JoLLE@UGA®
Journal of Language & Literacy Education

Scholars Speak Out

December 2019

Language, Literacy, and MECP2 Duplication Syndrome: Reframing and Reconsidering Learning for Children with Rare Disorders

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My husband and I learned that our son Charlie has a rare disorder at Disney World, purportedly the happiest place on Earth. At 18 months old, Charlie couldn't sit up on his own for much more than a few seconds, walk independently, or verbalize words like many children of the same age. We had been waiting for specialized test results for months. Just before we headed to the Magic Kingdom for the day, our developmental pediatrician called with a diagnosis: MECP2 Duplication Syndrome (MDS), a rare genomic disorder almost always found in boys (Peters et al., 2013). Sometimes referred to as the opposite of Rett Syndrome—a similar, more prevalent gene disorder that primarily affects girls (Miguet et al., 2018)—MDS was a mystery to our doctors, none of whom had heard of MDS before and had no other information to give us, other than to start searching for a specialist.

We soon learned about the specific medical features of MDS. Children with the disorder often experience numerous, concurrent medical symptoms, such as seizures, recurrent respiratory infections, muscle spasticity, and gastrointestinal issues, often resulting in death by the end of childhood (Sztainberg & Zoghbi, 2016). While some children experience these serious medical symptoms from birth, others have a sustained period of relative health before they regress, usually due to the onset of seizures (Peters et al., 2013). Charlie is part of this latter category, eventually learning to sit up on his own, walk short distances, and participate in many typical childhood activities. As we adjusted to the news of Charlie's diagnosis, we began to wonder what the future held for our son.

MDS, Research & Educational Opportunities

Once we located a neurologist with a dedicated MDS practice to help care for Charlie's medical needs, we started navigating the maze of special services for early education. We soon learned that finding ways for Charlie,

now almost four years old, to participate in the same high-quality literacy and language learning opportunities afforded to his neurotypical siblings would prove tricky. MDS has robbed Charlie of speaking words, although he babbles with intonation and expression. It has shortened his attention span, reduced his hand use, and resulted in low muscle tone that makes movement more difficult, all of which make using an assistive communication device or sign language challenging. These differences often mean Charlie is judged as less intelligent and less able to participate in mainstream learning environments and activities. In medical, school, and therapeutic contexts, Charlie has repeatedly been labeled as at risk for "not meeting developmental milestones," "not belonging" at certain schools, and even "lazy" in evaluations of his educational potential.

To most parents, these comments would be devastating (and certainly they are for us), but I argue this framing signals a fundamental misunderstanding about how to educate children like Charlie. For children with MDS who are minimally or not-yet-verbal, their learning differences and medical needs do not determine their ability or desire to learn. The absence of verbal language does not exclude them from their fullness as members of language and literacy communities. Shifting away from a deficit perspective, Charlie and other children with MDS could be reframed in educational settings by focusing on their strengths.

Most research on children with rare diseases is centered on a pathological, rather than educational, perspective (Darretxe, Gaintza & Monzon-Gonzalez, 2017). With the newness of MDS as a diagnosis (it was first isolated approximately 15 years ago), only ~200 cases have been reported in the medical literature (Peters et al., 2019). Charlie and other children with MDS are often classified as "mentally retarded" or "severe/profoundly intellectually disabled" in medical studies (Lotti, Geronzi & Grosso, 2019; Ramocki et al., 2009). While these labels may be necessary for treatment to progress in the medical field, they serve to limit and disable children with MDS when used deterministically in educational spaces.

Language, Literacy & the Potential to Learn

As children with MDS are socialized into contextually appropriate ways of using language and literacy to mediate their experience of the

world, the medical symptoms of MDS disrupt the process. Typical socialization practices may not work for children with MDS, presenting a challenge for the adults around them. There is a dearth of research on the cognitive worlds of children with MDS, but parents of children with MDS have echoed our desire to learn how to communicate effectively with our children. In the latest survey (n=48) of North American parents whose children are diagnosed with MDS, a "lack of effective communication" was one of their chief concerns (Peters et al., 2019). Similarly, in a recent French study (n=59), researchers reported that communication systems, including limited words and pictogram cards, were used successfully with MDS patients (Miguet et al, 2018). Building on parents' desire to communicate with their children, this study showed that MDS does not remove the ability to learn and manipulate symbols. Both studies suggest that children with MDS are capable learners and can benefit from a strengths-based approach to learning.

Our experiences with Charlie illustrate the differences between approaches to teaching children with MDS. In schools, language-based differences usually result in segregated classroom placement for children with MDS because a lack of verbal ability (or approximation through an assistive device or communication cards) is typically seen as a barrier to mainstream classroom success. Based in a belief in cognitive incapacity when compared to "normal" children, this "less able" perspective bleeds into the educational setting as diagnoses focused on pathology are shared with school personnel. The labels and (dis)abilities placed upon Charlie, for example, slate him to attend special needs preschool in his local school district, far from his home and his neighborhood school. The absence of verbal language and other skills negatively defines how educators view Charlie's ability to participate in mainstream classrooms, leading to his segregation away from neurotypical children.

Outside of the public school system, Charlie's abilities are framed much differently. He is accepted at home as a different kind of learner—not as "disabled." Building on his strengths, Charlie participates in his own way in the same activities as his neurotypical siblings while physical, occupational, and speech therapy sessions teach him how to engage in family life, academic learning, and the world around him. Charlie demonstrates the ability to mediate his experience of the world through communication and grasp beginning literacy skills, such as print

awareness, as measured by formal therapeutic evaluations and our personal assessments. As noted in Charlie's most recent occupational therapy evaluation, he "has developed preferred toys and will look to find them from a container with other toys or walk to another room to find the toy." Charlie's movements communicate his interests and allow us to understand his thoughts, including his favorite toys and books.

Literacy is an important part of Charlie's life. He loves to sit in our laps or at the table while we read to him. On his own, he happily turns the pages of his favorite books, often from the *Little Blue Truck* series. As he moves through the pages, Charlie studies the illustrations and attends closely to their details. He often vocalizes as if he is reading aloud, demonstrating his understanding of early literacy skills like directionality. At home and in therapy, Charlie is framed as a capable learner and communicator, even though cognition and understanding manifest differently for him. In formal educational settings, it could be transformative if Charlie and other children with MDS were understood through this type of lens.

Planning for the Future

An MDS diagnosis is becoming more common as an increasing number of families gain access to specialized genetic testing. Complicating the diagnosis, the severity of MDS symptoms fall along a spectrum due to variations in duplication size, which means not all children with the disorder have the same educational needs. As a result, a variety of practices and settings may be required to tailor learning appropriately. Further research can allow teachers and therapists to select contextually appropriate tools for teaching children with MDS.

At home and in therapy, we have found success by meeting Charlie where he is and using tools that match his needs and abilities. We teach Charlie explicitly to show us his thoughts and preferences in the absence of telling us. Now Charlie uses behavior and movement to communicate. We nurture literacy development through many of the same practices we use with Charlie's neurotypical siblings, including frequent opportunities to read and write and an emphasis on the enjoyment of favorite stories. When possible, we avoid educational settings in which Charlie is reduced to the sum of his differences, but we have hope for a time when that is

unnecessary.

Ultimately, we want others to see Charlie as we do: as a bright, capable, hard worker with a clear desire to learn. In school settings, educators could reconsider how to teach Charlie and other children with MDS by reframing their abilities to focus on their strengths rather than their differences. Through this new lens, children with MDS would no longer be segregated from their peers based on their (dis)abilities, and instead, they can become fully enabled members of their educational community.

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