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An Investigation of Social Communication Behaviors in Children with Siblings who have ASD

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Abstract

Autism spectrum disorder (ASD) is distinguished by deficits in social communication and the presence of restricted, repetitive, and stereotypic behavior. Research into familial patterns of ASD has indicated there is a behavioral phenotype in relatives, including siblings, that is milder but qualitatively similar to behaviors within ASD. This broader ASD phenotype (BAP) is characterized by sub-clinical impairments in cognition, language, and social communication, but more research is required to understand the developmental trajectory of these high-risk siblings (ASIBs). Thus, this study utilizes the Brief Observation of Social Communication Change (BOSCC), a behavioral coding assessment intended to track subtle change in social communication behaviors within ASD, to measure the developmental trajectory of ASIBs compared to that of typically developing (TD) children, hypothesizing that ASIBs would show more deficits over time compared to TD children. BOSCC assessments were completed at two time points for 16 ASIB and 15 TD participants; change scores were determined by subtracting the assessment 1 score from the assessment 2 score. ASIBs showed a decrease in BOSCC scores over time, while TD participants showed little change over time. However, there was no significant difference in assessment scores at either time point between ASIBs and TD children; yet, there was a significant difference between the change scores of these groups. This indicated that, while ASIBs may show more social communication impairment at a younger age, they may grow out of these deficits as they develop. Thus, the developmental trajectory of ASIBs appears unstable when compared to TD children.

Table of Contents

Introduction

ASD Symptomatology

Autism spectrum disorder (ASD) is a heterogeneous disorder characterized by impairments in social interaction and communication behaviors coupled with restricted, repetitive, and stereotypic behavior (American Psychiatric Association, 2013). There are two domains of behavioral features that define the ASD diagnostic criteria. The first is persistent deficits in social communication and social interaction across multiple contexts, which includes deficits in social-emotional reciprocity, in non-verbal communicative behaviors used in social interaction, and in developing, maintaining, and understanding relationships (American Psychiatric Association, 2013). The second is restrictive, repetitive patterns of behaviors, interests, or activities, manifested by at least two of the following: stereotyped or repetitive motor movements, use of objects, or speech; insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal/non-verbal behavior; highly restricted, fixated interests that are abnormal in intensity or focus; or hyper- or hypo-activity to sensory input or unusual interest in sensory aspects of the environment (American Psychiatric Association, 2013). Criteria must be present in the early developmental period, cause significant impairment in social, occupational, or other important areas of functioning, and should not be better explained by intellectual disability or global developmental delay (American Psychiatric Association, 2013).

ASD diagnoses have increased in prevalence within the past few decades, affecting approximately 1 in 59 children (CDC, 2018). However, it is unclear whether this increase reflects an increase in prevalence or an increase in awareness and detection of ASD. A sex bias in diagnosis exists as boys are diagnosed 4 times more commonly than girls (CDC, 2018). ASD often begins to emerge between ages 6-18 months (Szatmari *et al.*, 2016) and mean age of ASD

symptom recognition is between 16-20 months (Cassell *et al.*, 2007). However, there is a time lag between symptom recognition and diagnosis; diagnosis typically occurs between ages 4-6 (Szatmari *et al.*, 2016). This is consistent with the CDC's (2018) findings that median age of diagnosis for ASD is 52 months.

Etiology

The etiology of ASD is multifactorial with a strong genetic basis (Faras *et al.*, 2010). This is supported by twin studies which report autism prevalence rates in monozygotic and dizygotic twins are 90-100% and 10-33%, respectively (Faras *et al.*, 2010, Goldberg *et al.*, 2005) Additionally, the recurrence rate of ASD in younger siblings of individuals diagnosed with ASD is around 20%, which is much higher than prevalence rates in the general population (Ozonoff *et al.*, 2011).

Research into familial patterns of ASD has indicated there is a behavioral phenotype in relatives, including siblings, that is milder but qualitatively similar to behaviors within the two domains of ASD (Gerdts & Bernier, 2011, Toth *et al.*, 2007). This broader autism phenotype (BAP) encompasses behaviors related to the core diagnostic domains of ASD. Siblings within the BAP demonstrate subclinical deficits in social responsivity, communication abilities, and personality traits (Cassell *et al.*, 2007, Gerdts & Bernier, 2011). Siblings of children with ASD, who do not meet ASD or BAP thresholds, may be at risk for other learning disabilities and language disorders at higher rates than the general population (Gerdts & Bernier, 2011).

Broad ASD Phenotype

The BAP is characterized by sub-threshold impairments in personality, social behavior, and language that are typically present within relatives, especially siblings, of children diagnosed with ASD (Losh *et al.*, 2008). Research has indicated that as early as 12-18 months, siblings of children with ASD (ASIBs) exhibited significant differences in cognitive, motor, language, and social development when compared to low-risk typically developing (TD) children, with deficits in social communication being most prominent (Ozonoff *et al.*, 2014). In one study, 28% of ASIBs demonstrated atypical development compared to low-risk TD children by 36 months (Ozonoff *et al*., 2014).

However, it is difficult to distinguish whether these findings are truly indicative of the BAP or are merely early indicators of the full autism phenotype (Sucksmith & Hoekstra, 2011). Ozonoff *et al.* (2011) have suggested that among children later diagnosed with ASD, there was a period in early development where sub-threshold ASD features emerged but did not elicit an ASD diagnosis. Longitudinal research has been conducted to qualify the BAP as a distinct phenotype (Losh *et al.,* 2008). This research has indicated that ASIBs, who do not later receive a diagnosis of ASD, still present deficits across cognitive, language, and social communication domains.

Research assessing cognitive ability of ASIB and TD children has indicated that ASIBs show increased cognitive deficits when compared to their TD peers (Toth *et al.*, 2007, Ozonoff *et al*., 2014). When measuring performance on cognitive tasks, Toth *et al.* (2007) found that approximately 29% of ASIBs showed deficits in overall cognitive abilities, including composite IQ scores and visual reception, fine motor, and gross motor abilities, when measured against typically developing children of comparable age. Similar deficits in cognitive abilities were

identified in other research in children by 36 months of age (Ozonoff *et al.*, 2014, Georgiades *et al.*, 2013). While most cognitive deficits appeared in conjunction with other impairments, less than 10% of high-risk ASIBs showed cognitive delay alone (Ozonoff *et al.*, 2014). However, some studies have shown that these cognitive delays are short-lived. Gamliel *et al.* (2007) found that at 24 months, a subset of ASIBs showed significantly lower cognitive ability compared to TD children. These differences subsided over time, and by 54 months of age the siblings reached typical cognitive levels and there were no significant differences between the ASIB and TD groups. Overall, research has revealed there is much variability in cognitive function within the BAP.

In addition to cognitive deficits, there is significant research support for the presence of language deficits within the broad ASD phenotype. Toth *et al.* (2007) examined a verbal sample of ASIBs and reported that a significant portion of the ASIB group scored lower than the TD group in receptive language skills; many of the ASIBs also scored lower than average in expressive language skills. These results are echoed by Gamliel *et al.* (2007), who reported that receptive and expressive language remained areas of difficulty for ASIBs who otherwise displayed typical cognitive and language ability. ASIBs also demonstrate fewer symbolic behaviors and less frequent use of words and distal gestures (Toth *et al.*, 2007). Despite these findings, Pilowsky *et al.* (2003) and other researchers have found no significant deficits in language ability of ASIBs. While some research has supported the presence of language deficits within the broad ASD phenotype, there is variation in the severity and frequency of language impairment.

Research examining social communication development in ASIBS has identified a profile of social communication impairment more similar to individuals with ASD than typically

developing peers (Goldberg *et al.,* 2005, Toth *et al*., 2007, Ozonoff *et al*., 2014). Goldberg *et al.* (2005) reported that ASIBs differed significantly from TD group in measures of social interaction, but not differ significantly from the ASD group. This indicates impaired prosocial behavior in ASIBs that mimics impairments in ASD. As early as 18 months, Ozonoff *et al.* (2014) found that ASIBs scored significantly higher on the ADOS than TD children. The ASIB group showed social communication difficulties, including reduced eye contact, infrequent social initiations, and delayed onset of play (Ozonoff *et al.*, 2014). Toth *et al.* (2007) reported that ASIBs demonstrated fewer communicating and gesturing behaviors, as well as less frequent social use of gestures and words. While there were not significant differences in levels of social interaction, shared enjoyment, or joint attention between ASIB and TD groups, siblings responded less to smiling, requesting, and showing (Toth *et al.*, 2007). Overall, researchers have concluded that ASIBs showed lower overall rates of social communication and social-emotional functioning than TD children (Goldberg *et al.,* 2005, Toth *et al*., 2007).

Among those at high risk for developing ASD, deficits in social communication tend to emerge between 6 to 18 months of age (Szatmari *et al.*, 2016). Due to the phenotypic variability within these high-risk populations, researchers have become interested in the developmental trajectories of social communication behaviors. Georgiades *et al.* (2013) found that among 20% of ASIBs, social communication impairment exhibited at 12 months persisted until 36 months of age. This is supported by research by Ozonoff *et al.* (2014), which reported that high-risk siblings displayed impairment in social communication skills between 18-27 months, and these impairments persisted at 36 months. However, Chawarska *et al.* (2014) indicated that social communication impairment could not be used to reliably predict long-term outcomes for high-risk siblings; although 20% of ASIBs demonstrated deficits in social communication skills, over

70% were still classified with TD siblings. Further research is needed to identify the developmental trajectories of social communication behaviors in high risk populations.

Overall, research supports the presence of cognitive, language, and social communication deficits in a subset of ASIBs within the BAP. However, researchers disagree about the prevalence, intensity, and developmental trajectory of these deficits. Additional research is needed to further qualify subtle changes within these deficits over time. To augment these gaps in the field, this study utilizes the Brief Observation of Social Communication Change (BOSCC) to measure change over time in social communication behaviors in toddlers, with the prediction that high-risk ASIBs will demonstrate subclinical deficits within social communication behaviors that persist over time, while their TD peers will demonstrate few deficits within social communication behavior at either time point. The BOSCC is more sensitive to capturing subtle variations in social communication behaviors that are core to ASD diagnoses than diagnostic measures like the Autism Diagnostic Observation Schedule (ADOS) (Grzadzinski *et al.*, 2016).

The study aims to measure subtleties in behavior that could help refine the phenotype of the BAP in non-autistic siblings. The refinement of the BAP has important clinical implications. ASIBs who meet criteria for the BAP can be routinely monitored to detect if there is improvement or decline in cognitive, language, or social communication ability. Research has suggested that some children, who are later diagnosed with ASD, go through a period where they exhibit sub-threshold symptoms that do not elicit a diagnosis (Ozonoff *et al.,* 2011). Consistent monitoring of children who meet criteria for the BAP could lead to a quicker ASD diagnosis if their deficits worsen over time, which may help decrease the delay between ASD symptom recognition and diagnosis (Szatmari *et al.*, 2016). Alternatively, refining the BAP prevents a misdiagnosis of ASD for children who exhibit deficits in cognitive, language, or social

communication functioning but are below the ASD threshold and who may grow out of deficits they demonstrate early on.

Methods

Participants

This study included 31 male toddlers. Participants comprised two groups, typically developing (TD) children and children who have a sibling with ASD, but have not received an ASD diagnosis themselves (ASIB). Participants were each seen at two time points. At the first assessment, participants were an average of 24.76 months old (SD= 1.46). ASIB participants were 24.33 months old (SD= 0.80) and TD participants were 25.51 months old (SD= 1.86). At the second assessment, participants were an average of 41.58 months old (SD= 8.55). ASIB participants were 44 months old (SD= 8.21) and TD participants were 39 months old (SD=5.21). See Table 1 for sample demographics.

Two independent samples t-tests were conducted to determine if there were significant differences between the ages of ASIB and TD participants at each the first and second assessment. There was no significant difference between the ages of ASIB and TD participants at the first ($t(18.78)= -1.68$, $p= 0.11$) or second assessment ($t(25.6)= 2.04$, $p> 0.05$).

Three scales, visual reception (VR), receptive language (RL), and expressive language (EL), from the Mullen Scales of Early Learning (MSEL), were used as a measure of development of the ASIB and TD groups. Mullen scores were determined at each assessment, so the average of the VR, RL, and EL scores was found for each participant (Table 1). Three independent samples t-tests were conducted to determine if there were significant differences in the VR, RL, and EL scores between the ASIB and TD populations. There were no significant

differences between the VR scores ($t(26.23) = -0.70$, $p = 0.49$), RL scores ($t(26.70) = -1.36$, $p = 0.19$) or EL scores ($t(24.10) = -1.36$, $p = 0.19$). Two participants were not included in the MSEL analysis (1 TD, 1 ASIB) because the MSEL was not collected at their assessment visit.

Measures

Diagnoses of Autism Spectrum Disorder

The Autism Diagnosis Observation Schedule-2 (ADOS-2) is a diagnostic measure that aims to elicit behaviors that are indicative of an autism spectrum disorder. The ADOS assesses communication, social interaction, and play within standardized activities. These activities aim to provide structured and unstructured contexts within which interactions between the child and examiner can occur. This allows the examiner to identify the occurrence of typical and atypical behaviors. There are four ADOS modules designed to evaluate children of differing developmental and language levels. Examiners select a module that is appropriate for a child's expressive language ability and chronological age. For the purposes of this study, the ADOS will be used to inform the clinical best estimate of ASD in all participants. Additionally, clips from ADOS assessments will be compiled to generate BOSCC videos; description of the BOSCC to follow.

Social Communication Change

The Brief Observation of Social Communication Change (BOSCC) is a behavioral coding measure used to rate social communication and restricted and repetitive behaviors, often seen as part of the behavioral presentation of ASD. The BOSCC is intended to measure change over time by comparing two BOSCC total scores from different time points. Higher scores on the BOSCC indicate more impaired social communication, while lower scores indicate more

advanced social communicative skills. This study will use BOSCC scores from two subsequent assessments to determine a change score (e.g. Time 2-Time 1 = change score). Therefore, a positive change score indicates a decrease, in either frequency or quality, of social communication behaviors, while a negative change score indicates an increase, in either frequency or quality, of social communication behaviors. The BOSCC coding scheme will be applied to video clips of specific tasks that take place within the ADOS; these tasks have been designated by the authors of the measure (Kitzerow *et al.*, 2015). Tasks include free play, play with bubbles, play within a birthday or bath time scheme, and play with a balloon. Joint attention tasks or snack time may be added when the aforementioned tasks are shorter than necessary. BOSCC videos are composed of two segments, segments A and B. Segment A is composed of the free play and bubbles tasks; a joint attention task is added when when the free play and bubbles tasks do not total 6 minutes. Segment B is composed of either the birthday or bath time task and the balloon task; a snack clip is added when these tasks do not total 6 minutes. Segments A and B total 12 minutes of codable behavior. Each segment is watched and scored twice; there are 15 individual criteria scored 0-5 in each segment. The scores for the first 12 items are summed together to produce a segment score for each A and B. The segment A and B scores are averaged to produce a total score for the assessment. The three criteria not averaged into the total score include activity level, disruptive behavior/irritability, and anxious behavior. Few children exhibit these behaviors, yet they are included on the BOSCC to determine whether the observation provides a valid representation of the child's behavior (Grzadzinski, 2016).

Developmental Level

The Mullen Scales of Early Learning (MSEL) is a measure that assesses cognitive and motor ability in children under the age of 5, typically to evaluate their intellectual development and readiness for school. There are five domains encompassed in the MSEL: gross motor (GM), visual reception (VR), fine motor (FM), expressive language (EL), and receptive language (RL). In this study, the visual reception, expressive language, and receptive language scales were used to compare the cognitive and language abilities of the ASIB and TD populations. The visual reception scale uses non-verbal problem solving tasks to measure cognitive ability. The expressive language scale measures language production, while the receptive language scale measures language processing and understanding. Standardized VR, EL, and RL T-scores have an average of 50 and a standard deviation of 10.

Procedures

This study will utilize data from an ongoing longitudinal study conducted by the USC Neurodevelopmental Disabilities Lab examining ASD symptomatology in infants and toddlers at risk for developing autism. As part of the parent study, participants are seen up to 7 times, beginning as early at 9 months of age with diagnostic evaluations for ASD beginning at 24 months, with annual visits occurring thereafter. Participants were selected when their initial ADOS assessment ruled out a diagnosis of ASD. The BOSCC coding scheme was applied to two subsequent ADOS assessments, including the initial ADOS assessment at 24 months. The subsequent assessment occurred when the child was less than 60 months old. For each BOSCC assessment, a total score, the average of the two segments of the BOSCC, was derived.

In order to use the BOSCC protocol, inter-rater reliability was achieved. To attain inter-rater reliability, two independent raters must agree on 80% of the codes in Segments A and B and must differ by no more than 3 points in the total score for three consecutive BOSCC videos. To maintain inter-rater reliability, 20% of the videos coded in this study were recoded by the primary coder, who is research trained on the BOSCC.

Research Question

How do siblings of children with an ASD, who do not meet criteria for ASD, exhibit change in social communication behaviors over time compared to typically developing children during the toddlerhood to preschool years?

Data Analysis

To answer the research question, 62 BOSCC videos were available for coding and analysis. There were 32 videos featuring an ASIB participant and 30 videos featuring a TD participant. 14 of these videos required an additional segment (i.e. joint attention or snack) to meet the 12-minute threshold. During coding, a BOSCC total score was calculated for each video. Once two videos were coded for a participant, a BOSCC change score was determined. Three independent samples t-tests were run using R to determine if there were significant differences between the first assessment scores, second assessment scores, and change scores of the ASIB and TD populations.

Results

Average assessment and change scores for ASIB and TD populations are presented in Table 2. The differences between first and second assessment scores for both populations are displayed in Figure 1. Results of the three independent samples t-tests are shown in Table 3.

There was not a significant difference between total scores for the first assessment ($t(28.51)= 1.60$, $p= 0.12$) nor was there a significant difference between total scores for the second assessment ($t(28.94)= -0.27$, $p= 0.79$) . However, there was a significant difference between change scores for ASIB and TD participants, with $t(28.44)= -2.48$, $p= 0.02$.

Discussion

This study aimed to investigate how ASIB children develop social communication skills over time compared to typically developing children, in hopes to further define the broad autism phenotype. It was hypothesized that the ASIB group would demonstrate subclinical deficits within social communication behaviors that persisted over time and differentiated them from their typically developing peers. However, this hypothesis was not supported, as infant siblings of children with ASD did not show significant differences in social communication behavior at either time point compared to typically developing children.

While the BOSCC scores between groups at Assessment 1 were not significantly different, the ASIB group did score higher than the TD group, indicating that the ASIBs initially showed more social communication deficits than the TD participants. By Assessment 2, BOSCC scores, and thus social communication behaviors, were comparable between the two groups. The social communication development shown by ASIBs led to a significant difference between the BOSCC change scores of the two groups, indicating the ASIB group developed more typical

social communication patterns over time while the TD group displayed little to no change in social communication during the time period.

These results suggest that ASIBs may show impairment in social communication at a younger age, however, these impairments may lessen with time and ASIBs may reach typical levels of social communication as they develop. Gamliel *et al.* (2007) supports this, reporting that infant ASIBs grew out of early cognitive and language deficits by 54 months. The results of this study and of Gamliel *et al.* (2007) indicate that the developmental trajectory of social communication skills in ASIB infants is unstable when compared to low-risk typically developing children. In this sample, TD children seemed to develop sufficient social communication skills by the age of 2. These social skills appeared stable, as assessment scores did not differ much between the two time points for the TD group. This is expected, as the skills that are measured on the BOSCC align with the developmental abilities of typically developing children.

It appears contradictory that assessment scores did not differ significantly at the first or second time point, while the overall change scores did differ significantly. This is most likely attributable to the instability within social communication behaviors of the ASIB group. Many ASIBs fall within the broad ASD phenotype, which has been characterized by deviations from typical cognitive and language ability and social communication behaviors (Ozonoff *et al.*, 2014, Toth *et al.*, 2007, Gamliel *et al.*, 2007). This sample of ASIBs exemplified the variability present within this phenotype. Throughout this study, ASIB participants demonstrated varying degrees of social communication ability. These variations produced a large standard deviation from the average BOSCC Assessment 1 score in the ASIB group. Thus, it was difficult to detect a non-significant difference in the Assessment 1 total scores. It is assumed that the change scores did

differ significantly because the BOSCC was able to capture the changes in social communication behaviors that occurred over time. This signifies the importance of longitudinal research in examining ASIBs and those at high risk for developing ASD.

No ASIB participant began any course of medication during their participation in this experiment. Thus, medication can be ruled out as a confounding variable. It is unknown whether any ASIB participants were involved in other treatment programs, as this information was not available for analysis. If ASIB participants did receive treatment, this could account for decrease in BOSCC total scores from Assessment 1 to Assessment 2, which has thus far been attributed to ASIBs late development of typical social communication patterns.

There was much group variation within the TD sample as well, although this variation is assumed to be due to age-dependent differences within individual development, as average assessment scores for the TD group remained stable over time.

Limitations of this study include small sample sizes, which exacerbated group variability, making it difficult to detect significant differences among group assessment scores. It is advised that future studies include more participants so that variation within each group can be minimized. It is also recommended that future studies include more time points to examine the stability of ASIB social communication deficits and the stability of TD development. Szatmari *et al.* (2016) found that, due to variability of developmental trajectories, ASIBs with high-familial risk for ASD should be monitored over multiple time points throughout the first three years of life.

A portion of high-risk siblings do become diagnosed with ASD. Ozonoff *et al.* (2011) found within ASIB populations, ASD diagnoses determined at 18 or 24 months were extremely stable, with 93% and 82% accuracy, respectively, when measured again at 36 months. Contrary

to this, Chawarska *et al.* (2014) observed that a large portion of siblings could not be reliably diagnosed with ASD at 18 months, although they received an ASD diagnosis later on. Taken together, these studies show that 40% of high-risk siblings, who later receive ASD diagnoses, are symptomatic by 18 months, and within these siblings, diagnoses based on comprehensive assessment are 93% stable (Szatmari *et al.*, 2016). Thus, Szatmari *et al.* (2016) recommends that siblings who display clinical ASD symptoms by 18 months should be referred for a diagnosis, while siblings who display subclinical symptoms be monitored periodically until age 3. Clinicians must be sensitive to emergence of symptoms over time to properly diagnose their patients. They must also be careful when diagnosing in infancy and toddlerhood, as research shows that siblings can grow out of atypical behaviors exhibited in early childhood (Gamliel *et al.*, 2007). The BOSCC is an important tool in tracking the developmental trajectories of ASIBs, as it detects subtle changes in social communication behaviors over time. Many screening protocols only examine a single point in time, and may not be sensitive enough to capture these changes.

Figures and Tables

Table 1

Sample Demographics

	ASIB (n=16)	TD (n=15)
	Mean (SD)	Mean (SD)
Age		
Assessment 1	24.33 (0.80)	25.21 (1.86)
Assessment 2	41.58 (8.21)	39.00 (5.21)
Mullen Scales of Early Learning		
Visual Reception		
Assessment 1	46.73 (12.86)	50.93 (11.67)
Assessment 2	49.87 (15.34)	52.00 (12.41)
Average of Assessments	48.30 (13.62)	51.46 (10.65)
Receptive Language		
Assessment 1	44.53 (15.17)	53.00 (14.26)
Assessment 2	43.0 (14.39)	47.29 (13.25)
Average of Assessments	43.87 (12.86)	50.46 (13.30)
Expressive Language		
Assessment 1	37.07 (11.02)	45.71 (13.64)
Assessment 2	40.60 (11.67)	43.86 (14.59)
Average of Assessments	38.83 (9.99)	44.79 (13.30)

Table 2

Means and Standard Deviations of BOSCC Total Scores and Change Scores

		First Assessment	Second Assessment	Change Score
	n	Mean (SD)	Mean (SD)	Mean (SD)
ASIB	16	19.91 (7.62)	14.75 (8.30)	-5.16 (5.74)
TD	15	15.37 (8.13)	15.53 (8.13)	0.17 (6.18)

Figure 1

ASIB Changes in Assessment Scores vs. TD changes in Assessment Scores

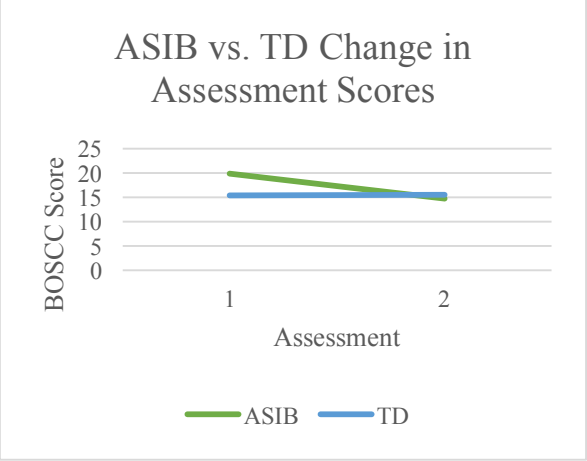

Table 3

T-test for BOSCC Total Scores and Change Scores

	t	*df*	*p*
First Assessment	1.60	28.51	0.12
Second Assessment	-0.27	28.94	0.79
Change Score	-2.48	28.44	0.02

Table 4

Problems with BOSCC videos

	Joint attention added	Snack added	Both added	Timing problems	Lighting problems	Audio problems	Other problems
ASIB	3.13%	3.13%	7.81%	3.13%	1.56%	3.13%	1.56%
TD	N/A	6.25%	1.56%	N/A	3.13%	N/A	1.56%

Resources

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