Hypotonia, Jaundice, and Chiari Malformations: Relationships to Executive Functions

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Hypotonia, Jaundice, and Chiari Malformations: Relationships to Executive Functions

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This article postulates that movement and action control are the underpinning of executive functioning. We selectively examine brain regions that have traditionally been almost exclusively understood as critical to the control and expression of movement—namely, the basal ganglia and the cerebellum. We first describe the relationship between movement and cognition. This is followed by a review of common developmental disorders that are known to exhibit abnormal executive functions and movement anomalies. Against that background, we examine hypotonia, neonatal jaundice, and Chiari I malformation, and we demonstrate why these are “at-risk” factors for neurodevelopmental disorders that can feature both motor control and executive function abnormalities. Our goal is to prepare the clinical neuropsychologist for gathering information about these features of a child’s birth and developmental histories, while using this as a framework for interpreting test results and applying test data in a useful, practical way to guide descriptive diagnosis and treatment.

Key words: Chiari I malformation, executive functioning, hypotonia, neonatal jaundice

INTRODUCTION

This selective review article examines hypotonia, neonatal jaundice, and Chiari I malformation as “at-risk” factors for neurodevelopmental disorders with specific emphasis on deficits in executive functions. Upon initial thought, these three issues appear to have very little in common. However, they are all linked together because of their association with possible deficits in brain regions involved in the control of movement. The control of movement, or action control, is a critical underpinning for the development of executive function (see Koziol & Lutz, 2013). According to Von Hofsten (2004, 2007, 2009), all movement in children is purposive and goal-directed action that foresees events in the world and stresses the critical importance of anticipation, a fundamental aspect of executive function. It has been proposed that the brain did not develop for the purpose of thinking or cognition (Stout, 2010). Instead, the functional architecture of the brain evolved and developed to meet the needs of interactive behavior (Cisek & Kalaska, 2010). Pezzulo (2011) proposed that all knowledge, both procedural and declarative, is grounded in sensorimotor interaction. The needs of interactive behavior required the evolving and developing brain to select for anticipatory control mechanisms to acquire purposeful movements and procedures. This procedural knowledge, when simulated, imagined, or in other words, when reflected upon or thought about, can lead to the development of declarative knowledge. The internal manipulation of this information can generate executive function. Koziol, Budding, and Chidekel (2012) significantly expanded upon this viewpoint and defined the putative neuroanatomic underpinnings of this process. They reviewed specific cortico-cortical, cortical-basal...
ganglia, and cerebro-cerebellar interactions as critical supporting systems for cognition while illustrating how cognition develops from the motor system in order to control it. At this time, there appears to be overwhelming evidence to support the suggestion made by Diamond (2000) that cognitive and motor systems are not separate as has been traditionally thought, as all adaptive behavior is dependent upon the interactions of frontal-striatal and cerebro-cerebellar ensembles.

There is a very substantial literature (well beyond the scope of this selected review) that relates childhood motor development to cognition and executive functioning. For example, the infant sucking reflex, an early index of action control, has been described as predictive of neurodevelopmental outcomes. Early sucking and feeding, and the later development of speech and language, all involve brainstem, subcortical, and cortical neural networks that overlap and interact and that later support executive control (Poore & Barlow, 2009). Roebers and Kauer (2009) reported that cognitive and motor control were significantly interrelated in a normative sample of young school-aged children, while the ability to respond to the speed and accuracy demands of all the tasks they administered constituted an important aspect of executive functioning. Piek, Dawson, Smith, and Gasson (2008) and Durston, van Belle, and de Zeeuw (2011) investigated the developmental trajectory between motor behavior and cognitive ability and demonstrated a predictive relationship between gross motor control and the later development of “processing speed” and “working memory” as assessed by the Wechsler Intelligence Scale for Children-Fourth Edition (WISC-IV).

Abnormalities in motor development have been consistently related to a range of neurodevelopmental disorders. Michel, Roethlisberger, Neuenschwander, and Roebers (2011) demonstrated a very notable stability of motor coordination impairments and persistent “executive functioning” deficits in 5- to 7-year-old children. Developmental coordination disorder (DCD) seldom occurs in isolation and is frequently comorbid with attention-deficit hyperactivity disorder (ADHD; Barkley, 2006). It has been reported that as many as half of all children diagnosed with ADHD have poor motor coordination and fulfill diagnostic criteria for DCD (Barkley, DuPaul, & McMurray, 1990; Piek, Pitcher, & Hay, 1999; Pitcher, Piek, & Hay, 2003). In addition, DCD is frequently associated with the cerebellar cognitive affective syndrome (CCAS) described by Schmahmann (2004) and Marien, Wackenier, De Surgeloose, De Deyn, and Verhoeven (2010). CCAS is characterized by deficits in cognition, specifically within the areas of attention, executive functioning, and language, as well as problems with affective regulation. Cerebro-cerebellar networks have been consistently implicated in DCD and ADHD (Ashtari et al., 2005; Bledsoe, Semrud-Clikeman, & Pliszka, 2011; Mackie et al., 2007; Marien et al., 2010; Pasini, D’Agati, Pitziante, Casarelli, & Curatolo, 2012; Zwicker, Missiuna, Harris, & Boyd, 2010). Goulardins, Marques, Casella, Nascimento, and Oliveira (2012) found that children diagnosed with ADHD-Combined type differed in comparison with their typically developing peers on a wide variety of motor parameters. Anxiety disorders in children, independent of comorbidity with ADHD, also demonstrate motor impairment upon formal motor system evaluation (Skirbekk, Hansen, Oerbeck, Wentzel-Larsen, & Kristensen, 2012). In another investigation, teachers used a motor observation checklist to identify students with ADHD and autism relative to normal controls (Efstratopoulou, Janssen, & Simons, 2012). Children with autism and Asperger syndrome consistently exhibit motor impairment (Green et al., 2009; Mostofsky et al., 2009; Shetreat-Klein, Shinnar, & Rapin, 2012). Acknowledging a variety of motor abnormalities in these two disorders, Nayate, Bradshaw, and Rinehart (2005) were struck by the abnormalities in gait within the autism spectrum disorder (ASD) patient populations. They explained these motor anomalies were a manifestation of disruptions within basal ganglia and/or cerebellar systems and proposed that these diagnoses might even be classified as movement disorders. Nicolson, Fawcett, and Dean (2001) and Nicolson and Fawcett (2007) have documented the relationships between motor control and specific learning disabilities. Therefore, the relationship between movement and cognition is undeniable.

HYPOTONIA

Hypotonia is defined as a state of low muscle tone (A.D.A.M. Medical Encyclopedia, n.d.). However, hypotonia is not a specific diagnosis or disease. Instead, it is a symptom that can arise from a variety of etiologies. For example, there are numerous congenital and/or genetic disorders as well as infections, toxins, metabolic and autoimmunity disorders, and neurological conditions that can generate the symptom of hypotonia. Attempting to diagnose the underlying cause is often difficult and sometimes unsuccessful (Harris, 2008). There is a group of infants in which hypotonia appears idiopathic. Idiopathic hypotonia is not well researched and not well understood (Strubhar, Meranda, & Morgan, 2007). Hypotonia occurs in degrees and can be classified as transient, mild, moderate, or severe. The term “floppy infant syndrome” is often used when low muscle tone is noticed in infancy. When these babies are held, they give the impression of “ragdolls.” They seem limp, head movements are often ataxic-like, they often have difficulties in “latching on” when mothers attempt to feed them, and because their mouth muscles are weak and poorly controlled, they can demonstrate a disrupted suck–swallow
pattern or cycle, which we have already identified above as “at-risk” factors for an increased likelihood of neurodevelopmental, cognitive disorders. Early initial symptoms of hypotonia can include difficulties or lateness in lifting the head while lying on the stomach, problems in “rolling over,” and problems in sitting unassisted. Problems with balance, crawling, and walking can all be manifestations of low muscle tone. Problems in following movement with the eyes, pointing toward objects, grasping toys, switching an object from one hand to another, and self-feeding are issues that demonstrate problems in both gross- and fine-motor control. This makes good anatomic sense because aspects of these artificial distinctions are mediated by combinations of the four motor tracts of the human body (Kolb & Whishaw, 2008). Children are often discovered to be hypotonic at around the age of 2 years because of delays in these different areas of motor development. However, low muscle tone might be first noticed even later—for example, at school age when a child might be experiencing difficulties with printing or handwriting. To our knowledge, there are no reliable estimates on these seemingly serendipitous later-onset identifications of hypotonia.

Strubhar et al. (2007) used questionnaires in a retrospective study to examine outcomes of mild and globally impaired hypotonia. In the minimally impaired group, 59% were described as poorly coordinated, and 35% were characterized as clumsy walkers. Almost 60% were described as having difficulties with handwriting, and 70% had difficulties with math. Although the percentage of children was not specified, most were described as having learning problems. In this regard, while 58% were described as inattentive and 53% were termed distractible, 20% had a diagnosis of ADHD and 56% were diagnosed with a language disorder. In a 5-year follow-up investigation, Chaudhari and colleagues (2010) found persistent language impairment continuing through the age of 5 years. Pediatricians typically make referrals to occupational and/or physical therapists for a systematic motor evaluation to identify low muscle tone. Therefore, neuropsychologists are usually not the first referral of clinical choice. This underscores the need for the clinical neuropsychologist to routinely ask questions about “floppy” in infancy, hypotonia, low muscle tone, and early participation in occupational and/or physical therapy and for what reasons, during the course of the developmental clinical interview with parents. The answers to these questions can provide important clues upon referral for neuropsychological evaluation.

Hypotonia and the Cerebellum

The cerebellum is believed to be the most common driver of hypotonia. However, as summarized by Njokiktjien (2010), functional neuroimaging studies reveal a “crossed”

neocortical-cerebellar diaschisis. This occurs through the reciprocal connections of the cerebro-cerebellar circuitry system (see Schmahmann and Pandya, 1997a, for a review of the cerebro-cerebellar circuitry system; for a very brief summary, see Koziol & Lutz, 2012). For example, with cerebral involvement, there is a depression of activity within the cerebellum; with cerebellar involvement, there is a corresponding reduction of cerebral activity or recruitment. The vestibulocerebellum is presumably mature at the time of full-term birth; throughout the course of development, the cerebro-cerebellar circuitry system very closely resembles what is observed in adults (De Quiros & Schrager, 1979; Power, Fair, Schlagger, & Petersen, 2010; Rorke & Riggs, 1969). The cerebellum already exhibits its well-established neuroanatomic architecture at the time of birth (Altman & Bayer, 1997). Between the ages of 7 and 11 years old, adult volume can be already achieved, with peak size usually reached by approximately 15 years in males and by about 11 years in females (Giedd & Rapoport, 2010; Power et al.). However, preterm birth and its frequent co-occurrence with low birth weight and/or jaundice (to be discussed in the next section of this article) is often associated with structural abnormalities within the cerebellum, sometimes within focal regions (Limperopoulos et al., 2005; Messerschmidt et al., 2005; Petrini et al., 2009; Riva, Usilla, Saletti, Esposito, & Bulgheroni, 2011). Broadly speaking, the developmental program of the cerebellum can be disrupted by preterm delivery (Haldipur et al., 2011).

The vestibulocerebellum is important for postural control, and the vermis is important for “elementary” motor functions such as aspects of tone, coordination, equilibrium and balance, and the “quantity” of motor activity, such as hypokinesias and hyperkinesias (Njokiktjien, 2010). The lateral regions of the neocerebellum are critical for the learning of new procedures and the acquisition of automaticity (Imamizu, Kuroda, Miyauchi, Yoshiocka, & Kawato, 2003; Molinari et al., 1997). Improvement in performance is a manifestation of motor (and cognitive) adaptation, which is mediated through the cerebro-cerebellar circuitry system, while the long-term retention of motor (and cognitive, or thought) sequences is retained within the cerebral cortex (Galea, Vazquez, Pasricha, Orban de Xivry, & Celnik, 2010). Therefore, the cerebellar pathology that generates hypotonia could easily be the cause of slow learning, especially those actions that require quick adaptation to environmental changes, because there would be less “transfer” of a presumably automated action to the same action within other somewhat different yet similar circumstances. These functions help explain many of the findings of Strubhar et al. (2007), described earlier, including the inattention, distractibility, and failures in adaptation, which are aspects of executive control. Similarly, these functions can serve to explain the attention, executive function, and
language disturbances often characteristic of the CCAS. While hypotonia can be a cause of developmental dyspraxia (Njioiktjien), this is also consistent with reviews of DCD (Green, Baird, & Sugden, 2006; Marien et al., 2010; Zwicker, Missiuna, & Boyd, 2009; Zwicker et al., 2010).

We have emphasized that both procedural and declarative knowledge are grounded in action or behavior. Because hypotonic children are less active, they interact less with their environment. This means they have less experience with sensorimotor interaction than their peers. Similarly, when interacting, they perform and learn more slowly. Based upon these two features of the behavior profile of hypotonic children, it would be predicted that the “off-line” simulation, imagination, or reflection about behavior would be a more restricted process than it is for typically developing children. This would negatively impact upon the development of executive functioning within this population. This group of children might learn more by observing others instead of acting on their own. How this might affect the quality of executive function development remains to be studied.

### JAUNDICE

Neonatal jaundice is characterized by elevations in bilirubin levels. Severe cases of bilirubin neurotoxicity are associated with the development of the full Kernicterus syndrome, which can be fatal. These cases are now relatively rare since treatment guidelines for hyperbilirubinemia have been implemented (Hansen, 2011). Nevertheless, neonatal jaundice in varying degrees of severity remains frequent and is estimated to occur in approximately 60% of births (Shapiro, 2003). As recently reviewed and summarized by Koziol, Budding, and Chidekel (2013), moderately elevated levels of bilirubin in otherwise healthy infants are often associated with movement abnormalities, and this raises concern as to whether or not these anomalies are associated with the later development of learning, attention, and executive function deficits. Voeller (2004) considers jaundice an “at-risk” factor for the subsequent development of ADHD. A relationship between elevated serum bilirubin levels and hearing impairment has been established (De Vries, Lary, Whitelaw, & Dubowitz, 1987; Shapiro & Popelka, 2011). There is a growing clinical literature that relates hyperbilirubinemia with neurodevelopmental problems such as ADHD, ASD, central auditory processing disorders (CAPDs), nonspecific, generalized learning difficulties, and nonprogressive developmental delays (Johnson & Bhutani, 2011; Shapiro; Soorani-Lunsing, Woltl, & Hadders-Algra, 2001). These can be considered partial Kernicterus syndromes, but they are more often referred to as bilirubin-induced neurologic dysfunction, or BIND (Johnson & Bhutani).

### The Neuropathology of Jaundice

The neurotoxicity of bilirubin has a particular affinity for certain brain regions. The globus pallidus (both the internal and external segments, or GPi and GPe, respectively) and the subthalamic nucleus can be affected (Johnston & Hoon, 2000). These regions are sometimes referred to as “intermediate structures” of the basal ganglia because they project to other structures within the basal ganglia (Middleton, 2003). Hyperbilirubinemia can also affect the vermis, the dentate nucleus, and the Purkinje cells of the cerebellum, while certain regions of the hippocampus can similarly be involved (Connolly & Volpe, 1990; Shapiro, 2003; Soorani-Lunsing et al., 2001; Turkel, 1990; Volpe, 2001). Although reviewing all of the possible, multiple mechanisms responsible for this pathology is well beyond the scope of this article, one hypothetical reason concerns the fact that some of these brain regions are spontaneously active. Therefore, they require more oxygenated blood perfusion to maintain activity so that bilirubin might have a differential toxic affect within these areas. These same studies cited earlier have also reported brainstem abnormalities, including the oculomotor and cochlear nuclei, and the vestibular nuclei of the cerebellum, which receives projections from regions of the auditory system. These regions of involvement correlate with the visual gaze and oculomotor abnormalities observed in Kernicterus, as well as with the increased incidence of CAPD that can be observed in BIND (De Vries et al., 1987; Rance et al., 1999; Vohr et al., 1990).

### Simplified Review of Basal Ganglia Circuitry and Functions

Alexander, DeLong, and Strick (1986) initially identified the five prototypical frontal-striatal (basal ganglia) circuits, and Middleton and Strick (2001) later added two prototypical sensory-perceptual circuits reciprocally connecting the temporal and parietal lobes with the basal ganglia system. All of these circuits are topographically organized and segregated, and they function as parallel processes, so that to understand the operational mechanisms of one circuit allows for an understanding of all connectivity profiles. How to translate motor behavior to cognitive, motivational, and affective analogues has previously been described by Koziol and Budding (2009). In a meta-analytic review of functional magnetic resonance imaging (fMRI) studies, Arsalidou, Duerden, and Taylor (2012) generated a comprehensive “topographical map” of how body and eye movements, working memory and planning functions, emotional and reward processes, and somatosensory functions were regionally represented.

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within the basal ganglia. The segregated operations of these circuitry profiles explain how attention and action/behavioral selection become highly focused and maintained. There are four additional integrative networks of the basal ganglia that explain how information flows between circuits, which is necessary for modifying previously learned behaviors and for the development, acquisition, and implementation of new behaviors (Haber, 2010, 2011; Haber & Calzavara, 2009; Yin, Ostlund, & Balleine, 2008). Two of these integrative networks are of relevance to our discussion because they feature the GPi and GPe as network “hubs.” (The reader who is unfamiliar with the functional neuroanatomy of the basal ganglia is referred to Utter & Basso, 2008, or Middleton, 2003.)

Because the basal ganglia are key players in modulating such a wide range of functions, it can be tempting to conclude that they subsume multiple, diverse roles. However, the basal ganglia can currently be best understood as serving a unified, integrative role (Cockburn & Frank, 2011). The basal ganglia are an inhibitory system that dynamically and adaptively selects or “gates” the flow of information through its cortical-striatal-pallidal-thalamic-cortical loops. Broadly speaking, activity within the direct pathway releases behavior, and activity within the indirect pathway inhibits behavior. This gating function is essentially a reinforcement or instrumental learning system. Activity within the direct pathway always leads to positive reinforcement, associations, and the performance of the behavior in question; activity within the indirect pathway always leads to negative outcomes and their avoidance. The neural systems governing these behaviors that are strongly and repeatedly co-activated are strengthened. According to stimulus context and generalizability, the basal ganglia release those actions that have a high probability of generating a positive outcome; they avoid selecting actions that have been associated with negative outcomes. So the frontal-striatal system learns what to do as well as what it should not do, which of course, depends upon the stimulus context in question. Therefore, on the basis of selective damage to the globus pallidus, which influences behavioral release by modulating thalamic activity, it would be predicted that focused and sustained attention and behavior would be affected, as would the learning of new procedures and “habits.” Activation of the subthalamic nucleus (STN), sometimes referred to as the “hub” of the hyperdirect pathway, immediately stops all behavior (Aron & Poldrack, 2006; Frank, 2006). This pathway has been implicated in impulse control disorders. Broadly speaking, based upon these functions, one would predict that neurotoxicity within these basal ganglia regions would be associated with many of the symptoms of ADHD and conduct disorder.

The basal ganglia gate cognition the same way they select behavior—on the basis of activity within the direct and indirect pathways. For example, within the executive function of working memory, which is essentially cognitive control, the temporary storage of information to guide behavior is supported by prefrontal-parietal circuitry (Derrfuss, Brass, & Yves von Cramon, 2004). Cortical-basal ganglia interactions through the direct and indirect pathways literally “let in” task-relevant information while “keeping out” distracting information, which allows for appropriate information maintenance, manipulation, and updating (Aron & Poldrack, 2006; Awh & Vogel, 2008; Frank, Loughry, & O’Reilly, 2001; McNab & Klingberg, 2008). Because pallidal activity is critical to the proper functioning of these pathways, deficits in executive control would be predicted on the basis of bilirubin neurotoxicity. Although a working-memory deficit might easily occur in the absence of a diagnosis of ADHD, it also might not even be detected in early school-aged children and might not be noticed until cognitive demands increase as in “middle school” (Denckla & Reader, 1993; Hazy, Frank, & O’Reilly, 2006). While the hippocampus can also be affected by bilirubin neurotoxicity, its role in the formation of declarative and episodic memory formation is well established (Squire, Stark, & Reader, 1993; Hazy, Frank, & O’Reilly, 2006). This has obvious implications for a child’s performance in any situation that requires the learning, retention, and recollection of factual knowledge (for a comprehensive review of hyperbilirubinemia and cognitive dysfunction, see Koziol et al., 2013).

**CHIARI I MALFORMATION**

Chiari malformations represent a group of disorders described by the Viennese physician Hans Chiari in 1891 (Loukas et al., 2008). Chiari malformations are structural defects in the brain, identified when part of the cerebellum is located below the foramen magnum (Kumar, Chumas, Peckham, Guthrie, & Murray, 2010). Chiari malformations are usually classified as falling within one of four categories (labeled Types I to IV). Classification depends upon the degree of displacement and the etiology of the malformation as originally proposed by Chiari and as reviewed by Loukas et al., who cite Chiari’s paper describing these conditions, which was written in his native language. Chiari I malformation, presumably and usually the most benign form of the condition, is identified when there is a cerebellar tonsil herniation at least 5 mm below the plane of the foramen magnum (Saletti et al., 2011). On occasion, the brainstem is involved in this malformation and syringomyelia is frequently associated (Elster & Chen, 1992). Approximately 59% of pediatric cases are asymptomatic as reported by Saletti et al. Presentations are considered heterogeneous, and most investigations of the condition concern neurosurgical studies (Brockmeyer, 2011). With the development and
proliferation of neuroimaging techniques, it is believed that asymptomatic forms of the condition are identified more frequently, even throughout adulthood (Massimi, Caldarelli, Frassanito, & Di Rocco, 2011). The diagnosis has become controversial at least in part because there is no correlation between the degree of herniation and the level of disability of the presenting patients (Labuda, Loth, & Slavin, 2011). The most common presenting symptom, occurring nearly 70% of the time in children and adults, is a headache within the occipital region and/or neck pain (Saletti et al.). However, Chiari can present with an extremely wide variety of symptoms. These symptoms can be generated by the cerebellum, from the spinal cord, the cranial nerves, and/or because of pressure dynamics of the cranium and spinal column (Luciano, 2011). This range of symptoms can also seem nonspecific. The diagnostic methodology and the treatment approaches for Chiari I malformations are varied and controversial (Tubbs, Lyerly, Loukas, Shoja, & Oakes, 2007). Currently available data on cognitive and behavioral disorders in Chiari I malformation are considered inconclusive (Riva et al., 2011).

Koziol and Budding (2009) presented a case of a 13-year-old boy with a Chiari I malformation. This patient’s presentation featured herniation of the right cerebellar tonsil as well as a posterior fossa arachnoid cyst that reportedly did not have notable mass effect upon the cerebellum (pp. 337–347). In terms of Diagnostic and Statistical Manual of Mental Disorders—Fourth Edition diagnosis, he fulfilled criteria for ADHD, obsessive-compulsive disorder, and bipolar disorder. He was first treated with a variety of psychopharmacological interventions during the course of several years, but improvement was never better than minimal. He began to experience episodes of extreme lethargy. He was then referred for an MRI study, which identified his Chiari malformation. His psychiatric symptoms consisted of a range of symptoms of ADHD, significant, repetitive obsessions concerning contamination fears, and severe mood swings.

The WISC-IV (Wechsler, 2003) was administered as a part of the neuropsychological evaluation. The WISC-IV results revealed a Verbal Comprehension Index of 114, a Perceptual Reasoning Index of 102, a Working Memory Index of 97, but a Processing Speed Index of only 75, which is an obviously differential test finding often associated with executive dysfunction. The efficiency of his problem solving on the Tower of London Test (Culbertson & Zillmer, 2001) was only at the 18th percentile ranking and he achieved only one category on the Wisconsin Card-Sorting Test (Heaton, Chelune, Talley, Kay, & Curtis, 1993). On the Brown-Peterson Technique (Auditory Consonant Trigrams; see Spreen & Strauss, 1998; Strauss, Sherman, & Spreen, 2006), his performances were well more than a standard deviation below the mean for the 9- and 18-second delay intervals, and he was never able to perform anywhere near the level of his age-matched cohorts on any task that required the inhibition of prepotent responses. On a continuous performance test, he was only slightly inattentive as indexed by errors of omission, but errors of commission were significantly elevated. On a word list-learning and memory task, his learning slope was shallow; acquisition was slow but incremental. Although he did not learn as much information as his peers, what he did learn was retained in delayed conditions. Visuoconstructual skills were considered unimpaired by WISC-IV subtest standards, although his copying of the Rey Complex Figure (Rey, 1959) was below the 1st percentile ranking; this pattern of spatial cognition is often interpreted as a manifestation of executive control deficit.

Therefore, his test protocol featured many signs that could be characterized as executive dysfunction. He exhibited deficits in inhibition, working memory, planning and problem solving, and in speed of novel information processing. His cognitive assets concerned his general level of intelligence and his semantic memory. A motor examination was not administered. The overall pattern of neuropsychological test results is consistent with what is observed in the CCAS (Schmahmann, 2004; Schmahmann & Sherman, 1998). However, the case was initially presented to demonstrate the lack of localization properties of neuropsychological tests.

Riva et al. (2011) presented two cases of children with Chiari I malformation, a 5-year-old boy with language delay and a 15-year-old girl who additionally had an arachnoid cyst in the left temporal-lobe area. Both of these children required decompression surgery. Postsurgically, the boy’s language functioning was considerably improved, but his attentiveness, motor difficulties, and general behavioral control deteriorated. The girl’s pattern of recovery took an opposite course. Although initially she was easily distracted but had good language skills, after surgery, her attentional skills significantly improved, but her language skills deteriorated. Although these cases present contrasting pictures, the girl’s arachnoid cyst in the left temporal region may have affected her language processing.

In any event, in Chiari malformation I, the cerebellum is at least somewhat compressed and crowded inside the posterior fossa. The cerebro-cerebellar circuitry system is highly specialized and segregated (see Schmahmann and Pandya, 1997a, for a review). The prefrontal cortex, frontal motor regions, the superior temporal sulcus, the parietal lobes, and the paralimbic cortices all have specified, reciprocal connectional profiles with the cerebellum. Based upon this neuroanatomic geography and the pattern of diaschisis we described earlier, one would predict specific deficits dependent upon the region of cerebellar involvement. This prediction has been supported by a variety of studies, although this has not been systematically
investigated in Chiari I malformations (see Koziol & Budding, 2009, Chap. 5 and 11, for a review of these studies).

Finally, Kumar et al. (2010) have conducted perhaps the most systematic study to date on Chiari I using diffusion tensor imaging (DTI) and neuropsychological testing. The small sample included 10 patients ranging in age from 18 to 36 years old, which is hardly a “pediatric” population. Neuropsychological test results were interpreted as abnormal in all Chiari I patients compared with controls. However, the most striking findings concerned microstructural abnormalities in different brain regions and white-matter tracts using DTI, even when these areas appeared normal on conventional MRI. DTI metrics in Chiari I identified abnormalities in the genu, splenium, fornix, cingulate, putamen, and thalamus as compared with control subjects. These structural changes could easily be associated with neuropsychological abnormalities. When all the findings we reported are taken together, in aggregate, the conclusion is clear that the field is in need of large-scale, prospective, longitudinal studies to assess the impact of Chiari I malformation on neuropsychological functioning.

SUMMARY

This article reviewed a variety of subcortical brain regions and structures that traditionally have been understood as only participating in movement. However, by definition, purposive movement involves action control, and action control is executive functioning. A child enters the world with only basic and seemingly primitive sensory and motor capacities, and deficits in these early functions very frequently predict neurodevelopmental disorders. These disorders are almost routinely characterized by executive function deficits as measured by neuropsychological tests. Hypotonia is often considered a benign condition. However, because it can significantly restrict levels of activity, we propose that low muscle tone can significantly impact upon the development of executive function, because all knowledge that eventually guides functioning is grounded in interactive behavior. Neonatal jaundice is a frequent condition evident at birth or shortly thereafter. Although this condition might be easily managed from the point of view of medical stability, bilirubin has an affinity for attacking certain brain regions that are critical to the acquisition of new behaviors and that are key to the development of inhibitory and working-memory functions that eventually guide independent, functional adaptive behaviors. Therefore, these features of a child’s developmental history should never be overlooked or underestimated because they can easily represent a key for establishing the framework for understanding a child’s adaptive difficulties. Finally, Chiari I malformations are poorly understood and in need of much greater systematic investigation than has been the case up until now. The brain regions involved in this condition are highly sensitive and can easily contribute to deficits in the development of self-regulation or executive function.

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