Key Points

1. Prior to MRI's Chiari was not widely studied
2. In 1999, Dr. Milhorat published a landmark study on Chiari and syringomyelia
3. Study entailed 364 people and looked at symptoms, MRI findings, and family history
4. Found that over half of the patients had been told by a doctor that they suffered from a mental problem
5. Trauma was a precipitating factor for 24% of the participants
6. Headaches, eye disturbances, vestibular problems, brain stem/cerebellar problems, and abnormal spinal cord function were common symptoms
7. MRI's revealed that the most common finding was a lack of space behind the cerebellar tonsils
8. MRI also revealed that Chiari patients had on average a smaller posterior fossa volume
9. Length of herniation - once the definition of Chiari - did not correlate with measured disability or presence of SM
10. 21 families had two or more close relatives with CM/SM

Definitions

autosomal dominant - a genetic trait that is produced when only one copy of a gene is present

autosomal recessive - a genetic trait that is expressed only when two copies of a gene are present

basilar invagination - condition, sometimes associated with Chiari, where the C2 vertebra is displaced upward, potentially compressing the brainstem

cerebellar tonsils - portion of the cerebellum located at the bottom, so named because of their shape

cerebellum - part of the brain

Looking Back: Milhorat Redefines Chiari

In May of 1999, Dr. Thomas Milhorat, then with the State University of New York Health Science Center in Brooklyn, put Chiari on the map by publishing a landmark study in the journal Neurosurgery. With the growing popularity of MRI's, Chiari was beginning to receive more attention from the neurosurgical community, and Milhorat's study propelled this trend by providing a detailed, comprehensive look at the clinical and radiological characteristics of over 300 Chiari patients, and by challenging some of the long-held beliefs about the disease.

Milhorat, of course, went on to establish the Chiari Institute in Long Island and treat hundreds of Chiari patients, while his paper is perhaps the most cited in the recent Chiari medical literature. Given the impact it had, Chiari & Syringomyelia News thought it would be worthwhile to take a look back at this classic study and what it revealed.

Milhorat and his colleagues prospectively looked at 364 symptomatic Chiari patients between 1994 and `1997. The team collected a complete medical and family history from each patient along with a checklist of symptoms. The doctors performed complete physicals and neurological exams and each patient underwent an MRI. Some people received additional testing such as cine-MRI's, sleep studies, and vestibular testing. Each person's functioning was measured using a common scale (the Karnofsky disability scale), which creates a score of between 0-100.

The group consisted of 275 women and 89 men. The average age of diagnosis was 31 years, however most had reported having symptoms for years prior to that. In fact, 37% reported having life-long problems, such as headaches or balance problems. Illustrating the struggle many go through before being accurately diagnosed, more than half (57%) had been told by at least one doctor that they had a mental or emotional problem. Interestingly, 24% of the group reported that a traumatic event had precipitated symptoms (see Figure 1). Other common precipitating factors included infection, coughing/sneezing, and pregnancy. Syringomyelia was present in 65% of the patients, while 42% had some degree of scoliosis and 12% also had basilar invagination.

The research team found, not surprisingly, that headache was the most commonly reported symptom, with 81% of the patients suffering from the classic symptom (see Figure 2). However, in addition, the team found that many patients also suffered from visual problems, vestibular problems, spinal cord problems, and compression of the brain stem, cerebellum, or cranial nerves. Common individual symptoms included headache, pressure behind the eyes, dizziness, trouble swallowing, and sleep apnea.

While the documentation of symptoms was certainly worthwhile, a more powerful result of the study came from the MRI results. Despite the traditional definition of Chiari as a tonsillar herniation of greater than 3mm-5mm, Milhorat showed that the length of herniation actually was not related to the clinical situation. Specifically, his team found that the herniation size did not correlate with the disability scale or the presence of syringomyelia. In fact, 15 of the 32 patients with a herniation of less than 5mm acutally had syringomyelia.

What Milhorat's team did find is that in every patient, the CSF space behind (and to the side) of the cerebellar tonsils was reduced, or even blocked completely. In a subset of patients who underwent a cine-MRI, there was evidence that CSF flow was restricted around the cerebellar tonsils. In further study, the posterior fossa volume for 50 patients was quantitatively measured and compared to 50 age and gender matched control subjects with no neurological abnormalities. They found that the Chiari patients had significantly smaller posterior fossas than the control group, indicating that Chiari is a condition where the crowding is due to a small posterior fossa region, rather than a too large brain.

Finally, the team - specifically Dr. Marcy Speer of Duke University - looked at the familial aspects of the disease. Forty-three patients (12%) reported having at least one close relative with Chiari and/or syringomyelia. The families of twenty-one patients with two or more affected family members agreed to undergo further study. In studying these families, Dr. Speer found evidence of a genetic component to Chiari consistent with either autosomal dominant or recessive inheritance (see Definitions). However, the authors also state that the finding could be explained by exposure to a common environmental factor as well.

Since this study was published, many of the findings have been confirmed by other researchers. Several studies have found no correlation between the size of the herniation and clinical symptoms or outcome; further research has confirmed that on average Chiari patients have smaller posterior fossa volumes; and further genetic studies are adding evidence that at least some Chiari cases have a genetic basis.

Unfortunately, new information is often slow to spread in the medical community and many doctors are basing their diagnoses and treatments on outdated and incorrect information. For too many patients, the status-quo remains; which means going years without a proper diagnosis and probably being told at least once that there's nothing wrong with them, it's all in their head.

located at the bottom of the skull, near the opening to the spinal area; important for muscle control, movement, and balance

cerebrospinal fluid (CSF) - clear liquid in the brain and spinal cord, acts as a shock absorber

Chiari malformation - condition where the cerebellar tonsils are displaced out of the skull area into the spinal area, causing compression of brain tissue and disruption of CSF flow

cine MRI - type of MRI which can show CSF flow

dysphagia - trouble swallowing

familial - tending to occur among family members

magnetic resonance imaging (MRI) - diagnostic device which uses a strong magnetic field to create images of the body's internal parts

posterior fossa - depression on the inside of the back of the skull, near the base, where the cerebellum is normally situated

scoliosis - bnormal curvature of the spine

syringomyelia (SM) - neurological condition where a fluid filled cyst forms in the spinal cord

syrinx - fluid filled cyst in the spinal cord

tonsillar herniation - descent of the cerebellar tonsils into the spinal area; often measure in mm

vestibular - elating to the balance system; more specifically to the vestibule of the inner ear

Source

Milhorat TH, Chou MW, Trinidad EM, Kula RW, Mandell M, Wolpert C, Speer MC. Chiari I malformation redefined: clinical and radiographic findings for 364 symptomatic patients. Neurosurgery. 1999 May;44(5):1005-17.

Table 1
Precipitating Factors

Factor	# of Patients
None	193
Trauma	89
Infection	27
Coughing, Sneezing	24
Pregnancy	16
Other	15

- Trauma includes whiplash and direct blows to head/neck
- Other includes sexual intercourse, epidural anesthesia lumbar puncture, and air travel

Table 2
Common Symptom Categories

Category	# of Patients (%)
Spinal Cord Function	305 (84%)
Headache	296 (81%)
Visual Problems	283 (78%)
Vestibular	269 (74%)
Brain Stem, Cerebellum, or Cranial Nerves	191 (52%)

Selected Common Symptoms:

Headache: intense pressure in back of head made worse by certain activities

Spinal Cord: abnormal sensations; muscle weakness

Visual: pressure behind eyes; floaters and flashing lights

Vestibular: dizziness; pressure in ears

Brain Stem/Cerebellum/Cranial Nerves: dysphagia, sleep apnea