

Desperate for Answers

South Carolina teen finds them with DCH neurosurgeon

AT THE AGE OF 17, MACKENZIE MATHIS OF SPARTANBURG, S.C., IS A JUNIOR IN HIGH SCHOOL, AT THE TOP OF HER CLASS AND LOOKING FORWARD TO COLLEGE WHERE SHE WILL CONTINUE HER STUDIES TO BECOME A DANCER. WITHIN MONTHS, WHAT BEGAN AS A PERSISTENT HEADACHE SOON HAS HER UNABLE TO WALK AND HER DOCTORS MYSTIFIED.

- My Journey to Walk Again -

September 2007

I have a headache that won't stop. I have never had a headache like it. My doctor thinks it is because of stress. Nothing is helping the pain, so I am learning to work through it and keep going.

October 2007

A month passes, but the headache remains. My parents take me to an ENT and a neurologist. Numerous tests including a CT scan are performed all with the same result - normal. Why is this happening to me? I find strength in my faith.

January 2008

The headache is worse, never letting up, but I continue to dance.

Dancing has been my passion since I started in second grade. My joints are loose and allow me to be very flexible when I dance. When I am down, or stressed, dancing is a release for me and something I love - despite the headache.

March 2008

More months, more tests, no answers. I was at school when I suddenly feel like someone has hit me. My headache instantly gets much worse and I collapse into my friend's arms. I am taken to the emergency room. They give me a double dose of morphine that doesn't help. I am sent home with the headache, dizzy and having problems concentrating. We get the MRI results - fully prepared to hear that I have a brain tumor - but the MRI is clear - nothing is wrong.

April 2008

I have now made countless trips to neurologists. No one knows what is wrong. One doctor gives me pain killers that make me feel sick. I can't take them anymore. I think I am getting depressed. Why doesn't anyone know what is wrong with me?

June 2008

Summer comes. The dance continues. I am dancing 3 to 5 days each week, but some days, my headache is too bad and I can't. I have started a summer job working as a nanny. On and off, I am noticing strange tingling and numbing sensations from my forearms to my fingertips probably from picking up the baby.

August 2008

Back to school
My senior year starts and I begin sending in college applications. I am still dancing but feeling worse. The numbness I felt over the summer in my arms returns and becomes constant. There is a constant pressure in my head - like it could explode.

September 2008

Writing is becoming difficult. My current pediatric neurologist still can't tell me anything after more tests. He is starting to think it is fibromyalgia but tests don't confirm it. We keep researching non-stop on the Internet looking for the answer. My blood tests come back normal but I feel terrible. I am starting to drift further behind at school.

October 25, 2008

I try to get out of bed but my feet feel tight, tingly and numb. My worst fear - that my legs would feel numb like my arms - has happened. Tonight, I go to my sister's play. I walked in but I couldn't walk out to the car. My friend has to carry me. My legs are strong from dancing, but tonight they feel so weak. I know in my heart that this is something big and it is not going away soon.

Overnight I went from dancing hours each week to not being able to walk. It is a slap in the face.

October 27, 2008

It is a week before my 18th birthday and my mom is trying to find me a new neurologist. I am walking so slowly and painfully.



November 3, 2008

Happy 18th birthday

My 18th birthday is spent in bed. It is too hard to get out. I have an appointment with a new neurologist tomorrow. I am hoping and praying he has answers.

November 4, 2008

The new doctor listens to my symptoms and has no new answers. The doctor takes my mother into the hall and tells her that I am faking my condition to get attention. He gives me a prescription for anti-depressants. Now, I am livid! Why would I fake this and give up everything I love and my future?

November 11, 2008

My mom calls a doctor from my church who agrees to review my files. He thinks I might have a connective tissue disorder - Ehlers-Danlos syndrome and recommends we see a new neurologist.

Late November 2008

The puzzle is starting to come together. The new neurologist wants to run more blood tests that sometimes can show abnormalities related to a connective tissue disorder. New blood tests come back showing an abnormality and I am diagnosed with Ehlers-Danlos syndrome. My connective tissues do not function as they should and cannot hold my skeleton in place properly. This affects all parts of my body, including my head and neck. The neurologist thinks I may also have a Chiari Malformation because of this condition.

We write the Chiari Institute in New York and they call the next day to schedule our appointment 2 weeks later. I am so excited I won't have to wait much longer to finally get answers!

Journal of Mackenzie Mathis

December 15, 2008

I am very excited to go to NYC! I have never been there before and never even been on a plane! My first appointment is tomorrow, and I am getting a lot of tests - MRIs, CTs, etc. The next day I have appointments with the neurologist and neurosurgeon.

December 19, 2008

Finally a diagnosis.

The doctors confirmed that I have craniocervical instability. I am a bobble head! My head is settling down on my neck pressing on my cerebellum and brain stem causing the problems I am experiencing.

The doctors feel I also have a tethered spinal cord (another diagnosis common to people who have connective tissue disease). The spinal cord is connected to the bottom of the spine by a cord of connective tissue, if this is not functioning properly, the spinal cord can be pulled downward, causing neurological symptoms and pain. The good news is surgery can be performed to correct both of these issues.

The doctor speaks to a geneticist in Baltimore who is an expert in connective tissue disorders. They are going to try medicine before making a decision about further treatment. I am scheduled to go to Baltimore in February to see the geneticist.

January 3, 2009

Christmas has come and gone. My joints still seem to be getting looser and the doctors don't know why. All of the joints in my fingers are dislocating more often, which is painful, so I am getting silver ring splints. I am also having more difficulty walking and climbing stairs because my ankles, knees, and hips are constantly sliding out of place. I have to walk slower and concentrate on trying to hold them in joint as much as possible.

January 5, 2009

I have to go get my neck brace adjusted. I hate wearing it right now, because it pushes my jaw out of joint, which makes me lightheaded and causes my face to go numb. I am scheduled for a tilt table test - which I have heard will make me feel sick.



January 11, 2009

The tilt table results are in and I am diagnosed with postural orthostatic tachycardia syndrome or POTS for short. When I go from laying down to sitting to standing my blood pressure drops dramatically and my body compensates by racing my heart. This is due to Ehlers-Danlos. I am going to the cardiologist Monday. Hopefully he can figure out some medicine to keep it under control.

January 21, 2009

The cardiologist puts me on a medicine that will increase my blood volume to help my heart maintain my blood pressure without having to beat so fast.

February 10, 2009

We are in Baltimore! The geneticist says that I am one of the most hyper-mobile people she has ever seen. The doctor confirms the diagnoses of cranial instability and tethered spinal cord that were made in NY. She is going to give them the OK for me to have the tethered cord release surgery. She feels many of my symptoms will diminish or disappear with this procedure. She also feels I will eventually need neck fusion surgery to stabilize my head/neck. The surgery is scheduled for April 21 at the Chiari Institute.

March 7, 2009

I can't stop thinking about the surgery...I am still not sure what I think about it...I don't want to do it, but I know I have to!

March 30, 2009

My pink chair
I finally got my own wheelchair! I was strangely excited to get it! I have come a long way from October, when I would not even say the word wheelchair, to now, when I was actually excited to get my own! I guess it helps that it is PINK! I must say, it is pretty cool, or at least as cool as a wheelchair can get!



April 19, 2009

I am in NY for surgery at the Chiari Institute. I need to get an upright MRI before surgery that will show the effect of gravity and what my brain and some of my spine look like when I am upright. The tech gets frustrated that I am moving during the MRI. I didn't think I was moving, but maybe I was breathing too much. So he starts again and I try to breathe less.

My mom and I noticed how low I had settled down and mention it to him. He said he noticed the same thing in the films and was coming to raise the seat. He had to raise the seat 6 INCHES to get me back to where I had started!!! In 30 minutes I shrunk 6 inches!!! He let me look at my films. We were shocked to see my spine completely curved even though I was sitting STRAIGHT!

My odontoid bone was sticking into my brainstem REALLY far! My esophagus was extremely bent and almost completely closed! My neck was SO compressed! My Chiari was worse! And my spinal cord was too posterior in some places and too anterior in others! This MRI looked NOTHING like the supine MRI I had in December! If I hadn't been present at both, I would have thought they were done on two completely different people!!!

April 20, 2009

We got the call 12 hours before my surgery - it was cancelled. We were told the surgeon had a family emergency so we headed home. The Chiari Institute would call to reschedule.

May 28, 2009

The Chiari Institute still hasn't called. I haven't had a full day of school since December so I won't graduate. I will have to go back in August and repeat my senior year.

I am going to have an EEG to check for seizure activity. Our neurologist here in S.C. feels I am having what are called absence seizures, which are episodes where I stop and stare. It is as if I just "check out" for a few seconds at a time.

June 9, 2009

After almost two months of waiting to hear something definite from the doctors in NY, we felt it was time to investigate other options. My symptoms are not improving, and we know from the experience of others with this condition that there can be lasting effects if I don't get help quickly.

Our geneticist in Baltimore suggested I see a neurosurgeon she works closely with - Dr. Fraser Henderson at Doctors Community Hospital in Lanham, Md. We sent all of my records and MRIs to him last week. He called my mom on Saturday, and we see him Monday, June 15.

June 17, 2009

We flew up to Maryland to meet Dr. Henderson. I love him. I told him what is really important to me is that I get back in school on time so I can go to college and he said he wants that too. Dr. Henderson spent lots of time with us talking about our options. We decide to have the tethered cord release surgery in a week.

June 26, 2009

First surgery
I rolled into the hospital in my snazzy pink wheelchair ready to go. Everyone was great. After the 3 1/2 hour surgery to release the pressure on my spinal cord, I woke up. The nurses were wonderful.

I was in the hospital for 5 days. As I started to recover, we realized that although some of my symptoms were gone, my headache remained. The tethered cord has been pulled tight so long it was loose now - it had been stretched. I would need the spinal fusion surgery too to stabilize my head.

July 6, 2009

We stay in a hotel as I try to gather my strength. We talk to Dr. Henderson every day. I am still feeling dizzy and my headache is constant. Dr. Henderson schedules spinal fusion surgery for July 10.

July 10, 2009

My surgery lasts just over 6 hours. During surgery Dr. Henderson creates more room at the base of my skull for my brain stem and cerebellum. My Chiari malformation has pushed the bottom of my cerebellum down into my spinal column. Dr. Henderson cuts out a piece of my skull measuring 2x4 cm. To stabilize my head in relation to my neck, he fuses my head to my neck bones down to the third cervical vertebra using plates, screws, and rods as well as bone from two of my ribs right below my left shoulder blade.

I wake up from the surgery without a headache for the first time in 2 years.

July 14, 2009

I leave the hospital and walk without any pain. In two days we head home to South Carolina.

August 3, 2009

Life is good.
I still have no headache. I am walking and ready to sell my wheelchair and throw away the handicapped tags! My heart rate returned to a normal rate and rhythm after the first surgery. My joints seem much stronger, and the aching and tingling has subsided. I am able to think more clearly. I just finished reading a novel!

August 18, 2009

I started my second senior year. Although I wanted to go to school for dance, now I want to raise awareness and work with people like me.

September 2009

I am doing something I didn't think I would do again.
I can dance.



Fraser Henderson, Sr., M.D.

Patient: Mackenzie Mathis
DOB: 11/03/90

Patient Background:

18-year-old female from South Carolina presents with daily, severe debilitating headaches; dizziness that remands her to bed; poor eye coordination; pain when moving her eyes to read; intolerance to sound; difficulty swallowing; trouble finding words; difficulty breathing at night; insomnia; abdominal problems; and weakness, pain and numbness in arms and legs.

Diagnosis:

Ehlers-Danlos Syndrome (EDS)

A collection of genetic disorders that affect collagen, a protein that adds strength and elasticity to connective tissue including skin, muscles and ligaments. Patient's EDS resulted in weaker-than-normal ligaments, extremely loose joints and a downward pull on the spinal cord—resulting in a chiari malformation.

Chiari Malformation

A structural defect in the part of the brain that controls balance (cerebellum). This defect pushes the cerebellum and brainstem downward, creating pressure that can cause a range of symptoms including dizziness, muscle weakness, numbness, vision problems, headache, and problems with balance and coordination.

Treatment:

This patient required two surgeries. The first involved untethering the spine—removing some of the bone at the base of the spine and then, using a surgical microscope, identifying and removing portions of ligament that was pulling the spinal cord down.

The second surgery, a cranial spinal surgery, normalized the relationship between the skull base and the upper spine in order to alleviate the brain stem deformation. This procedure required the use of small screws and rods as well as a fragment of rib and then placing these in the skull and upper vertebrae to provide long-term stability and fusion.

Overall Prognosis Following Surgery:

Majority of patient's symptoms are much improved—providing alleviation of pain as well as increased mobility, endurance and strength. Patient is able to undertake activities that she previously was prohibited or limited in doing due to her condition.

DOCTORS COMMUNITY HOSPITAL

Symptoms and Progress to Date:

Constant, daily headache
Progress: Resolved.

Dizziness
Progress: Improved.

Gait Disturbance/ Inability to walk long distances without a wheelchair
Progress: Patient's legs are much stronger. Patient can walk and rise up on her toes and stand on one foot.

Fatigue
Progress: Improving steadily.

Neck pain
Progress: Only post-surgical pain reported.

Hypermobility in small and large joints
Progress: Much improved. Patient's shoulders are not sliding in and out of joint.

Pain
Progress: Joint pain is much improved. Back pain is much improved, and patient can walk inclines normally and pain-free.

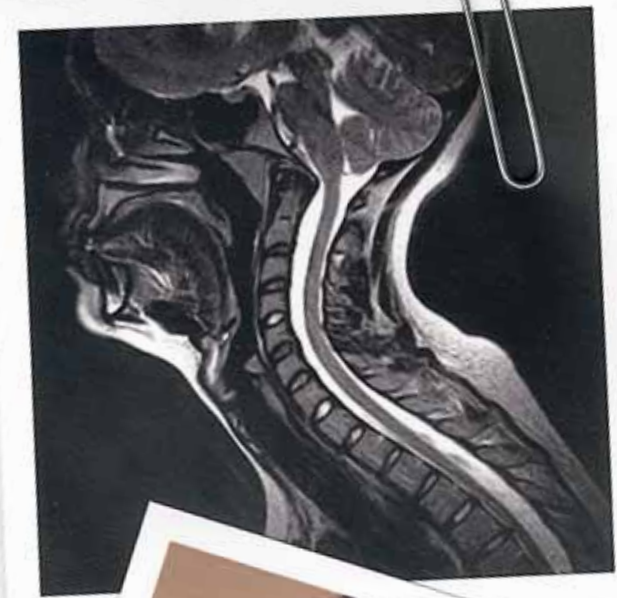
Foot pain/ numbness
(inability to wear closed shoes)
Progress: Patient remarks that she has more feeling in feet and can wear closed-toe shoes.

Arm/hand pain, weakness and numbness
Progress: Much improved. Now patient is able to pick up a gallon-size pitcher of liquid and pour it out. Overall weakness also is much improved.

Involuntary eye movement
Progress: No pain when shifting eyes from left to right or up and down following surgery.

Muscle spasms/twitching
Progress: Improved.

Difficulty climbing stairs
Progress: Much improved.



MATHIS, MACKENZIE

Trouble finding words
Progress: Steadily improving.

Inability to stay warm/sensitivity to temperature changes
Progress: Patient seems to be adjusting much better and notes that feet are actually warm at times.

Difficulty swallowing pills and food
Progress: Improved.

Dr. Fraser Henderson is a board-certified neurosurgeon on staff at Doctors Community Hospital.