Her Chiari seems to have resolved itself.

Heather, on the other hand, anticipates surgery. Chiari has impacted her thyroid, leading her to put on weight, which makes the headaches worse. Frim tells her that she should lose the weight to relieve the symptoms, but the cycle is a catch-22.

“Chronic pain destroys their lifestyle and quality of life,” Frim said. “Everyone has a story, but you don’t know it unless you listen.”

Take, for example, Lisa Newcomb. Lisa and her sons, some of Frim’s first Chiari patients, have been seeing the pediatric neurosurgeon for almost 10 years. She also has worked with Frim as a volunteer for several years, meeting new patients’ families to explain Chiari, share her story and answer questions. Lisa, now in her early 40s, has suffered from Chiari symptoms her whole life.

Lisa remembers going on routine grocery shopping trips as a young mother, weak and in pain, wondering if the other mothers who were loading bags into their cars felt the same way. She had learned to live with the pain—how to circumvent the worst of headaches by not bending over to pick up a dropped towel or crawling at times to keep from passing out.
Ironically, before Frim, doctors had told Lisa that her health problems were only in her head. Other Chiari patients have had similar experiences: neurologists and neurosurgeons who attributed the symptoms to stress, hormones or psychological problems.

It wasn’t until Lisa’s first son was born that she started to find answers. Early on, she knew Brandon, now 15, wasn’t like other children. He’d pound the back of his head. He’d vomit more often than normal. Once he learned to crawl, he’d climb onto his dresser and pull down the shades. His parents became especially alarmed when he began to have spells in which he’d lose consciousness.

“I told my husband something was wrong with Brandon’s brain,” Lisa said. As Brandon grew older and could communicate, his parents better understood the extent of his symptoms.

In spite of his infirmities, Brandon learned to speak and use the toilet earlier than most boys, but by age four he became incontinent. Nausea spells began again. Pain in his legs kept him from walking. And he suffered from dysarthria, a speech disorder.

“He lived his life in a dark room,” his mother said. “He went from being an articulate kid to having lost all his speech.”

Despite long-known evidence of Chiari, the scientific community is only beginning to understand it. In 1883, Scottish physician John Cleland, chairman of anatomy at Glasgow University, published “Contribution to the study of spina bifida, encephalocele, and anencephalus” in the Journal of Anatomy and Physiology. It’s believed to be the first public observations of the deformity.

In 1891, Austrian pathologist Hans Chiari advanced Cleland’s findings and classified different variations. In cadavers, he noticed four variations, which he categorized as types I, II, III and IV.

The four types involve the cerebellum, but that’s where the similarities end:

• In Chiari malformation Type I, the brain tissue protruding from the bottom of the cerebellum, called “cerebellar tonsils,” extends beyond the structure at the bottom back of the brain (the foramen magnum). This extra brain tissue exerts pressure on the brainstem, which affects speech, swallowing, vomiting and other functions, and often impedes the flow of cerebral spinal fluid, causing a host of other possible symptoms.

• Type II, which always accompanies open spina bifida, consists of tonsils longer than those in Type I and also involves the brainstem, fourth ventricle and the vermis, another part of the cerebellum separate from the tonsils.

• Types III and IV are extremely rare. In Type III, the cerebellum and brainstem descend into the spine and an external sac, creating a fluid-filled cyst. A small or underdeveloped cerebellum characterizes Type IV.

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—David Frim, Section Chief of Pediatric Neurosurgery
In terms of understanding the condition, “we are where Pasteur was when he found out that you could pasteurize milk,” Frim said.

“We know that if you operate, most of the patients get better. We don’t know why,” he said. “We don’t know why some people have the same looking MRIs and don’t have any symptoms at all. We don’t know why some children of parents with Chiari are affected and some aren’t. We don’t know why some people have headaches and some don’t. We don’t know any of those things.”

Researchers are beginning to believe Chiari is genetic. Frim likens the condition to dwarfism. The genes can be passed from generation to generation, but unless it’s a dominant gene, it doesn’t show itself. Duke University has sponsored a study to collect blood samples, but researchers are still looking for the gene.

Here and there, other institutions also are supporting studies. For example, scientists at the University of Wisconsin-Madison use MRIs to observe the flow of fluid out of the base of the skull. At the University of Washington, researchers study fluid flow in the syrinx and spinal cord.

Frim’s research team currently consists of a research nurse, lab technician, nurse psychologist and testing technician. In one study, they examine the cognitive impact of hydrocephalus and shunts. They’ve expanded this study to include Chiari patients.

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The team found that the intelligence levels of children with Chiari fall within the same range as those of the general population. However, Chiari patients more frequently exhibit problems with various executive functions, such as problem-solving or decision-making.

“A lot of the Chiari children seem to have some issues in school,” Frim said. He hopes one day he and his team will be able to suggest learning strategies so educators can tailor lesson plans for Chiari patients.

In a second study, Frim compares ventricular cranial shunts to lumbar peritoneal shunts. Both are used to relieve cerebrospinal fluid pressure. The latter seems to break more often. Frim would like to study many other aspects of Chiari treatment too, but for now, he spends his typical 12-hour day treating patients.

Medical professionals do not know how many people have Chiari. The total could exceed 250,000. “If there are no symptoms, then we don’t take care of them,” he said. “It may be a disease state that is more prevalent than we think.”

Frim is one of a small number of pediatric neurosurgeons who treats Chiari. According to families with Chiari, many doctors have never heard of the condition.

“We tend to see the patients who get ignored for a while,” said Frim, who nicknamed the University of Chicago Medical Center the “Chiari capital of the Midwest.” Some families have “long, torturous stories” of seeing doctor after doctor, Frim said. He added that neurologists and neurosurgeons in private practice tend to shy away from treating it because the process is complicated, time-consuming and expensive.
Those neurosurgeons who understand Chiari tend to accept decompression surgery as the treatment, but they disagree about how to do it. “There’s not a uniform approach,” Frim said.

Neurosurgeons debate the finer points. They disagree on how much bone should be removed in the decompression surgery. And there’s the question of whether to operate at all: At what point do the pain and debilitating symptoms outweigh the risks of surgery?

Then there’s the question of size. How large should the opening be? Should the dura be opened? And finally, the most hotly disputed issue: If the dura is opened, what should be used to patch it? Some physicians use grafts of the patient’s own tissue. Others use synthetic materials. Frim uses a piece of the pericranium—the lining of the patient’s own skull bone, which prevents any sort of reaction to the graft placement.

For some patients, relief from decompression surgery is permanent; for others the procedure provides only temporary relief.

Decompression surgery is an art that Frim has refined. To begin the delicate procedure, Frim and his team position the sedated patient on his or her stomach as if in a massage therapist’s chair. This position moves the brain ever so slightly towards the face, taking pressure off the hind of the head. The doctors shave a patch of hair at the base of the head, mark a line of incision about six centimeters long, and commence. Carefully monitoring all vital signs, they work their way to the skull. Along the way, they remove part of the pericranium, which eventually will regenerate itself, to use as a patch.

Once arriving at the skull, they slowly and deliberately remove fragments of the bone like an archeologist picking dirt from an ancient artifact. Each piece is no larger than a clove. In time, they have an opening in the bone that will allow the extra brain tissue to fit better. Then Frim opens the dura to give the cerebellar tonsils as much room as possible and to allow the best possible fluid flow in and out of the skull. To repair the opening over this extra space, they place the pericranium and assiduously begin to stitch, making it a watertight seal. The patch is then covered with the layers of muscle and tissue that had been opened, until the last layer, the skin, is closed. This patch gives the brain more room.

Chiari patients often have these related conditions:

- **Hydrocephalus** is an excessive buildup of cerebrospinal fluid in the brain. A Chiari malformation can block the normal flow of this fluid, resulting in pressure that can cause mental defects and/or an enlarged or misshapen skull. Left untreated, severe hydrocephalus can be fatal. The disorder can occur with any type of Chiari malformation, but is most commonly associated with Type II.

- **Spina bifida** is the incomplete development of the spinal cord and/or its protective covering. The bones around the spinal cord don’t form properly, leaving part of the cord exposed and resulting in partial or complete paralysis. Patients with Type II Chiari usually have myelomeningocele, a form of spina bifida in which the bones in the back and lower spine don’t form properly and extend out of the back in a sac-like opening.

- **Syringomyelia, or hydromyelia**, is a disorder in which a cerebrospinal fluid-filled tubular cyst, or syrinx, forms within the spinal cord’s central canal. The growing syrinx destroys the center of the spinal cord, resulting in pain, weakness and stiffness in the back, shoulders, arms or legs.

- **Tethered cord syndrome** occurs when the spinal cord attaches itself to the bony spine. This progressive disorder causes abnormal stretching of the spinal cord and can result in permanent damage to the muscles and nerves in the lower body and legs.

- **Spinal curvature** is common among patients with syringomyelia or Chiari Type I. Two types of spinal curvature can occur: scoliosis, a bending of the spine to the left or right; and kyphosis, a forward bending of the spine.

Source: National Institutes of Health
Frim’s technique differs from others in that he uses the pericranium. Frim has found that synthetic matter can cause damage from a “foreign body reaction.” The body sometimes rejects synthetic patches and those harvested from other species, such as pigs. More than one adult patient has come to him seeking relief after another neurosurgeon operated. He has had to tell some of these patients that he can’t do anything for them.

For some patients, decompression surgery provides the relief they need for a normal life. Unfortunately, others need more help. In some children, the tonsils at the back of the skull scar together and cause fluid flow restriction or brainstem pressure. These children might need repeated brain surgeries as the years pass to reconstruct additional space and restore flow.

Another complication is that Chiari frequently is associated with syringomyelia, a fluid-filled pocket that forms in the spine. This type of fluid collection is usually well treated by the decompression surgery. However, in a small number of patients, the brain becomes unable to absorb all the spinal fluid and the fluid pressure becomes too high, causing additional symptoms. This is called Chiari pseudotumor syndrome. Frim sometimes recommends a lumbar peritoneal shunt to remove the excess fluid. This shunt, which he places in the middle of the back, drains fluid from the spinal cord into another part of the body where it is absorbed. The shunt has a valve to help maintain the correct amount of cerebro-spinal fluid pressure, which helps relieve the symptoms.

The Sosenheimer boys have met with Frim several times to insert and adjust their shunts. One of the boys’ shunts coiled inside him after he toppled from a four-wheeler. Frim temporarily has banned them from playing on such vehicles.

Leading a normal childhood is difficult with Chiari. Lisa Newcomb knows how they feel: ‘A miracle for me is if Noah [her 10-year-old son with Chiari] can get out of bed, get dressed and be up for an hour,’ she said.

Parents of children with Chiari know that plans can change in the blink of an eye. Before driving to Chicago for Kedrick’s latest MRI, Heather had to let him lie down for 45 minutes. “Every day is so different,” she said.

Families with Chiari use the Internet to educate and support one another. “That’s what has really saved me—the connections with the other families,” Heather said. “Trying to help raise money for research, knowing we might not reap the benefits but knowing it will help other families who will have to go through it. That’s been my main, number one coping mechanism.”

Frim suggests three prescriptions to raise awareness of the condition: One, create an organization that provides information and advocates for patients along the lines of the American Cancer Society. Two, invest in an infrastructure that supports Chiari education for doctors and nurses. Three, fund a research organization with dedicated resources.
Some children are lucky. Their symptoms abate, sometimes even go away, when they stop growing.

The physician met the Sosenheimers when Kedrick was in intensive care after his first surgery. “Kedrick had to lie flat for a long time,” Kahana said. “He was a little sad, so we were looking for ways to entertain him.”

For children, distraction is as effective an analgesic as narcotics, she said. “You have to figure out who they are—that little piece of them that can be enchanted for the moment.”

Kahana arranged for Kedrick to make bracelets for Baylie for Brains, a grassroots fund-raising effort started by the mother of another Chiari patient. (See story at right.) Kahana hung a sign on the door: “Don’t come in without $5.” The sign intrigued. Many people visited.

“He forgot about being sad,” Kahana said. “It’s okay not to fix them. They forgive you for that, but you need to listen to them.”

Some children are lucky. Their symptoms abate, sometimes even go away, when they stop growing. In November, Brandon and Noah Newcomb underwent what should be their last Chiari operations to remove their shunts. Their mother—who credits a rigorous program of nutritional supplements—said that now, after numerous surgeries over 10 years, at ages 15 and 10 they appear to be fully recovered. Drake and Hunter Sosenheimer also have found relief. Now they wish the same for Kedrick.