

Cognition

Introduction

Matthew W. Mosconi, PhD presented on the topic of cognition, learning, and development in individuals with PMS. His presentation, “What We Think We Think About Thinking in PMS” provided an overview of the importance of assessing cognition in individuals with PMS, current limitations of studying cognition in this population, review of existing literature, and proposed practice parameters.

Cognition is a relatively general term that refers to any mental process. Intellectual disability (ID) refers to a diagnosis given to individuals who demonstrate significant impairments in intellectual and adaptive functioning. Developmental disability (DD), in contrast, is a diagnosis given to individuals who are 5 years old or younger and demonstrate significant delays in at least two areas of functioning (e.g., language, problem solving, or motor skills). Thus, DD is used for younger children who fail to meet expected developmental milestones, whereas ID is typically reserved for children older than 5 years old, as intellectual functioning is less variable and more stable at this age.

Cognition and learning are important areas of research in PMS as it can inform the development of effective therapeutic interventions. Cognition and learning also can be used as an outcome measure to assess the effectiveness of behavioral and pharmaceutical treatment or to assess developmental changes longitudinally. Yet, examining cognition and learning in individuals with PMS has several limitations. For example, standardized tests used to measure cognition are often not suited for individuals with PMS due to social, behavioral, speech, and motor challenges associated with this disorder. Thus, standardized tests may under-estimate (or over-estimate) true cognitive functioning in individuals with PMS. Other methods of assessing cognition and learning in individuals with PMS that take these additional challenges into account could be used, but they are not standardized, thus reducing validity.

Review of existing literature from 2008 – 2016, which included individuals ranging from 5 months to 42 years old, has revealed some consistent findings. However, all findings should be interpreted with caution due to limitations of age range within each study, small sample sizes, and cognitive tests used. In a study completed by the Mt. Sinai group (n = 30), investigators reported that individuals with PMS demonstrated reductions in nonverbal IQ. However, there was considerable heterogeneity among the group, such that some individuals had IQs in the average range as well as the mild to moderate ID range, but the majority had IQs in the severe to profound ID range. In another study, a pattern of relative strengths and weaknesses emerged. For example, verbal and language skills were more robustly affected in individuals with PMS.

Additionally, attention and executive functioning skills appear to be more impaired compared to visuo-spatial and visuo-motor skills in a small sample of adults with PMS (n = 7). Also, there is some evidence supporting a relationship between cognitive ability and deletion size, such that individuals with greater-sized deletions have more severe cognitive impairments. However, this finding is inconsistent across the literature. Lastly, with regard to longitudinal findings, development of cognitive and language skills are most prominent in early years, with general slowing of development of these skills at approximately 6 years old. This data suggests that intervention services may be most beneficial in the early stages of development for individuals with PMS.

Due to the unique challenges associated with assessing cognition and learning in individuals with PMS, a few practice parameters were recommended. First, intellectual and developmental skills should be assessed using standardized measures suitable for individuals with severe intellectual disability. Second, due to highly prevalent gait and motor abnormalities in individuals with PMS, these motor abnormalities should be carefully assessed and considered in the context of overall functioning. Also, appropriate referrals to occupational and physical therapists should be considered.

Identified Problems

- 1. Traditional methods of teaching skills are not effective for my child with PMS. I do not know which strategies myself, therapists, teachers, and family members should use to help my child learn and develop important cognitive and daily living skills.**

Although a small portion of individuals with PMS does not demonstrate intellectual disability, the majority of individuals reportedly have severe to profound intellectual disability. Thus, individuals with PMS may not naturally "pick up" certain concepts and traditional teaching practices used with typically developing children are less appropriate (and effective) with this population. Therefore, different strategies and methods must be used with this population. However, well-trained and educated therapists and teachers who are familiar and knowledgeable about working with severely disabled individuals are not usually accessible to our families, and few parents themselves have received any appropriate training in these areas. Therefore, there is a large gap in the system between having evidenced-based methods that may be effective at developing cognitive and other development skills in individuals with PMS, but few individuals who are able to provide these services. Additionally, because these methods typically require advanced training through schooling or intensive workshops, more readily accessible methods that can be provided by a larger group of providers are not available. Although a few of these more simple methods used to enhance learning and development may be used by teachers, therapists, and parents (e.g., using visual versus verbal prompting), there is poor general awareness of these methods within the

PMS community. Thus, parents largely rely on word of mouth from other parents. This, too, is a great limitation to the overall goal of enhancing skills and cognition in individuals with PMS. According to our Group discussion data, parents asked about which were the most effective strategies to use at home (n = 47), at school (n = 26), and at therapies (n = 41) to enhance learning. This was rated to be a severe concern, as parents ranked it 4.6 out of 5. Parents also addressed additional specific questions regarding such strategies, such as whether certain strategies were best for a specific age- or developmental-level and what were the most affordable but most effective options. In addition, parents indicated concerns related to the knowledge and familiarity of appropriate interventions that their children’s teachers and therapists had. Other parents recognized the importance of finding a good motivator to be used in therapies and other educational opportunities; however, they were unsure how to specifically use motivators to help learning and how to identify a good motivator if one has not already been found. For example, one parents noted, “food driven learning is a huge motivation. How do we exploit this or find the driver for that so it can be applied?”

2. Standardized test measures are not a fair way to measure my child’s cognitive skills.

Due to developmental, cognitive, motor, and other limitations, standardized test measures to assess cognitive level may not be appropriate for individuals with PMS. Thus, estimates of developmental or intellectual level are just that, estimates, and unlikely capture true capabilities. Although certain providers and researchers may prefer certain developmental or intelligence tests over others because of reduced floor effects (i.e., able to capture lower IQs) or reduced verbal demands, currently there is no “gold-standard” cognitive test for individuals with PMS. About 40 parents indicated this as a primary concern for them, and ranked it to be a very high concern (4.7 out of 5). For example, parents indicated they were “concerned about getting accurate responses from a nonverbal person” and questioned whether “cognitive assessments are a true indicator of a child’s skills”. Other parents said, “the current tests just do not work”. Additionally, parents indicated that aside from test limitations due to cognitive and motor abilities, that their child often demonstrated significant behavioral challenges during testing, such as task refusal, which impacted results.

3. My child has not be able to retain the skill he/she has learned OR My child is not improving in his/her therapies OR I am concerned my child will not be able to learn after 6 years old.

The attainment and retention of skills appears to be another area of concern for parents of individuals with PMS. Over thirty parents indicated specific concerns related to the waxing and waning and reduced retention of cognitive skills and rated it to be a significant concern (4.6 out of 5). For example, one parent shared that his child was not able to “retain any learned skills with respect to intellectual, cognitive, and motor abilities”. Similarly, another parent shared her experience, “We learn a skill, we lose a

skill, and we learn it and lose it". Parents expressed a need for understanding why their children had difficulty attaining and retaining skills, and they wanted specific strategies to prevent it. Additionally, parents noted that they were concerned about research findings of reduced cognitive increases after the age of 6 years old. Approximately 20 parents identified this as a moderate to severe concern (4.3 out of 5). For example, one family shared, "with our age group of 10-14 years olds and the research has now shown us that the 'window of learning success' closes around 6 years of age, what sort of therapy goals or successes should we be pushing for". Families in this age group seem to be concerned whether they "missed the boat" on cognitive development for their children.

Proposed Solutions

1. Development of parent and provider tool-kit on identified effective strategies for learning in PMS.

In order to improve the availability and accessibility of effective treatment strategies for individuals with PMS, a three-tiered approach should be applied. First, parent and provider tool-kits should be made readily available that include basic teaching strategies known to be effective in individuals with developmental disabilities. The tool-kit also should provide helpful "troubleshooting" strategies that may help parents and providers overcome common behavioral challenges that may occur during learning. For example, these troubleshooting strategies may target escape/avoidance behavior. Because this tool-kit is aimed at both parents and providers, the format should be lay-friendly and easily accessible. The primary aim of this tool-kit is to empower parents to feel knowledgeable and capable of working with their children with PMS to help in the learning process. A secondary aim is to enhance providers' knowledge and skills when working with individuals with PMS. Additionally, the tool-kit is aimed at empowering parents to speak with their children's teachers and therapists about potential effective strategies to enhance their children's learning. The second level is a continuation of the first, such that it provides parents an avenue to identify the most effective strategies for their children and enhance communication with providers. Again, a primary aim of this approach is to empower parents to work collaboratively with providers to identify individual-specific goals and strategies to enhance learning for their children. Additionally, this may be particularly helpful within the development of Individualized Education Plans (IEPs) within the school district. As part of this approach, deliverables to parents would include information about the Individuals with Disabilities Education Act (IDEA), template letters for schools, scripts to speak with therapists/teachers, and samples of target goals and strategies. At the final level, increase training for teachers and therapists should be a larger goal of the developmental disability community. For example, additional training during their schooling should be available for all teachers, not just those focusing on the special education population. Lastly, funding to support

parent and teacher/therapist participation in local workshops and conferences should be made available to further increase accessibility and knowledge.

2. Development of more appropriate standardized measures of developmental/cognition/intelligence in individuals with PMS.

Current standardized measures often are not appropriate for individuals with PMS. Thus, the development of adapted versions of current standardized measures or of a new standardized measure to assess developmental/cognitive functioning should be conducted. Such a measure would not be restricted to use of individuals with PMS, but to all individuals with significant limitations that impact the administration and interpretation of traditional standardized protocols. For example, an adapted version may have more appropriate norms to estimate functioning level. Or, have alternative methods of administration for individuals with significant motor or language delays (i.e., technology-based administration). The measure should include a wide range of skills across verbal and nonverbal domains. As part of this measure, the manual should include recommendations based upon scores within specific sections (i.e., a child receiving a standard score < 70 in a motor domain should be referred to an occupational and/physical therapist; it is recommended that a child with a standard score < 70 in working memory should be presented with simple one-step instructions provided in multiple forms—e.g., written, picture, verbal).

3. Increased awareness and understanding of cognition and development in PMS.

Challenges related to the attainment and retention of skills may in part be aided with the development of the above-mentioned tool-kit (see Proposed Solution 1). As part of this approach, it also will be important for teachers and therapists to work with parents to provide them with the tools to practice interventions at home. As repetition and generalization of skills is necessary for individuals with PMS, it is thus important for therapeutic practices to continue into the home. This does not necessarily mean parents need to "drill" their children or set up a "miniature school" at home, instead, parents should be provided with basic tools that can be incorporated into daily activities and routines, such as meals or bed time. Furthermore, increased research examining the cognitive and developmental profile of individuals with PMS will help parents and providers to better understand best strategies to use during the learning process. It also may help parents prepare for specific challenges during the learning process. For example, if more evidence confirms that cognitive development slows after the age of 6, greater emphasis should be placed on attaining early intervention and avenues to improve access to these services should be determined. Additionally, with regard to this finding, research investigating ways to extend this apparent "developmental window" also should be a priority.