**Jennell McIntosh**

**Module 4 Assignment**

**Arnold Chiari Malformation**

**Introduction:**

**Chiari malformations are structural defects of the posterior fossa of the skull as well as the cerebellum that involve part of the cerebellum extending into and sometimes through the foramen magnum into the spinal canal.1 There are four types of Chiari malformation but Chiari type II malformation is most often referred to as an Arnold Chiari malformation (ACM). I will be focusing on Chiari type II malformation specifically as well as it’s impact on pediatric patients. While Type 1 Chiari malformations are the most common of the four types, ACM is the most often diagnosed in children.2 The ACM is almost always found along with myelomeningocele, the most common form of spina bifida, and some research even states that it is present in every child with myelomeningocele.2-4 The exact epidemiology of ACM is unknown and due to its strong connection with myelomeningocele, much of the research regarding ACM focuses on the epidemiology of myelomeningocele. Myelomeningocele affects about 0.5-1 out of every 1,000 births in America.5**

**Pathology/Pathophysiology:**

Arnold Chiari malformation occurs due to abnormalities in fetal development and along with myelomeningocele it is often associated with hydrocephalus.1,6 The abnormalities during fetal development lead to a smaller than normal posterior fossa which in turn causes the cerebellum’s vermis and tonsils as well as the fourth ventricle to extend into the foramen magnum and spinal canal as they grow and develop.7,8 Many theories have been researched as to what exactly happens during fetal development to facilitate Arnold Chiari malformation but the most widely accepted theory involves an open neural tube allowing leakage of CSF through it’s caudal end which in turn leads to stretching of “embryonic vesicles” and bony defects that create a smaller posterior fossa.7 This lack of adequate space for the cerebellum and fourth ventricle to grow causes them to develop abnormally and also disrupts the flow of CSF.7,8 These abnormalities also compress the pons and medulla of the brainstem and often push these lower brainstem structures downward.3,9 The overcrowding of structures in the posterior fossa specifically displaces the vermis of the cerebellum down below the foramen magnum to form was is called a “peg” that can be moved down as low as the upper thoracic spine level.4 The overall lack of space creates a cerebellum that is smaller in size overall as well as a decrease in the amount of gray matter in the inferior posterior cerebellar lobes and the anterior lobes.3 Although it is not fully understood or the most widely accepted, the “molecular genetic theory” is based on the notion that abnormalities in genetic programming lead to the associated changes in the growth and development of the cranial structures involved in ACM.2 However it comes to be, the obstruction to the flow of CSF created by the overcrowding of the posterior fossa often leads to hydrocephalus and this further compresses the contents of the posterior fossa.3 This environment can lead to changes in many body systems that differ with the age of the child at symptom onset. These changes as they occur in infants as well as older children are detailed below.

*Symptomatic ACM in Neonates and Infants:* In infants with ACM, the **cardiopulmonary system** is often greatly affected. The most common sign of ACM in neonates is inspiratory stridor and it can be fatal.2,4,7 This stridor is often due to abnormal functioning of the brainstem, particularly the pons and medulla as well as abnormal functioning of the vagus nerve.4 Infants with ACM are also at an increased risk of prolonged expiratory apnea with cyanosis (PEAC) which can slow their heart rate and potentially be fatal.4,10 PEAC is often observed during a painful or startling experience and is caused by dysfunction of the vagus nerve causing paralysis or paresis of the vocal cord.4 The **gastrointestinal system** is also often compromised in infants with ACM. Dysfunction of the glossopharyngeal and vagus nerves causes “neurogenic dysphagia” which increases the risk of aspiration and failure to thrive.2,4,7 Changes to the **neuromuscular system** seen in infants with ACM include hypotonia, sensory deficits like dysesthesia, paresis or quadriparesis, nystagmus, spasticity, and bowel and bladder dysfunction.4,7 Changes in the **musculoskeletal system** include torticollis, dysmetria of eye movements, ataxia, and arm weakness.3

*Symptomatic ACM in Older children:* In older children with ACM the symptoms progress more gradually and are often less severe than that of infants. The most common symptoms include weakness of the arms and spasticity.4 Other changes that affect the **neuromuscular system** include ataxia of the arms and trunk and sensory impairments.4,7Additional changes in the **musculoskeletal system** include ataxia, disorders of eye movement, atrophy of hand muscles, changes in fine motor skills like handwriting, neck pain, and occipital headaches.7 Older children with ACM can also experience sleep disordered breathing secondary to brainstem abnormalities impacting the **cardiopulmonary system**.10,11

When hydrocephalus is present with ACM, which is the case about 70% of the time, symptoms are worsened and often become progressive.3,8,11 Hydrocephalus increases intracranial pressure and a ventriculoperitoneal (VP) shunt is often placed to reroute excess fluid. Shunt malfunction is often the cause of progressively worsening symptoms in children with ACM and shunt function should be checked with any change in symptoms.3,4 Hydrocephalus worsens symptoms of ACM when not well managed because the extra pressure acts to push the already ill positioned brainstem and cerebellar structures further downward.4 Some other conditions associated with ACM at any age that may be linked to progressive symptoms include tethered cord syndrome and syringomyelia.3,4,7,8 Tethered cord syndrome is characterized by tethering connections of the spinal cord to the surrounding tissues at the distal end of the cord and there is a theory that this traction on the spinal cord is a possible cause of ACM.1,6,12 Although this theory has been challenged and isn’t widely accepted, there is evidence that untethering of the spinal cord can relieve some “mild to moderate symptoms” of ACM.12 Syringomyelia involves a CSF-filled cyst called a syrinx forming adjacent to the spinal cord and as the cyst grows it can cause sensory loss, weakness, scoliosis, back pain, and lower motor neuron dysfunction.1,2,4

**Body Structure and Function Impairments:** The many changes to body systems described above can lead to a multitude of impairments. Some potential impairments are listed below.

* **Decreased arm strength 1,8**
* **Dysphagia (seen often in infants) 2,7,8,13**
* **Neck pain and headaches 13**
* **Respiratory issues such as stridor, apnea, and increased risk of aspiration (seen often in infants) 2,7-9**
* **Nystagmus, visual problems, involuntary eye movement 7-9,13**
* **Hearing loss or tinnitus 1,13,14**
* **Hypotonia 7.8**
* **Ataxia 7,8**
* **Dizziness, balance difficulties, poor coordination 1,13,14**
* **Dyesthesia in extremities 13**
* **Seizures 8**
* **In the presence of myelomeningocele, may see paralysis of the areas below the spinal cord opening.1**
* **Sleep disorders such as sleep apnea and REM sleep behavior disorder 11.15**
* **If syringomyelia is present, may see hand weakness, scoliosis, sensory impairment, spasticity, loss of bowel and bladder control, or paralysis if severe. 1,2,13**
* **Many of these symptoms can be worsened in the presence of hydrocephalus. 7**

**Activity Limitations and Participation Restrictions:**

 Due to it’s association with myelomeningocele, ACM can be diagnosed prenatally and it is most commonly diagnosed by early childhood. Many patients with ACM have an IQ within the normal range for their age and are able to function independently but this is not always the case.2,17 Some potential activity limitations for children with symptomatic ACM include: difficulty with feeding and respiration leading to failure to thrive, difficulty with self care skills, difficulty with fine motor skills, difficulty with tasks that require extended attention, limitations in functional mobility, and limitations in age-appropriate play skills.1,4,13,16 Due to these potential activity limitations, children with ACM may have restrictions in their ability to participate. Some possible participation restrictions include: independence with self care and ADLs, certain recreational and leisure activities, education, social interactions, and certain community outings.11,16,17 Decreased arm strength, mobility, and coordination as well as visual issues and dizziness may limit their participation in certain recreational sports and leisure activities.11 Restrictions in the educational setting may be due to trouble with fine motor skills, attention deficit, cognitive deficits, and limitations in functional mobility and self care.11,17 Henriques Filho and Pratesi found a link between sleep disordered breathing and attention deficit in children with ACM while Vinck studied the effects of ACM as well as hydrocephalus on cognition.11, 17 Vinck et al found that children with ACM performed poorly on cognitive tasks that required “visual analysis and synthesis, verbal memory, and verbal fluency.”17 Restrictions in social interactions may stem from potential hearing loss as well as these children being seen as different from their typically developing peers. The level of restriction in community outings depends on each child’s level of functional mobility and the presence and effects of myelomeningocele or syringomyelia.11

**Interventions:**

 The evidence is limited in regards to interventions and approaches specifically for children with ACM but there is extensive evidence regarding children with myelomeningocele and hydrocephalus as well as the impairments created by ACM. I chose to focus my search more on the treatment of vestibular issues such as nystagmus, dizziness, and poor coordination in pediatric patients like those with ACM. Vestibular rehabilitation therapy in children is similar to that of adults but activities are modified to be fun and playful to maintain the motivation and attention of pediatric patients. The major difference in treating vestibular impairments in pediatric patients lies in the ability to examine and assess for the self-reported symptoms.18 In screening of these patients for vestibular issues, physical therapists can look for the presence of behaviors such as the child holding on their caregiver, refusing to stand up, and wanting to be held as well as the observations of difficulty with postural control and balance during play and gait.18 Interventions should focus on fun ways to work on gaze stabilization, habituation, adaptation, balance, substitution, and improving motor skills.18,19 In their 2003 study, Rine et al studied the effects of an exercise based intervention to improve postural control on the motor development and mobility of children with sensorineural hearing loss and vestibular dysfunction.20 They found that an exercise intervention that involved activities focused on improving eye hand coordination, visual motor ability, balance, visual awareness, and somatosensory awareness improved the postural control, sensory organization ability, and motor development of participants after 12 weeks.20 The literature highlights that when working with pediatric patients on vestibular impairments, it’s important to match interventions to the child’s level of cognition as well as use toys and games that interest them.18 Using toys and games that interest the child to work on gaze stabilization as well as swings to add body movement can help keep children ready to cooperate and give their best effort in therapy.18 These activities can also be made educational by having patients identify letters or read books to work on their visual stabilization. The American Physical Therapy Association even offered a continuing education course last year on pediatric vestibular rehabilitation and has plans to offer another in December this year.21 Clinically I haven’t had much experience with pediatric vestibular impairments but what I’ve learned from the evidence is that it’s possible to adapt vestibular rehabilitation interventions used for adults into fun games and activities for pediatric patients.

**Sources:**

1. **National Institute of Neurological Disorders and Stroke. Chiari Malformation Fact Sheet. National Institute of Neurological Disorders and Stroke website.** <https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Fact-Sheets/Chiari-Malformation-Fact-Sheet>**. July 27, 2017. Accessed February 28, 2018.**
2. Khoury C. Chiari malformations. UpToDate website. <https://www.uptodate.com/contents/chiari-malformations>. September 27, 2017. Accessed March 1, 2018.
3. Salman MS. Posterior fossa decompression and the cerebellum in Chiari type II malformation: a preliminary MRI study. *Childs Nerv Syst* 2011;27(3):457-462. doi:10.1007/s00381-010-1359-8.
4. Stevenson KL. Chiari Type II malformation: past, present, and future. *Neurosurg. Focus* 2004;16(2):E5.
5. Shaer CM, Chescheir N, Schulkin J. Myelomeningocele: a review of the epidemiology, genetics, risk factors for conception, prenatal diagnosis, and prognosis for affected individuals. *Obstet. Gynecol. Surv.* 2007;62(7):471-479. doi:10.1097/01.ogx.0000268628.82123.90.
6. Cincinnati Children’s Hospital. Chiari Malformation. Cincinnati Children’s Hospital website. <https://www.cincinnatichildrens.org/health/c/chiari-malformation>. July 2016. Accessed February 28, 2018.
7. Poretti A, Boltshauser E, Huisman TAGM. Chiari malformations and syringohydromyelia in children. *Semin. Ultrasound CT MR* 2016;37(2):129-142. doi:10.1053/j.sult.2015.12.001.
8. McLone DG, Dias MS. The Chiari II malformation: cause and impact. *Childs Nerv Syst* 2003;19(7-8):540-550. doi:10.1007/s00381-003-0792-3.
9. American Association of Neurological Surgeons. Chiari Malformation. American Association of Neurological Surgeons website. <http://www.aans.org/Patients/Neurosurgical-Conditions-and-Treatments/Chiari-Malformation>. Accessed February 28, 2018.
10. Alsaadi MM, Iqbal SM, Elgamal EA, Gozal D. Sleep-disordered breathing in children with Chiari malformation type II and myelomeningocele. *Pediatr. Int.* 2012;54(5):623-626. doi:10.1111/j.1442-200X.2012.03660.x.
11. Henriques Filho PSA, Pratesi R. Sleep disorder: a possible cause of attention deficit in children and adolescents with Chiari malformation type II. *Arq Neuropsiquiatr* 2009;67(1):29-34.
12. Mehta VA, Bettegowda C, Amin A, El-Gassim M, Jallo G, Ahn ES. Impact of tethered cord release on symptoms of Chiari II malformation in children born with a myelomeningocele. *Childs Nerv Syst* 2011;27(6):975-978. doi:10.1007/s00381-010-1294-8.
13. Physiopedia. Arnold Chiari Malformation. Physiopedia website. <https://www.physio-pedia.com/Arnold_Chiari_Malformation>. June 6, 2017. Accessed March 1, 2018.
14. National Organization for Rare Disorders. Chiari Malformations. National Organization for Rare Disorders website. <https://rarediseases.org/rare-diseases/chiari-malformations/>. 2014. Accessed March 1, 2018.
15. Dauvilliers Y, Stal V, Abril B, et al. Chiari malformation and sleep related breathing disorders. *J. Neurol. Neurosurg. Psychiatr.* 2007;78(12):1344-1348. doi:10.1136/jnnp.2006.108779.
16. Meeker J, Amerine J, Kropp D, Chyatte M, Fischbein R. The impact of Chiari malformation on daily activities: A report from the national Conquer Chiari Patient Registry database. *Disabil. Health J.* 2015;8(4):521-526. doi:10.1016/j.dhjo.2015.01.003.
17. Vinck A, Maassen B, Mullaart R, Rotteveel J. Arnold-Chiari-II malformation and cognitive functioning in spina bifida. *J. Neurol. Neurosurg. Psychiatr.* 2006;77(9):1083-1086. doi:10.1136/jnnp.2005.075887.
18. Rine RM, Wiener-Vacher S. Evaluation and treatment of vestibular dysfunction in children. *NeuroRehabilitation* 2013;32(3):507-518. doi:10.3233/NRE-130873.
19. Medeiros IRT, Bittar RSM, Pedalini MEB, Lorenzi MC, Formigoni LG, Bento RF. Vestibular rehabilitation therapy in children. *Otol. Neurotol.* 2005;26(4):699-703.
20. Rine RM, Braswell J, Fisher D, Joyce K, Kalar K, Shaffer M. Improvement of motor development and postural control following intervention in children with sensorineural hearing loss and vestibular impairment. *Int. J. Pediatr. Otorhinolaryngol.* 2004;68(9):1141-1148. doi:10.1016/j.ijporl.2004.04.007.
21. American Physical Therapy Association. Pediatric Vestibular Rehabilitation: A Competency Based Course. <http://www.apta.org/Courses/2017/10/PediatricVestibularRehab/>. May 19, 2017. Accessed March 17, 2018.