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Developmental Functioning of Infants and Toddlers with Autism and Down Syndrome

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DEVELOPMENTAL FUNCTIONING OF INFANTS AND TODDLERS WITH AUTISM AND DOWN SYNDROME

A Thesis

Submitted to the Graduate Faculty of the Louisiana State University Agricultural and Mechanical College in partial fulfillment of the requirements for the degree of Master of Arts

in

The Department of Psychology

by

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Abstract

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in social communication, as well as restricted interests and repetitive behaviors. Approximately 10% of individuals with ASD also have comorbid genetic or chromosomal conditions, like Down Syndrome (DS). While it was once believed that DS and ASD rarely co-occurred, it has been demonstrated that it is not uncommon for children with DS to also meet criteria for ASD. Due to the difficulties in differentiating between impairments associated with intellectual disability (ID) and ASD symptomology, DS often leads to delayed or misdiagnoses of ASD. This can interfere with early intervention services and appropriate educational placements. While prior research has compared developmental functioning in children with ASD and DS, no studies have examined the impact of ASD risk and DS on developmental functioning in infants and toddlers under the age of 3. Utilizing the Baby and Infant Screen for Children with Autism Traits-Part 1 (BISCUIT-Part 1) and the Battelle Developmental Inventory, Second Edition (BDI-2), the current study aimed to expand the existing literature by investigating the effects of ASD risk and DS on developmental functioning in infants and toddlers. The current study compared three groups consisting of 46 toddlers each: ASD screen positive only without co-occurring DS (ASD+), DS screen negative (DS-), and DS screen positive (DS+). The results of the current study revealed significant group differences in the overall developmental functioning, as well as each developmental subdomain of the BDI-2. These findings support the need for early screening and identification of ASD among those with genetic conditions, like DS.
Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in social communication and social interaction and the presence of restricted, repetitive behaviors (American Psychiatric Association, 2013). Since the 1990’s, prevalence rates of ASD have increased substantially (Matson & Kozlowski, 2011; Christensen et al., 2016). ASD currently effects approximately 1 in 54 children in the United States, according to the Centers for Disease Control and Prevention (“Autism Spectrum Disorder (ASD): Data & Statistics”, 2020). Early diagnosis and intervention are crucial to improving outcomes for children with ASD (Woods & Wetherby, 2003, Koegel et al., 2014). The importance of early diagnosis and intervention are well-established and have been supported by several studies (Volkmar, 2014; Rotholz et al., 2017; Elder et al., 2017). While experienced clinicians can diagnose ASD in children as young as 18 months of age (Ozonoff et al., 2015; Zwaigenbuam et al., 2016), diagnosis can be delayed up to three years after the presentation of the initial concerns to a clinician. Many children do not receive an ASD diagnosis before the age of 5 (Shattuck et al., 2009; Zuckerman et al., 2015). This is especially concerning as research has indicated that interventions implemented before 4 years of age are related to improvements in cognition, language skills, and adaptive behavior (Dawson et al., 2010; Vivanti et al., 2016). Thus, it is necessary for children to receive an ASD diagnosis as early as possible to improve their outcomes.

Several barriers may contribute to a delayed diagnosis of ASD, including the presence of comorbid conditions. In recent years, co-occurring genetic and chromosomal conditions have been documented in approximately 10% of children with ASD (Davis et al., 2017; Oxelgren et al., 2019). One such condition associated with ASD is Down Syndrome (DS), a genetic condition
that is caused by a full or partial trisomy of chromosome 21. It is characterized by distinctive facial features, intellectual disability, and increased occurrence of comorbid medical conditions, such as celiac disease, hearing loss, and congenital heart defects (Molloy et al., 2009; Moss et al., 2012; Centers for Disease Control and Prevention, 2015). It is the most common cause of genetic intellectual disability and occurs in about 1 in 700 births each year (Centers for Disease Control and Prevention, 2015). While ASD is comorbid in only a portion of individuals with DS, autism symptomology is important to consider as ASD is typically diagnosed later and underdiagnosed compared to children without DS (Gray et al., 2011). Due to difficulties in differentiating between behaviors associated with ASD and communication and cognitive impairment associated with intellectual disability, it can be exceedingly challenging to diagnose DS-ASD (Ji et al., 2011).

Since Down Syndrome can confound identification and treatment of ASD, the current study seeks to further an understanding of DS, ASD, and developmental functioning. Although previous research has compared developmental functioning between children with ASD only and DS only (Loveland & Kelley, 1988; Rodrigue et al., 1991), very few studies have investigated developmental functioning in children with DS at risk for ASD. Diagnosis and treatment strategies for children with comorbid DS-ASD may be informed by a better understanding of developmental functioning in these children. Using a sample of infants and toddlers will also expand the research to help understand whether differences in developmental functioning are present early in life. Thus, the present study looks to examine the impact of DS on developmental functioning in toddlers. Comparisons of the overall developmental functioning, as well as the subdomains of developmental functioning (i.e. Adaptive, Personal-Social, Communication, Motor, and Cognitive) will be examined in toddlers at risk for ASD only, with
DS at risk for ASD, and with DS who are not at risk for ASD. The results of the current study will have clinical implications for the assessment of ASD in toddlers and young children.

**Autism Spectrum Disorder**

**History of ASD**

Leo Kanner is credited with first describing the condition currently known as autism spectrum disorder in 1944. In his publication entitled “Autistic Disturbances of Affective Contact”, Kanner provided a detailed account of 11 children who presented with a similar set of symptoms unique from any existing conditions (Kanner, 1944). While variations existed in the severity and manifestation of the symptoms, there were commonalities in the core deficits exhibited by the children. Some of these key features included: abnormal speech patterns, repetitive behaviors with an insistence on sameness, and difficulty relating to other people (Kanner, 1944). In regard to communication, 3 out of the 11 children Kanner observed did not have the ability to speak. The children that did speak exhibited echolalia, or the repetition of words and phrases, and did not use words in a functional manner to convey their thoughts (Kanner, 1944). Another important shared feature was the “obsessive desire for the maintenance of sameness” (Kanner, 1944, p. 245). This strict adherence to routine led to distress in the children when there were changes in the environment. However, among these symptoms, Kanner (1944) emphasized “extreme autistic aloneness”, or the inability to relate to others, as being the most characteristic of these children.

Due to their presenting symptoms, Kanner believed children with this condition may have been erroneously thought to have schizophrenia or ‘feeble-mindedness’ in the past (Wolff, 2004). However, he believed the symptoms to be distinct from schizophrenia, as they seemed to
manifest from birth and the social deficits generally appeared to improve throughout development (Barahona-Corrêa & Filipe, 2016).

One year later, Hans Asperger published a paper entitled ‘Die Autistischen Psychopathen im Kindesalter’ (‘Autistic Psychopathy in Childhood’) (Asperger, 1944/1991). However, English speaking researchers were not made aware of the paper until nearly 40 years later, as it was published in German (Wing, 1981). It was later made available in English in 1991 (Asperger, 1944/1991). Like Kanner, Hans Asperger used the same term, autism, to describe the case studies of boys with commonalities in social difficulties, characteristic stereotypies, and special interests (Asperger, 1944/1991; Barahona-Corrêa & Filipe, 2016). Similar to Kanner’s descriptions, Asperger described the severe social problems as the core feature of the disorder (Asperger, 1944/1991). While the children Asperger observed had adequate linguistic skills, they struggled to use speech in a functional manner. Additionally, they exhibited narrow interests, often at the exclusion of other activities, behavioral problems, rigid thinking, and motor clumsiness. Asperger also noted that the traits he observed were seen almost exclusively in boys, stating that “the autistic personality is an extreme variant of male intelligence” (Asperger, 1944/1991).

The initial descriptions of autism, provided by Kanner and Asperger, created confusion among professionals attempting to differentiate childhood autism from schizophrenia. Eugen Bleuler originally coined the term “autism” to refer to the concept of social withdrawal in patients with schizophrenia (Evans, 2013; Barahona-Corrêa & Filipe, 2016). For years following the descriptions of autism as a childhood disorder, confusion persisted as professionals struggled to differentiate it from schizophrenia. It was erroneously believed that the behaviors exhibited by children with autism were early onset indicators of schizophrenia (Boucher, 2017). However,
both Kanner and Asperger emphasized that childhood autism and schizophrenia were distinct conditions (Barahona-Corrêa & Filipe, 2016; Kanner, 1944). As Kanner noted, children with childhood autism expressed “extreme autistic aloneness” from birth often “failing to develop the usual amount of social awareness” (Kanner, 1944, p. 242). This differs from those with schizophrenia who develop a change in behavior and withdraw from previously established relationships (Kanner, 1944; Barahona-Corrêa & Filipe, 2016). Kanner also described a difference in the onset of the symptoms manifested in both conditions. While individuals with autism evinced an earlier onset of symptoms that were present from the beginning of life, those with schizophrenia typically demonstrated a gradual onset of symptoms (Kanner, 1944). In addition, the prevalence of childhood autism was reported to be significantly higher in males than females. This differed from schizophrenia which had similar prevalence rates in both males and females (Kanner, 1944; Rutter, 1968).

Following the descriptions of autism, several ideas were posited in an attempt to account for the etiology of the condition. In Kanner’s description of autism, he noted that among the children he observed, “there [were] very few really warmhearted fathers and mothers” (Kanner, 1944). He believed that many of the parents lacked interest in other people and appeared to be cold in nature (Kanner, 1944). Subsequently, Kanner published a paper entitled “Problems of Nosology and Psychodynamics of Early Infantile Autism” in 1949. In this paper, Kanner described that autism in children may be related to a lack of maternal warmth. He believed that children with autism were raised in emotional refrigerators, withdrawing from social interactions to seek comfort in their aloneness (Kanner, 1949). Bruno Bettelheim aided in the acceptance of the “refrigerator mother” theory in his book “Empty Fortress: Infantile Autism and the Birth of the Self” (1967). While the idea that autism is caused by disturbed mother and child
relationships was disproved by the 1970’s, it contributed greatly to the further isolation of families with children with autism (Boucher, 2017). Behavioral theories have also been proposed to explain the etiology of autism. For example, Ferster (1961) suggested that conditions in a child’s home could initiate schedules of reinforcement which increase the behaviors associated with autism (Hixon et al., 2008). However, other researchers suggested physiological causes of autism. These researchers completed studies which demonstrated that people with autism had abnormalities in their brain structure and functioning (Hutt et al., 1965; Rimland, 1964; Rutter et al., 1968). While many early hypotheses on the etiology of autism have since been disproven, subsequent research has supported underlying neurobiology as being related to autism (Ha et al., 2015; Nickl-Jockschat et al., 2012). More recently, genetic factors have been shown to contribute to the etiology of ASD, along with environmental effects (Bai et al., 2019; Karimi et al., 2017).

**Diagnostic Criteria for ASD**

While diagnostic criteria for ASD has significantly changed over time, Kanner’s initial descriptions included several features that remain part of the diagnostic criteria for the disorder (Volkmar & Reichow, 2013). Autism was first acknowledged as a diagnosis in the *Diagnostic and Statistical Manual of Mental Disorders, Third Edition (DSM-III)* in 1980 and was referred to as infantile autism (American Psychiatric Association, 1980; Volkmar & McPartland, 2014). Infantile autism, childhood-onset pervasive developmental disorder (PDD), and atypical PDD were included as a class of disorders called pervasive developmental disorders, distinct from schizophrenia (Volkmar & Reichow, 2013; Harris, 2018). The criteria for infantile autism included an onset before 30 months of age, pervasive lack of responsiveness to other people, unusual responses to the environment, deviant language development, and the absence of delusions and hallucinations that characterize schizophrenia (APA, 1980; Volkmar & Reichow,
In order to be diagnosed with childhood-onset PDD, disturbances in social relationships had to develop after 30 months of age but prior to age 12. Atypical PDD included individuals that did not meet criteria for either infantile autism or childhood-onset PDD, but still had atypical development across several domains (APA, 1980; Spitzer & Cantwell, 1980). However, the diagnostic criteria set forth in the DSM-III were quite restrictive, as they were better applied to diagnosing young children with severe impairments (Volkmar & Reichow, 2013).

In the *Diagnostic and Statistical Manual of Mental Disorders, Third Edition-Revised* (*DSM-III-R*; American Psychiatric Association, 1987), the name of the disorder changed and included a subthreshold category (APA, 1987; Harris, 2018). The term infantile autism was changed to Autistic Disorder, while the childhood onset-PDD and atypical PDD subtypes were reclassified as Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS) (APA, 1987; Harris, 2018). The criteria for Autistic Disorder included symptoms involving impairments in social interaction, verbal and nonverbal communication, and the presence of a significantly restricted area of interests. While Autistic Disorder included a specifier if the onset was after 36 months of age, generally the requirement was that the disorder would emerge in infancy or early childhood. The criteria for PDD-NOS were similar to the criteria for PDD in the DSM-III; individuals that were impaired across developmental domains but did not meet criteria for Autistic Disorder would be classified as PDD-NOS (APA; 1987). Although the DSM-III-R addressed the lack of developmental orientation in the DSM-III, it tended to over-diagnose those individuals with cognitive impairments and under-diagnose those without cognitive impairments (Volkmar & Reichow, 2013). This led to increases in the false positive rates of diagnosis (Harris, 2018).
The Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV, American Psychiatric Association, 2000) began with the goal to increase specificity and sensitivity across various ages and intellectual quotient ranges (Harris, 2018). Additionally, changes were made to the classification of the disorder. The categories of PDD in the DSM-IV included Autistic Disorder, Rett’s Disorder, Childhood Disintegrative Disorder, Asperger’s Disorder, and PDD-NOS (APA, 2000; Harris, 2018). These diagnostic categorizations continued to be used in the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revised (DSM-IV-TR; American Psychiatric Association, 2000). The criteria for Autistic Disorder included impairments in social interaction, communication, and the presence of restricted, repetitive, and stereotyped patterns of behavior. Similar to the DSM-III, PDD-NOS described individuals that were impaired across developmental domains but did not meet criteria for Autistic Disorder. Asperger’s Disorder was made up of individuals with deficits in social interaction and the presence of restricted repetitive and stereotyped patterns of behaviors, interests, and activities; however, the criteria for Asperger’s Disorder did not include deficits in communication or language. In order to meet criteria for Rett’s Disorder or Childhood Disintegrative Disorder, clinically significant loss of previously acquired skills had to be present. However, individuals with Rett’s disorder experienced a loss of previously acquired hand skills between the ages of 5 and 30 months of age, loss of social skills, uncoordinated gait, and impaired expressive and receptive language development. Conversely, to meet criteria for Childhood Disintegrative Disorder, children experienced typical development for the first two years of life, followed by a significant loss of previously acquired skills in areas such as expressive or receptive language, social skills, bowel or bladder control, play, or motor skills, before 10 years of age. At least two abnormalities in communication, social interaction, or
restricted, repetitive behaviors also had to be present for the diagnosis of Childhood Disintegrative Disorder.

In the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5; American Psychiatric Association, 2013) several key changes took place. A decision was made to eliminate the diagnostic subcategories that made up PDD in the DSM-IV-TR. Rather, autism was conceptualized as a spectrum of disorders called Autism Spectrum Disorder (Regier et al., 2010; Barton et al., 2013). Thus, Autistic Disorder, Asperger’s Disorder, and PDD-NOS became part of autism spectrum disorder. Rhett’s disorder is currently considered a neurological condition and Childhood Disintegrative Disorder is no longer categorized as a separate developmental disorder (Volkmar & Reichow, 2013). In past editions, ASD was thought to have consisted of a triad of features, including deficits in social interaction, impaired social communication, and restricted and repetitive behaviors (Barton et al., 2013; APA, 2000); however, in the DSM-5, the diagnostic criteria were reduced to two domains, restricted and repetitive behaviors and social communication deficits (APA, 2013; Volkmar & McPartland, 2014).

The ASD criteria designated for the DSM-5 are also more stringent than previous diagnostic criteria (Barton et al., 2013). For example, in the DSM-5, individuals must meet all three criteria in the social-communication domain and two of the four criteria in the category of restricted and repetitive behaviors (APA, 2013). In the social-communication domain, impairments must be present in social-emotional reciprocity (e.g., reducing sharing of interests or failure to initiate or respond to social interactions), nonverbal communication used for social interaction (e.g., abnormalities in eye contact), and developing, maintaining, and understanding relationships (e.g., absence in interest in peers or difficulties sharing in imaginative play).
Regarding restricted, repetitive patterns of behavior, abnormalities must be present in two of the four areas including: stereotyped or repetitive motor movements (e.g., lining up toys or echolalia), insistence on sameness or inflexible adherence to routines (e.g., extreme distress at small changes), highly restricted, fixated interests that are atypical in intensity (e.g., strong attachment to certain objects), or hyper- or hypo-reactivity to sensory aspects of the environment (e.g., atypical visual exploration of lights). Additionally, three levels of symptom severity can be specified for ASD. An ASD Level 1 diagnosis indicates that the deficits related to ASD require support. Level 2 indicates that the deficits require substantial support and level 3 specifies that the impairments require very substantial support (APA, 2013).

**Prevalence of ASD**

While ASD was once considered rare, affecting approximately 4 in 10,000 children (Ritvo et al., 1989; Kirby et al., 1995), prevalence rates have been substantially increasing since the 1990’s (Christensen et al., 2016). According to the Centers for Disease Control and Prevention (CDC), approximately 1 in 54 children in the United States are currently identified with ASD (“Autism Spectrum Disorder (ASD): Data & Statistics”, 2020). There are several proposed explanations for the increased prevalence rates of ASD including better identification and screening tools, earlier diagnosis, increased awareness of ASD, better access to services, and changes to the diagnostic criteria for ASD (Rice et al., 2015; Neggers, 2014). With each updated publication of the Diagnostic and Statistical Manual of Mental Disorders, changes have been made to the symptoms included, number of symptoms required for an ASD diagnosis, and the diagnoses classified as ASD (Neggers, 2014; Kites et al., 2013). Many researchers have suggested that these changes to the diagnostic criteria may account for the increased prevalence rates of ASD (King & Bearman, 2009). Furthermore, the age of diagnosis of ASD has decreased
in recent years which may contribute toward increasing rates of ASD. Experienced clinicians can now diagnose ASD in toddlers as young as 18 months old (Ozonoff et al., 2015; Zwaigenbaum et al., 2016).

ASD also reportedly presents across all racial identities and socioeconomic strata (Durkin et al., 2017). However, it is well-established in prior research that ASD is considerably more prevalent among males than females (Fombonne, 2009; Werling & Geshwind, 2013). Multiple studies, completed both in the United States and internationally, have found a 4:1 male to female gender difference (Fombonne, 2009; Werling & Geschwind, 2013; Idring et al., 2015).

**Comorbid Conditions**

In addition to the core features of ASD, it is common for children with ASD to also have co-occurring psychiatric conditions (Belardinelli et al., 2016). While research has reported varying prevalence rates, it is estimated that approximately 70% of children with ASD have at least one co-occurring psychiatric condition (Rosen et al., 2018; Siminoff et al., 2008). Anxiety, mood disorders, obsessive compulsive disorder (OCD), attention deficit hyperactivity disorder (ADHD), and oppositional defiant disorder (ODD) are among the most common comorbid conditions in children with ASD (Rosen et al., 2018; Siminoff et al., 2008; Postorino et al., 2016). Medical conditions such as gastrointestinal problems, sleep disorders, and seizures are also significantly more prevalent in individuals with ASD compared to the general public (Gurney et al., 2006; Isaksen et al., 2012; Mazurek et al., 2012; Mazurek & Petroski, 2015). Additionally, co-occurring genetic and chromosomal conditions, like Fragile X syndrome, Rett syndrome, Tuberous Sclerosis, and Down Syndrome, have been documented in approximately 10% of children with Autism Spectrum Disorder (ASD) (Davis et al., 2017; Oxelgren et al., 2019, American Psychiatric Association, 2013). Identifying these comorbid
symptoms is crucial to ensure that children receive appropriate intervention services and educational placements.

**Down Syndrome**

**History of Down Syndrome**

In 1866, John Langdon Down, an English physician, first described what is currently known as Down Syndrome (Down, 1866; Mégarbané et al., 2009). While other physicians (e.g., Jean-Étienne Esquirol and Édouard Séguin) may have reported accounts of people with DS prior to Down’s publication, Down is credited with being the first person to group them together based on phenotypic similarities (Mégarbané et al., 2009; Starbuck, 2011; Stratford, 1996). In his essay, he identified a specific group of individuals with similar features. He noted the characteristic appearance and shared facial features among the individuals, stating “…that when placed side by side, it is difficult to believe that the specimens compared are not children of the same parents” (Down, 1866, p. 260). Based on his observations, Down believed that these individuals, with what is currently known as Down Syndrome, differed from others with cognitive impairments (Down, 1866; Starbuck, 2011). He referred to the group of patients as ‘Mongoloids’, as he believed they resembled people from Mongolia (Down, 1866; Mégarbané et al., 2009). Sadly, by the beginning of the 1900’s, the term ‘Mongolism’ was the accepted term used to refer to what is now called Down Syndrome (Mégarbané et al., 2009).

Throughout the 20th century, people with Down Syndrome were marginalized members of society. It was not uncommon for children with Down Syndrome to be institutionalized and often not provided with the appropriate treatment for co-occurring medical conditions, such as congenital heart defects, vision impairments, or intestinal problems. With the rise of the eugenics movement in America, the Supreme Court case *Buck v. Bell* (1927) established that
forcible sterilization in individuals with intellectual disabilities was not unconstitutional (U.S. Supreme Court, 1927; McCabe & McCabe, 2011).

While Down Syndrome was first described in 1866, the cause of the condition remained unknown until the 1950’s. In 1932, Waardenburg and Davenport suggested that Down Syndrome may be the result of a chromosomal abnormality (Allen, 1974; Davenport, 1932). Years later, the advancements in the science of genetics and the discovery of karyotype techniques led to a greater understanding of Down Syndrome. In 1959, French geneticist Jerome Lejeune and his colleagues discovered that Down Syndrome resulted from the presence of an additional chromosome. The additional chromosome was subsequently labeled Number 21 (Lejeune et al., 1959; Mégarbané et al., 2009). As a result, the term Trisomy 21 began to be utilized in the medical community to describe individuals with the condition (Mégarbané et al., 2009).

In 1961, nineteen geneticists, including John Langdon Down’s grandson, presented a letter to the Lancet. They objected to the term ‘Mongolism’ being used to refer to people with Trisomy 21 (Mégarbané et al., 2009; Allen et al., 1961). In addition to the pejorative nature of the term, it suggested the existence of a racial bias for the condition. As a result, the geneticists suggested that the condition be termed “Langdon-Down anomaly” or “Down Syndrome” instead (Ward, 1999). In 1965, the country of Mongolia submitted an informal request to the World Health Organization (WHO) to stop using the terms “Mongolism” and “Mongoloids” to refer to people with Trisomy 21. After this request, the WHO adopted the recommendation and the name change was confirmed. While Down Syndrome became the universally acceptable term for Trisomy 21, regrettably use of the term “Mongolism” persisted in scientific literature into the 1970’s (Howard-Jones, 1979).

**Prevalence of Down Syndrome**
Down Syndrome is the most common chromosomal disorder ("Birth Defects: Data and Statistics", 2019; Presson et al., 2013; Shin et al., 2009). According to the Centers for Disease Control and Prevention (CDC), Down Syndrome affects nearly 1 in 700 infants ("Birth Defects: Data and Statistics", 2019; Mai et al., 2019). Over the past three decades, the prevalence rates of Down Syndrome have risen steadily. In a cross-sectional study of infants with Down Syndrome, researchers found that there was a 31.1% increase in the babies born with Down Syndrome from 1979 to 2003. In 1979, approximately 9 out of 10,000 infants were born with Down Syndrome, compared to 11.8 out of 10,000 infants in 2003 (Shin et al., 2009). Moreover, the prevalence of Down Syndrome also dramatically increases with maternal age. Thus, older mothers are more likely to give birth to a child with Down Syndrome (Mai et al., 2013; NDSS, 2020a). Despite the increasing prevalence rates of Down Syndrome, the life expectancy for those with the condition has improved significantly. In 1960, the average life expectancy for individuals with Down Syndrome was just 10 years of age. Currently, however, the average life expectancy for those with Down Syndrome is approximately 60 years old (Presson et al., 2013; NDSS, 2020a).

**Types of Down Syndrome**

There are three different types of Down Syndrome. Though caused by varying chromosomal abnormalities, the resulting physical and behavioral characteristics are similar (Centers for Disease Control and Prevention, 2019). Complete trisomy 21 is the first type of chromosomal change that can result in Down Syndrome. This occurs when there is an error in the process of cell division, called nondisjunction (NDSS, 2020a). Nondisjunction arises in an embryo when there are three copies, as opposed to two copies, of chromosome 21. Either prior to or during conception, a pair of the 21st chromosomes in the egg or sperm do not separate properly (NDSS, 2020a). People with complete trisomy 21 have a total of 47 chromosomes.
Complete trisomy 21 is the most common cause of Down Syndrome and accounts for nearly 95% of the cases (NDSS, 2020a; CDC, 2019).

Another type of Down Syndrome is called translocation. Translocation accounts for nearly 2 to 4 percent of cases of Down Syndrome (Shin et al., 2010; NDSS, 2020a). Translocation occurs when a full or partial copy of the 21st chromosome breaks off during cell division and attaches to another chromosome in the body, typically chromosome 14 (NDSS, 2020a). Individuals affected by a translocation have two copies of chromosome 21, as well as the additional copy attached to another chromosome (United States National Library of Medicine, 2020). While Down Syndrome is not typically inherited, translocations can be genetically inherited from parents that do not have Down Syndrome themselves. Either parent may have the genetic material from his or her 21st chromosome rearranged on another chromosome in the body. While this would not result in symptoms or characteristics of Down Syndrome in the parents, it could result in passing on a translocation to children (Mayo Clinic, 2018).

Mosaicism, also known as Mosaic Down Syndrome, arises when nondisjunction of chromosome 21 occurs in only a portion of the body’s cells after fertilization (Down Syndrome Association of Central Ohio, 2019). When Mosaicism is present, not every cell in the body is the same. Rather, some of the cells contain 46 chromosomes, while others have 47 chromosomes. In the cells with 47 chromosomes, an additional chromosome 21 is present (NDSS, 2020a). Mosaicism is the least common form of Down Syndrome and accounts for approximately 1% of the cases (NDSS, 2020a). While the symptoms of Mosaic Down Syndrome may resemble both complete trisomy 21 and translocation, there may also be variations in the symptoms depending on the number of cells with an extra chromosome present in the body (NDSS, 2020a; Parker, 2010).
**Etiology of Down Syndrome**

While there are different types of Down Syndrome, all individuals with Down Syndrome have an extra copy, either full or partial, of chromosome 21 (NDSS, 2020a). The extra chromosome accounts for the characteristics associated with Down Syndrome (NDSS, 2020a). Most frequently, the presence of the extra chromosome is caused by an error in the cell division process. Generally, the cell division error occurs randomly during the formation of an egg or sperm. Researchers investigating cell division errors have indicated that the majority of the time, the extra copy of chromosome 21 comes from the mother’s egg. In approximately 5% of cases, the extra copy of the 21st chromosome results from the father’s sperm. However, on rare occurrences, cell division errors may take place while the embryo grows, after fertilization (Parker et al., 2010).

While all forms of Down Syndrome result from chromosomal abnormalities, only 1% of cases of Down Syndrome can be inherited. In about one-third of individuals with Down Syndrome resulting from a translocation, there is an associated hereditary component. In these cases, either parent is a carrier for a chromosomal translocation (NDSS, 2020a). When parents are carriers of the translocation associated with Down Syndrome, they are at an increased risk for having a child with Down Syndrome (Mayo Clinic, 2018).

Additionally, the most well-known and only certain risk factor related to Down Syndrome is advanced maternal age. The association between maternal age and Trisomy 21 has been replicated in various populations and studies (Coppedè, 2016; NDSS, 2020a; Morris et al., 2003). After a maternal age of 35, the risk of having a child with Down Syndrome increases substantially alongside maternal age (Coppedè, 2016; Morris et al., 2002). For example, a 35-year-old woman has a 1 in 350 chance of conceiving a child with Down Syndrome. However, the
odds drastically increase as maternal age increases. By the age of 45, women have a 1 in 30 chance of conceiving a child with Down Syndrome (NDSS, 2020a). Maternal age has not been shown to be associated with Down Syndrome that is the result of translocation (NDSS, 2020a). Currently, there are no known behavioral or environmental risk factors associated with Down Syndrome (Mayo Clinic, 2018; NDSS, 2020a).

**Down Syndrome and Autism Spectrum Disorder**

**Prevalence**

It was once believed that Down Syndrome and ASD rarely presented together (Kent et al., 1999; Moss et al., 2012), as individuals with DS were and continue to be thought of as especially friendly, without impairments in social interaction (Rasmussen et al., 2001; Ji et al., 2011). However, research has cast doubt upon this widely-held belief and even suggested that individuals with DS may be at an increased risk for developing ASD (Kent et al., 1999; Hepburn et al., 2008). ASD is exhibited in DS at higher rates than would be expected by chance. The prevalence rates of ASD in people with DS are estimated to be anywhere from 5 to 41% (Kent et al., 1999; Hepburn et al., 2008; DiGuiseppi et al., 2010; Channell et al., 2015). Moreover, Warner et al. (2014) found that approximately 38% of those with Down Syndrome met the cut off score for ASD on a screening test. While ASD is comorbid in only a portion of individuals with DS, autism symptomology is important to consider, as ASD is typically diagnosed later and underdiagnosed compared to children without DS (Gray et al., 2011). Due to difficulties in differentiating between behaviors associated with ASD and communication and cognitive impairment associated with intellectual disability, it can be exceedingly challenging to diagnose DS-ASD (Ji et al., 2011). However, delayed or missed diagnoses of ASD can have implications for children with DS, as the symptoms can be severe and these children may not be eligible for
early intervention services or appropriate educational placements (Howlin et al., 1995; Ji et al., 2011; Gray et al., 2011).

**Cognitive Functioning**

Down Syndrome is associated with some degree of intellectual disability, making it the most common genetic cause of intellectual disability (Haydar & Reeves, 2012; Centers for Disease Control and Prevention, 2015). The severity of intellectual disability in those with DS ranges from profound to borderline intellectual functioning, with the average intellectual quotient (IQ) in DS being approximately 50 (Haydar & Reeves, 2012; Chapman & Hesketh, 2000; Grieco et al., 2015). Cognitive functioning in those with DS changes across the lifespan and varies depending on the presence of conditions like ASD, seizures, sleep disturbances, and psychiatric disorders (Grieco et al., 2015; Gasquoine, 2011). Individuals with DS typically exhibit weaknesses in specific areas of cognitive functioning. For example, research has indicated that individuals with DS show weaknesses in concentration and memory tasks compared to typically developing peers (Weijerman & de Winter, 2010). They also have been shown to have more difficulties processing verbal information, as opposed to visual information. While they typically make progress in acquiring non-verbal cognitive abilities, verbal abilities may decline throughout adolescence and adulthood (Grieco et al., 2015). As adults with DS age, they develop an increased risk for dementia, cognitive decline, and Alzheimer’s disease (Head et al., 2012; Grieco et al., 2015).

While intellectual disability is not always associated with ASD, they do co-occur frequently. Recent studies reported that as many as 30% of individuals with ASD also have intellectual disabilities (Polyak et al., 2015; Baio et al., 2018). However, this estimate may be low, as the subset of the ASD population with intellectual disabilities and minimal verbal
abilities are underrepresented in ASD research (Thurm et al., 2019). When intellectual disability is present, the deficits in social communication and the presence of repetitive behaviors and restricted interests that characterize ASD lead to greater adaptive impairments than those associated with intellectual disability solely (Thurm et al., 2019). For example, research has supported that adaptive behavior skills are lower than anticipated based on IQ in people with ASD (Ashwood et al., 2015). Cognitively, children with a comorbid DS-ASD diagnosis showed greater cognitive impairments on both verbal and nonverbal developmental quotient measures compared to individuals solely diagnosed with ASD or DS. However, the ASD symptoms appeared to be less severe in the children with a comorbid DS-ASD diagnosis than they did for children with an ASD-only diagnosis (Hamner et al., 2019). Hamner et al. (2019) also found that children with co-occurring DS-ASD exhibited more deficits in cognitive functioning, followed by those with DS only and ASD respectively.

**Motor Skills**

Children with DS exhibit delays in motor skill development, as they typically have poor balance, hypotonia (i.e., low muscle tone), and ligamentous laxity (i.e. ligament looseness that accounts for increased joint flexibility), making it more difficult to interact with the environment (Kim et al., 2017). Infants with DS often meet motor milestones, such as rolling over, sitting up, standing, and crawling, in the same chronological order but at later ages than their typically developing peers (Palisano et al., 2001; Malak et al., 2015). Moreover, children with DS generally attain gross motor skills (e.g., walking, standing, running) at twice the age of their typically developing peers. For example, research has shown that children with DS on average begin to walk at 24 months of age, with a range occurring between 14 and 42 months. However, typically developing children begin to walk at an average age of 13 months, with a range

19
between 9 and 17 months (Malak et al., 2013; Frank & Esbensen, 2014). While motor skills are not a core feature of ASD, children with ASD often exhibit clumsiness and overall deficits in motor skills. Children with ASD also are often delayed at reaching motor milestones, like walking and crawling (Lloyd et al., 2013). Even fine motor skills, like pointing and clapping, can be challenging for toddlers and children with ASD (Gernsbacher et al., 2008).

**Social and Communication Skills**

As previously discussed, delays in meeting motor milestones are expected among infants with DS, as they experience physiological differences that impact their muscle tone and postural control (Kim et al., 2017; Ferreira-Vasques & Lamônica, 2015). In turn, delays in motor skills inhibit opportunities to interact with the environment and develop language and social skills (Ferreira-Vasques & Lamônica, 2015). Children with DS have been characterized as highly social and affectionate (Rasmussen et al., 2001; Ji et al., 2011). While there is individual variability in social skills among children with DS, research has suggested that specific areas of social functioning, like social orientation and social engagement, can be strengths (Fidler et al., 2008). For example, Channell et al. (2014) found that when language abilities were controlled, children with DS performed at the same developmental level on emotion knowledge as the typically developing controls. Additionally, research has suggested that children with DS form interpersonal relationships much like their typically developing peers; adults with DS also have fewer social problems and lower antisocial behavior than individuals with other learning disabilities (Collacott et al., 1998; Freeman & Kasari, 2002; Næss et al., 2017).

However, children with DS are at a greater risk for impairments in social functioning compared to typically developing children (Næss et al., 2017; Hepburn et al., 2008). For example, children with DS show a significant weakness in planning, problem solving, and
playing with objects (Næss et al., 2017; Fidler et al., 2014). In regard to communication, individuals with DS generally have stronger receptive language than expressive language (Martin et al., 2010). For example, phonology and syntax skills have proven to be more difficult (Martin et al., 2010).

Inherent to ASD, individuals experience deficits in social communication and social interaction. These deficits include a reduction in the sharing of interests and emotions with others, difficulties developing and maintaining relationships, and abnormalities in eye contact and the use of gestures (APA, 2013). Children with ASD often have difficulty developing expressive and receptive language skills. While some children with ASD may be unable to speak, others may be able to speak about specific topics in detail. Individuals with ASD often have difficulties using language in a functional manner to communicate with other people (National Institute of Deafness and Other Communication Disorders, 2020). A study by Volkmar et al. (1987) found that older children with ASD exhibited more delays in adaptive social behaviors compared to their developmentally delayed peers with matched ages and IQ’s. People with ASD also exhibited more significant delays in communication skills and social skills, such as interpersonal relationships and sensitivity towards others, in comparison to adolescents and adults with a DS diagnosis (Loveland & Kelley, 1988).

**Adaptive Skills**

In general, children with DS obtain adaptive skills more slowly than typically developing peers. In addition to delays in acquiring adaptive skills among those with DS, there also tends to be a lower ceiling (Duijn et al., 2010). While communication and motor skills tend to be areas of weakness for individuals with DS, daily living skills have been shown to be an area of overall strength (Duijn et al., 2010; Tingey et al., 1991; Dykens et al., 2006).
In addition to the core deficits of ASD, impairments in adaptive behaviors, such as socialization, communication, and daily living skills, are often associated with ASD (Kanne et al., 2011; Chatham et al., 2018). Previous research suggests that children with ASD may have more deficits in adaptive skills compared to children with DS (Rodrigue et al., 1991; Loveland & Kelley, 1988). While cognitive ability may impact adaptive behavior, cognitive ability cannot fully explain adaptive behavior problems in those with ASD (Chatham et al., 2018; Charman et al., 2011). Research on the association between adaptive behavior and ASD symptomology has been variable. For example, Klin at al. (2007) found a weak relationship between adaptive behavior and ASD symptomology. However, Perry et al. (2009) examined children with ASD under the age of 6 and found a strong negative relationship between ASD symptom severity and adaptive behavior. Additionally, Kanne et al. (2011) found a positive association between adaptive behavior, as measured by the *Vineland Adaptive Behavior Scales (2nd edition)*, and IQ. IQ accounted for approximately 55% of the variance in adaptive skills beyond that which was predicted by ASD symptom severity and age. Previous research suggests that children with ASD may have more deficits in adaptive, social, and communication skills compared to children with DS (Rodrigue et al., 1991; Loveland & Kelley, 1988). Children with comorbid DS-ASD and DS only exhibited significant strengths in receptive communication skills and adaptive daily living skills, compared to children with ASD only (Dressler et al., 2011). Studies have found that compared to individuals with a comorbid DS-ASD diagnosis, those with DS only have higher IQ scores (Carter et al., 2007; Molloy et al., 2009), language abilities, and adaptive skills (Molloy et al., 2009).
Purpose

As the rates of ASD rise (Christensen et al. 2016; “Autism Spectrum Disorder (ASD): Data & Statistics”, 2020), the need for effective early diagnosis and intervention continues to increase. Approximately 10% of children with ASD have also been identified as having a co-occurring genetic or chromosomal condition (Davis et al., 2017; Oxelgren et al., 2019). Down Syndrome is one such condition associated with ASD. It was historically believed that ASD and DS seldom presented together, as those with DS were thought to possess strengths in social skills (Rasmussen et al., 2001; Fidler et al., 2008). However, research suggests that children with DS are at an increased risk, compared to their typically developing peers, for impairments in social functioning and the development of ASD (Næss et al., 2017; Hepburn et al., 2008; Kent et al., 1999).

DS can be diagnosed prenatally through chronic villus sampling (CVS) and amniocentesis or after birth (NDSS, 2020b). However, ASD can be reliably diagnosed in children as young as 18 months of age (Ozonoff et al., 2015; Zwaigenbuam et al., 2016). Diagnosis of ASD is often delayed up to three years after the initial concerns are presented to a clinician, with the majority of children being diagnosed as late as 5 years of age (Shattuck et al., 2009; Zuckerman et al., 2015). Interventions implemented before 4 years of age, however, are related to improvements in cognition, language skills, and adaptive behavior, so this can be especially problematic (Dawson et al., 2010; Vivanti et al., 2016). Thus, it is necessary for children to receive an ASD diagnosis as early as possible to improve their outcomes.

Research suggests that individuals with DS are at an increased risk for developing ASD compared to their typically developing peers (Kent et al., 1999; Hepburn et al., 2008); however, shared elements of symptomology and developmental functioning deficits may contribute to
difficulties in identifying ASD in those with DS. Namely, research indicates that dual diagnoses of DS-ASD can be exceptionally challenging to recognize as behaviors associated with ASD and communication and cognitive impairments associated with intellectual disability can appear to be similar in nature (Ji et al., 2011). Due to the increased risk of the comorbid occurrence of ASD and DS, it is crucial for clinicians to be able to accurately recognize those with both conditions to allow for appropriate early intervention services and educational placements (Howlin et al., 1995; Ji et al., 2011; Gray et al., 2011).

While prior research has compared developmental functioning between children with ASD only and DS only (Loveland & Kelley, 1988; Rodrique et al., 1991), very few studies have investigated developmental functioning in children with DS at risk for ASD. Using a sample of infants and toddlers will also expand the research to help understand whether differences in developmental functioning are present early in life. Thus, the current study looks to examine the impact of DS on developmental functioning in toddlers. Comparisons of the overall developmental functioning, as well as the subdomains of developmental functioning (i.e. Adaptive, Personal-Social, Communication, Motor, and Cognitive) will be examined in toddlers at risk for ASD only, with DS at risk for ASD, and with DS who are not at risk for ASD. The results of the current study will have clinical implications for the assessment of ASD in toddlers and young children.
Method

Participants

Participants consisted of 138 toddlers, aged 17 to 36 months old (\(M = 26.13, SD = 4.62\)). The data for the current study were extracted from an existing database of children enrolled in EarlySteps, Louisiana’s statewide early intervention program. Under the Individuals with Disabilities Education Act, part C, children are eligible to participate in EarlySteps if they are under 36 months of age or if they have a developmental delay or a medical condition that is likely to result in a developmental delay. These disabilities and conditions include cerebral palsy, epilepsy, deafness, down syndrome, premature birth and blindness, among others. Only children between 17 and 36 months at the time of assessment were included in the final analyses.

The participants were assigned to groups based on whether they met criteria for DS or were elevated on an ASD screener, the Baby and Infant Scale for Children with Autism-Traits (BISCUIT-Part 1; Matson et al., 2007). Down Syndrome diagnoses were provided through caregiver reports of previous diagnoses, made by medical professionals, on the demographic section of the BISCUIT (Matson et al., 2007). The BISCUIT-Part 1 was completed by a caregiver informant of the participants. Information regarding screening results was made based on the predetermined cutoff score for the BISCUIT-Part 1. While the BISCUIT assesses autism symptomology in infants and toddlers through caregiver report, it does not provide a clinical diagnosis. For the purpose of the analyses, the participants were assigned to one of three age and gender matched groups: an ASD screen positive only group without co-occurring DS (ASD+, \(n = 46\)), a DS screen negative group (DS-, \(n = 46\)), and a DS screen positive group (DS+, \(n = 46\)).
Figure 1. Participant Inclusion Chart

The dataset consisted of 27,408 total cases. Participants missing data, such as data from either the BISCUIT-Part 1 or BDI-2 developmental subdomains, were excluded from the
analyses. Participants that screened negative on the BISCUIT-Part 1 without a co-occurring DS diagnosis were excluded from the analyses. If participants received multiple assessments, the initial assessment was included in the analyses. Participants that screened positive on the BISCUIT-Part 1 with co-occurring neurological, developmental, and genetic conditions (e.g., seizures, cerebral palsy, intellectual disability, traumatic brain injury, Fragile X Syndrome, chromosomal deletion syndromes) were excluded as these conditions were found to further impact developmental functioning (Burns & Matson, 2018; Razak et al., 2020). The total cases excluded from the analyses can be found in Figure 1.

There were 6,243 cases including in the matching procedure (i.e., 6,058 ASD+, 130 DS-, and 55 DS+). Due to the unequal sample sizes across the groups, the groups were age and gender matched to the smallest group (i.e., DS+ group), which resulted in 46 participants per group and 138 participants total. The matching process was utilized due to age and gender having been found to impact developmental functioning in prior research. The cases were randomly assigned when drawing case matches and matched without replacement. All cases were matched exactly for age and gender.

The total sample consisted of 78.26% males (n = 108) and 21.74% females (n = 30). Regarding participant ethnicity, 34.78% were African American (n = 48), 62.96% were Caucasian (n = 68), 9.42% were Hispanic (n= 13), 4.35% were listed as Other ethnic identity (n =6), and 2.17% did not report their ethnic identity (n = 3). Demographic information for the groups is presented in Table 1.

Table 1. Group Demographic Information

<table>
<thead>
<tr>
<th></th>
<th>ASD+ (n = 46)</th>
<th>DS+ (n = 46)</th>
<th>DS- (n = 46)</th>
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<tbody>
<tr>
<td>Mean Age (months)</td>
<td>26.13</td>
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<td>26.13</td>
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<tr>
<td>SD (months)</td>
<td>4.62</td>
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Gender

<table>
<thead>
<tr>
<th></th>
<th>Male</th>
<th>Female</th>
</tr>
</thead>
</table>
|       | 36   | 10     | (78.26%) (21.74%)  
|       | 36   | 10     | (78.26%) (21.74%)  
|       | 36   | 10     | (78.26%) (21.74%)  

Ethnicity

<table>
<thead>
<tr>
<th>Ethnicity</th>
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<th>Hispanic</th>
<th>Other</th>
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<td>18</td>
<td>4</td>
<td>4</td>
<td>1</td>
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<td></td>
<td>(41.30%)</td>
<td>(39.13%)</td>
<td>(8.70%)</td>
<td>(8.70%)</td>
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<tr>
<td></td>
<td>19</td>
<td>22</td>
<td>3</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>(41.30%)</td>
<td>(47.83%)</td>
<td>(6.52%)</td>
<td>(2.17%)</td>
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<tr>
<td></td>
<td>10</td>
<td>28</td>
<td>6</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>(21.74%)</td>
<td>(60.87%)</td>
<td>(13.04%)</td>
<td>(2.17%)</td>
<td>(2.17%)</td>
</tr>
</tbody>
</table>

Missing Data 1 (2.17%)  

Measures

**Baby and Infant Screen for Children with aUtIsm Traits—Part 1 (BISCUIT-Part 1)**

*The Baby and Infant Screen for Children with aUtIsm Traits—Part 1 (BISCUIT-Part 1; Matson et al., 2007)* is an informant report measure that is made up of three different sections which assess ASD symptomology, comorbid psychopathology, and challenging behaviors which occur in children aged 17-37 months. The *BISCUIT-Part 1* is the diagnostic section of the measure which consists of 62 items. These items are related to nonverbal and verbal communication, socialization, and repetitive, restricted behaviors and interests. The items are rated on a 3-point scale (0= ‘not different or no impairment’, 1=’somewhat different or mild impairment’, 2=’very different or severe impairment’). This measure includes cut off ranges for the total score on the *BISCUIT-Part 1*. A score of 0-16 is considered to fall in the “No ASD/Atypical Development” range, a score of 17-38 places a toddler in the “Possible ASD” range, and a score of 41-124 falls in the “Probable ASD” range. Thus, a score of 17 or higher is
classified as in the “at risk range” (Matson et al., 2009). For the purposes of this study, individuals are considered in the DS screen positive group if they have a score of 17 or higher and are considered to be in the “at risk” range. The DS screen negative group will have a score lower than 17 and will be considered to be in the “No ASD/Atypical Development” range.

The BISCUIT-Part 1 demonstrated an overall correct classification rate of .89 and internal reliability of .97 (Matson et al., 2009). The BISCUIT-Part 1 also exhibited convergent validity with the Personal-Social subdomain of the Battelle Developmental Inventory, Second Edition (BDI-2) and the Modified Checklist for Autism in Toddlers (M-CHAT) (Matson et al., 2011), as well as divergent validity with the Adaptive and Motor subdomains on the BDI-2 (Matson et al., 2011). The current study utilizes the BISCUIT-Part 1 demographic form and the total score. The demographic form will be used to gain information regarding participants’ developmental and medical history.

**Battelle Developmental Inventory, Second Edition (BDI-2).**

The Battelle Developmental Inventory, Second Edition (BDI-2; Newborg, 2005) is a measure used to assess development skills in children from birth to 7 years and 11 months old. The BDI-2 consists of 450 items which evaluate five separate subdomains including: Adaptive (ADP), Personal-Social (P-S), Communication (COM), Motor (MOT), and Cognitive (COG). All of the individual items are scored on a 3-point Likert scale. A rating of 0 indicates that the child has no ability in the skill being assessed. A rating of 1 indicates that the child has an emerging ability in the skills area being assessed. A rating of 2 indicates that the child has an ability at the skill which is being evaluated. The ratings for the items are combined for the score in each subdomain and the total developmental quotient (DQ). The mean of the scores is 100 and the standard deviation is 15.
The BDI-2 was found to have adequate test-retest reliability with estimates of .90 or above for each subdomain and the total DQ score (Newborg, 2005). Internal consistency, determined by the split-half method, showed reliability coefficients of .98 to .99 for the subdomain and total DQ scores (Newborg, 2005). The BDI-2 was also shown to be correlated with several other developmental scales including the Bayley Scales of Infant Development, the Denver Developmental Screening Test-II (DDST), the Vineland Social Emotional Early Childhood Scales, and the Wechsler Preschool and Primary Scale of Intelligence (WPPSI) (Newborg, 2005). In the current study, the total DQ and the five subdomain scores will be utilized to assess the participants’ developmental functioning.

**Procedure**

Before the data was collected, the study was approved by the Louisiana State University Institutional Review Board and the Office for Citizens with Developmental Disabilities (OCDD) of the State of Louisiana IRB. The data for this study will be taken from a database stripped of identifying information, such as name and date of birth. Both IRB’s determined that informed consent from caregivers of the participating children was not necessary, as the database was stripped of identifying information prior to receipt by the researchers. Data collected between 2008 and 2019 were included in the final statistical analyses.

Both the BISCUIT-Part 1 and the BDI-2 were administered as part of a larger assessment battery which included child observations and caregiver interviews. They were administered by service providers who held a degree, certification, or licensure in fields such as physical therapy, occupational therapy, psychology, speech-language pathology, and special education. All of the providers are trained and competent in administering the study’s measures and evaluating children.
**Statistical Analysis**

A statistical power analysis program, G*Power 3.1, was utilized to determine the sample sizes needed for the three groups to establish adequate power. A power of .80 (Field, 2013), alpha level of .05, and medium effect size of $\eta^2 = .30$ were used to find the appropriate sample size. The results of the a priori power analysis suggested that using at least 111 total participants, or approximately 37 participants per group, would produce the appropriate power for a one-way ANOVA, and a total sample size of 33 would yield the appropriate power for a MANOVA.

Statistical analyses were conducted with the use of SPSS 26.0. Bivariate and multivariate analyses were conducted to investigate the following research questions: (1) does overall developmental functioning differ based on group (i.e., ASD+, DS-, DS+)? (2) does group impact functioning across the five developmental subdomains (i.e., Adaptive, Personal-Social, Communication, Motor, and Cognitive). Since the groups (i.e., ASD+, DS-, DS+) were age and gender matched to the smallest group, bivariate comparisons of the variables were not conducted. However, a chi-square analysis was conducted to evaluate differences in ethnicity between the groups, as the groups were not matched for ethnicity.

In the present study, an analysis of variance (ANOVA) was conducted to determine the relationship between group and overall developmental functioning. The groups (i.e., ASD+, DS-, and DS+) served as the independent variable (IV) and scores from the overall total developmental quotient of the BDI-2 functioned as the dependent variables (DV). Specific group differences were further investigated with a Games-Howell post-hoc test.

In order to investigate the relationship between group and developmental subdomains, a multivariate analysis of variance (MANOVA) was run with group as the IV and scores from the five developmental subdomains (i.e., Adaptive, Personal-Social, Communication, Motor, and
Cognitive) of the *BDI*-2 as the DV. Following the MANOVA, separate ANOVA’s were conducted to assess developmental differences between the groups. A Bonferroni correction was applied to account for the multiple comparisons, which resulted in an adjusted value of $p = .01$. Then, the ANOVA’s were followed up by a Games-Howell post-hoc test to account for the violation in homogeneity of variance (Field, 2013; Laerd Statistics, 2018).

**Results**

The results of the chi-square analysis indicated that there were no significant differences between the groups based on ethnicity, $\chi^2 (6) = 9.69, p = 0.138$. Preliminary assumption checking for the ANOVA indicated that there were no outliers in the data as assessed by inspection of a boxplot for values greater than 1.5 box-lengths from the edge of the box. Additionally, the total DQ was normally distributed for the groups (i.e., ASD+, DS-, DS+), as assessed by both the Shapiro-Wilk’s test ($p > .05$) and graphical Normal Quantile-Quantile (Q-Q) plots. The assumption of homogeneity of variances was violated, as assessed by Levene’s test for equality of variances ($p < .001$). Since Levene’s Test for Equality of Variances was significant ($p < .001$), the Game’s Howell post-hoc tests were used. Significant differences were found across groups for *BDI*-2 total DQ scores, $F (2, 86.72) = 18.69, p < .001$. Results from the Games-Howell post-hoc test indicated that all of the groups had significantly different total DQ scores on the *BDI*-2 (ASD+ and DS-, $p = .011$; all other groups $p < .001$). The ASD+ group had the highest average *BDI*-2 total DQ scores, followed by the DS-, and then the DS+ group.

Descriptive statistics for the *BDI*-2 total DQ scores across groups are provided in Table 2.

Prior to conducting the MANOVA, preliminary assumptions were examined. Five total univariate outliers were identified for the Communication and Cognitive subdomains of the *BDI*-2, as assessed by inspection of a boxplot for values greater than 1.5 box-lengths from the edge of
the box. None of the outliers were found to be due to data entry mistakes and consequently were included in the sample (Laerd, 2018; Tabachnick & Fidell, 2007). Additionally, no extreme outliers were identified and as a result winsorization was not performed. Based on observations of the graphical Normal Q-Q plots and histograms, it was determined that the Communication subdomain scores were not normally distributed for each group but rather strongly positively skewed. With respect to Communication subdomain scores, there was a mean score of 62.91 ($SD = 10.64$), skewness of 1.82 ($SE = .21$), and kurtosis of 3.00 ($SE = .41$). Thus, a logarithmic transformation was applied to the Communication subdomain to determine whether the assumption of normality could be met for these data. The transformation was found to impact the model, improve on the normality of the data, and reduce the strong positive skew. Therefore, all the statistical analyses were conducted using the transformed data.

Following the transformation, there were four total univariate outliers identified for the Communication and Cognitive subdomains of the *BDI-2*. Normal Q-Q plots and histograms continued to demonstrate non-normality for the Communication subdomain across groups. However, the skewness and kurtosis values were improved for the variable, with skewness of 1.49 ($SE = .21$) and kurtosis of 1.58 ($SE = .41$). Using Mahalanobis distance, there was one multivariate outlier in the DS- group ($p < .001$), using the critical value of 20.52 based on the number of dependent variables. There was no multicollinearity, as assessed by Pearson’s correlation. Correlations between the dependent variables in the study ranged between $r = .257$ and $r = .585$. There were also linear relationships detected between the BDI-2 subdomain scores in each group as assessed by scatterplots. The Box’s M Test of Equality of Covariance Matrices was statistically significant ($p < .001$), meaning that the assumption of homogeneity of variance-covariance matrices was violated. However, given the equal sample sizes across groups (i.e., $n =$
46), this violation is not considered concerning (Tabachnick & Fidell, 2007; Laerd, 2015). Pillai’s Trace, rather than Wilks’ Lamba, was interpreted as it is less sensitive to violations of this assumption (Field, 2013; Laerd Statistics, 2015). There was not homogeneity of variance for the Communication, Motor, or Cognitive subdomains ($p < .001$ for Communication, $p = .007$ for Motor, and $p < .001$ for Cognitive). Thus, the ANOVA’s were followed up by the Games’s Howell post-hoc tests (Field, 2013; Laerd Statistics, 2018).

According to Pillai’s Trace, there was a significant effect of group on developmental subdomains, $V = .623$, $F(10, 264) = 11.94$, $p < .001$, partial $\eta^2 = .311$. Separate one-way ANOVA’s were conducted and a Bonferroni correction ($p = .01$) was applied to account for multiple comparisons. Significant differences were found across groups in each developmental subdomain: the Adaptive subdomain $F(2, 135) = 16.52$, $p < .001$, $\eta^2 = .197$, the Personal-Social subdomain $F(2, 135) = 6.22$, $p = .003$, $\eta^2 = .084$, the Communication subdomain $F(2, 135) = 8.04$, $p = .001$, $\eta^2 = .11$, the Motor subdomain $F(2, 135) = 49.65$, $p < .001$, $\eta^2 = .424$, and Cognitive subdomain $F(2, 135) = 12.38$, $p < .001$, $\eta^2 = .155$.

Results from the Games- Howell post-hoc analysis indicated statistically significant group differences among each of the five developmental subdomains. For the Adaptive subdomain, statistically significant differences were found between the ASD+ and DS+ groups ($p < .001$) and DS- and DS+ groups ($p < .001$). In regard to the Personal-Social subdomain, the ASD+ and DS- ($p = .024$) and DS- and DS+ groups ($p = .003$) significantly differed from one another. The ASD+ and DS+ groups ($p = .002$) were significantly different from one another in the Communication subdomain. Regarding the Motor subdomain, statistically significant differences were exhibited between the ASD+ and DS- groups ($p < .001$), ASD+ and DS+ groups ($p < .001$), and DS- and DS+ groups ($p = .027$). Lastly, the ASD+ and DS+ groups ($p <
.001) and DS- and DS+ groups ($p = .015$) differed significantly from one another in the Cognitive subdomain. Descriptive statistics for the five developmental subdomain scores on the BDI-2 are presented in Table 2 and Figure 1.

**Table 2. Group Differences on the Developmental Domains**

<table>
<thead>
<tr>
<th></th>
<th>ASD+</th>
<th>DS-</th>
<th>DS+</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$M$</td>
<td>$M$</td>
<td>$M$</td>
</tr>
<tr>
<td></td>
<td>(SD)</td>
<td>(SD)</td>
<td>(SD)</td>
</tr>
<tr>
<td>BDI-2: Total DQ Scores</td>
<td>74.24+Δ</td>
<td>67.93+Δ</td>
<td>61.80+Δ</td>
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<tr>
<td></td>
<td>(12.47)</td>
<td>(7.28)</td>
<td>(8.38)</td>
</tr>
<tr>
<td>Adaptive</td>
<td>76.63</td>
<td>73.04</td>
<td>64.52+Δ</td>
</tr>
<tr>
<td></td>
<td>(10.81)</td>
<td>(10.75)</td>
<td>(9.54)</td>
</tr>
<tr>
<td>Personal-Social</td>
<td>81.39Δ</td>
<td>87.04+Δ</td>
<td>79.96Δ</td>
</tr>
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<td></td>
<td>(10.74)</td>
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<td>(16.60)</td>
<td>(10.76)</td>
<td>(11.63)</td>
</tr>
<tr>
<td>Cognitive</td>
<td>75.50</td>
<td>71.15</td>
<td>67.28+Δ</td>
</tr>
<tr>
<td></td>
<td>(10.16)</td>
<td>(5.88)</td>
<td>(7.13)</td>
</tr>
</tbody>
</table>

+Significantly differs from ASD+, $p<.05$
ΔSignificantly differs from DS-, $p<.05$
Significantly differs from DS+, $p<.05$

**Figure 1. Estimated Mean Groups Scores across BDI-2 Subdomains**
Discussion

The current study examined the relationship between group (i.e., ASD+, DS-, DS+) and overall developmental functioning, as well as functioning across five developmental subdomains on the BDI-2 in toddlers. Significant differences were found between all groups for the BDI-2 total DQ scores, such that the DS+ group exhibited lower overall functioning ($M = 61.80$) than toddlers in the DS- ($M = 67.93$) and ASD+ groups ($M = 74.24$).

Both the Adaptive and Cognitive developmental subdomains showed the same pattern of differences among the groups in relation to one another. In the Adaptive subdomain, toddlers in the DS+ group were found to demonstrate lower adaptive skills ($M = 64.52$) than toddlers in the DS- ($M = 73.04$) and ASD+ groups ($M = 76.63$). It was expected that DS+ group would have less proficient adaptive skills than the DS- or ASD+ groups. This finding was in alignment with previous studies which suggest that elevated ASD symptoms in individuals with DS are associated with significantly lower adaptive behaviors, conceptual skills, and daily living skills (Channell et al., 2019; Dressler et al., 2011; Molloy et al., 2009). While few studies have compared the adaptive functioning of individuals with ASD only and DS only, Dressler and colleagues (2011) found that individuals with DS scored significantly higher than individuals with ASD on the adaptive daily living skills subdomain of the Vineland Adaptive Behavior Scales. Children with DS tend to experience delays in attaining adaptive skills, along with a lower ceiling compared to typically developing peers (Duijn et al., 2010). However, adaptive daily living skills have been found to be an overall area of strength for infants and toddlers with
DS compared to communication and socialization skills (Tingey et al., 1991; Dykens et al., 2006). In the present study, no significant difference in adaptive functioning was found between the ASD+ and DS- groups, which differed from previous research (Rodrique et al., 1991; Loveland & Kelley, 1988; Dressler et al., 2011). This may be explained by the older and larger age range utilized in previous studies. The Dressler and colleagues (2011) study examined both children and adults with DS, ASD, and DS and ASD between the ages of 6 and 34, whereas the Rodrique and colleagues (1991) study looked at children and adults between 2 and 19 years of age.

Similarly, in the Cognitive developmental subdomain, the toddlers in the DS+ ($M = 67.28$) group displayed significantly lower cognitive skills than the toddlers in the DS- ($M = 71.15$) or ASD+ ($M = 75.50$) groups. It was predicted that the DS+ group would have the lowest overall cognitive functioning, as DS is associated with some degree of intellectual disability ranging in severity from mild to profound (Haydar & Reeves, 2012; Centers for Disease Control and Prevention, 2015; Grieco et al., 2015). Additionally, children with DS and comorbid ASD have shown greater cognitive impairments on both verbal and nonverbal quotient measures compared to children with either DS only or ASD only (Hamner et al., 2019; Molloy et al., 2008; Carter et al., 2007). For example, Capone and colleagues (2005) found no individuals with DS only to be in the severe to profound range of intellectual disability, but nearly 87% of those with comorbid ASD and DS fell into the severe to profound range of intellectual disability (FSIQ = 0-39). Similarly, Channell and colleagues (2019) suggested that individuals with DS at high risk for developing ASD were more likely to perform at or below the floor on the Kaufman Brief Intelligence Test, 2nd Edition compared to individuals with DS at low risk for developing ASD.
While intellectual disability does not always co-occur with ASD, it has been reported to be prevalent in nearly 30% of individuals with ASD (Polyak et al., 2015; Baio et al., 2018). Previous researchers have suggested that cognitive ability may impact adaptive behavior in children, with a positive relationship between IQ and adaptive functioning (Kanne et al., 2011; Thurm et al., 2019). A recent study investigated adaptive functioning among those with ASD only, ASD and ID, and ID between the ages of 1.5 and 18 years of age. The ASD and ID group was found to have the lowest overall adaptive and conceptual skills. However, the group differences were not maintained when the groups all consisted of individuals with moderate to severe intellectual disability (Bradbury et al., 2021). This suggests that cognitive functioning may influence adaptive functioning among individuals with DS and ASD. The results from the Adaptive and Cognitive subdomains indicate that impairments in these domains are more strongly influenced by the DS diagnosis than scoring at-risk for ASD.

Conversely, in the Personal-Social subdomain, individuals in the DS- (M = 87.04) group exhibited significantly higher social skills than the ASD+ (M = 81.39) or DS+ (M = 79.96) groups. These results were expected because, as previous researchers suggested, individuals with ASD exhibited lower social skills than their peers with DS only (Rodrique et al., 1991; Dressler et al., 2011). Specific areas of social functioning, like social engagement and social orientation, have been found to be areas of strength among those with DS (Fidler et al., 2008). Prior researchers suggested that individuals with DS form social relationships similarly to typically developing peers (Collacott et al., 1998; Naess et al., 2017). However, the Personal-Social subdomain reflects a core diagnostic feature of ASD, such that social deficits are required for an ASD diagnosis. Both of the groups at risk for ASD had lower scores than the DS group not at
risk for ASD, suggesting that the impairments in social skills were more strongly influenced by the ASD diagnosis compared to the DS diagnosis.

In the Motor subdomain, the toddlers in the ASD+ (M = 92.46) group had significantly higher motor skills than the toddlers in the DS- (M = 72.28) and DS+ (M = 66.15) groups. The toddlers in the DS- (M = 72.28) group also had significantly higher motor skills than those in the DS+ (M = 66.15). Children with DS typically exhibit delays in motor skills, such as poor balance, hypotonia (i.e., poor muscle tone), and ligament laxity (i.e., ligament looseness that accounts for increased joint flexibility). Additionally, prior studies have found that infants with DS acquire gross motor skill milestones, like walking, standing, and running, at nearly twice the age of their typically developing peers (Malak et al., 2013; Frank & Esbensen, 2014). Motor skill impairments are not a core diagnostic feature of ASD, though children with ASD can be delayed in reaching motor milestones, like crawling and walking (Lloyd et al., 2013). Both of the groups of toddlers with diagnoses of DS had lower motor skill abilities than those at risk for ASD, suggesting that the DS diagnosis is related to lower motor functioning. The co-occurrence of risk for ASD and DS had negative impacts on motor functioning beyond that which was accounted for the DS diagnosis only. To date, no prior studies comparing motor functioning between children with DS and ASD have been conducted.

In the Communication subdomain, the ASD+ (M = 66.39) group demonstrated higher communication skills than the DS+ (M = 59.03) group. Like the Personal-Social subdomain, the Communication subdomain reflects a core diagnostic feature of individuals with ASD. ASD is characterized by deficits in social communication and social interaction. Children with ASD typically experience difficulties developing both expressive and receptive language skills. Utilizing language in a functional manner to communicate with others can particularly be
challenging to those with ASD (National Institute of Deafness and Other Communication Disorders, 2020). Prior researchers investigating communication skills among those with DS and comorbid ASD and DS found that children with comorbid ASD and DS had both poorer receptive and expressive language skills compared to their peers with DS only (Molloy et al., 2008; Dressler et. al., 2011). Additionally, children with comorbid ASD and DS reportedly acquired their first words significantly later than children with DS only (Warner et al., 2014; Bradbury et al., 2021). Even compared to their peers with ASD only, children with comorbid ASD and DS were found to experience more deficits in verbal communication skills, like use of pronouns, social chat and neologisms (Warner et al., 2014). The present study did not find significant differences between the communication skills of the DS- and DS+ groups or the ASD+ and DS- groups. However, prior research examining group differences in communication skills among individuals with ASD, DS, and ASD and DS used an older and larger age range.

Across each of the five developmental subdomains, the toddlers in the DS+ group exhibited the poorest functioning among the groups. While autism symptoms are more prevalent among those with DS with more severe and profound intellectual disabilities, all individuals with DS are at an increased risk for developing ASD (Rachubinski et al., 2017). However, the developmental profile of those with comorbid ASD and DS is not well established or understood (Channell et al., 2019). The genetic etiology of ASD in DS has only recently been explored, though it is likely that genetic factors contribute to the increased risk of the co-occurrence of DS and ASD. As previously described, DS is caused by trisomy of human chromosome 21 (Hsa21), which likely leads to the elevated expression of Hsa21 genes. An overexpression of these genes has been found to cause a range of neurological abnormalities, including learning and memory difficulties, as well as synaptic plasticity impairments. Researchers suggest that elevated
symptoms of ASD in those with and without DS may be due to overexpression of Hsa21 genes. However, trisomy of Hsa21 cannot be the sole explanation for ASD in DS, as only a portion of those with DS are diagnosed with comorbid ASD. One explanation for the increased prevalence of ASD among those with DS is the presence of additional genetic variants, which may act as genetic modifiers, among those with trisomy Has21 (Rachubinski et al., 2017). Future research should further investigate genetic factors contributing to ASD among those with DS.

To the best of the author’s understanding, there are no prior studies that examined developmental functioning in those at risk for ASD only and those with DS at risk for ASD under the age of 3. Despite an increased risk for ASD symptomology among individuals with DS, ASD is often diagnosed later in individuals with DS (Bradbury, 2021; Howlin et al., 1995). This may in part be due to the developmental or behavioral impairments in children with DS being attributed to the DS or intellectual disability diagnosis, rather than a co-occurring ASD diagnosis (Bradbury et al., 2021; Howlin et al., 1995). For example, rigid and repetitive behaviors that are characteristic of ASD are also commonly observed in individuals with intellectual disabilities, making it very challenging to distinguish between behaviors associated with comorbid ASD from those associated with DS only (Channell et al., 2019; Glennon et al., 2017). However, it is imperative that ASD symptomology be identified in those with DS to ensure appropriate early intervention and educational services.

The present study, as well as prior studies, found that those with co-occurring ASD symptomology and DS had more cognitive, adaptive, and motor deficits than those with DS only (Warner et al., 2017). An understanding of the impact of ASD risk and DS on developmental functioning should inform clinical practice through improved screening methods among those with pre-existing genetic conditions, like DS. Previous researchers examining the refusal rates of
ASD screening among toddlers in an early intervention program found that having a previously diagnosed medical condition made toddlers more susceptible to screening refusal. Specifically, children with DS were approximately three times more likely to have an ASD screening refused by caregivers than toddlers without the diagnosis (Matheis & Matson, 2015). As a result, it is vital for clinicians to inform caregivers of the increased risk for ASD in those with DS at the time of screening. Improving screening rates among those with DS, particularly in early intervention programs, is crucial to earlier diagnosis and entry into ASD related intervention services.

While the current study can significantly contribute to present literature, there are limitations to consider. The information on the BISCUIT-Part 1 demographic form, used to confirm the DS diagnoses, was provided through caregiver report. Therefore, the DS diagnoses were based solely on caregiver report measures and were not validated through medical records. Additionally, diagnostic confirmations of ASD through comprehensive diagnostic assessments were not completed in the study. Findings of the present study are based on ASD risk that was determined from a caregiver report and should be interpreted as such. Future studies may consider using other informants, such as teachers or clinicians, to determine ASD risk. As previously stated, the current study recruited participants from EarlySteps, Louisiana’s statewide early intervention program, so the findings of the study may not be representative for other populations.

The findings of the present study provide preliminary indication that ASD risk in toddlers with DS negatively impacts developmental functioning overall and across developmental subdomains in toddlers under 3 years of age. Future studies should seek to investigate both developmental functioning and ASD symptom severity among toddlers with diagnostic
confirmation of ASD, DS, and DS and comorbid ASD, as opposed to ASD risk based off of screeners. While the sample of participants were too young to reliably diagnose intellectual disabilities, future research should be completed to investigate the impact of intellectual disability (ID) on developmental functioning and autism symptomology among those with DS and ASD. For example, future researchers may consider examining group differences among those with ASD and DS, ASD and ID, and ID only as cognitive functioning has been found to impact adaptive behavior (Bradbury et al., 2021). Additionally, future studies involving toddlers should further clarify strengths and weaknesses of those with ASD only, DS only, and ASD and DS within specific subdomains of adaptive functioning measures, like the *Vineland Adaptive Behavior Scales*, to further individualize specific target goals for early intervention services.
References


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Vita

Celeste Tevis, born in Youngstown, Ohio, received her Bachelor of Science in psychology with minors in social work and sociology from Grove City College in Grove City, Pennsylvania. Following her undergraduate career, Celeste provided applied behavior analysis (ABA) therapy and job coaching services as a behavior technician to children and adolescents with autism spectrum disorder. She also worked as a research assistant in a gender studies lab at Slippery Rock University in Pennsylvania. Celeste is currently a third-year-graduate student under the supervision of Dr. Johnny L. Matson. Her clinical and research interests include early identification and treatment of autism spectrum disorder and developmental disabilities. She plans to receive her Master of Arts in clinical psychology in May 2022.