The Effects of Singing vs. Speech on Joint Engagement in a Young Child with Phelan-McDermid Syndrome

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ABSTRACT

Phelan-McDermid syndrome (PMS) is a rare genetic disorder typically caused by loss or disruption of the distal long arm of chromosome 22. Children with PMS exhibit severe to profound communication deficits, and the majority meet the criteria for Autism Spectrum Disorder (ASD). To date, no language or communication intervention studies focused on children with PMS exist in the literature despite a steady increase in diagnosed cases. Anecdotally, it has been reported that music facilitates learning and engagement in children with PMS. The purpose of this clinical case study is to investigate the effects of singing vs. speaking on joint engagement, a critical prerequisite for later language development, in a young child with PMS. Given previous research on children with ASD, we predicted that the participant's joint engagement would improve in response to intervention. We also hypothesized that her joint engagement scores would be higher in the singing condition. A simultaneous-treatments approach was utilized to compare the effects of singing and speech on joint engagement during 16 sessions over a 10-week period. Videotapes of teaching trials were analyzed to measure the participant's visual attention and general behavior. Results indicated that the participant's joint engagement increased over time in both the singing and speaking intervention conditions. The singing condition appeared to have a slight advantage in eliciting visual attention, but over time both singing and speaking promoted increases in joint engagement. Limitations of the current study, recommendations for clinical practice, and implications for future research are discussed.

Introduction

Phelan-McDermid syndrome (PMS), or 22q13 deletion syndrome, is a genetic disorder typically caused by loss or disruption of the distal long arm of chromosome 22 (Phelan & McDermid, 2012; Richards et al., 2017). Although PMS is rare, it is one of the more frequently occurring microdeletion syndromes (Zwanenberg, Ruiter, van den Heuvel, Flapper, & Ravenswaaij-Arts, 2016). Children with PMS exhibit moderate to profound global developmental delays (Costales & Kolevzon, 2015; Oberman et al., 2015; Zwanenburg et al., 2016). Major features of PMS include hypotonia, moderate to profound intellectual disability, severe to profound communication delays, and high comorbidity with autism spectrum disorder (Phelan & McDermid, 2012; Rankine et al., 2017; Richards et al., 2017). Neonatal hypotonia is one of the first presenting signs of PMS, leading to problems with feeding, delayed motor milestones, and delayed speech development (Phelan & McDermid, 2012). Physical features may include subtle facial characteristics, such as a wide nasal bridge, deep-set eyes, and long eyelashes (Zwanenberg et al., 2016). Children with PMS tend to grow at typical rates and be of normal size. Motor milestones, such as sitting up, crawling and walking are generally delayed (e.g., the average age of walking is 27 months; Phelan & McDermid, 2012). Common medical issues include seizures, renal abnormalities, gastroesophageal reflux, cyclic vomiting, and cardiac defects (Costales & Kolevzon, 2015; Phelan & McDermid, 2012; Zwanenberg et al., 2016). The significant physical, cognitive and communication deficits associated with PMS require interventions in the areas of medicine, physical therapy, occupational therapy and speech-language pathology. These interventions must begin early in the child's life and be delivered with sufficient intensity to bring positive and lasting outcomes (Phelan-McDermid, 2012). The severity of the disorder and subsequent need for multiple interventions are socially and emotionally challenging for affected individuals and their families (Zwanenberg et al., 2016).

The first case reporting deletion of the distal long arm of chromosome 22 was in 1985 (Watt et al., 1985). The syndrome was later described as 22q13.3 deletion and named Phelan-McDermid Syndrome in 1998 (Phelan-McDermid Syndrome Foundation, 2020). In 2012, Phelan and McDermid reported that more than 600 cases of PMS had been diagnosed globally. Since then the rate of diagnosis has rapidly increased (Zwanenberg et al., 2016). Recently, the Phelan-McDermid Syndrome Foundation reported having 2,255 members worldwide and growing at a rate of nearly one new family per day (Phelan-McDermid Syndrome Foundation, 2020). Due to the subtlety of the physical features, lack of knowledge about the syndrome, and complexity of genetic testing, PMS is likely underdiagnosed or misdiagnosed as cerebral palsy, idiopathic autism, idiopathic intellectual disability, Angelman Syndrome, Velocardiofacial Syndrome, Fragile X, or other developmental disorders (Costales & Kolevzon, 2015; Phelan & McDermid, 2012; Richards et al., 2017). It is predicted that the number of children diagnosed with PMS will continue to increase as more healthcare professionals become aware of the syndrome and diagnostic techniques for genetic testing improve (Costales & Kolevzon, 2015; Zwanenberg et al., 2016).

Of particular interest to speech-language pathologists and other special educators is that children with PMS present with significantly impaired communication abilities (Richards et al., 2017). While most characteristics associated with PMS tend to vary in severity depending on the size or type of deletion (e.g., intellectual disability), significant speech and language impairment is a consistent feature across all individuals (Rankine et al., 2017; Richards et al., 2017). Infants with PMS often babble at the expected age and some toddlers develop a limited expressive vocabulary; however, loss of language skills at 3 or 4 years of age is common (Phelan-McDermid, 2012). Zwanenberg et al. (2016) assessed 34 children with PMS, ages 8 months to 14 years of age, across a variety of developmental domains (i.e., adaptive behavior, cognition, motor skills, language). As a group, poorest performance was observed in the domain of language development.

Another characteristic of this syndrome is that a high comorbidity exists between PMS and Autism Spectrum Disorder (ASD; Oberman et al., 2015; Phelan-McDermid, 2012; Richards et al., 2017). ASD is characterized by deficits in social communication, and restricted and repetitive behaviors or interests (American Psychiatric Association, 2013). Children with PMS commonly exhibit social anxiety, reduced social interaction, self-stimulatory behaviors, reduced eye contact and other characteristics commonly associated with ASD (Phelan & McDermid, 2012). Richards et al. (2017) assessed 30 individuals with PMS ages 4 to 37 and found that 87% met the criteria for ASD. PMS is estimated to account for between 0.5% and 2.0% of individuals with ASD and intellectual disability (Costales & Kolevzon, 2015).

Prelinguistic Communication in Children with ASD

As noted above, the majority of individuals with PMS are also diagnosed with ASD (Richards et al., 2017). A core deficit in children with ASD is delayed or absent joint attention behaviors (Adamson et al., 2019; Murza, Schwartz, Hahs-Vaughn, & Nye, 2016; Thorup et al., 2018). Joint attention is generally defined as the sharing of attention between two individuals to the same object or event and monitoring each other's attention during the interaction (Bottema-Beutel, 2016, Ellis Weismer & Kover, 2015). For example, a child points to a dog, looks at her mother who says, "A dog!", then turns her gaze back to look at the dog. Previous research has found that joint attention behaviors in infants and young children are powerful predictors of later language ability (Adamson et al, 2019; Bottema-Beutel, 2016; Ellis Weismer & Kover, 2015; Murza et al., 2016; Thorup et al., 2018). Children with ASD who exhibit limited joint attention are at risk for poorer language outcomes because they have fewer opportunities for making associations between spoken language and the environment (Adamson et al., 2019; Conway et al., 2018). Impairment in joint attention is thought to be a causal factor in the high proportion of children with ASD who are minimally verbal or nonverbal (Tager-Flusberg, Paul, & Lord, 2005).

Given the robust literature connecting joint attention and language acquisition, Tenenbaum et al. (2017) stated, “If we can capture the attention of the child with autism...we may be able to improve language acquisition in this population (p. 1803).” Other researchers have also advocated for interventions that increase joint attention skills in young children with ASD to prevent the “negative developmental cascade” (Adamson et al., 2019, p.2) of joint attention deficits on early word acquisition (Bottema-Beutel, 2016; Edmunds et al., 2017; Ellis Weismer & Kover, 2015. For example, Kasari et al. (2014) found that even minimally verbal children with ASD exhibited language gains when interventions directly supported episodes of joint attention. Murza et al. (2016) conducted a meta-analysis of joint attention interventions for children with autism. They analyzed 16 research studies that met their criteria for inclusion. The results indicated that all joint attention interventions were effective, with effect sizes ranging from .345 to .719. Given the variations in dosage, participant characteristics, and agents of intervention (i.e., caregiver or therapist), Murza et al. (2016) concluded that while interventions designed to increase joint attention are effective, it remains unclear which approaches are best suited for which subgroups of children with ASD.

Recent research has focused on the development of joint engagement, which emerges earlier than joint attention (Adamson et al., 2019; Adamson et al., 2021; Kaale et al., 2018; Shih et al., 2021). Joint engagement refers to a child and adult interacting together while focused on the same object or activity (Shih et al., 2021). During joint engagement the child is aware of the adult but does not overtly monitor or direct the adult’s attention to an event or object (Kaale et al., 2018). Typically developing children exhibit joint engagement behaviors before their first birthday (Striano & Bertin, 2005). Similar to the research on joint attention, children with ASD exhibit delays in joint engagement which are negatively correlated with later expressive language skills (Adamson et al., 2019; Adamson et al., 2021; Shih et al., 2021). Given the relationship between early joint engagement and later language development, researchers suggest that children with ASD should receive early intervention designed to increase joint engagement behaviors, such as increased visual attention and sustained interaction with adults around a shared activity (Adamson et al., 2019; Kaale et al., 2018; Patterson et al., 2014).

Musical Interventions for Children with ASD

Given the significant communication and social deficits observed in children with PMS, interventions that promote social and linguistic development are critically needed. Anecdotally, children with PMS are described as having a “passion for music,” and caregivers have reported that music and singing help their children learn (Unique: Understanding Chromosome Disorders, 2016, p. 7). Similar sentiments have been reported by caregivers of children with ASD (e.g., Schwartzberg & Silverman, 2017). Numerous empirical studies have examined the effect of music on behavior and social engagement in children with ASD. For example, Finnigan and Starr (2010) found that a musical intervention (singing familiar songs accompanied by guitar) was more effective than a non-music intervention (speaking the lyrics) in promoting socially responsive behaviors (e.g., eye contact; turn-taking) and decreasing avoidant behaviors (e.g., gaze aversion, moving away) in a three year-old girl with ASD. LaGasse (2014) randomly assigned 17 children with ASD to either a music therapy group or a no-music social skills group. Children participated in 10 group treatment sessions over five weeks. Results indicated that children in the music therapy group exhibited increased joint attention and eye gaze toward persons compared to children in the no-music group. Research reviews by Wigram and Gold (2006) and Lim (2012) concluded that musical interventions for children with ASD were effective in promoting social engagement and positive behavior. To date, no research studies exist examining the affects of music on joint attention or joint engagement in young children with ASD and/or PMS.

Research Questions

The purpose of this clinical case study is to systematically examine the effects of singing and speaking on the joint engagement of a toddler with PMS. Although severe to profound delays in communication development are consistently observed across children with this syndrome, to our knowledge no communication or language intervention research focused on children with PMS currently exists in the literature. In addition, none of the previous research on the effectiveness of interventions for increasing joint attention or joint engagement utilized music or singing. The specific research questions include:

1. Will a toddler with PMS show increases in joint engagement in response to intervention?

2. Will the participant's response to intervention differ between the singing and speaking conditions?

Given previous research on the positive effects of intervention on joint attention and joint engagement in children with ASD, we predicted that our participant's joint engagement behaviors would increase over the course of the study. Also, given previous empirical and anecdotal evidence on the positive effects of music on engagement in children with ASD, we predicted joint engagement scores to increase more rapidly in the singing condition and remain higher across the study when compared to the speaking condition.

Methods

Participant

Molly (not her real name), a 29-month-old female with PMS and ASD, was recruited from a university speech and hearing clinic where she was receiving services to improve her communication and feeding skills.

Medical History

Molly was diagnosed with hydronephrosis while *in utero*, a common condition in individuals with PMS. Hydronephrosis is the swelling of a kidney caused by a blockage or obstruction which prevents the free flow of urine from the kidney to the bladder. Molly successfully underwent kidney surgery at four months of age. At approximately five months of age Molly was recognized as having low muscle tone (i.e., hypotonia), a primary characteristic of PMS (Phelan, Rogers, & Boccuto, 2005; 2018). A subsequent MRI of Molly's brain revealed abnormal results (e.g., non-specific hypoplasia of the cerebral white matter, mild prominence of the ventricles). Given the combination of hydronephrosis, hypotonia and abnormal MRI results, genetic testing was completed resulting in a diagnosis of PMS. Due to the high prevalence of autism in PMS, Molly was evaluated for ASD at 15 months of age and the results were inconclusive. She was re-evaluated at 18 months and diagnosed with ASD.

Molly has a history of chronic colds, sinus infections, and ear infections. She received pressure- equalizing (PE) tubes at 7 months of age. An Auditory Brain Stem Response (ABR) test at 10 months old indicated normal functioning of the auditory nerve, suggesting that if Molly had hearing loss it would be conductive in nature. Behavioral hearing tests had been inconclusive in terms of identifying the presence or degree of hearing loss; however, her mother expressed concerns about Molly's ability to hear.

Feeding Skills

Molly's feeding and swallowing abilities have been monitored since birth. At one year of age a videofluoroscopic swallow study indicated swallowing and feeding abilities within normal limits. Molly was later diagnosed with oral phase dysphagia (i.e., overstuffing her mouth with food, decreased bolus control, residuals in mouth after swallowing, food and liquid loss). Similar to other children with PMS, Molly's feeding challenges are due to hypotonia (van den Engle-Hoek, de Groot, de Swart, & Erasmus, 2015). Molly's feeding goals at 29 months were improved endurance for chewing food, taking appropriately sized bites, and learning to use a straw. Her mother reported that Molly enjoys eating and her diet consists of a variety of foods. Consistent with descriptions of children with PMS, Molly was born at a normal birth weight and is growing normally in terms of height and weight.

Speech and Language Development

Molly's mother reported that she started babbling soon after her first set of PE tubes were placed at 7 months old. She produced CVCV patterns (e.g., baba, mama) and some vowel sounds. According to her mother, Molly stopped babbling at 14 months old. A regression in speech and language development is commonly observed in children with PMS (Phelan, Rogers, & Boccuto, 2005; 2018). Molly's communication skills were evaluated at the university clinic when she was 17 months of age. At that time, she exhibited minimal communication, although her interest in people was evidenced by her desire to crawl toward both her caregivers and unfamiliar people. In addition, her communicative behaviors included some imitative gestures (e.g., clapping following a clinical model) and spontaneous nonword vocalizations consisting of prolonged vowels. She did not produce any consonant-vowel combinations. The standardized tests administered were the *Vineland Adaptive Behavior Scales – Second Edition* (VABS-II; Sparrow, Cicchetti, & Balla, 2005) and the *Receptive-Expressive Emergent Language Test – Third Edition* (REEL-3; Bzoch, League, & Brown, 2003). The VABS-II is a standardized caregiver interview designed to assess individuals from birth through adulthood on a variety of developmental and functional domains. On the VABS-II, Molly obtained an Adaptive Behavior Composite Score (a composite of communication, daily living skills, socialization, and motor skills domains) of 53 (mean = 100, SD = 15; < 1st percentile). On the VABS-II subdomains Molly received a standard score of 64 (1st percentile) on Communication, a standard score of 63 (1st percentile) for Socialization, a standard score of 71 (3rd percentile) for Motor Skills, and a standard score of 21 for Daily Living Skills (<0.1st percentile). The REEL-3 is a standardized assessment of language development that utilizes a caregiver interview format. Molly's receptive language standard score on the REEL-3 was <55 (< 1st percentile), and her expressive language standard score was <58 (< 1st percentile).

Current Therapies

The current clinical study began one year after her initial evaluation when Molly was 29 months old and starting her fourth semester of twice-weekly speech-language therapy at the university clinic. She was also receiving speech, language and feeding therapy three times per week at a community agency. In addition, she was receiving special education once per week, occupational and physical therapy twice per week, and 6 hours of Applied Behavior Analysis (ABA) therapy per week. Molly was continuing to be monitored by a regional clinic specializing in AAC and was using a TouchChat iPad app with 15 symbols per screen. Molly's goals for the semester at the university clinic included increasing receptive vocabulary by correctly identifying body parts and common objects (represented by real objects and pictures) and following one-step directions. By the end of the semester she was able to identify 3 body parts and identify objects within a field of 2 with 73% accuracy. She was able to demonstrate comprehension of *up*, *down*, *in*, and *out* by moving her body or manipulating toys with verbal and gestural cues from the clinician.

Her expressive language goals were to increase her communication through vocalizations, signs/gestures, and/or AAC symbols. Progress in this area included production of three new phonemes (/l/, /p/, /w/), use of six gestures (e.g., wave, point), and use of eight new symbols on the iPad (e.g., *play*, *I want*, *puppet*). Her social language goals included engaging in vocal reciprocity and turn-taking. In most sessions Molly engaged in fewer than 10 vocal turns, but in three sessions she engaged in more than 10 turns with the clinician. Expressive language was facilitated through the use of TouchChat on an iPad, AAC symbols presented on cards, signs, gestures, and words.

Therapy approaches and activities implemented during the semester included facilitated play, focused stimulation, indirect language stimulation, and direct language teaching. Physical movements, such as swinging, use of a therapy ball, and postural changes were used to facilitate speech sounds and communication. A visual schedule using symbols from Touch2Chat was used to represent the activities for each session. As each activity was completed, Molly removed the symbol card and placed it into a bucket. Molly was especially motivated by food and music. Singing was frequently used during therapy to gain her attention and musical toys were used as rewards for her participation in intervention activities.

This study was approved by the Institutional Review Board of Marquette University. The research goals and procedures were described to Molly's caregivers through verbal conversations and written materials. Written informed consent for Molly's participation was obtained from her mother.

Procedures

Two intervention approaches were delivered in a simultaneous-treatments approach (Meline, 2009). Baseline data were collected during two sessions at the end of the fall semester prior to the start of intervention. The intervention sessions began in the spring semester during 16 regularly scheduled therapy sessions at the university clinic over 10 weeks. Intervention trials consisted of the examiner either singing or speaking a script based on the song "This Land is Your Land." The examiner sung the script in the singing condition and used normal speech in the speaking condition. The words of the traditional song were replaced with more repetitive, predictable language that focused on color names: "This color's ____. We call it ____. I say ____, you say ____. This color's ____. It's a color. This one is called ____." The examiner held a two-inch laminated square of the corresponding color close to her face while singing or speaking. Her current therapies included the use of picture cards to represent various vocabulary words and concepts (e.g., on her AAC device, or when teaching new vocabulary words). We chose unfamiliar, advanced color names (i.e., copper, fuchsia, turquoise, silver) because our interest was in isolating the effects of singing and speech on Molly's joint engagement during a typical therapeutic format. Using basic color names that were perhaps familiar to Molly may have influenced her behaviors during the intervention trials. In each of the 16 sessions, Molly was presented with 4 singing trials and 4 speaking trials. The order of the conditions (i.e., singing, speech) was randomized across sessions.

During the interventions Molly was positioned to maintain an upright position (e.g., seated in a cube chair with an assistant supporting her back). The examiner was positioned directly in front of Molly seated on the floor. Prior to starting each teaching trial, the examiner gained Molly's attention through calling her name and cueing her that the activity was about to begin. During the intervention, the examiner used animated facial expressions and precise articulation when singing or speaking the script. In addition, she leaned into Molly, positioning the color card in Molly's direct line of sight. If Molly moved her head or looked away, the examiner followed Molly with the color card and her body, attempting to put the card and her face back into Molly's direct line of sight in order to regain her attention. The examiner reinforced Molly when she re-engaged in the activity through eye contact, head nods, and smiles. In addition to providing postural support, the assistant behind Molly helped to physically redirect her when needed (e.g., bringing her back to the cube chair when she attempted to crawl away).

Measures and Data Analysis

Molly's level of joint engagement was based on her visual attention and general behavior during each intervention trial (i.e., presentation of the script through singing or speaking). Every trial lasted for at least 10 seconds; therefore, in order to be consistent across trials we analyzed the first 10 seconds of each intervention, starting when the examiner sung or spoke the words, "This color's…" Each 10-second video was split into five equal segments (two seconds each), and each segment was scored separately. For each two-second segment, Molly was given a point for joint visual attention if she was looking at the clinician's face and/or the colored square for the entire two seconds. A similar procedure was used for scoring her behavior. For each two-second segment, Molly was given a point if she was clearly content and showed no signs of distress (e.g., crying, whining, crawling away from the activity). Each 10-second teaching trial could then result in a joint attention or behavior score of 0 to 5 points. Four trials were presented in each intervention condition per session, resulting in a maximum score of 20 points for each condition (i.e., 5 points possible per trial multiplied by 4 trials in each condition per session).

Reliability

Four intervention sessions (25%) were randomly selected and independently scored by a second examiner for joint visual attention and behavior. Inter-rater scoring agreement for attention was 91.80% (147 agreements/160 judgements), and 100% for behavior (160 agreements/160 judgements).

Results

The scores for joint engagement (i.e., visual attention and behavior) were graphed and visually analyzed for trend and variability of performance across the 16 intervention sessions (Horner et al., 2005; Kennedy, 2005). In addition, scores were compared across the singing and speech interventions to determine if Molly's performance differed between conditions. Overall, Molly's scores increased over time for both visual attention and behavior, indicating that her joint engagement improved in response to intervention. Her scores for visual attention increased more gradually over time, with scores at or near ceiling in the final two intervention sessions for both the singing and speaking conditions (see Figure 1). Molly's behavior was variable during first three intervention sessions; however, by the fourth intervention session her scores were highly consistent, with perfect scores of 20 during the last nine intervention sessions in both the singing and speaking conditions (see Figure 2).

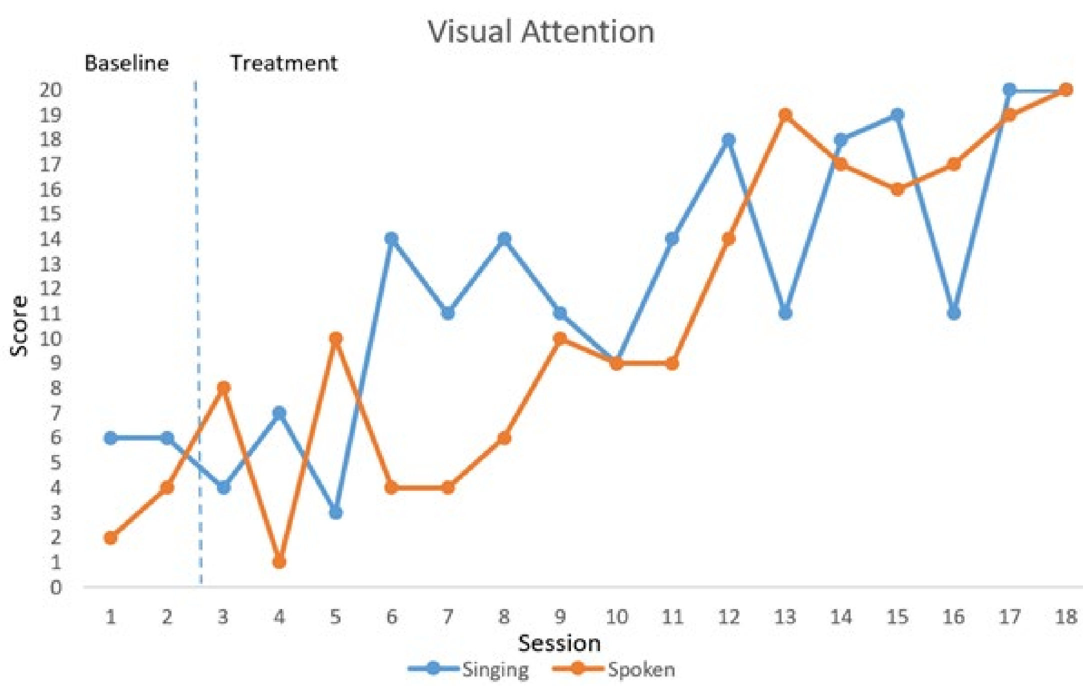

Figure 1. Visual joint engagement scores for the singing and speaking conditions.

When comparing Molly's joint engagement across conditions, her visual attention scores in the singing condition were higher in both baseline sessions and in 10 (of 16) intervention sessions; however, in the majority of sessions the differences between singing and speaking scores were small or negligible. Molly's behavior scores were higher in the singing condition in both baseline sessions, but scores were similar across conditions during the intervention sessions.

Discussion

The purpose of this case study was to examine the effects of singing vs. speaking on the joint engagement behaviors of a young child with PMS. We were interested in examining the effects of singing on Molly's joint engagement given the anecdotal impressions by her caregivers and clinicians that music facilitated her engagement and learning. Previous research on the use of musical interventions for children with ASD has shown that singing and music increases engagement (e.g., sustained attention, appropriate interaction), social interaction and positive behavior (LaGasse, 2014; Lim, 2012; Wigram & Gold, 2006). In light of this previous research, and also the robust relationship between joint attention, joint engagement, and language acquisition, we were interested in examining the effect of music on Molly's joint engagement skills.

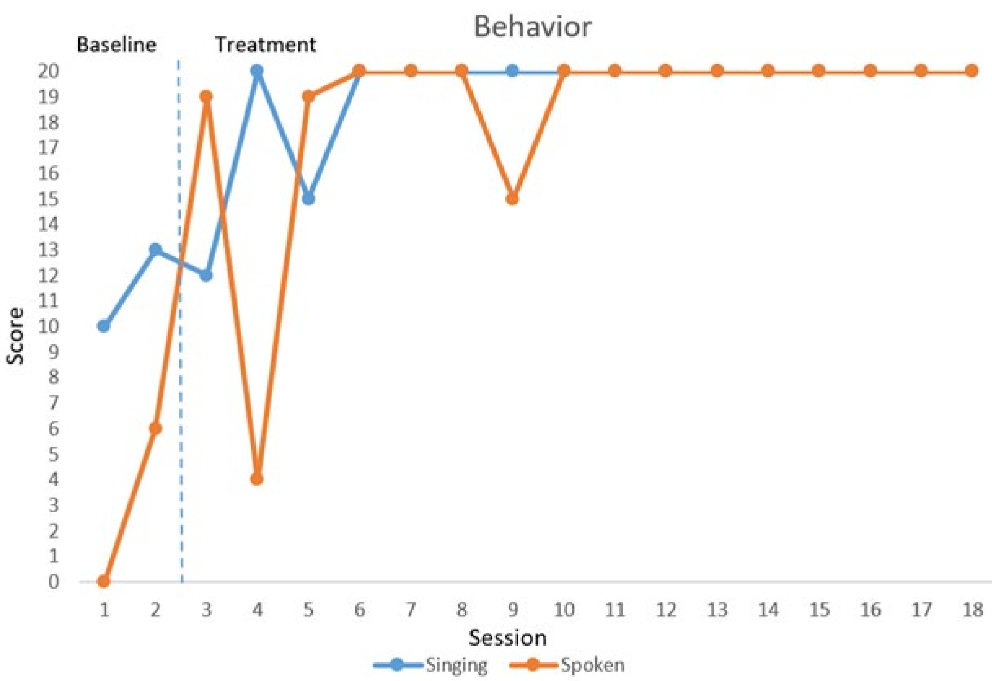

Figure 2. Behavior scores (max = 20) during intervention sessions for the singing and speaking conditions.

Our first research question asked whether our participant would show improvements in her joint engagement skills in response to intervention. We predicted that her joint engagement, as measured by visual attention and general behavior, would increase in both the singing and speaking conditions, and the data supported this hypothesis. These results suggest that the intervention implemented in the current research was effective in increasing the joint engagement behaviors in a young child with PMS. Next, we predicted that the participant's joint engagement scores would increase more rapidly in the singing condition and remain higher than her scores in the speaking condition. This hypothesis was based on the participant's history of responding positively to music (according to caregivers and clinicians), and previous research suggesting that children with ASD exhibit increased social engagement during musical interventions. During the baseline sessions the participant's visual attention and behavior scores were higher in the singing condition than in the speaking condition. Perhaps the baseline sessions reflect her initial positive response to music, which could explain the anecdotal impressions by caregivers and clinicians. During the intervention sessions her scores were more variable, with a slight advantage observed for the singing condition with regard to visual attention. Over time, both singing and speaking appeared to be effective in eliciting joint engagement. We think this is good news for caregivers and clinicians given that communication occurs more often through speaking than through singing. In terms of clinical implications, caregivers and therapists may want to start with singing or music to facilitate joint engagement when introducing a new skill or language target. Next, input could progress to predictable speech, followed by more natural, spontaneous spoken language.

Limitations and Future Directions

The current findings should be interpreted with caution given that the results are based on a single subject over the course of 10 weeks. Also, the scoring of visual attention may have been affected by the angle of the camera, and judging the participant's behavior may have involved subjectivity on the part of the raters. In addition, Molly exhibited erratic behavior scores in the first few intervention sessions, and occasional dips in her visual attention in later sessions. These fluctuations may have due to health issues that caused discomfort on a particular day (e.g., ear infections), lack of sleep the night before, etc. The day-to-day variability in children with PMS and ASD underscores the importance of looking at trends over time. Despite these potential challenges, scoring was highly reliable. It should be noted that the spoken input utilized in this study was based on a script, and therefore more predictable and routine than spontaneous speech. Research comparing predictable, routine speech to less structured, spontaneous speech on children's joint engagement is warranted. In addition, research examining the long-term effects of interventions for joint engagement on language learning in children with PMS is greatly needed.

Conclusions

Results of the current research are promising in terms of improving the joint engagement of young children with Phelan-McDermid syndrome (PMS). Children with PMS consistently exhibit severe to profound deficits in speech and language skills. Joint engagement is an early emerging set of prelinguistic behaviors that provide the foundation for later communication and language development. This study is a novel contribution to the current literature given that to the best of our knowledge, no research exists on the effectiveness of communication interventions for children with PMS.

We hope that this clinical case study increases awareness of Phelan-McDermid Syndrome (PMS), or 22q13 deletion syndrome, among early childhood professionals. Early diagnosis and intervention is critically important for children with PMS, especially given the high comorbidity with ASD. Historically, PMS has been underdiagnosed or misdiagnosed as other neurodevelopmental disorders due to lack of familiarity with the syndrome and subtlety of the physical features (Phelan & McDermid, 2012; Richards et al., 2017). In addition, PMS has several characteristics in common with other genetic syndromes, such as global developmental delay, hypotonia, and communication delays (Costales & Kolevzon, 2015), further complicating accurate identification. Phelan et al. (2005, 2018) recommend that any child with neonatal hypotonia, absent to severely delayed speech, global developmental delay, and minor dysmorphic facial features should be suspected of having PMS.

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