Dandy Walker Malformation (DWM) is a congenital malformation of the cerebellum, posterior fossa, and the fourth ventricle that can physiologically present in different forms or variants.1,2 DWM is caused by abnormalities in the development of the cerebellum when a child is an embryo and genetic abnormalities have been associated with DWM.1 Due to the variation of DWM presentation, there isn’t one specific combination of genetic abnormalities and environmental factors that is known to cause DWM but some genetic causes of DWM include a deletion of chromosome 3q24.3, deletion of chromosome 6p25, deletion of chromosome 13q32.2-q33.2, or duplication of chromosome 9p.1 Chromosome 3q24.3 is where the first genes for DWM were found to be located and these genes are ZIC1 and ZIC4.1 In their 2015 retrospective study of the National Birth Defects Prevention Study, Reeder et al noted that it’s not clear how many cases of DWM are caused by just genetic abnormalities so they decided to look into environmental factors associated with DWM diagnoses.2 They found that a maternal history of infertility and non-Hispanic black ethnicity were associated with an increased risk of DWM and concluded that more research would be beneficial to “uncover potentially modifiable risk factors” in an effort to prevent DWM.2 DWM is also not always seen in isolation; it can be a part of other genetic disorders and present along with other birth defects like heart defects and sternal defects.1 Some other potential causes of DWM that have been studied include maternal viral infections passed on to the embryo, embryonic exposure to toxins or medications, and the presence of diabetes in the mother.2,3

The classic form of DWM has three key pathological features that impact function: 1) a small, abnormal, or absent vermis; 2) a large cyst in the fourth ventricle causing it to be dilated; and 3) an enlarged posterior fossa of the skull.1,2,3 The cerebellum helps to coordinate movements with help from sensory information, plays an important role in motor learning of skilled movements, and it helps to coordinate posture, equilibrium, and eye movements.4 Neuroanatomical evidence has also shown that the cerebellum plays a role in cognition and emotional behavior and in DWM the malformation of the cerebellum an lead to developmental issues due to it’s role in cognition and behavior.5 The vermis specifically receives sensory information from proximal muscles to coordinate movements and works to coordinate eye movements.4 The abnormality, decreased size, or absence of the vermis in DWM impairs the cerebellum’s ability to coordinate movement and regulate eye movement. The cyst and dilation of fourth ventricle can lead to cerebrospinal fluid blockage and buildup that in turn can cause hydrocephalus and increased intracranial pressure (ICP).1,3,6 Cerebrospinal fluid is supposed to flow through the ventricles and subarachnoid spaces to protect the brain and the excess is normally absorbed back into the brain’s circulation to maintain optimal intracranial pressure.6 The posterior fossa of the skull houses the cerebellum and the brainstem and in DWM this fossa enlargement can involve the presence of a cyst in the fossa.7 The presence of a cyst in the posterior fossa can cause issues similar to that of a posterior fossa tumor such as brain stem compression and herniation.7,8

The variation in how DWM can present creates an array of possible clinical manifestations of the disorder that can negatively impact activity and participation. The abnormalities of the cerebellum, fourth ventricle, and posterior fossa can cause ataxia, poor muscle tone, poor balance, developmental delays of motor function, language, and cognition, and issues with eye movement like nystagmus.1,3,5,9 When hydrocephalus is present with DWM it can also cause neurological impairments, seizures, irritability, vomiting, poor feeding, abnormal head growth, lethargy, and in really severe cases respiratory failure.1,3,10 Patients with severe hydrocephalus are often treated with a ventriculoperitoneal shunt to help drain excess CSF.3 As infants, poor feeding can weaken the emotional bond between mother and child and create a more stressful environment for new parents.11 Seizures, irritability, lethargy, and poor feeding can limit a child’s ability to explore their environment when they’re young and their ability to participate in social activities like family outings or school trips as they age. Difficulties with language are often present in patients with DWM as a product of their developmental delay and this can negatively influence social interaction with family and friends as well. Cerebellar cognitive affective syndrome has been seen in conjunction with cases of DWM.12 Cerebellar cognitive affective syndrome is caused by damage to the neural circuitry in the vermis and the posterior cerebellum.13 The syndrome is characterized by difficulty with language, impulsivity, attention difficulties, and disinhibition and these issues can limit social interactions and independence as patients with DWM age.13 Cognitive delays and cerebellar cognitive affective syndrome can also limit participation in regular education classes and require special education or educational modifications for a child.

The DWM impairments that can be addressed specifically by physical therapy include ataxia, low muscle tone, increased muscle tone, poor balance, and delays in motor function. All of these impairments limit a child’s ability to achieve motor milestones when they should. The cerebellum also plays a major role in learning new skilled movements and achieving motor milestones requires just that.4 The inability to coordinate movements like rolling or sitting when appropriate can lead to muscle weakness due to less use. As children with DWM age these impairments can be an impediment to independent ambulation and functional mobility. Difficulty learning new skills can cause patients with DWM to avoid new activities and therefore lead to decreased strength and endurance. This motor learning deficit along with poor muscle tone, poor balance, and ataxia can also limit participation in physical activity and negatively impact overall physical health in children of all ages with DWM.

There is little to no evidence available regarding specific physical therapy interventions to help improve the functional mobility of patients with DWM but physical therapy can help these patients achieve motor milestones, improve strength, improve coordination, and learn new motor skills. In their 2017 case report, Ipek, Akyolcu and Bayar discuss the physical therapy and rehabilitation of a young girl with Joubert Syndrome and an improvement in her motor function was noted after a year of physical therapy based in the theory of neurodevelopmental treatment.14 Joubert syndrome is similar to DWM in that it’s a rare genetic disorder characterized by “brain malformation, hypotonia, breathing abnormalities, ataxia, oculomotor apraxia, and developmental delay” as well as decreased size of the vermis.14 Neurodevelopmental Treatment (NDT) can be defined as a “holistic and interdisciplinary clinical practice model informed by current and evolving research that emphasizes individualized therapeutic handling based on movement analysis” for the treatment of individuals with neurological disorders.15 The presentation of children with DWM can vary greatly so the individualized handling utilized in NDT can facilitate motor milestone achievement. The framework of NDT doesn’t have to be limited to time in the physical therapy clinic; parents and caregivers can be educated on ways to handle their child in play at home to further improve their motor function. *Positioning for Play: Interactive Activities to Enhance Movement and Sensory Exploration* offers some great, easy to understand positions for families to try at home with their children. This manual was designed for use with children who have developmental delays and details ways for caregivers to help children improve in many different developmental positions.16 Another physical therapy intervention that can help children and youth with DWM is body weight support (BWS) on the treadmill and over ground for gait training. In their 2008 case report, Cernak et al described the effect that BWS locomotor treadmill training for 4 weeks followed by 4 months of home BWS training had on a 13-year-old non-ambulatory girl with severe cerebellar ataxia.17 After 6 months, the girl was able to walk 152 meters with a U-walker and supervision and she had also developed the strength to transfer independently.17 Children with DWM have difficulty learning new motor skills so physical therapy as the become adolescents can also focus on helping them learn new skills that interest them and help them be physically active. Including task-specific training for skills of interest has been shown to improve motor performance and participation of adolescents with developmental coordination disorder (DCD) and DCD is similar to DWM in that it involves coordination impairments that interfere with motor function.18 I think that task-specific training can also help youth with DWM improve their ability to participate in extracurricular activities and social interactions while also helping them increase their strength and endurance.

Because of their difficulties with coordination, children and youth with DWM may be reluctant to participate in physical activity or sports for leisure. I would recommend Special Olympics or any recreational sporting activity to children with DWM to improve their chances of getting and staying physically active. The Special Olympics can provide an environment for physical activity to take place, a support system for those affected and their families, and a space for increased social interaction with children of varying abilities. For informational and support resources, I would encourage families to get connected with the Dandy-Walker Alliance, the Hydrocephalus Association, and the National Organization for Rare Disorders (NORD).1,3,19 Among many other things the NORD has assistance programs to help patients with medical care.1 The Dandy-Walker Alliance has created a Child Information Form for parents and caregivers to use to inform teachers about their child’s diagnosis and their specific needs.3 The Hydrocephalus Association has a peer support hotline that can educate and provide social support.19 These organizations are great resources because they advocate for research, provide informational resources, and provide support groups and forums to children and families dealing with DWM.

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