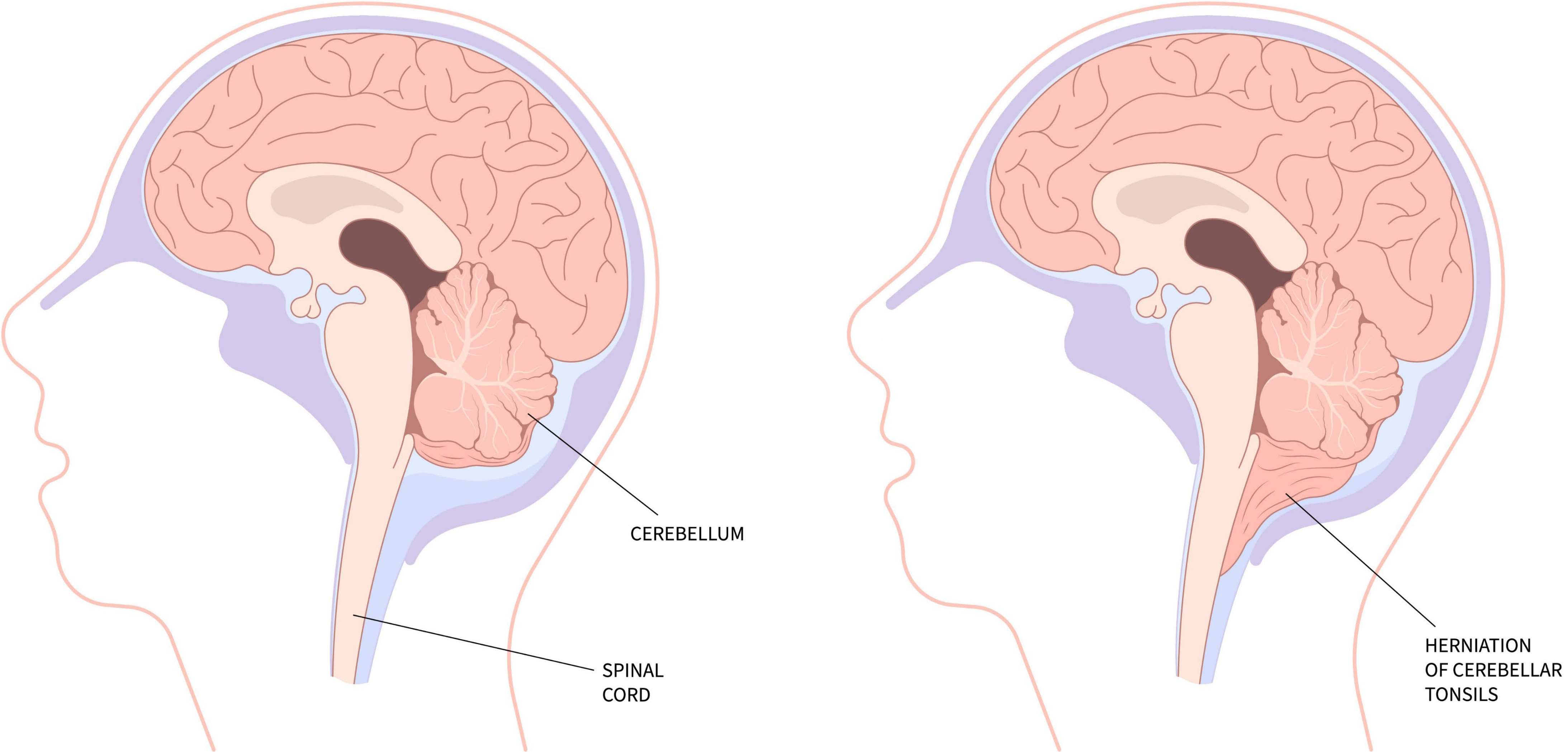
CONNECTING THE DOTS

Between Chiari & EDS

Courtesy of Chiari Bridges

NORMAL

CHIARI MALFORMATION



CONGENITAL
defined

Structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life.

ACQUIRED
defined

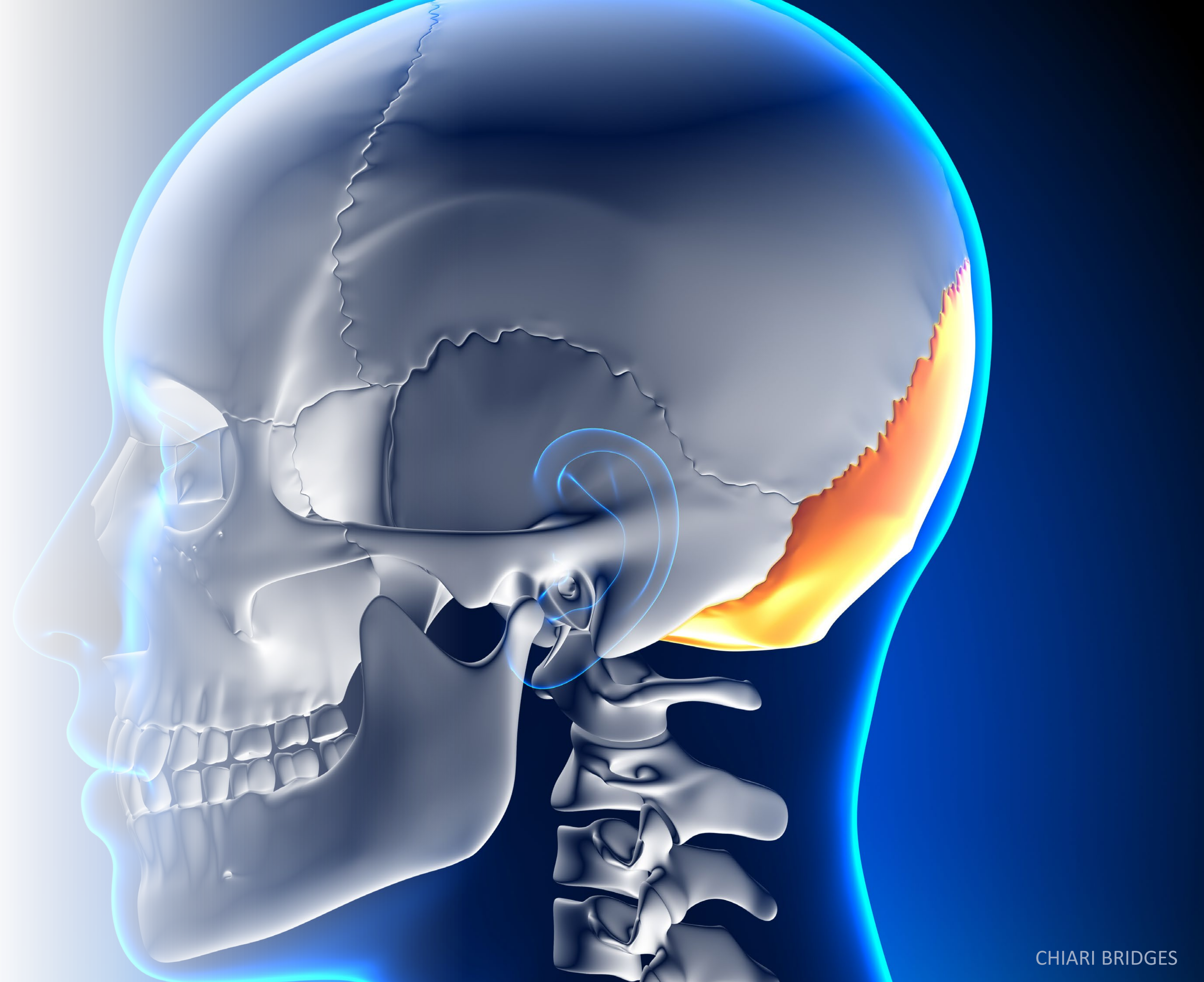
Conditions that are **NOT INHERITED** or present at birth (congenital) but developed after birth.

Congenital Assumption & Failed Decompressions

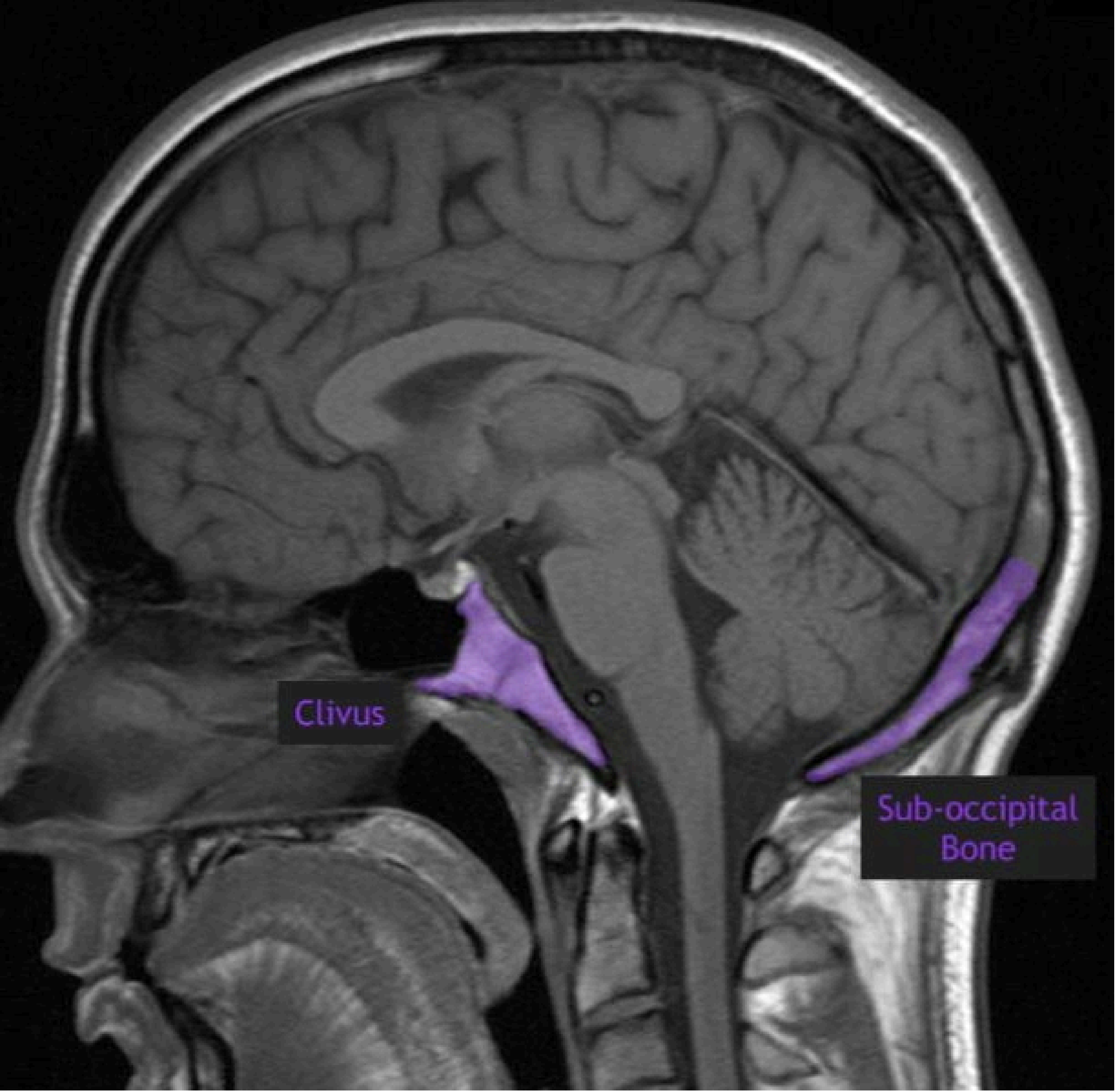
Congenital Chiari → Small Posterior Fossa

Acquired Chiari → Pathological Conditions

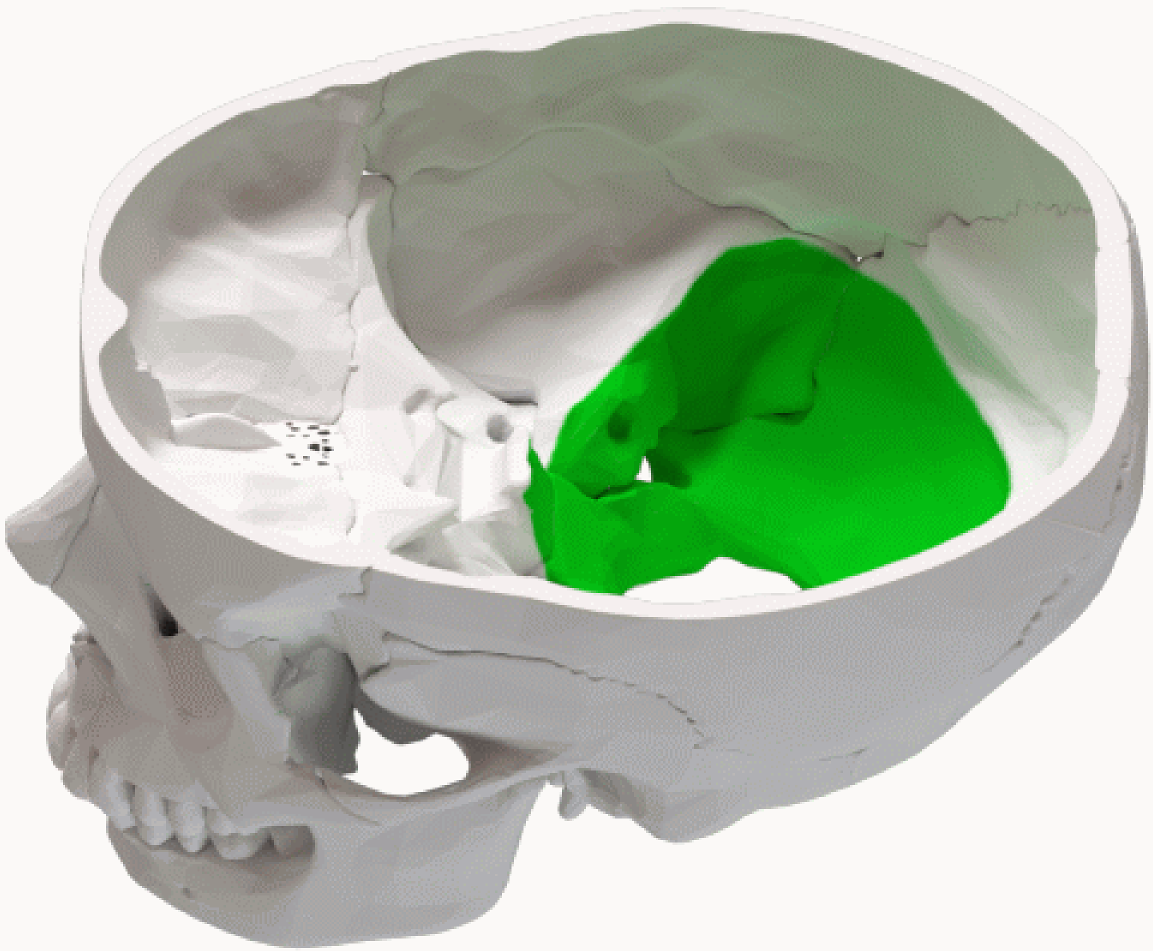
The Occipital Bone



The Foramen Magnum

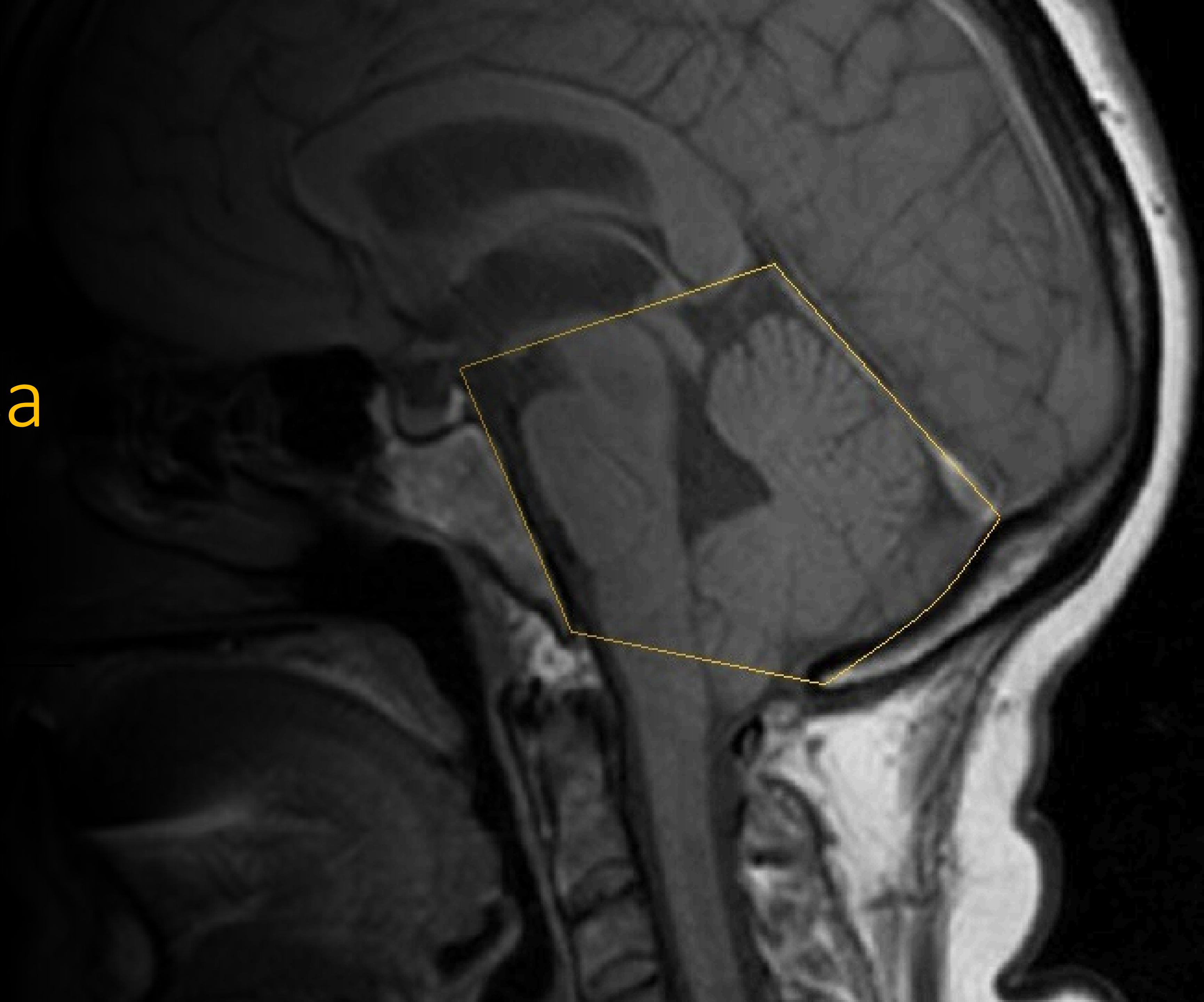


The Foramen Magnum

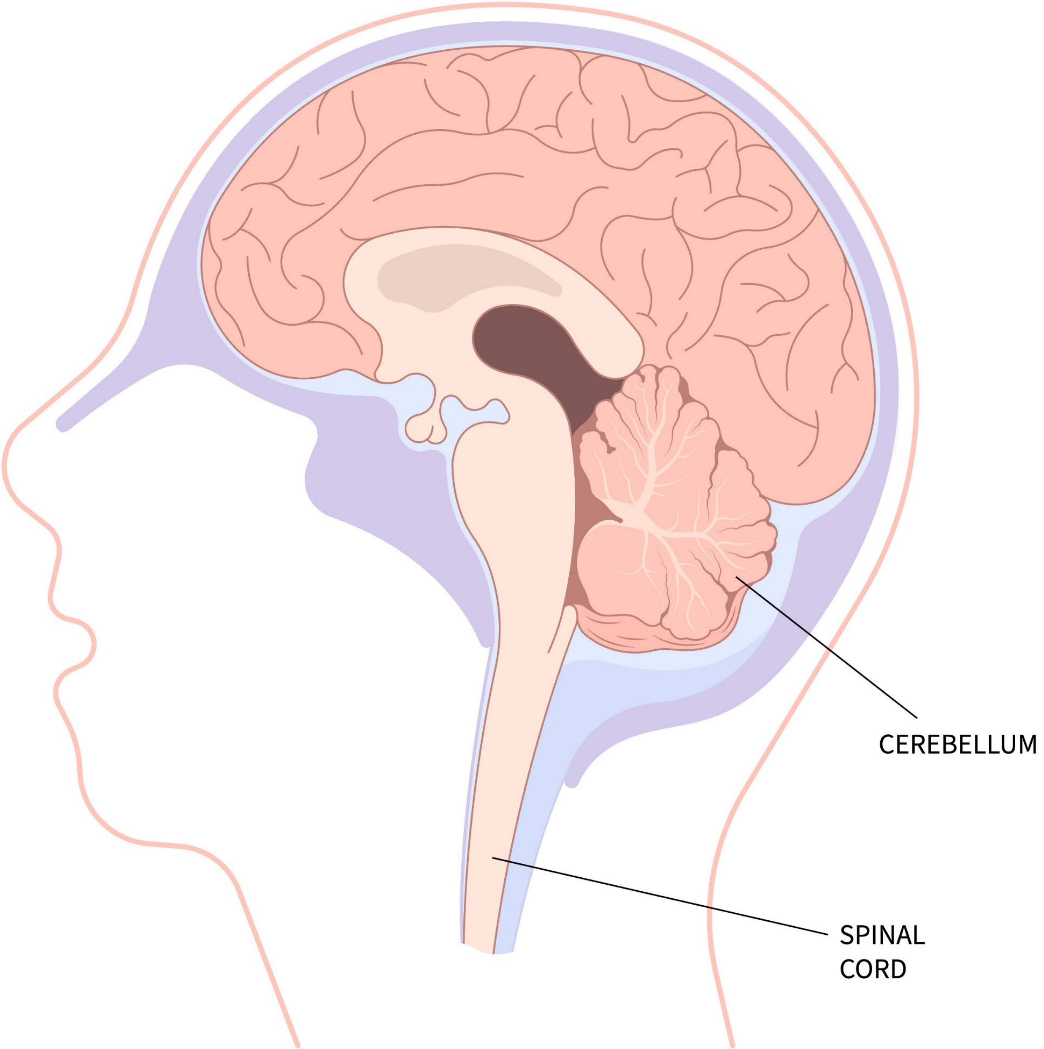


Posterior Fossa

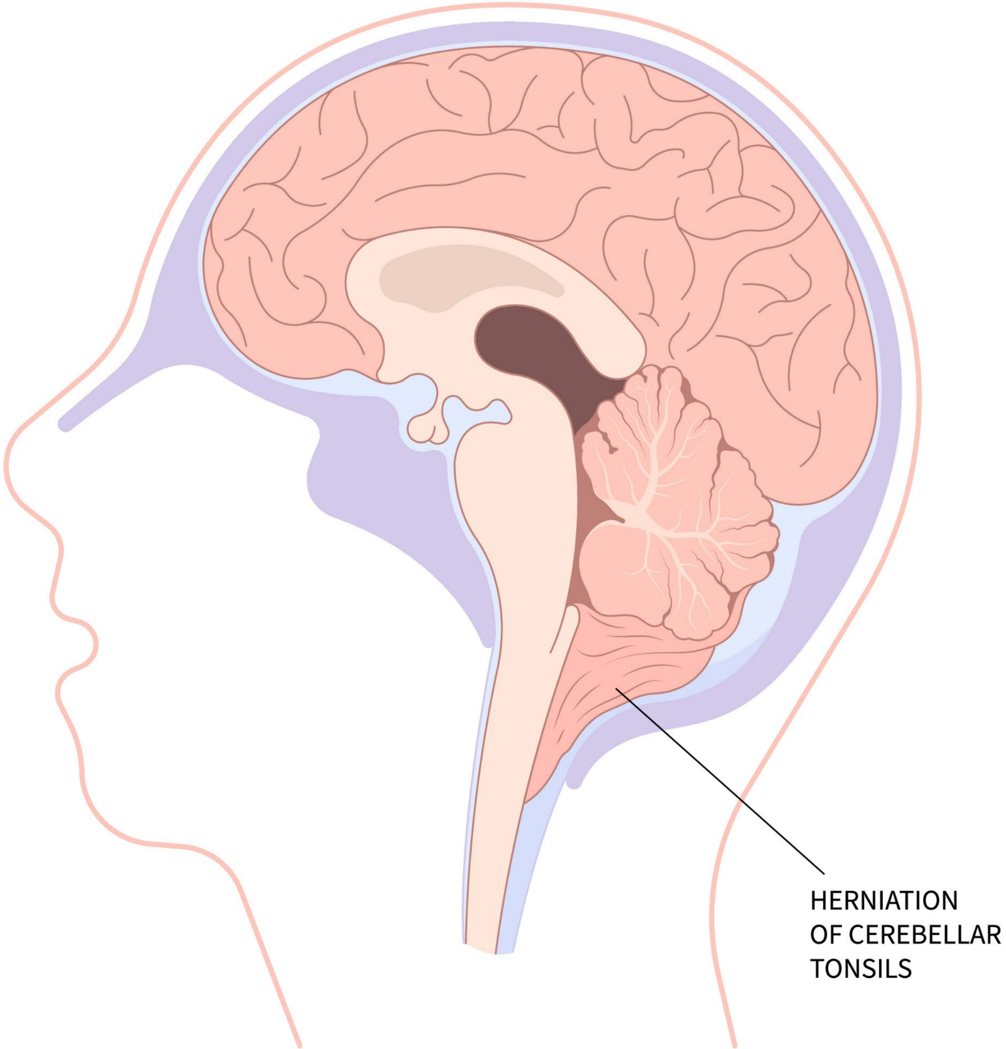
Posterior Fossa
Hypoplasia



NORMAL

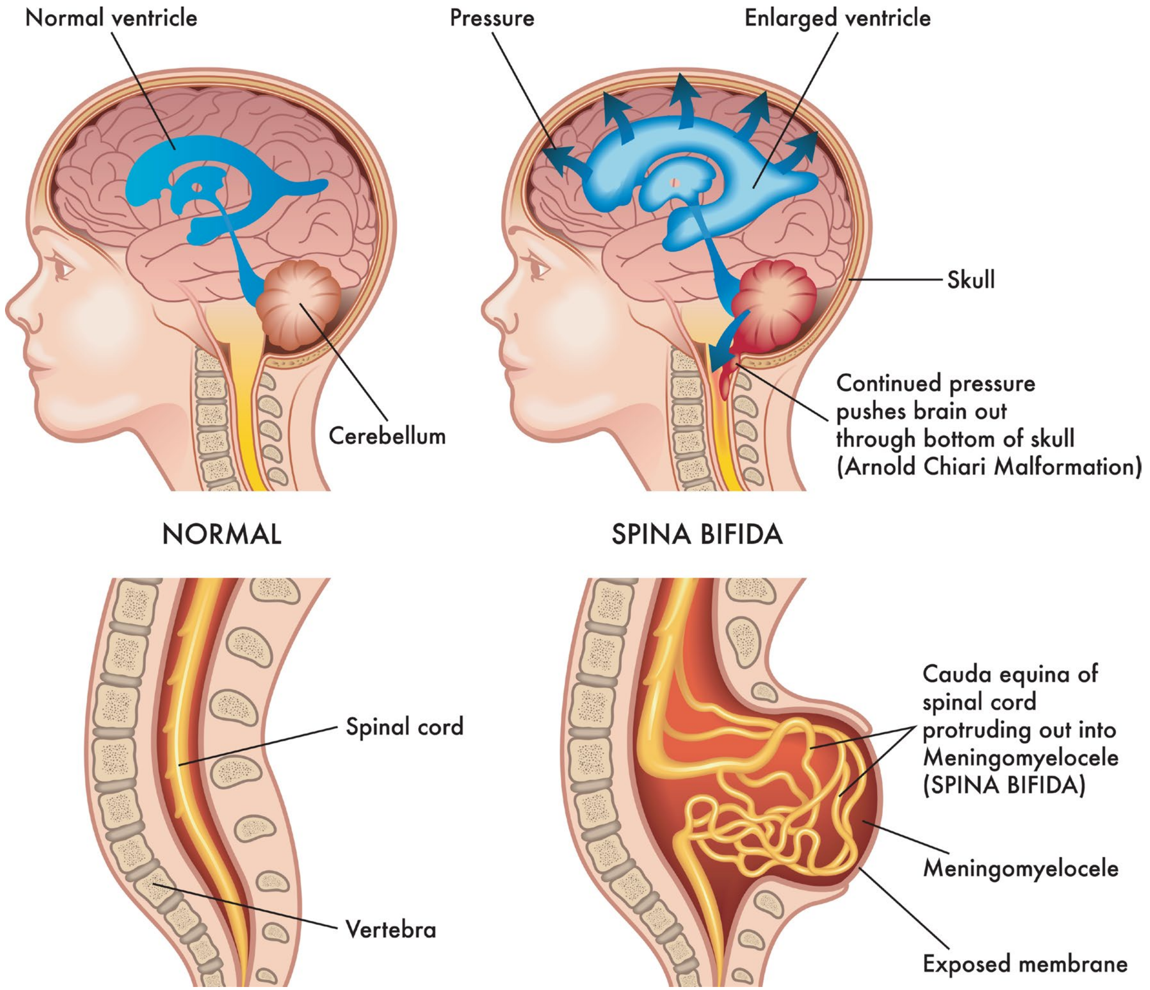


CHIARI MALFORMATION

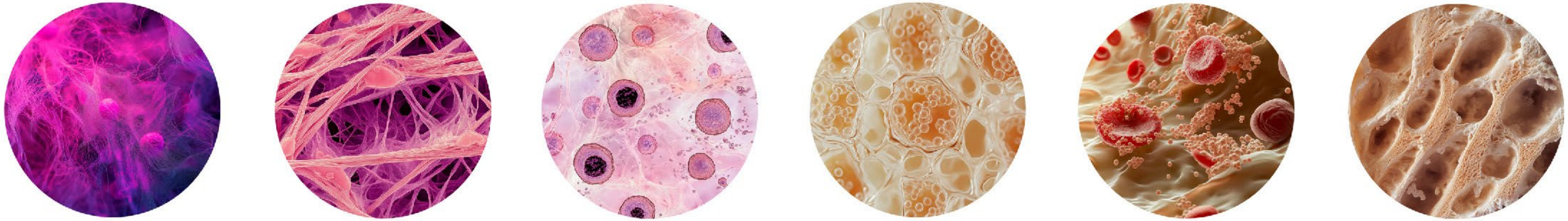


Arnold Chiari
Chiari 2 Malformations

- **Spina Bifida (myelomeningocele)**
- **Tethered Cord Syndrome**
- **Hydrocephalus induced IH**



TYPES OF CONNECTIVE TISSUE



Loose | Dense | Cartilage | Adipose | Blood and Lymph | Bone

Connective tissues provide strength and flexibility to structures such as ...

- Ligaments, Cartilage, Bursa
- Cutaneous Tissue (skin)
- Adipose tissue (fat)
- Muscles
- Organs
- Blood
- Lymph nodes
- Bones
- Blood vessels
- Nerves

What are Heritable Disorders of Connective Tissues (HDCTs)?

There are over 200 identified Heritable Disorders of Connective Tissue (HDCTs), conditions such as:

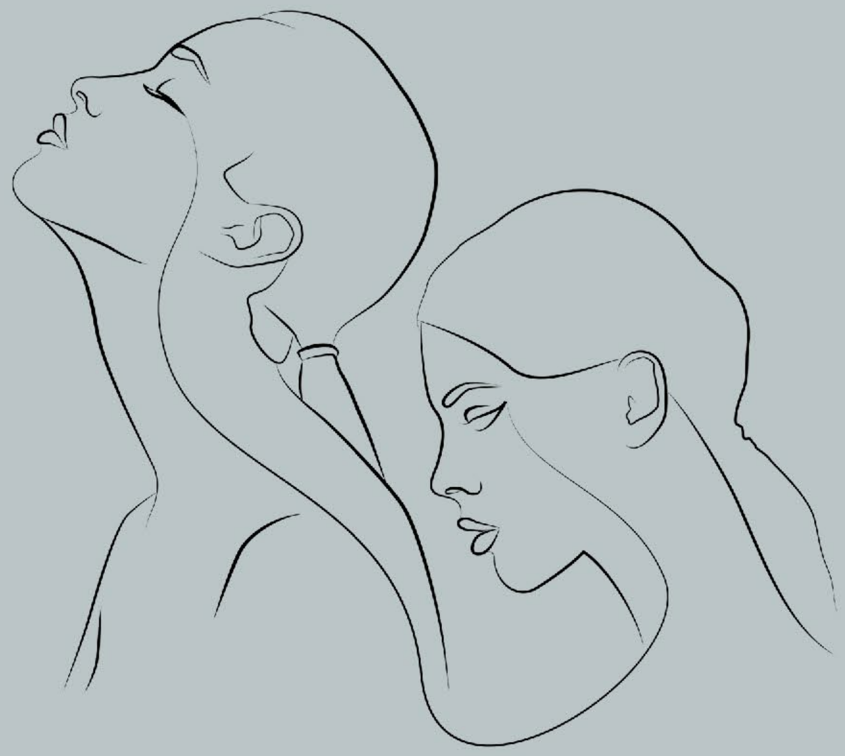
- **Ehlers-Danlos Syndrome *(pronounced: ee-lers dan-low)***
- Marfan Syndrome
- Loeys-Dietz Syndrome
- Stickler Syndrome
- Osteogenesis Imperfecta
- Pseudoxanthoma Elasticum
- Epidermolysis bullosa

*All involve one of the many **proteins** that make up our body's connective tissue.*

Ehlers-Danlos Syndromes (EDS) are a group of hereditary connective tissue disorders involving defects in our collagen.

There are fourteen known subtypes of Ehlers-Danlos syndromes (EDS).

- Classical EDS (cEDS)
- Classical-like EDS (clEDS)
- Cardiac-valvular EDS (cvEDS)
- Vascular EDS (vEDS)
- Arthrochalasia EDS (aEDS)
- Dermatosparaxis EDS (dEDS)
- Kyphoscoliotic EDS (kEDS)
- Brittle Cornea Syndrome (BCS)
- Spondylodysplastic EDS (spEDS)
- Musculocontractural EDS (mcEDS)
- Myopathic EDS (mEDS)
- Periodontal EDS (pEDS)
- Hypermobile EDS (hEDS)
- Classical-like EDS2 (clEDS2) – new formerly unnamed sub-type involving the AEPB1



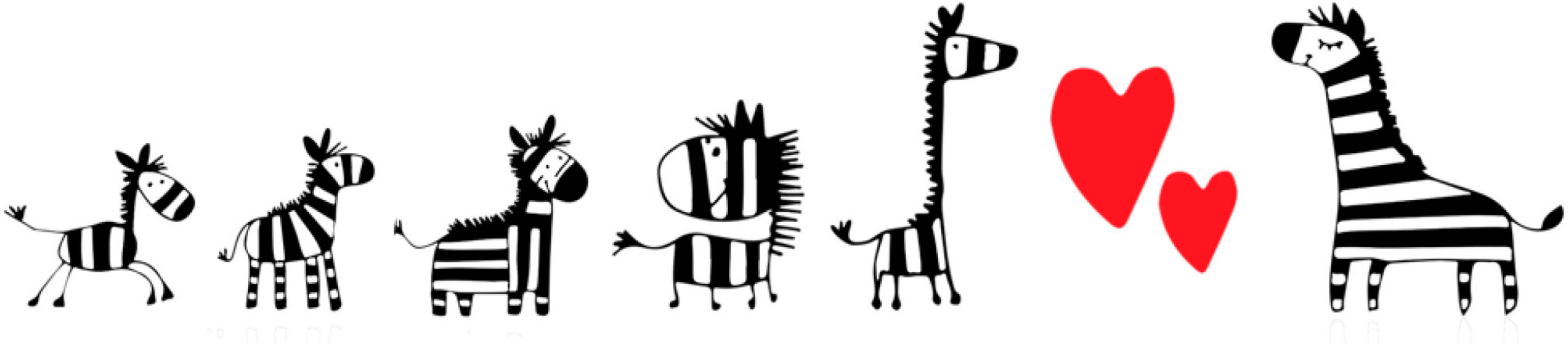
Crossover Symptoms

While each sub-type of the Ehlers-Danlos Syndromes (EDS) has its own characteristics and symptoms, it's important to remember that EDS symptoms are known to crossover from one sub-type to another.

So, while symptomology can help narrow down a diagnosis, it is not unusual for someone with one sub-type, to have symptoms or complications that seem more consistent with another subtype.

Hereditary

Ehlers-Danlos Syndromes (EDS) are known to be hereditary. Some are known to be autosomal recessive (where both parents must have the mutated gene to pass it down to a child) and others are autosomal dominant (where only one parent need have the mutated gene to pass it down to the child). It's important to remember that you can't control what you pass down to your children genetically, so it should NEVER be allowed to become a blame game!



ABOUT COLLAGEN

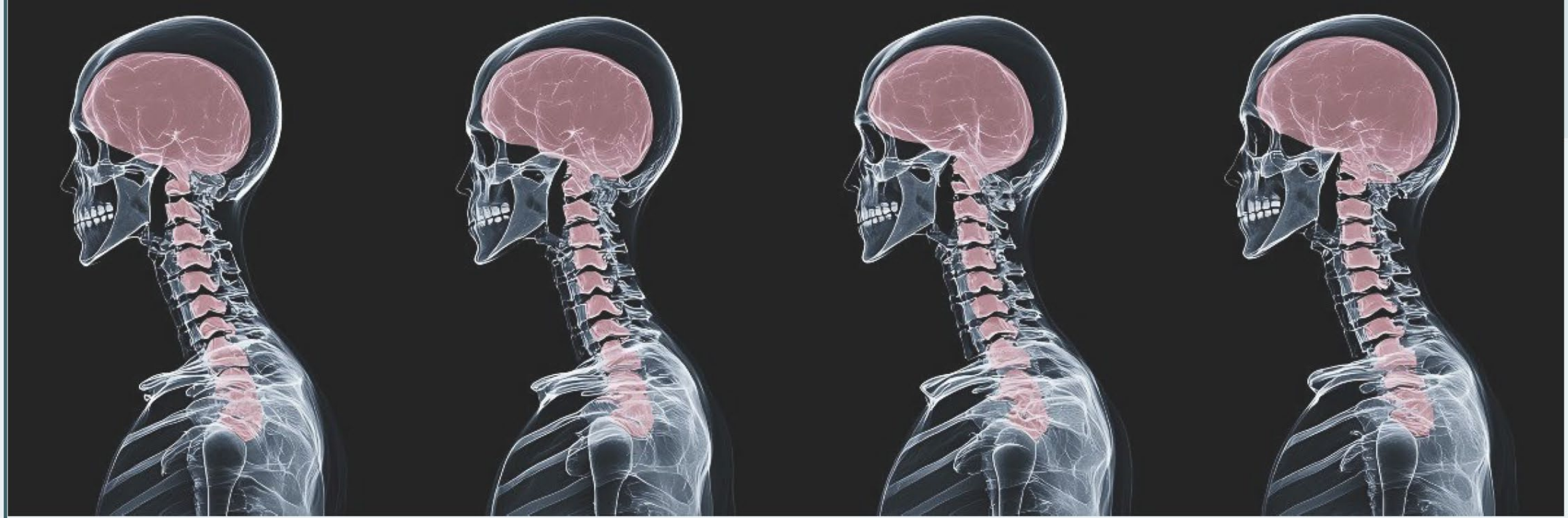
Collagen is a Structural Protein that Makes Up 30% of the Proteins in the Human Body

Collagen is often likened to the "**fibrous glue** that holds us together at a cellular level," providing structure to our organs, muscles, bones, skin, blood vessels, and connective tissues.

Imagine the difference between something bonded together with **Gorilla Glue** versus watered down **Elmer's Glue**. The creation using Elmer's Glue wouldn't be as strong. The consistency of Elmer's Glue could cause anything depending on it to lose its form and then gravity will cause it to prolapse/herniate.

Structure Gone Awry

- Ehlers-Danlos Syndromes (EDS) are known to involve a mutation in our body's collagen, but it's not as simple as it sounds. EDS doesn't just affect our skin and joints, but everything in our bodies that was designed to interact with that collagen. That is why we tend to have so many problems throughout our bodies that you would think couldn't be related. It really is our connective tissues that connect the issues.
- Our Intervertebral Discs are comprised of collagen, therefore the discs of the Ehlers-Danlos Syndromes (EDS) patient, tend to dehydrate, thin, bulge, slip, and herniate into the spinal cord. The average person might have 1-2 disc problems in their lifetime, EDS patients tend to have many discs bulging and herniated at any given time, often in different levels of the spine. This condition is known as Degenerative Disc Disease (DDD).



Connecting EDS & Chiari Malformations

The same theory can be used to explain why many of us with EDS have prolapsed cerebellar tonsils.

- EDS is known for causing organ prolapses and your cerebellum really is just a complex organ.
- Therefore, if you have EDS and Chiari, it should be considered an Acquired Chiari Malformation until all possible EDS related comorbidities are ruled out!
  - *Even if posterior fossa hypoplasia is confirmed, it is irresponsible not to give adequate attention to all comorbid conditions that can push/pull the cerebellar tonsils down, creating an Acquired (Secondary) Chiari Malformation.*

Studies have shown that Ehlers-Danlos Syndromes (EDS) can have many neurological and spinal manifestations, to include:

- Intracranial Hypertension (IH/IIH)
- Chiari Malformations (CM)
- Atlantoaxial & Craniocervical Instability (AAI/CCI)
- Segmental Kyphosis & Instability
- Tethered Cord Syndrome (TCS)
- CSF Leaks causing Spontaneous Intracranial Hypotension (SIH)
- Dystonia & Other Movement Disorders
- Neuromuscular Complications
- Acquired Chiari Malformations

Chiari Malformation Type 1 (CM1) has been reported as a comorbid condition in hypermobile EDS (hEDS). The precise incidence of the CM1 and EDS association is unknown, but the female to male ratio is higher in the CM1 and EDS subgroup (9:1) than in the general CM1 population alone (3:1).

-Dr. Fraser Henderson

- It is a matter of concern that so many surgeons find herniated cerebellar tonsils, diagnose it as a Chiari Malformation and assume it to be congenital.

- They offer relief from decompression surgery, without checking for any pathological conditions that might be causing this hernia of the cerebellar tonsils.

- Most of the literature simply says that an Acquired Chiari is rare without ever defining what would constitute an Acquired Chiari Malformation.

- **If you have EDS and Chiari, it should be considered an Acquired Chiari Malformation until all possible EDS related comorbidities are ruled out!**

  - *Even if posterior fossa hypoplasia is confirmed, it is irresponsible not to give adequate attention to all the other comorbid conditions that are considered pathological to Acquired Chiari Malformations.*

Hypermobility

- It may seem counter-intuitive, but many hypermobile people also suffer from stiffness and tightness in their joints and muscles. Muscle stiffness and tightness is caused by the extra work muscles need to do to try and keep a hypermobile person's joints stable. We affectionately refer to them as *Stiff Zebras*.

“When you hear hoofbeats, think horses, not zebras!”

In medical school, students are taught to not neglect the obvious, with the analogy of, *"when you hear hoofbeats, think horses, not zebras."*

This philosophy poses a problem for those with Ehlers-Danlos syndromes, as it's not that our condition is rare, but that it's rare that doctors know anything about it; and patients often go years or decades pursuing answers to our numerous symptoms.

Multisystemic Manifestations

Because collagen is so abundant in the human body, it often affects multiple systems and a multidisciplinary approach to is often needed:

- Coordinate care
- Maximize treatment
- Reduce medical visits and inappropriate surgical treatments
  - Scar tissue problems

HYPERMOBILE EHLERS-DANLOS CONNECTS THE DOTS

INTRACRANIAL PRESSURE PROBLEMS

- Idiopathic Intracranial HYPERtension*
- Spontaneous Intracranial HYPOtension

CRANIOVERTEBRAL ABNORMALITIES

- Craniocervical Instability
- Atlantoaxial Instability
- Basilar Invagination & Cranial Settling

SPINAL CORD PROBLEM

- Tethered Cord Syndrome
- Occult Tethered Cord
    - Scoliosis

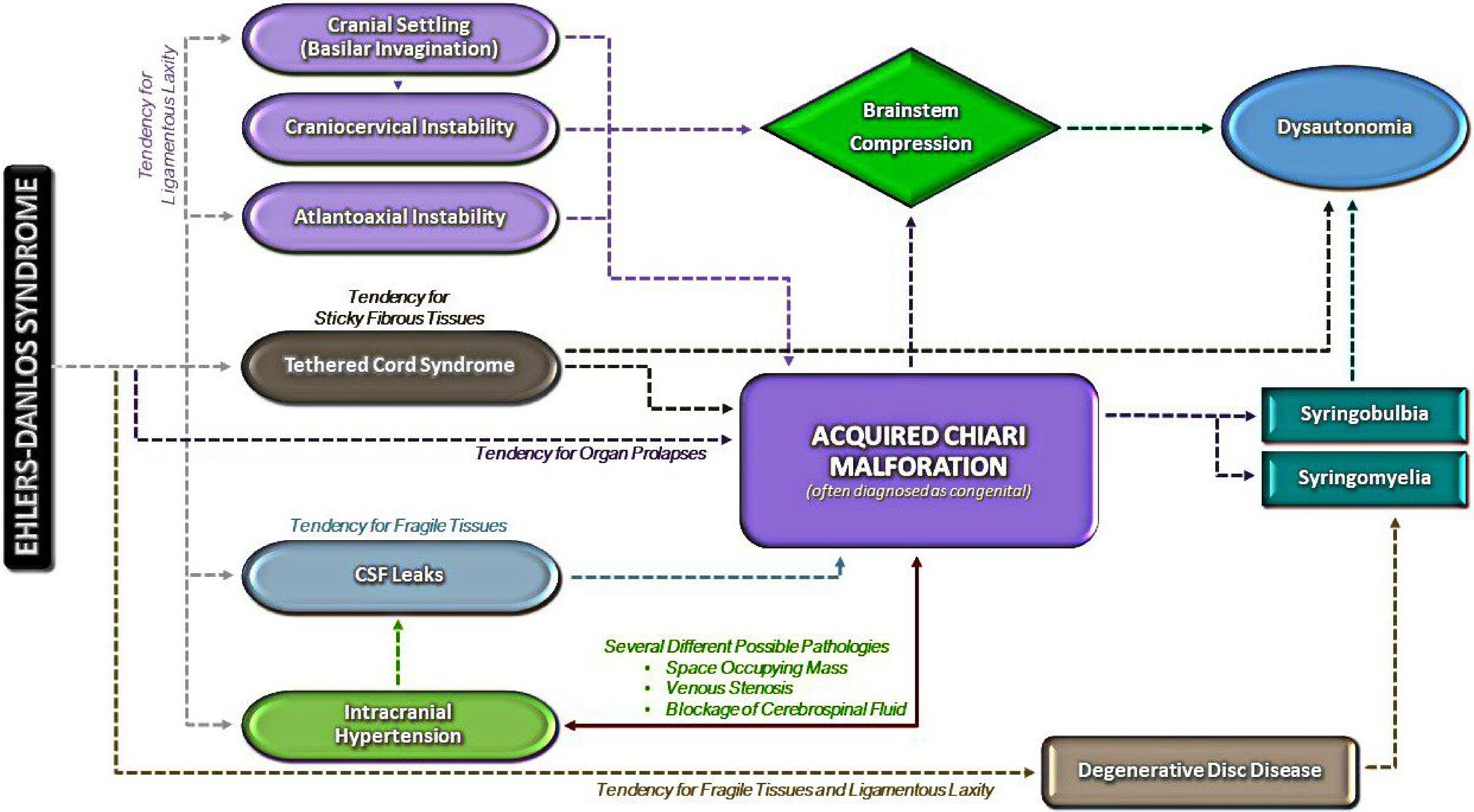
Sleep Apnea

Studies show that those with Ehlers-Danlos Syndromes (EDS) have a higher prevalence of symptomatic Obstructive Sleep Apnea (32% vs 6% in the general population).

For this reason, we recommended that **EVERYONE** with EDS is tested for Sleep Apnea and use their CPAP **RELIGIOUSLY!**

EDS Related Pathologies:

- Craniocervical Instability (CCI)
- Basilar Invagination (BI) & Impression
- Tethered Cord (TCS)
- CSF Leak (SIH)
- Intracranial Hypertension (IH/IIH)


EHLERS-DANLOS SYNDROME
Tendency for Ligamentous Laxity
Cranial Settling (Basilar Invagination)
Craniocervical Instability
Atlantoaxial Instability
Brainstem Compression
Dysautonomia
Tendency for Sticky Fibrous Tissues
Tethered Cord Syndrome
Tendency for Organ Prolapses
ACQUIRED CHIARI MALFORATION
(often diagnosed as congenital)
Syringobulbia
Syringomyelia
Tendency for Fragile Tissues
CSF Leaks
Several Different Possible Pathologies
• Space Occupying Mass
• Venous Stenosis
• Blockage of Cerebrospinal Fluid
Intracranial Hypertension
Degenerative Disc Disease
Tendency for Fragile Tissues and Ligamentous Laxity

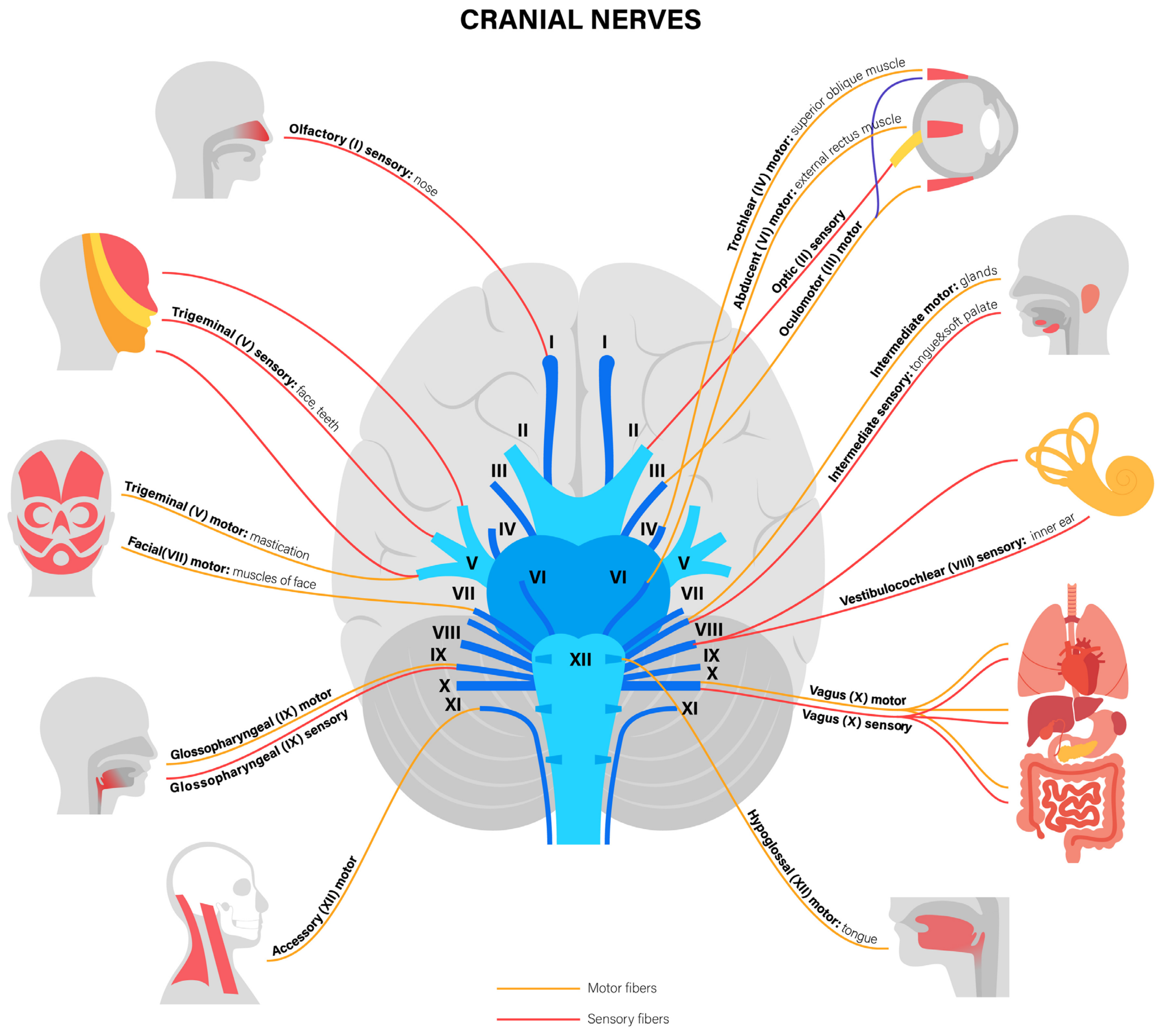
Does Size Matter?

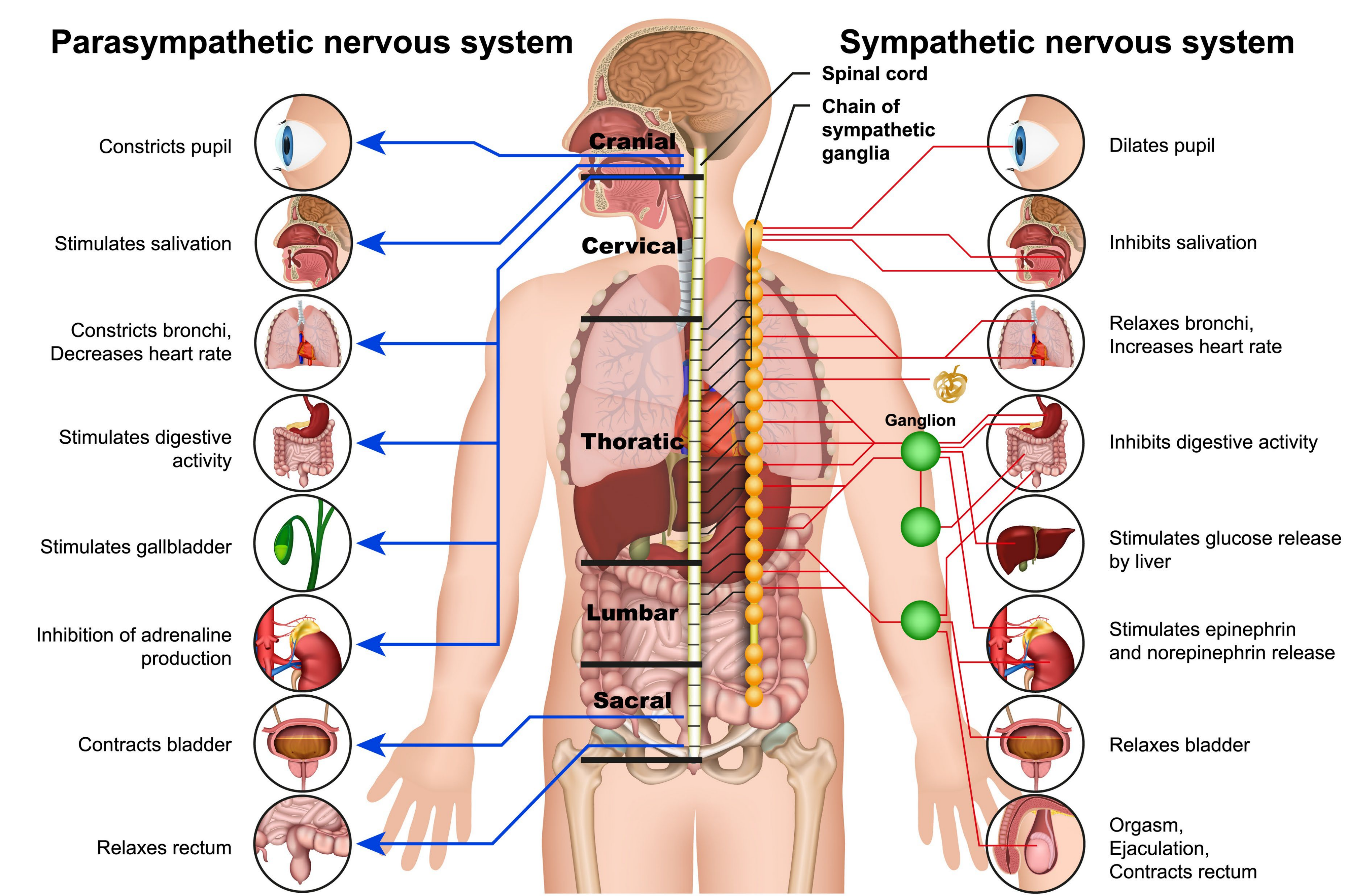
How many millimeters does it take to block the flow of cerebrospinal fluid?

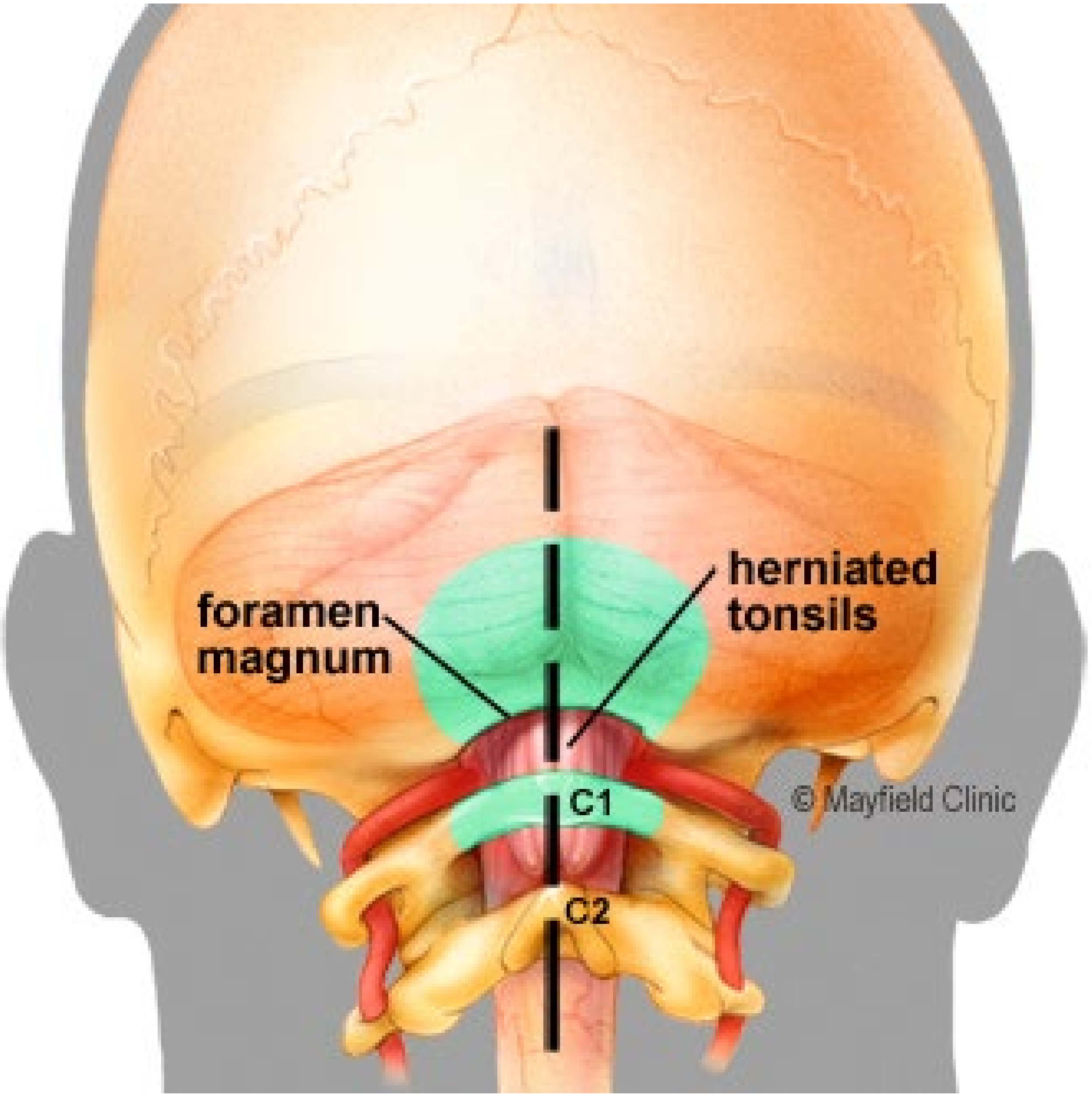
PRIMARY SYMPTOMS OF CHIARI

- **Occipital Pain**: pain at the occiput (base of the skull and upper neck), worsened by Valsalva maneuvers and being upright for a prolonged period.
- **Equilibrium Problems**: vertigo, dizziness, nausea/vomiting, balance, gait, and overall proprioception problems (heights, peripheral vision).
- **Cognitive Decline**: brain fog, memory issues often due to inflammation.
- **Throat and Esophageal Problems:** dysphagia, hoarseness, hiccups
- **Sleep Problems:** insomnia, Myalgic encephalomyelitis/chronic fatigue syndrome (ME/CFS) central/obstructive sleep apnea
- **Muscle Spasms:** eye twitching, painful neck spasms, restless leg/limbs
- **Numbness in the arms/hands**
- **Hormone Related Problems**
- **Autonomic Dysfunction (Dysautonomia): POTS, tachycardia, neurocardiogenic syncope...**

CRANIAL NERVES




Parasympathetic nervous system
Sympathetic nervous system
Spinal cord
Chain of sympathetic ganglia
Cranial
Cervical
Thoratic
Lumbar
Sacral
Ganglion
Constricts pupil
Stimulates salivation
Constricts bronchi,
Decreases heart rate
Stimulates digestive activity
Stimulates gallbladder
Inhibition of adrenaline production
Contracts bladder
Relaxes rectum
Dilates pupil
Inhibits salivation
Relaxes bronchi,
Increases heart rate
Inhibits digestive activity
Stimulates glucose release by liver
Stimulates epinephrin and norepinephrin release
Relaxes bladder
Orgasm,
Ejaculation,
Contracts rectum



POSTERIOR FOSSA DECOMPRESSION

DURAPLASTY

- Controversial
- Pseudomeningocele

CAUTERIZATION VS. RESECTION

- What's the difference?

LAMINECTOMY

- Posterior Arch of C1
- C1/C2 combination
- Craniocervical Instability

CRANIECTOMY VS. CRANIOTOMY

- Removal of the only bone opposing the prolapse
- Titanium mesh plates

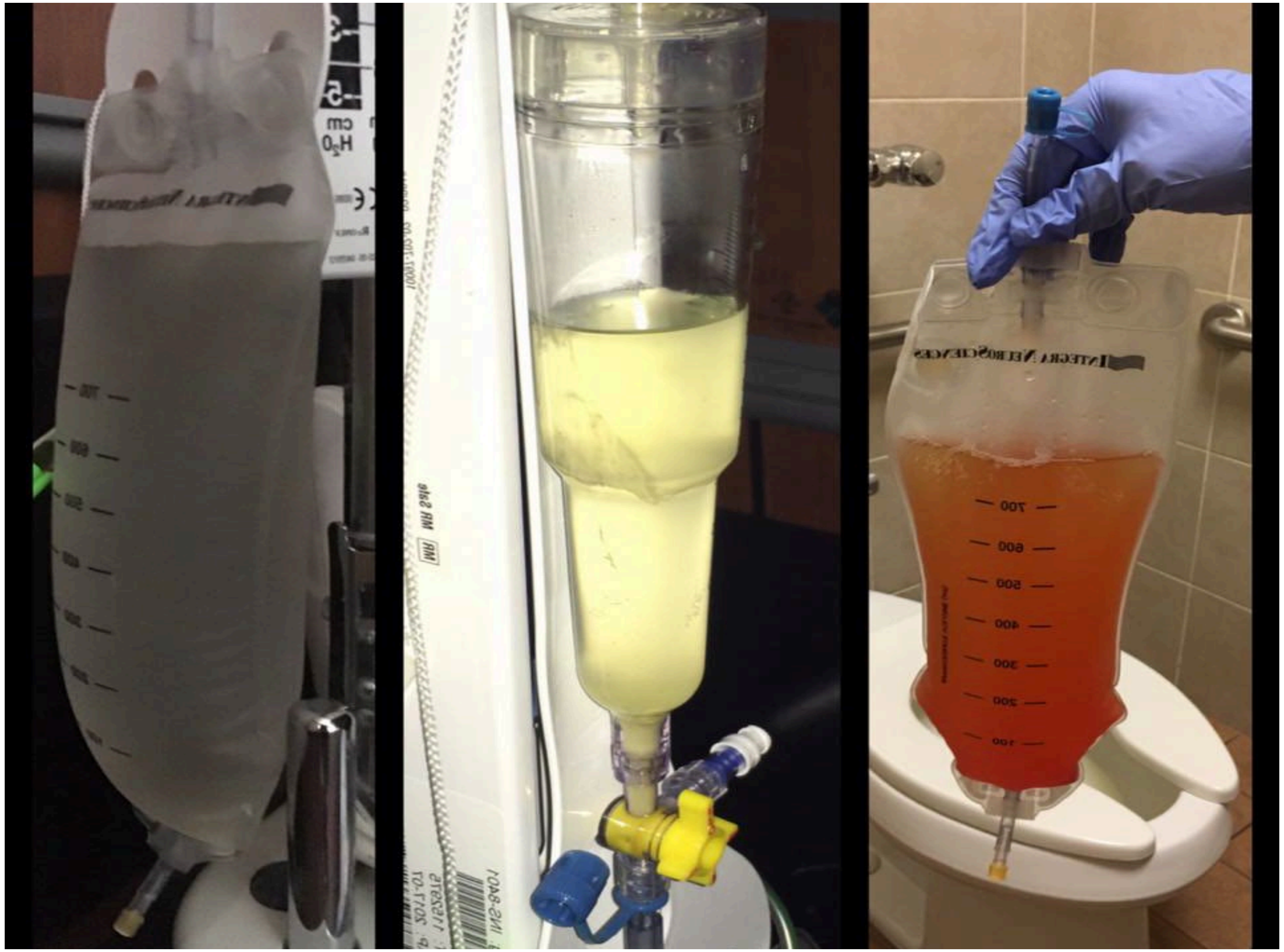
Chicago Chiari Outcome Scale

Details of the CCOS, as reported in Aliaga et al.

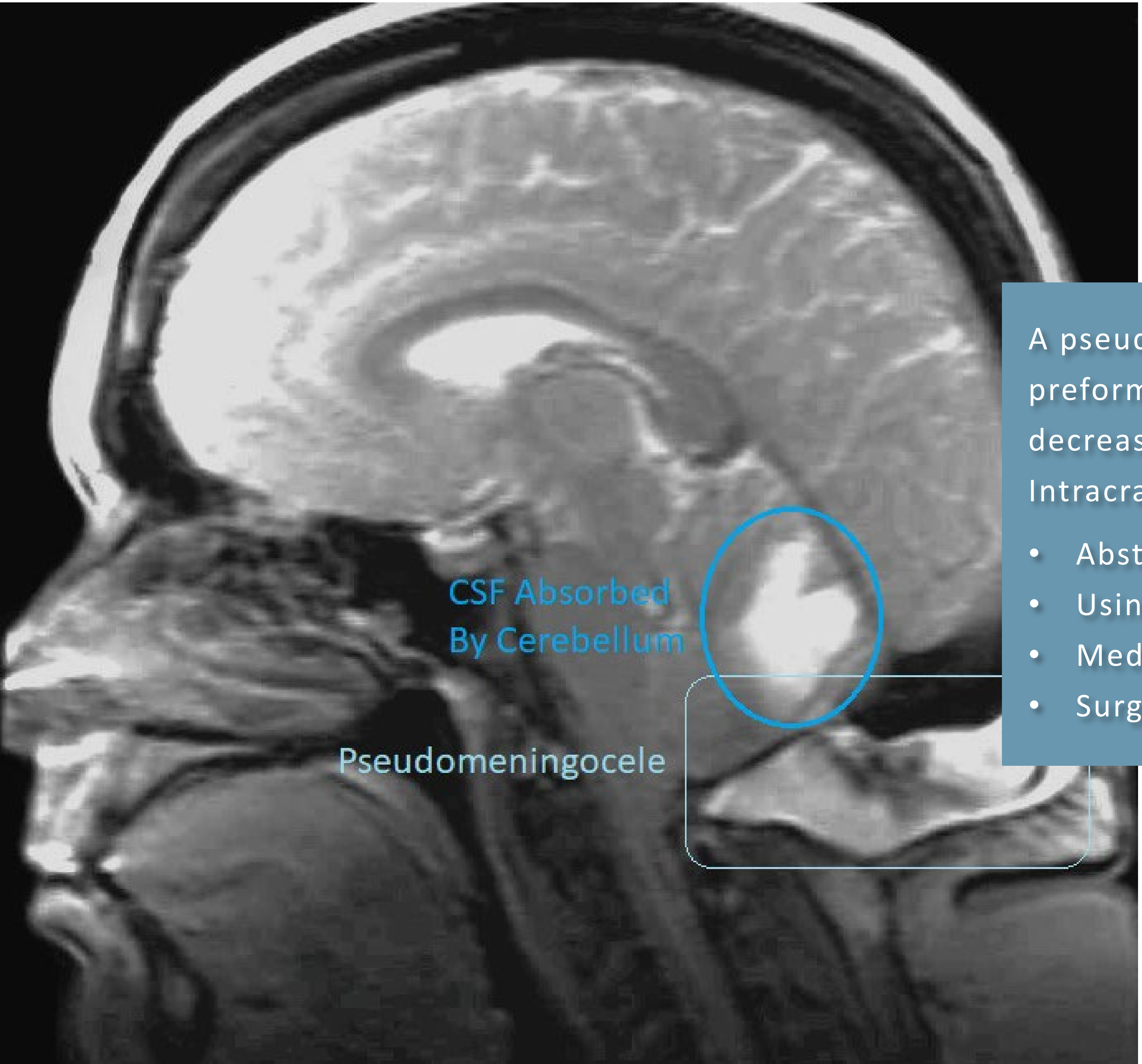
Score	1	2	3	4
Pain	Worse	Unchanged and refractory to medication	Improved or controlled with medication	Resolved
Non-pain	Worse	Unchanged or improved but impaired	Improved and unimpaired	Resolved
Functionality	Unable to attend	Moderate impairment (<50% attendance)	Mild impairment (>50% attendance)	Fully functional
Complication	Persistent complication, poorly controlled	Persistent complication, well-controlled	Transient complication	Uncomplicated course
Location of the cerebellum	Completely slumped and dysfunctional	Slumping and mostly dysfunctional	Slightly slumping and somewhat functional	Buoyant brain and fully functional

Purple Chiari Bridges addition.

COMPLICATIONS OF DECOMPRESSION



- INFECTIONS
  - Surgical site infections
  - Aseptic meningitis
  - Bacterial meningitis
- BLEEDING & ANESTHESIA-RELATED COMPLICATIONS
  - Excessive blood loss
  - Anesthesia risks
- CSF LEAKS
  - Pseudomeningocele
  - Spinal leaks
- BONY REGROWTH
- SCAR TISSUE & ADHESIONS
- OCCIPITAL NEURALGIA
- CRANIOCERVICAL INSTABILITY
- CEREBELLAR SLUMPING
- FAILED DECOMPRESSION



DECREASE YOUR RISK OF COMPLICATIONS

A pseudomeningocele is caused by **DURAPLASTY** being preformed while your cranial pressure is high. You can decrease the risk of a pseudomeningocele by bringing your Intracranial Pressure (ICP) down **BEFORE DECOMPRESSION**.

- Abstaining from caffeine
- Using non-hormonal forms of birth control
- Medicinal Options: Topamax, Diamox
- Surgical Options: Shunt, Stent

Statistics

Recent statistics are proving Ehlers-Danlos to be far more common than initially believed (just a few years ago the prevalence was believed to be 1 in 5,000 to 1 in 20,000 depending on the subtype). The most recent numbers published on its prevalence is 1 in 2,500 to 1 in 5,000 people depending on the subtype (1.5-3 million people worldwide).

A Cure for Ehlers-Danlos Syndromes

There are no known cures for Ehlers-Danlos syndromes (EDS). There is no way to change/repair our mutated collagen.

We deal with one symptom at a time as our bodies try to interact with our faulty collagen as if it's normal. We typically have a multitude of medical specialists (for each manifestation) and a box of braces to try and help keep our joints in place.

"At this point in time, I put EDS in the category of being in the top three or four most severe pain problems. A lot of people for example think that cancer pain is the worst pain, but let me assure you that many EDS patients have pain far beyond any cancer patient I've ever seen. And so it's one of the pain problems that is severe, has been very troublesome, many physicians are afraid of the disease and of the kind of pain that EDS patients have."

-Dr. Forest Tennant

INSIST ON
EXPLORING AND TREATING
ALL PATHOLOGICAL COMORBIDITIES
BEFORE DECOMPRESSION!

YOU HAVE A RIGHT TO BE A PART OF THE DECISIONS REGARDING YOUR TREATMENT OPTIONS, AND
YOU'RE WORTH FIGHTING FOR!