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Executive Functioning Among the Karyotypes in Turner Syndrome and Implications for Interventions

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EXECUTIVE FUNCTIONING AMONG THE KARYOTYPES IN TURNER SYNDROME AND IMPLICATIONS FOR INTERVENTIONS

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ABSTRACT

EXECUTIVE FUNCTIONING AMONG THE KARYOTYPES IN TURNER SYNDROME AND IMPLICATIONS FOR INTERVENTIONS

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Turner syndrome (TS) is a genetic disorder seen in phenotypically female (pf) individuals who have either a complete or partial absence of the second sex (X) chromosome. TS includes different karyotypes, and it presents with a variety of phenotypic and genotypic features. In general, the neuropsychological profiles for individuals diagnosed suggest that TS can contribute to challenges in various aspects of daily life, including social and emotional functioning. With regard to academic performances, individuals with TS often present with relative strengths in a range of verbal abilities and relative weakness in visual-spatial/perceptual abilities, nonverbal memory, motor function, processing speed, executive function, attentional abilities, and poor mathematics performance. Most studies of individuals with TS have noted variable executive functioning (EF) abilities. To date, there have been few studies of the relative EF strengths and weaknesses related to the specific karyotypes of TS. In order to clarify neurocognitive profiles among the different TS karyotypes, this study aimed to (a), review and analyze completed neuropsychological test data; (b), recode test data to aggregate specific EF components for comparison across karyotypes; and (c) identify distinct cognitive strengths and deficits in EF associated with TS so as to inform and promote early interventions and remediation that may improve educational and later life outcomes. This study presents descriptive statistics for the retrospective TS sample for each EF measure and domain. There was not sufficient evidence to support executive dysfunction in TS. However, over 50% of the retrospective sample was

diagnosed with ADHD. Information on how to assess for executive functioning during a neuropsychological evaluation is included. As weaknesses in selective attention were found in the retrospective TS sample, appropriate interventions are included. Lastly, this study presents the recommendations put forth by the American Academy of Clinical Neuropsychology (AACN) regarding consistent labeling of test performance. This dissertation is available in open access at AURA (https://aura.antioch.edu) and OhioLINK ETD Center (https://etd.ohiolink.edu).
Keywords: Turner syndrome, executive functioning, ADHD, selective attention

Dedication

This dissertation is dedicated to all individuals with Turner syndrome. Your perseverance and optimism are truly inspiring.

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CHAPTER I: INTRODUCTION

Individuals diagnosed with Turner syndrome (TS) have a number of neurocognitive weaknesses that can contribute to challenges in various aspects of their lives, including in academic, social, and emotional functioning. To date, researchers have compiled general neuropsychological profiles for individuals diagnosed with TS. However, there are limited studies exploring executive functioning (EF) for particular strengths and weaknesses related to the specific karyotypes of TS. EF deficits can impact all aspects of behavior and directly affect cognitive functions related to adaptive and strategic thinking, the ability to plan and carry out cognitive tasks, time management, self-control, organization, and effective self-monitoring (Baron, 2018). Current and future studies of TS can help clarify specific EF strengths and weaknesses among the different TS karyotypes, so as to better promote and inform earlier intervention for individuals with TS and to ensure better academic and later life outcomes.

Turner Syndrome

TS is a chromosomal disorder seen only in phenotypically female (pf)[1] individuals who have a partial or complete absence of the second sex (X) chromosome (Fechner, 2020; Hutaff-Lee et al., 2013; Mauger et al., 2018; Ross et al., 2000). TS affects approximately 1 in 2,000–2,500 live female births worldwide, and it is estimated that more than 70,000 individuals in the United States have TS (National Organization for Rare Disorders [NORD], 2019). There are no racial or ethnicity factors that determine etiology (NORD, 2019). Though TS can be

[1] The term phenotypically female (pf) is being used in an effort to use more inclusive language when discussing this population. (pf) will be used to describe that biologically all TS individuals are female, but may identify with a gender other than female.

diagnosed before or shortly after birth, mild cases may remain undiagnosed until adolescence or even into adulthood (Fechner, 2020).

TS can present with a range of phenotypic and genotypic features, and it varies greatly from one individual to another (Fechner, 2020). Most symptoms of TS occur because of the loss of specific genetic material from one of the X chromosomes (Fechner, 2020), although symptoms may also be caused by some cells containing Y chromosome sequences in rare cases (Gravholt et al., 2017; Gürsoy & Erçal, 2017), which can be treated with various interventions. Treatment involves a multidisciplinary approach and several specialized fields including, but not limited to, genetics, endocrinology, reproductive endocrinology, cardiology, behavioral health, neuropsychology, nephrology, and otolaryngology (Gravholt et al., 2017).

Variants of Turner Syndrome

Karyotyping in TS ranges from complete monosomy to forms of mosaicism, in which there is a typical cell line or an atypical second (or third) cell line (Gravholt et al., 2017).

Monosomy X (45,X)

Approximately 50% of individuals with TS are missing the X chromosome in all cells (45,X or monosomy X; Fechner, 2020; Gürsoy & Erçal, 2017). However, because 99% of monosomy X fetuses end spontaneously, living monosomy X individuals must have a form of mosaicism for another cell line (Held et al., 1992).

Structural Abnormalities of the X Chromosome

Isochromosome X [46,X,i(X)]. Isochromosomes occur when one arm of a chromosome is missing and is replaced by an identical version of the other arm and, therefore, consists of either two short arms or two long arms (Gürsoy & Erçal, 2017). Isochromosome occurs in about 15–18% of TS cases, with or without mosaicism (Gürsoy & Erçal, 2017).

Ring Chromosome [46,X,r(X)]. Ring chromosomes [r(X)] occur when the ends of a chromosome break off and the long and short arms fuse together to form a ring (Gürsoy & Erçal, 2017). A ring X chromosome is found in approximately 6% of individuals with TS (Gürsoy & Erçal, 2017). This phenotype can be highly variable depending on the size of the ring chromosome and the deletions of the long and short arms (Gürsoy & Erçal, 2017). However, structural brain abnormalities, intellectual disability, learning disability, and autism spectrum disorders are more frequently found in individuals with ring chromosome than in individuals with a monosomy X karyotype (Leppig et al., 2004).

Deletion (Xp or Xq). The frequency of Xp deletion in TS occurs in approximately 2% of cases (Gürsoy & Erçal, 2017). Although this phenotype exhibits variability in partial deletions, the observable, clinical features of TS are observed especially in individuals with deletion of the entire short arm of the chromosome (Gürsoy & Erçal, 2017).

Mosaicism

Approximately 30% of those with TS have mosaicism, where some cells have the complete number of chromosomes (46,XX) and others are missing an X chromosome (Fechner, 2020).

45,X/46,XX. This is the most common form of mosaicism and occurs in about 15% of TS cases (Gürsoy & Erçal, 2017). These individuals generally have a typical phenotype rather than the usual features associated with TS (Gürsoy & Erçal, 2017).

45,X/46,XY. In rare cases, some cells have one copy of the X chromosome, while other cells have one copy of the X chromosome with some Y chromosome material. About 5–10% of individuals with TS have cells that contain Y chromosome sequences (Gravholt et al., 2017; Gürsoy & Erçal, 2017). Though the amount of Y chromosome material is not enough to cause

the development of any male features, it is associated with increased risk of developing gonadoblastoma—tumors of the internal sex organs—and other gonadal tumors (Gürsoy & Erçal, 2017).

45,X/47,XXX; 45,X/46,XX/47,XXX. This karyotype is extremely rare and occurs in about 3–4% of individuals with TS. This variant of TS has an extra X chromosome (triple X; 47,XXX), and typically accompanies a milder phenotype, with observed learning disabilities, attention deficits and behavioral disorders (Tartaglia et al., 2010). While triple X syndrome in the non-mosaic state is associated with decreased intellectual ability, Sybert (2002) found no significant difference between individuals with monosomy X and those with 45,X/47,XXX and 45,X/46,XX/47,XXX regarding intellectual disability.

Purpose of the Present Study

With extensive attention paid to research exploring medical treatments for TS, learning disabilities in individuals newly diagnosed and living with TS are often overlooked (Levitsky, 2013). In fact, a survey completed by Sandberg et al. (2018) found that while individuals with TS and their families rated both biomedical and psychosocial research as "very important," an audit of peer-reviewed literature showed that 91% of published studies focused on the biomedical aspects of TS while only 9% concentrated on psychological variables.

Currently, many protocols, assessments, and interventions used for individuals with TS are based on other neurodevelopmental disorders. Interventions directed toward identified learning or cognitive problems related to EF improve educational and life outcomes, and early EF intervention can help to maximize social, educational, and vocational potential for individuals with TS. With further empowerment, opportunities for ongoing development of mastery and a

positive self-concept become available to a greater number of people with this rare genetic condition.

The primary objective of this study is to identify distinct cognitive strengths and deficits in EF associated with TS with a goal of clarifying, creating, or updating neurocognitive profiles among the different TS karyotypes. The present study stands to push the range of neurocognitive rehabilitation to include more people with TS. This includes possibilities for new neuropsychological developments and the increased promotion of existing early interventions, including targeted interventions and the most suitable accommodations by karyotype.

Research Questions and Overall Hypotheses

1. What are the differences in EF domains (goal setting, sustained attention, selective attention, inhibition, and working memory) between the different TS karyotypes—monosomy X, mosaicism, isochromosome X, ring X chromosome, and deletion of an X chromosome—as measured by neuropsychological test measures evaluating EF for (pf) individuals diagnosed with TS?
2. What are the differences in EF domains (goal setting, sustained attention, selective attention, inhibition, and working memory) and age range (children, adolescence, adults) in the different TS karyotypes (monosomy X, mosaicism, isochromosome X, ring X chromosome, and deletion of an X chromosome)

Based on the available research on neurocognitive functioning, and more specifically executive functioning, in TS, expected findings include executive dysfunction across karyotypes, specifically in the areas of attention and working memory. Compared to all karyotypes, it is also hypothesized that those with mosaicism TS may have less executive dysfunction compared to other karyotypes, and that ring X chromosome TS may have more executive dysfunction, as this

karyotype is linked more with intellectual disability, learning disabilities, and brain abnormalities. In terms of age, it is predicted that (pf) individuals diagnosed with TS will continue to have relative difficulties on measures of EF into early adulthood (Romans et al, 1998), and that (pf) individuals diagnosed with TS will have more executive dysfunction in early adulthood than during latency.

CHAPTER II: REVIEW OF THE LITERATURE

Early Neurodevelopment in Turner Syndrome

Of the limited data available, infants and toddlers with TS have associated developmental delays across all domains, including fine motor, gross motor, executive functions, attention, and language skills (Green et al., 2015; Hutaff-Lee et al., 2018). Furthermore, increased medical problems and repeated surgical interventions in the first few years of life can have negative impacts on early development. As a result, it can be difficult to distinguish which aspects of a TS developmental profile may be related to medical vs. neurodevelopmental features (Hutaff-Lee et al., 2018). For example, one study (Marino et al., 2012) found that infants with medical complications that require surgical intervention (i.e., congenital heart disease), which is often seen in variants of TS (Gravholt et al., 2017), are at an increased risk for neurodevelopmental problems. Middle ear disease and subsequent conductive, sensorineural, or mixed hearing loss is often observed in children with TS (Alves & Oliveira, 2014; Kubba et al., 2017), which can impact speech and language development, as well as social functioning. Because there is a lack of information on the early neurodevelopment in TS, early developmental evaluations are typically informed by protocols used for other populations at risk for developmental delay (Hutaff-Lee et al., 2018). With increasing frequency, literature on children with developmental disorders confirm improved outcomes with early interventions (Gravholt et al., 2017).

Turner Syndrome and Cognitive Functioning

Individuals with TS have significant heterogeneity in gene expression that accompanies a variety of phenotypes. Along with myriad physical features and medical conditions (i.e., short stature, webbed neck, characteristic facial features, infertility, and cardiovascular, renal, endocrine and auditory abnormalities; Bondy, 2007; Hutaff-Lee et al., 2018), there is also a

characteristic neurocognitive profile that can contribute to challenges in various aspects of life, including academic, social and emotional functioning. Individuals with TS may have various types of specific learning disabilities and psychological challenges such as, relative strengths in verbal abilities and relative weaknesses in visual-spatial/perceptual abilities, nonverbal memory function, motor function, processing speed, executive function, and attentional abilities (Green et al., 2015; Hong et al., 2009; Hutaff-Lee et al., 2018; Mazzocco, 2006; Ross et al., 2000), as well as poor mathematics performances (Murphy et al., 2006).

TS, as well as other genetically anomalous conditions (e.g., Fragile X syndrome, 22q11.2 deletion syndrome), can be associated with a nonverbal learning disability (NLD; Broitman, 2011), a learning disability in which verbal skills are greater than perceptual skills (Inchaustegui, 2019). Individuals with NLD frequently exhibit cognitive and neuropsychological strengths in simple motor skills, auditory perception, and rote learning with concurrent deficits in tactile perception, visual perception and spatial organization, complex psychomotor skills, and the learning of novel material. This profile of strengths and weaknesses is often significant enough to cause academic, professional, social, and emotional difficulties (Inchaustegui, 2019).

Attention-deficit/hyperactivity disorder (ADHD) is also associated with TS, and approximately 25% of individuals with TS meet criteria for ADHD. ADHD is defined by the Diagnostic and Statistical Manual of Mental Disorders (5th ed., text rev.; DSM-5 TR) as a disorder of inattention, impulsivity, and/or hyperactivity (American Psychiatric Association [APA], 2022). Evidence shows that the effects of ADHD can broadly impair functioning across multiple domains, including academic, occupational, and social (Baron, 2018).

While the presence or absence of a deficit or learning disability based on a karyotype should never be assumed, those with a ring X chromosome tend to have higher degrees of verbal

and non-verbal difficulties than those with 45,X monosomy (Bray et al., 2011; Fechner, 2020; Kuntsi et al., 2000). This includes the highest risk of intellectual disability (Kubota, 2002), compared to individuals with monosomy X (Gravholt et al., 2017). Individuals with a ring X chromosome also obtained significantly lower scores on the Wechsler Intelligence Scale for Children, Fourth Edition Perceptual Reasoning and Working Memory indexes when compared to those with mosaicism (Bray et al., 2011). Individuals with mosaicism, however, tend to be less severely affected than those with monosomy X (Ross et al., 2000). Compared to other karyotypes, 45,X monosomy has been associated with higher levels of visual-constructive and visual-perceptual deficits, verbal episodic memory impairments and visual-spatial working memory difficulties (Buchanan et al., 1998; Mauger et al., 2018; Temple et al., 1996).

Neurodevelopmental function, including EF, can change throughout one's development and may improve with appropriate intervention. As an individual transitions into adulthood, managing the demands and responsibilities associated with higher education, career, and independent living can become more difficult, especially for those with deficits in executive functioning. While there is limited information on executive dysfunction in individuals with TS in adulthood, research in other chromosomal disorders, such as 22q11.2 deletion, suggest that there tends to be more EF difficulties in adulthood (Quintero et al., 2014).

Executive Functioning

The concept of executive functioning (EF) suggests a set of correlated yet separatable abilities that are mediated by the prefrontal and parietal parts of the brain (Collette et al., 2006). There are multiple EF models and varying ways in which the construct of EF is defined. As such, EF abilities are measured in different ways. In general, EFs are defined as a set of high-level interrelated processes that assist with goal-directed behavior, adaptation to environmental

demands, and which allows for control, organization, and direction in cognitive activities and the self-regulation of behavior (Anderson, 2008; Hong et al., 2009; Sattler & Walters, 2014).

Challenges in the area of EF can impact how well individuals are able to complete daily tasks and may result in significant impairment of everyday functioning. EF are essential for perception of and adaptation to the changing demands in an environment (Hong et al., 2009). EFs are key to decision making, distinguishing relevant from irrelevant material, following general rules, and making use of existing knowledge in new situations (Sattler & Walters, 2014). Anderson (2008) describes a variety of presentations of impaired EF, including:

> an inability to focus or maintain attention, impulsivity, disinhibition, reduced working memory, difficulties monitoring or regulating performance, inability to plan actions in advance, disorganization, poor reasoning ability, difficulties generating and/or implementing strategies, perseverative behavior, a resistance to change activities, difficulties shifting between conflicting demands, and a failure to learn from mistakes (Executive Function section, para. 2).

EF skills are critical for generating alternative solutions to problems, recognizing multiple approaches to a task, deciding which responses may be more effective, and knowing when mistakes have been made (Anderson, 2008). Deficits in these areas may cause an individual to struggle with recognizing connections between different concepts or how different parts of a problem, task, or idea fit together. A failure to understand increasingly complex information involving concepts that build upon each other, such as those seen in math and science, may be caused by weaknesses in EF (Anderson, 2008; Hong et al., 2009). Math is highly dependent on EF, and includes skills such as working memory, attention, and mental flexibility. Deficits in working memory can also cause problems with multitasking, and

individuals with poor working memory often forget or lose track of what they were doing or thinking. (Hong et al., 2009).

Executive dysfunction is often associated with academic, employment, and social concerns (Anderson, 2008). Thus, formal EF supports and interventions can be of a significant benefit to individuals with academic, adaptive, social and vocational difficulties (Sattler & Walters, 2014). Early intervention can be especially important to mitigate the EF problems that can persist over the lifespan (Baron, 2018).

Four-Domain Executive Control System

The executive control system, as defined by Anderson (2008), is a developmental neuropsychology model used for understanding the functional domains of EF. Anderson (2008) explains that the four domains of EF—attentional control, cognitive flexibility, goal setting, and information processing—are independent of each other and made up of discrete functions, while also operating together in a bidirectional and functional manner. In other words, the task determines the domain(s) being utilized. Thus, the EF domains maintain both independence and interrelatedness in its overall operational function as a control system.

Attentional Control. This domain requires an ability to focus on relevant themes and details (prioritizing), to regulate one's behavior and monitor one's thoughts and actions (self-regulation), and to inhibit thoughts and actions that are inappropriate to a situation (inhibition) (Anderson, 2008). Attentional control involves both selective attention—focus on specific stimuli while ignoring irrelevant or distracting stimuli—and sustained attention—focused attention for prolonged periods (Sattler & Walters, 2014).

Cognitive Flexibility. Cognitive flexibility involves processing multiple sources of information concurrently, the dividing of attention across tasks, and learning from one's mistakes

(Sattler & Walters, 2014). Set-shifting is part of this domain, and it includes the ability to alternate between different thoughts and actions and to devise alternative problem-solving strategies. Working memory is integral to this domain and involves the ability to hold and manipulate information in short-term memory (Anderson, 2008). Another key component is the ability to monitor and encode incoming information by replacing outdated content with new, more relevant information (updating; Sattler & Walters, 2014).

Goal Setting. Goal setting refers to the ability to devise a logical, systematic, and strategic plan to completing an activity. This includes the ability to plan and reason conceptually, monitor one's actions, and set appropriate goals (planning and goal setting). Organizing is also a part of this domain, which involves organizing ideas and information (Anderson, 2008).

Information Processing. The ability to process information fluently and quickly is central to this domain. Of key importance to this ability, as a domain of EF, is the effective synthesis of content for use in memory, which involves four stages that are relevant to the temporal nature of information processing: (a), immediate, perceptual processing of new information; (b), efficient capture of relevant, representational content for manipulation and timely use; (c) aptitude for maintenance of well-timed responses; and 4) delivery of adequate outputs of information (Anderson, 2008).

Individuals with TS are found to perform significantly less well than individuals without TS on measures of EF (Green et al., 2015; Mauger et al., 2018; Temple, 2002; Zinn et al., 2007). The impact EF deficits can have on learning greatly influences how a child or adolescent with TS may perform in school. Children and adolescents with TS have been shown to present with mathematical impairments which have significant associations with EFs (Mazzocco, 2006).

Further, EFs impact an individuals' ability to stay focused, plan ahead, strategize, and recall information in the classroom setting (Anderson, 2008).

Executive Functioning in Turner Syndrome

Deficits in EF, as well as structural and functional abnormalities in EF brain regions, have been found to occur in individuals diagnosed with TS (Bray et al., 2011; Green et al., 2015; Hutaff-Lee et al., 2018; Lepage et al., 2013; Mauger et al., 2018). Existing studies have shown mixed results regarding deficits in executive functioning (Mauger et al., 2018), with some studies showing weaknesses in specific EF domains and other studies showing no weaknesses in the same EF domain. A meta-analysis exploring EF in TS revealed evidence supporting executive impairment in individuals with TS. However, the authors argue that these results should be interpreted with caution due to discrepancies in the samples reported IQ and karyotype, as well as whether or not the participant had received growth hormone or estrogen therapy (Mauger et al., 2018).

Impairments in working memory have been the most widely reported area of impairment in this population (Bray et al., 2011; Green et al., 2015; Lepage et al., 2013; Mazzocco, 2006; Murphy et al., 2006) with Mauger et al. (2018) finding that working memory (measured by digit span tasks), along with higher-order EFs, were the most affected compared to other EF domains. Neuroimaging studies have evaluated the functional connectivity between frontal and parietal regions related to working memory. Executive control regions (prefrontal cortex), as well as parietal regions during processing of visuospatial information, are widely implicated in individuals with TS (Hart et al., 2006). Bray et al. (2011) revealed that weakened frontoparietal interactions were found in ten 45,X monosomic and 4 unknown mosaicism TS participants, ages 7–13 years old.

Buchanan et al. (1998) have identified indicators that in the general population, cognitive deficits are more pronounced during tasks with high demands, such as in activities involving working memory and visuospatial domains, and are reflective of selective impairment in the engagement of higher-order cognitive functions in the brain. For example, impaired performance is often observable on measures that combine visual perception and abstract reasoning and mental flexibility, such as on the Wisconsin Card Sorting Test (WCST), the Contingency Naming Test (CNT), and the Raven's Progressive Matrices (RPM; Hong et al., 2009). Tests of visual-spatial processing, which also require the use of other executive skills—including tests of working memory and visual attention, such as on the Motor-Free Visual Perception Test (MVPT), Embedded Figures Test (EFT), Spatial Relations Test (SRT), and Mental Rotation Test (MRT)—also capture impairments in higher order functioning, including EF (Hong et al., 2009).

When considering EF functioning, the use of visual-spatial functioning must be taken into consideration, as many EF test measures and tasks involve visual-spatial functioning, which is noted to be a deficit in individuals with TS (Hong et al., 2009). Many studies look at EF abilities in terms of visual-spatial deficits. Interestingly, Green et al. (2015) report attention, self-inhibition and EF deficits independent of visuospatial effects in monosomy X TS. Further, impairments in working memory have been found to occur in both executive non-verbal and verbal tasks in more recent years (Mauger et al., 2018). This is important to note as previous hypotheses have suggested that individuals with TS may have more difficulty on executive tasks with non-verbal stimuli or when they cannot use a verbal strategy to support executive processes (Temple et al., 1996).

Aspects of attention, including prioritizing, self-regulation, inhibition, selective attention, and sustained attention (Anderson, 2008), have been less often explored in the TS population. Of

available studies, a deficit in cognitive inhibition on the Stroop task has been highlighted by Temple et al. (1996). Researchers also found that a group of 45,X monosomic TS individuals had scored significantly lower than a control group of neurotypical subjects on both the Auditory Attention and Response Set subtest and the Visual Attention subtest of A Developmental Neuropsychological Assessment (NEPSY; Green et al., 2015). A meta-analysis by Mauger et al. (2018) confirmed that cognitive inhibition may be more impaired than other inhibitory control abilities, such as selective or focused attention and response inhibition.

Temple (2002) has studied EF impairments in individuals with TS to observe whether skills within domains of strength, such as verbal skills, are negatively impacted. They found that oral fluency performance was impaired in the TS participant group due to fewer category switches (Temple, 2002). The TS participant group also struggled with narrative production that involved preservation of a temporal framework (Temple, 2002).

While the debate continues as to whether processing speed is a component of executive functioning or a separate and unrelated entity, individuals with TS have been found to have a reduced processing speed (Hong et al., 2009). Temple (2002) hypothesized that decreased fluency and speed on rapid naming tasks in individuals with TS is related to deficits in information processing due to atypical ways of searching for and recalling words. TS participants showed significantly decreased rates in set-shifting, due to different executive processes involved in retrieval of verbal stores in individuals with TS (Temple, 2002).

Summary

Differences across karyotypes may result in variability in EF profiles. Unfortunately, the impact of the karyotype on EF has not been widely studied. Few studies compare monosomy X to other karyotype groups, and the information that is presented on monosomy X can be

inconclusive. A systemic review and meta-analysis, completed by Mauger et al. (2018), indicated significant EF impairments with varying effect sizes. Further, methodological differences across studies cause barriers in determining which EFs are impaired and to what extent. It is thus important to study the different groups and the EF profile of each type in TS to improve the understanding of TS and determine how to best intervene with individuals with TS to help to maximize social, educational, and vocational potential. The current study sought to this gap in the literature.

CHAPTER III: METHOD

Design

This study originally set out to be a quasi-experimental, between-group design. However, due to a small sample size, the study became exploratory research using a quantitative, descriptive design. Neuropsychological evaluation report data was used from retrospectively completed comprehensive neuropsychological evaluations conducted between 2008 and 2020 from (pf) individuals diagnosed with TS. Neuropsychological reports were obtained from Maple Leaf Clinic, a private psychology practice located in Wallingford, VT and via a recruitment flyer distributed to the Turner Syndrome Global Alliance, Turner Syndrome Colorado, Turner Syndrome Carolinas, and Turner Syndrome Society of the United States.

Participants

All participants included (pf) individuals who had been diagnosed with TS. There were 20 participants (N = 20) ranging in age from 6 to 30 years (M = 12.2, SD = 5.9). Three participants had a karyotype of monosomy X, five participants had mosaicism (one participant had 45,X/46,XX, one participant had 45,X/47,XXX, and three participants had an unknown mosaicism), and one participant had Ring X. The karyotype was unknown for 11 of the participants in the sample. At the time of testing, 14 participants were receiving special education services. Specifically, 11 were on an individualized education plan (IEP) and three were receiving 504 accommodations. It is unknown if one of the participants was receiving special education services in school.

Thirty-five percent of participants were diagnosed with ADHD and 85% with NLD either prior to neuropsychological testing or as a result of testing. Two participants (10%) were diagnosed with intellectual disability (ID) and three participants (15%) were diagnosed with

borderline intellectual functioning (BIF). Six participants (33.3%) had surgery during the first year of life which included aortic repair ($N = 5$) and vitrectomy to treat diabetic retinopathy ($N = 1$). The surgical history in the first year of life is unknown for two participants.

All participants were born female. Gender identity is unknown. The race and ethnicity of the participants was varied, with 12 (70%) identifying as White, one identifying as Black, one identifying as biracial, one identifying as Latinx, one identifying as South Asian, and one identifying as Other Pacific Islander. The race and ethnicity of one participant was unknown. This dissertation included two international participants, with 18 (90%) from the United States, and one each from Canada and Brazil.

All participants were required to have had a comprehensive neuropsychological evaluation with an accessible neuropsychological report completed no earlier than 2008. Evaluations were required to have been completed no earlier than 2008 to ensure that the most recent normative data for the Wechsler Adult Intelligence Scale, 4th Edition (WAIS-IV; Wechsler, 2008) was used. This dissertation specifically focused on gathering reports for individuals beginning at the age of 6 years, as this is when age-appropriate norms become available for many of the neuropsychological test measures used in this study, including the Wechsler Intelligence Scale for Children, 5th Edition (WISC-V; Wechsler, 2014). In an effort to eliminate the confounding variable of neurodegeneration associated with aging, the cutoff for participation in this study was set to age 40.

Participants were recruited via a TS data set of neuropsychological reports and neuropsychological testing protocols from retrospectively-completed comprehensive neuropsychological evaluations conducted at Maple Leaf Clinic between 2008 and 2020 (See Appendix A and B for the permission form from Maple Leaf Clinic). The Maple Leaf Clinic

approved the review of client files from a specified filing cabinet designated for clients that had a diagnosis of either NLD or TS. Approximately 60 client files were reviewed and those containing a diagnosis of TS were used for the purpose of this study. Twenty client files from the Maple Leaf Clinic were found to meet this study's criteria of having a TS diagnosis. Of those 20 client files, two participants were excluded due to the individuals being under the age of 6 years old.

Participants were also recruited through an advertisement written in English and Spanish - Latin American (See Appendix C and D) and shared to various TS societies and groups, including the Turner Syndrome Society of the United States, Turner Syndrome Global Alliance, Turner Syndrome Colorado, and Turner Syndrome Carolinas who shared them to their Facebook pages. The Turner Syndrome Society of the United States also shared the recruitment flyer in their newsletter. Four participants were recruited through the advertisement sent out to the various TS groups. It is unknown which TS Facebook post or newsletter the participants were recruited from, as this information was not asked on the Google Forms questionnaire participants were directed to in order to participate in the study. Two participants were excluded from the study as they had never had a neuropsychological evaluation completed.

Procedure

Neuropsychological reports were obtained through two, separate means: (a) retrospectively-completed neuropsychological evaluations from Maple Leaf Clinic in Wallingford, Vermont; and (b) recruitment advertisements posted on various TS Facebook pages, including the Turner Syndrome Global Alliance, Turner Syndrome Colorado, Turner Syndrome Carolinas, and Turner Syndrome Society of the United States, and in the Turner Syndrome Society of the United States newsletter.

Neuropsychological Reports Obtained from the Maple Leaf Clinic

The Maple Leaf Clinic approved the review of client files for this study. Participants recruited from the Maple Leaf Clinic signed an informed consent at the time of testing indicating that their test results may be used for future research without requiring further consent. All were advised at that time that their information would remain confidential. Participants were included if they met the above inclusion criteria. Anonymity of participants was maintained by assigning each participant an ID number. A separate database was used to link each participant's ID number to their name. This database was stored in a separate, password-protected folder on a password-protected flash drive. Only the project investigator had access to the passwords. Neuropsychological reports accessed from Maple Leaf Clinic remained onsite in a locked filing cabinet with access limited to Dr. Dean Mooney and the Maple Leaf Clinic administrative staff.

The client file, which included a neuropsychological report, various neuropsychological tests and measures, behavioral checklists, and demographic questionnaires, was reviewed for each participant. Available demographic, educational, medical, and special education/tutoring information was collected from each client file and included in a deidentified data base. Of the demographic information collected, each participant's TS karyotype was recorded. Eleven participants did not have a known karyotype included in their client file. It was decided that these participants would be included in the study once the recruitment period expired and there were not enough participants in each karyotype group to complete an accurate analysis examining EF strengths and weaknesses between the karyotype groups. Results on neuropsychological tests and measures determined to measure specific EF components (goal setting, sustained attention, selective attention, inhibition, and working memory) was also obtained from each client file.

Raw scores, standard scores, and qualitative descriptors of the EF neuropsychological tests and measures were included in a deidentified data base for each participant.

Neuropsychological Reports Obtained from Recruitment Advertisements

The recruitment flyer, provided in English (see Appendix C) and Spanish - Latin American (see Appendix D), was emailed to various TS groups, including the Turner Syndrome Global Alliance, Turner Syndrome Colorado, Turner Syndrome Carolinas, and Turner Syndrome Society of the United States. Each of the aforementioned groups posted the recruitment flyer to their Facebook page. The Turner Syndrome Society of the United States also placed the recruitment flyer in their newsletter. The recruitment flyer included the study purpose and participant inclusion information, as well as a request to complete of a brief questionnaire along with submission of a neuropsychological report completed since 2008. The flyer also included a link to the questionnaire on Google Forms and the project investigator's name and email address.

Once participants were directed to the Google Forms page, participants 18 years of age and over were asked to complete an electronic consent form, provided in both English (see Appendix E) and Spanish - Latin American (see Appendix F). They were advised that that their information would remain confidential. Participants under the age of 18 required their parent or guardian complete an electronic consent form provided in English (see Appendix G) and Spanish - Latin American (see Appendix H). Participants under the age of 18 were also asked to complete an electronic assent form, provided in English (see Appendix I) and Spanish - Latin American (see Appendix J). The informed consent and assent included that each individual agreed to participate in a research project, described the terms of the research project, confirmed that participation was voluntary, and that any participant could withdraw from the study at any time without penalty. All participants were advised that their information would remain confidential.

Consent and assent were documented via Google Forms and could be printed out by participants upon completion.

Demographic, educational, medical, and special education/tutoring information was collected via a Google Forms questionnaire, provided in English (see Appendix K) and Spanish - Latin American (see Appendix L) for participants recruited through the Turner Syndrome Global Alliance, Turner Syndrome Colorado, Turner Syndrome Carolinas, and Turner Syndrome Society of the United States Facebook pages and the Turner Syndrome Society of the United States newsletter. Participants over the age of 18 who had not been appointed a guardian were asked to complete the questionnaire themselves. Parents and guardians of participants under the age of 18 or of incapacitated persons were asked to complete the questionnaire for participants.

Participants, or their parent/guardian, were asked to submit a neuropsychological report via Google Forms or by fax to the project investigator via MetroFax. Neuropsychological reports that were collected via Google Forms were kept in a password-protected folder on a password-protected flash drive. Only the researcher had access to the passwords. No participants chose to fax neuropsychological reports via MetroFax. Anonymity of participants was maintained by assigning each participant an ID number. A separate database was used to link each participant's ID number to their name. This database was stored in a separate, password-protected folder on a password-protected flash drive. Only the project investigator had access to the passwords.

Demographic, educational, medical, and special education/tutoring information collected via a Google Forms questionnaire was entered into a deidentified data base for each participant. Results on neuropsychological tests and measures determined to measure specific EF components (goal setting, sustained attention, selective attention, inhibition, and working

memory) was also obtained from each neuropsychological report submitted via Google Forms. Raw scores, standard scores, and qualitative descriptors of the EF neuropsychological tests and measures were included in a deidentified data base for each participant.

Measures

Assessment of EF included scores from retrospective neuropsychological evaluation tests and measures with reliable and valid data pertaining to the Four-Domain Executive Control System (Anderson, 2008) and its specific components of EF (goal setting, sustained attention, selective attention, inhibition, and working memory). Although the Four-Domain Executive Control System (Anderson, 2008) included the components of prioritizing, self-regulation, set shifting, updating, planning, and organizing, there were no neuropsychological assessment measures used in any of the retrospective comprehensive neuropsychological reports that measure these components, and thus were unable to be explored in this study.

Behavioral assessment of EF also included subscale scores from the Achenbach Behavior Checklist (Achenbach, 2009). Subscales used, included Attention Problems and ADHD Problems. The original dissertation proposal called for use of the Behavior Rating Inventory of Executive Function (BRIEF; Gioia et al., 2000) and its subscales (Inhibit, Shift, Emotional Control, Initiate, Working Memory, Planning and Organization, Organization of Materials, and Monitor); however, with only one participant being administered this scale during their neuropsychological evaluation, this measure was excluded.

Neuropsychological Test Measures

Neuropsychological tests were administered to participants during the retrospectively-completed neuropsychological evaluation that were conducted. For the purpose of this dissertation, the available neuropsychological tests and measures in the retrospectively-

completed neuropsychological evaluation were determined to be a measure of a specific EF component using the Four-Domain Executive Control System defined by Anderson (2008) and available literature on the constructs measured by each of the neuropsychological tests. It is recognized that there is fluidity in which tests fit into specific EF domains. In several instances, a strong case could be made for equally appropriate assignment of a measure to another EF domain. Based on the available tests and measures found in the retrospectively-completed neuropsychological evaluations, the following domains were able to be examined in this study: goal setting, aspects of attentional control (e.g., sustained attention, selective attention, and inhibition), and cognitive flexibility (e.g., working memory). As participants were of different ages and tested by different evaluators at different sites, different versions of tests measuring the same construct were given based on test availability and appropriateness given the participants' age.

Sustained Attention. Attentional Control requires sustained attention, which is defined as focused attention for prolonged periods (Sattler & Walters, 2014). The Seashore Rhythm Test and Speech Sounds Perception Test were used in this dissertation as a measure of sustained attention. Despite these tests being of two differing lengths, they were determined to be a measure of both short and long sustained attention for this study's purposes.

Seashore Rhythm Test. The Seashore Rhythm Test, a component of the Halstead Reitan Neuropsychological Test Battery (HRNTB), consists of 30 pairs of rhymical patterns presented on a standardized recording (Reitan & Wolfson, 1992). The individual is required to listen carefully to the stimuli in each pair and to determine whether the second stimuli is the same as the first or different from it. The 30 pairs are organized into three subsections of 10 items each, with a progression of complexity of the rhythmic beat from one series of ten items to the next

(Reitan & Wolfson, 1992). Administration is approximately 7 minutes in length. The Seashore Rhythm Test was used in this study as a measure of short, sustained attention. The addition of the time frame (e.g., short) was added to differentiate it from the Speech Sounds Perception Test, a measure which also used to evaluate sustained attention (detailed below). The test has a total test score reliability of .78 (Bornstein, 1983). Limitations to the Seashore Rhythm Test include low reliability for each subpart as many scores are no different than chance responding (Charter & Webster, 1997). For the purpose of this study, the full test was used. The Seashore Rhythm Test is not suggested for individuals with a substantial amount of musical experience, as they tend to perform normally on the Seashore Rhythm Test, even in the context of known cognitive impairment (Karzmark, 2001). Given that the neuropsychological evaluations were given retrospectively, there is no way of knowing if the participants used in this study were screened for musical experience prior to being administered the Seashore Rhythm Test.

Speech Sounds Perception Test. The Speech Sounds Perception Test is a component of the HRNTB (Reitan & Wolfson, 1992). The test consists of 60 nonsense syllables presented on a standardized audio recording of approximately 17 minutes in length. The individual is asked to correlate the nonsense syllable with alternatives printed on the answer sheet (Reitan & Wolfson, 1992). The Speech Sounds Perception Test assesses the ability of the test taker to discriminate phonemes while depending on their attentional capacity and single word reading (Baron, 2018). The test was used in this study as a measure of long sustained attention. The addition of the time frame (e.g., long) was added to differentiate it from the Seashore Rhythm Test (detailed above). Bornstein (1982) examined the internal reliability in two independent samples and found that the split-half reliability was .74 and .87. Further, a significantly greater number of errors were found

on the first half of the test in both samples such that the largest number of errors occurred on subtests B and A (Bornstein,1982).

Selective Attention. Attentional control also involves selective attention, or the ability to focus on specific stimuli while ignoring irrelevant ones (Sattler & Walters, 2014). Various forms of a trail making test were used to measure selective attention, including the Delis-Kaplan Executive Function System (D-KEFS) Trail Making Test, the Progressive Figures Test, and Trail Making Test.

Delis-Kaplan Executive Function System - Trail Making Test - Condition 4: Number-Letter Switching. The Delis-Kaplan Executive Function System (D-KEFS) Trail Making Test (TMT) is normed for ages 8 to 89 years (Delis et al., 2001). It requires completion of five separate conditions. Only Condition 4: Number-Letter Switching, was used for the purpose of this dissertation. Condition 4: Number-Letter Switching is a timed paper-and-pencil test requiring the individual to draw a line while alternating back and forth between numbers and letters in sequence over two pages. It is used for individuals who have learned number and letter sequences in English. The maximum time limit is 240 seconds (Delis et al., 2001). The D-KEFS Technical Manual (Delis et al., 2001) reported low test-retest reliability on the TMT Condition 4.

Progressive Figure Test. The Progressive Figure Test was developed for children aged 5 to 8 years (Reitan & Wolfson, 2004). The test includes eight stimulus figures printed on an 8 ½ x 11-inch paper. Each figure includes a large outside geometric form with a different smaller internal geometric form. The child is required to trace a path from an internal small geometric shape to the similar larger external shape in the next sequence and then to use that figure's internal shape to direct movement to the next larger shape in the sequence until completing the trial (Reitan & Wolfson, 2004).

***Trail Making Test, Part B*.** The TMT was originally developed for adults and eventually incorporated into the HRNTB and adapted for children (ages 9 to 14) in an abbreviated version (Reitan & Wolfson, 1992). The TMT is a timed paper-and-pencil test for individuals who have learned number and letter sequences in English. The TMT has two parts, although only Part B was used in this study. Part B (15 numbers and letters for ages 9 to 14, and a total of 25 numbers and letters for ages 15 and above) requires that the individual draw a line while alternating back and forth between numbers and letters in sequence while simultaneously searching the page for the next stimulus item (Baron, 2018; Reitan & Wolfson, 1992). TMT Part B has been found to require divided attention (Lamberty et al., 1994), planning (Baron, 2018), cognitive flexibility (Bechtold et al., 2001), and the ability to maintain a complex response set (Lezak et al., 2012). The retest reliability on TMT Part B is good and has been noted as .86 (Wagner et al., 2011).

Inhibition. Another aspect of attentional control includes one's ability to inhibit thoughts and actions deemed inappropriate to a situation (Anderson, 2008). Stroop tests were used in this study as a measure of inhibition due to the tests' requirement of inhibiting a dominant response over a nondominant response in order to complete the task (Lezak et al., 2012). Stroop versions differ by colors presented, type and arrangement of stimuli, and order of presentation (Baron, 2018), but generally include a color name printed in a different color ink. Although, construct validity can be confounded by response shifting, sustaining attention, reading level, and naming ability, a study by Cox et al. (1997) supported the validity of the Stroop interference score as a measure of inhibition. The tests include three parts where the subject is asked to first name patches of color, then read words of colors, and lastly to name the color of the ink in which incongruent color names are printed (e.g., "red" printed in green ink; Baron, 2018; Lezak et al.,

2012). Various tests were used to measure inhibition, including D-KEFS Color Word Interference Test (Delis et al., 2001) and Stroop Color and Word Test (Golden et al., 2002).

Delis-Kaplan Executive Function System, Color Word Interference Test, Inhibition Trial. The D-KEFS is a battery of neuropsychological tests designed to measure executive functioning in children and adults (ages 8-89; Delis et al., 2001). The Inhibition Trial requires that the examinee read the word name of the items in rectangles while responding with the color ink of those words not in rectangles. The D-KEFS Inhibition Trial has low test-retest reliability at .57 (Delis et al., 2001).

Stroop Color and Word Test, Interference Condition. The Golden version of the Stroop Color and Word Test (Golden et al., 2002) is a three-color version (blue, green, and red) that includes three 45-second trials. Each trial includes a 100-item page. The word reading trial includes words of colors typed in black, the color naming trail includes "XXXX" in randomized color sequences, and the color-word trial includes color naming when the words are printed in nonmatching colored ink (e.g., the word "red" is printed in "green" ink and the correct response in "green"). The examinee is asked to read down the columns of stimuli on each trial as quickly as they can within the 45-second time limit (Baron, 2018; Golden et al., 2002). The interference condition is calculated by the following formula: INT = CW – (C x W) / (C + W). Test-retest reliability was reported by Franzen et al. (1987) to be .67.

Goal Setting. Goal setting is defined by Anderson (2008) as the ability to plan and reason conceptually, monitor actions, set appropriate goals, and organize ideas and information. The Category Test was used as a measure of goal setting for this study as the constructs assessed by the Category Test include concept generation, mental set shifting, rule learning, and problem solving (Baron, 2018).

Category Test. The Category Test is a component of the HRNTB (Reitan & Wolfson, 1992). It requires that the individual view a series of designs, discern commonalities among varying elements, form the basis of an organizing principle, and apply that principle to diverse stimulus configurations (Reitan & Wolfson, 1992). Reliability of the Category Test is very good at .98 (Shaw, 1966).

Working Memory. Anderson (2008) has included working memory, or ability to hold on and manipulate information in short-term memory, as a component of cognitive flexibility. Various span tests have been used as measures of working memory as they gauge how much information can be held after a single presentation and includes both verbal and visuospatial stimuli (Baron, 2018). Digit span tests require immediate repetition of orally-presented numbers composed of sequentially longer strings. To examine visual working memory, block or spatial span paradigms test immediate repetition of visuospatial stimuli (Baron, 2018).

Wechsler Intelligence Scale for Children, 4th Edition, Digit Span. The Digit Span (DS) subtest from the Wechsler Intelligence Scale for Children, 4th Edition (WISC-IV, Wechsler, 2003) is used for children ages 6 through 16. It has two parts, Digit Span Forward (DSF) and Digit Span Backward (DSB). On DSF, the examinee is asked to repeat a string of numbers in the identical order presented. In DSB, the examinee is read a sequence of numbers and is required to recall the numbers in reverse order (Wechsler, 2003).

Wechsler Intelligence Scale for Children, 5th Edition, Digit Span. Similar to the WISC-IV DS subtest, the Wechsler Intelligence Scale for Children, 5th Edition (WISC-V; Wechsler, 2014) DS subtest requires that the examinee recall a sequence of numbers, first in identical order (DSF) and then in reverse order (DSB). The WISC-V is appropriate for ages 6 through 16 (Wechsler, 2014).

Wechsler Adult Intelligence Scale, 4th Edition, Digit Span. The Wechsler Adult Intelligence Scale, 4th Edition (WAIS-IV; Wechsler, 2008), is used for adults ages 16 through 90. The WAIS-IV DS subtest also requires examinees to repeat a sequence of numbers in identical order (DSF) and then in reverse order (DSB; Wechsler, 2008).

Knox Cube Test. The Knox Cube Test requires tapping a sequence of blocks in a given serial order after observing an examiner tapping the sequence (Knox, 1914). It consists of four 1-inch wooden blocks spaced 2 inches apart in a row and attached to a piece of wood approximately 9 inches long (Baron, 2018).

Wechsler Intelligence Scale for Children, 4th Edition Integrated, Spatial Span. The Wechsler Intelligence Scale for Children, 4th Edition Integrated (WISC-IV Integrated; Wechsler, 2015) is an assessment of spatial working memory used for children ag 6 through 16. The Spatial Span (SSP) subtest requires the examinee to view the examiner tap a sequence of blocks and then repeat the sequence exactly (Spatial Span Forward), or in reverse order (Spatial Span Backward).

Behavioral Measures

Behavioral assessments of executive dysfunction were administered to participants during the retrospectively-completed neuropsychological evaluation that were conducted. For the purpose of this dissertation, specific subtests from the Achenbach System of Empirically Based Assessment (ASEBA) behavioral checklists were used (Achenbach, 2009). The ASEBA includes several self-report and informant behavioral checklists for school aged children and adults. Of the parent and teacher informant behavioral checklists, the Attention Problems and Attention Deficit/Hyperactivity Problems scales were used for this study. The Attention Problems measure is an empirically-based syndrome scale, which is grounded in factor analyses (Achenbach & Rescorla, 2001). The ADHD scale is a DSM-oriented scale comprised of items identified as

being very consistent with DSM-5 categories by experts across a variety of cultures (Achenbach & Rescorla, 2001).

Child Behavior Checklist for Ages 6-18. The Child Behavior Checklist for Ages 6-18 (CBCL/6-18; Achenbach & Rescorla, 2001) is a component of the ASEBA. It is a 113-item standardized rating scale that is completed by parents, and it measures a wide range of behavioral and emotional problems in children and adolescents (Achenbach & Rescorla, 2001). Test-retest reliability was reported to be very high for each item and was a .92 for Attention Problems and .93 for ADHD Problems (Achenbach & Rescorla, 2001).

Teacher Report Form for Ages 6-18. The Teacher Report Form for Ages 6–18 (TRF, Achenbach & Rescorla, 2001) is another component of the ASEBA. It is a 113-item standardized rating scale that is completed by teachers and is used to detect behavior and emotion problems in children and adolescents (Achenbach & Rescorla, 2001). Test-retest reliability was reported to be very high for each item and was a .95 for Attention Problems and .95 for ADHD Problems (Achenbach & Rescorla, 2001).

Adult Behavior Checklist. The Adult Behavior Checklist (ASR; Achenbach & Rescorla, 2003) is also a component of the ASEBA. It is a 126-item rating scale that is completed by someone well known to the adult. For this study, each participant checklist was completed by a parent. The ASR assesses competencies, adaptive functioning, personal strengths, and behavioral, emotional, social, and thought problems (Achenbach & Rescorla, 2003). Reliability is reported to generally be high for the ASR, with all test-retest correlations being in the .80s and .90s and being significant at $p<.01$. Reliability for the Attention Problems scale was reported at .91 and the ADHD problems scale was .84 (Achenbach & Rescorla, 2003).

Collecting Neuropsychological Test Data from Neuropsychological Reports

Raw and standard scores, as well as qualitative descriptors, were collected from the neuropsychological reports for each of the above-mentioned tests and measures. All standard scores were converted into z-scores. If only a raw score was provided, a standard score was calculated from the normative data provided in the test manual and converted into z-scores. For the Seashore Rhythm Test and Speech Sounds Perception Test, normative data from Findeis and Weight (1994) was used for ages 9–14, Fromm-Auch and Yeudall (1983) was used for ages 15–20, and Heaton et al. (2004) for ages 20 and older. Normative data for the Category Test was gathered from Knights (1966) for ages 6–8, Spreen et al. (1969) for ages 9–15, Fromm-Auch and Yeudall (1983) for ages 16–19, and Heaton et al. (2004) for ages 20 and older. Normative data for the Trail Making Test was used from Spreen et al. (1969) for ages 8–15, Fromm-Auch and Yeudall (1983) for ages 16–19, and Heaton et al. (2004) for ages 20 and older.

In some cases, only descriptors were used (e.g., “below the norm,” “severely below average,” “severely impaired,” “within normal limits,” and “impaired”). When only descriptors were used, Heaton et al. (2004) categorization descriptors were used to place scores in a category (e.g., severe, moderate/severe, moderate, mild/moderate, mild, low average, average, high average, superior, very superior). Based on the standardized scores provided in Heaton et al. (2004) for each category, a z-score was then assigned to each descriptor used in a neuropsychological report.

Analysis

For this study, analyses were completed using scores from neuropsychological tests and measures administered during retrospectively-completed neuropsychological evaluations of (pf) diagnosed with TS. The deidentified data set was downloaded to SPSS, which was the sole

statistical package used to conduct analyses. Various statistical procedures were utilized to explore how executive functioning presents in individuals with Turner Syndrome, particularly across karyotypes.

Analyses for Research Question One

A one-way ANOVA with *p* set at .05 was the analysis planned in order to identify differences in EF domains (goal setting, sustained attention, selective attention, inhibition, and working memory) between the different TS karyotypes—monosomy X, mosaicism, isochromosome X, ring X chromosome, and deletion of an X chromosome. A Tukey's honestly significant difference (HSD) test was also planned as a post hoc analysis. In order to control for participants who had a diagnosis of ID and BIF, an ANCOVA was also intended to be used with a post hoc analysis using a Tukey's HSD test. Due to the small sample size, there were not enough participants to make up a sufficient group for each karyotype. Therefore, these analyses were not completed.

Analyses for Research Question Two

In order to identify differences in EF domains (goal setting, sustained attention, selective attention, inhibition, and working memory) and age range (children, adolescents, adults) in the different TS karyotypes (monosomy X, mosaicism, isochromosome X, ring X chromosome, and deletion of an X chromosome) a 5 x 4 factorial ANOVA was planned with a *p* set to .05. A post hoc analysis was planned using Tukey's HSD test. In order to control for participants who had a diagnosis of ID and BIF, an ANCOVA was also intended to be used to with a post hoc analysis using a Tukey's HSD test. Due to the small sample size and insufficient participants in both the karyotype and age group, these analyses were not completed.

Exploratory Analysis of the TS Retrospective Sample

Due to a small sample size and the karyotype not being known for a number of participants, this study became an exploratory study that looked for patterns in the various EF domains in this TS retrospective sample. In order to look for EF patterns in the TS retrospective sample, means and standard deviations for this sample were compared to the normative data available for each EF neuropsychological test.

Means and standard deviations for each EF domain (sustained attention, goal setting, selective attention, inhibition, and working memory) and EF measure (Seashore Rhythm Test, Speech Sounds Perception Test, Category Test, TMT Part B, D-KEFS TMT Condition 4, Stroop Color-Word Test, D-KEFS Color Word Interference Test, WISC-IV DSF, WISC-V DSF, WAIS-IV DSF, WISC-IV DSB, WISC-V DSB, WAIS-IV DSB, WISC-IV Integrated SSF, WISC-V Integrated SSF, Knox Cube Test, WISC-IV Integrated SSB, and WISC-V Integrated SSB) were calculated. The means and standard deviations for the working memory indexes on the WISC-IV, WISC-V, and WAIS-IV were also calculated. Not every participant was administered the same neuropsychological tests and measures due to different versions of tests being given based on participants' ages, as well as evaluator preferences and test availability. Therefore, means and standard deviations were calculated for the number of participants who were administered that particular neuropsychological test.

Independent-samples Mann-Whitney U tests were determined to be the best non-parametric analysis to determine if there were significant differences between the TS retrospective raw test scores and means obtained from normative data. During data collection, it was discovered that some of the neuropsychological reports did not contain raw or standard scores, but rather qualitative descriptors (e.g., "below the norm," "severely below average,"

"severely impaired," "within normal limits," and "impaired") with no defined criteria for how scores were qualified. Therefore, a raw score was not available for comparison with the normative data for several of the EF measures. Raw scores were available for the Speech Sounds Perception Test and the Trail Making Test, Part B and were compared to the age-based normative data for each individual measure using an independent-samples Mann-Whitney U test.

Other non-parametric tests used as part of the exploratory analysis included a Kruskal-Wallis H Test to look at the relationship between age and scores on EF behavioral measures. In order to perform these analyses, participants were dived into three groups: (a) children ($N = 9$); (b) adolescents ($N = 8$); and, (c) adults ($N = 3$). Significance testing used a Pairwise Wilcox Test. A Kendall's Tau-B was also employed to examine the relationship between age and performance on EF behavioral measures and the relationship between surgery before the first year of life and score on EF measures.

It is important to note that due to the small sample size in this study, the power is negatively impacted and any conclusions drawn from these analyses should be considered with caution. It is also necessary to consider the impact that performing multiple hypotheses tests had on this research study and the probability that significant results would be found. Due to the high number of analyses performed, given the vast amount of data obtained from this population, there was a higher likelihood that a significant result would be found, and any significant result should be considered with caution.

CHAPTER IV: RESULTS

Demographics

A sample of 20 retrospective, comprehensive neuropsychological reports were collected for participants who had been diagnosed with TS, were between the age of 6 and 40 years old at testing, and had completed said neuropsychological evaluation no earlier than 2008. The sample consisted of 20 (pf) individuals who had been diagnosed with TS. The average age of participants was 12.2 (*SD* = 5.9). The karyotype of participants was unknown for over half of the participants (55%). The remaining participants included a karyotype of monosomy X (15%), mosaicism (25%), and ring X (5%). The sample predominately consisted of White individuals (70%) who lived in the United States (90%). Participants had diagnoses of ADHD (35%), NLD (85%), intellectual disability (10%), and borderline intellectual functioning (15%). Of those diagnosed with ADHD, 57% had an IEP and 14% received 504 accommodations (See Figure 1). Of participants, surgery during the first year of life (33.3%) included aortic repair (28%) and a vitrectomy to treat diabetic retinopathy (5%).

Descriptive Statistics

Table 1 reports the means and standard deviations for the retrospective sample for each EF measure and domain. Scores were given a qualitative descriptor based on the recommendations outlined in Guilmette et al. (2020) and are shown in Figure 2. Qualitative descriptors for the retrospective sample were also given for each EF domain, which are shown in Figure 3. The ASEBA behavioral checklists results are shown in Figure 4 for the attention problems and ADHD problems scales.

Comparison of Scores in the Retrospective vs. Normative Sample on the Speech Sounds Perception Test

Based on the independent-samples Mann-Whitney U test, participants within the retrospective TS sample (*Mdn* = 5.50, *n* = 10) were not found to be significantly different from the normative sample (*Mdn* = 4.60, *n* = 10) on the Speech Sounds Perception Test, *U* = 41.00, *z* = -.68, *p* = .52, with only a small effect size *r* = .15. While the groups were not significantly different, the TS retrospective sample made more errors on the Speech Sound Perception Test (mean rank = 11.40) than the normative sample (mean rank =9.60).

Comparison of Scores in the Retrospective vs. Normative Sample on the Trail Making Test, Part B

The results of the independent-samples Mann-Whitney U test indicated that there was no significant difference between the retrospective TS sample (*Mdn* = 82.00, *n* = 10) and the normative sample (*Mdn* = 49.20, *n* = 10) on the Trail Making Test, Part B, *U* = 28.00, *z* = -1.67, *p* = .10, with a small effect size *r* = .15. The TS retrospective sample performed faster (mean rank = 12.70) than the normative sample (mean rank = 8.30).

Relationship Between Age and ASEBA Behavioral Checklist Scores

A Kruskal-Wallis test was performed on the ASEBA Behavioral Checklist scores and age. The one significant result among the ASEBA Behavioral Checklist Scores was that age was negatively correlated with ADHD Problems reported on the Teacher Report Form, *H* (1, n = 13) = 4.82, *p* = 0.02. A post-hoc Pairwise Wilcoxon test revealed a significant difference between those whose ratings were in the clinically significant range compared to those within normal limits, *p* = 0.04.

Correlation Between ASEBA Behavioral Checklist Scales at Home Vs. at School

A Kendalls Tau-B test was performed on the ASEBA Behavioral Checklists comparing results of symptoms reported at home and the symptoms reported at school. Results are reported in Figure 5 and show a significant correlation between attention problems and ADHD problems at home, $r = 0.83$, $p < .05$.

Correlation Between Age Group and Performance on Executive Functioning Measures

A Kendalls Tau-B test found no significant correlations between age group and performance on executive functioning measures.

Correlation Between Surgery During the First Year of Life and Performance on Executive Functioning Measures

The results of a Kendalls Tau-B test revealed no significant correlations between having a surgery during the first year of life and performance on executive functioning measures.

CHAPTER V: DISCUSSION

The main objective of this study was to raise awareness of the neuropsychological characteristics associated with TS, as the majority of published studies on individuals with TS focus on the biomedical aspects. To do this, the present study investigated EF strengths and deficits in (pf) individuals with TS using a retrospective sample of neuropsychological reports. There is still little-known information on executive functioning in TS, especially as it relates to each individual karyotype. As such, a goal of this study was to clarify, create, and update information on the neurocognitive profiles among the different TS karyotypes.

A number of factors contributed to a change in the overall research design becoming exploratory, including a small sample size and an unknown karyotype for a number of participants. There were an insufficient number of participants represented in each karyotype group. Therefore, this study was unable to assess differences in EF strengths and weaknesses between the different karyotypes, as well as differences in EF in each age group between the different karyotypes as posited in research questions one and two. Despite these challenges, this study was able to examine differences between a TS sample and the general population by comparing mean scores of the retrospective TS sample and the normative population for the Speech Sounds Perception Test and the Trail Making Test. To examine the relationship between EF and age, scores on EF neuropsychological measures and scores on EF self-report measures were compared to age groupings. Finally, an additional analysis was conducted by examining the relationship between surgery during the first year of life and scores on EF measures.

It is important to note the methodological concerns and change in research design before continuing with interpretation of these results. Conclusions made from the results of this study are made with caution due to the small sample size and the statistical probabilities associated

with performing multiple hypotheses tests. The characteristics found in the retrospective TS sample that follow should be considered as exploratory for future research.

The main findings of this study were that the retrospective TS sample had more scores below normal limits in the selective attention domain than any other EF domain. Additionally, there were more reports of attention and ADHD problems reported at home than at school. Finally, teachers reported less ADHD problems as participants aged. When comparing means of the retrospective TS sample with the normative sample on neuropsychological tests, no significant differences were found. There was also no correlation between age group and EF or between surgery during the first year of life and EF.

Selective Attention in the Retrospective TS Sample

One of the aims of this study was to better understand EF strengths and weaknesses in TS. Given the retrospective TS samples' scores in each EF domain, it appears that individuals with TS may have more difficulty in selective attention than in any other EF domain. Problems with focusing while ignoring irrelevant stimuli would mean that individuals with TS would have more difficulties attending to tasks in situations where there are a lot of distractions, movement, or noises. This finding is important as it sheds light on the challenges individuals with TS may have in academic, vocational, and social settings.

The Impact of Emotional Functioning on Selective Attention in Individuals with TS

Individuals with deficits in selective attention can be easily drawn off task by extraneous, irrelevant stimuli, which can include external sights or sounds, as well as internal distractions, such as worry or rumination (Sohlberg & Mateer, 2001). It is important to consider the impact that anxiety and depression may have impacted the scores for the retrospective TS sample. The lifetime incidence of anxiety or depression in TS is as high as 52% (Schmidt et al., 2006).

Assessing for emotional problems, such as depression and anxiety in the retrospective sample was beyond the scope of this study and therefore not explored. However, it is possible that at least some of the participants in the retrospective sample were experiencing emotional problems at the time of testing, thus impacting scores on measures of selective attention. It will be important for individuals with TS to be evaluated, not only for EF weaknesses, but also for psychological complaints, such as depression and anxiety, to determine if weaknesses in selective attention occur independently or concurrently with emotional problems.

Attention and ADHD Problems at Home vs. at School

An interesting discovery from the current study is that teachers reported less problems associated with attention and ADHD at school than parents did at home. This appears to indicate that individuals with TS are exhibiting more problems associated with executive functioning deficits at home, such as difficulty concentrating, than they do at school. When considering the previous results that individuals with TS have more problems with selective attention, it can be inferred that individuals with TS have more difficulty focusing at home because there are more irrelevant stimuli to distract them. This is supported by the idea that children and adolescents with ADHD and attentional weaknesses do better in more structured environments, such as the traditional school setting (Fiese et al., 2002).

A traditional American school setting tends to be a highly structured environment while the home setting is typically less organized and involves more free time. One would expect that deficits in the ability to shift focus from one event to another would be greater and more obvious in the home setting where there is more irrelevant stimuli and less structure so as to know what one should be paying attention to. Further support for this finding is that the retrospective TS sample performed relatively well on measures of sustained attention, the ability to be vigilant

and respond consistently during a continuous activity (Baron, 2018). In other words, when individuals with TS are presented with minimal information and continuous prompting in a structured environment with clear and consistent goals, they may be able to perform better than they would without this structure.

ADHD Problems and Age

A significant correlation was found between age and reported ADHD problems at school. This finding would appear to suggest that as TS individuals age, behaviors associated with ADHD and executive dysfunction decrease in the school setting. In terms of age, it was predicted that individuals with TS would continue to have relative difficulty on measures of EF into adulthood based on relevant literature (Quintero et al., 2014; Romans et al, 1998). It is important to note that the age groups were not evenly distributed, as the adult group only had three participants. Therefore, these results are likely to be the result of a skewed distribution and a small sample of adults with TS.

Implications

Although this study was not able to examine EF as it relates to karyotype, it was successful in expanding the knowledge on the strengths and weaknesses in specific EF domains in individuals with TS. In the past, studies have only looked at overall cognitive functioning in TS or focused primarily on the medical complications associated with this chromosomal disorder. Knowing that selective attention may be a significant area of weaknesses in this population allows for future research to continue to examine this EF domain. This study additionally encourages clinicians to fully assess EF by examining all EF domains in order to obtain an accurate picture of the EF strengths and weaknesses in an individual with TS.

The literature thus far has not extensively explored attention as an EF weakness in TS. Individuals with TS and selective attention weaknesses would be distracted by irrelevant stimuli, such as noises or movement from peers sitting near them in the classroom or office, what may be occurring outside of a nearby window, or by their own thoughts or associations causing them to miss important information that is being relayed to them in the classroom, in a meeting, or at home (Sohlberg & Mateer, 2001). Students may miss important information being relayed to them about new concepts or instructions for how to complete an assignment. Further, distractions can interfere with deepening of understanding and making connections between already learned concepts. Problems with selective attention also impact social functioning, such as difficulty holding a conversation without getting distracted (Sohlberg & Mateer, 2001). Further, activities of daily living may also be impacted, such as difficulties with grocery shopping or preparing a meal due to children playing in the background.

Interventions for Weaknesses in Selective Attention

As the goal of this study was to raise awareness of the psychological issues pertaining to TS, it is also important to provide psychoeducation to individuals with TS, their families, their educators, and their providers. Knowledge of the cognitive strengths and weaknesses typical in TS offers opportunities to learn how to use strengths and overcome weaknesses in order to develop mastery, empower, and build a positive self-concept, thus lowering the risk of emotional problems. The following recommendations are encouraged to begin as early as possible, as early interventions have confirmed improved outcome sin children with developmental disorders (Gravholt et al., 2017).

This study sheds light on the learning difficulties that may be present in individuals with TS. With 35% of the retrospective TS sample having a diagnosis of ADHD and 85% having a

diagnosis of NLD, it will be important for individuals with TS, their families, their educators, and their providers to know about how learning differences can be present in TS and potentially impact academic success. Learning to be a good advocate for oneself or a family member with TS will be essential when navigating school systems, education law, neuropsychological evaluations, accommodations, and transitions to college.

One thing individuals with TS and their families can advocate for is to ask for a comprehensive neuropsychological evaluation that includes EF tests and measures in order to determine EF strengths and weaknesses. One should not assume that every comprehensive neuropsychological test battery is the same. In fact, one of the retrospective neuropsychological reports obtained for this study did not include any measures of executive functioning at all. A comprehensive neuropsychological evaluation should generally include tests that measure language, visual-spatial, attention, executive functioning, learning and memory, processing speed, motor, and sensory-perceptual capacities (Baron, 2018). Given the still developing research on the executive functioning profile in TS, neuropsychological evaluations should examine all aspects of EF, including the following domains: inhibition, selective attention, sustained attention, set-shifting, problem-solving, and working memory. Further, it is recommended that a behavioral checklist rating scale be completed by the individual with TS, a parent/guardian, and/or a teacher, which can provide insight into some behaviors associated with attentional weaknesses and executive dysfunction.

This study highlighted differences in executive functioning behaviors at home and at school. It is critically important that effective interventions are implemented across school, home, and community settings to reduce functional impairment related to symptoms related to ADHD and attentional weaknesses. Parent psychoeducation is often recommended as an early

intervention and an effective way to bridge interventions between home and school environments. Dahl et al. (2020) found that psychoeducation as an intervention led to improvement in ADHD symptoms and behavioral problems. Researchers have posited that improvement may be due to parents' greater knowledge about the ways ADHD influences their child's behavior and better adherence to treatment recommendations (Dahl et al., 2020).

An important part of managing weaknesses in selective attention and symptoms of ADHD is to use environmental modifications (Betker, 2017). Because deficits in selective attention make it difficult to attend to the correct thing, it will be important to reduce the number of extra stimuli an individual might see or hear when they are trying to focus on a particular task. For example, during homework time, the house should be as quiet as possible. It will be necessary for the TV/radio to be off or in a different room with the person watching/listening using headphones so there is no sound. The individual with attentional weaknesses may also benefit from noise cancelling headphones or ear defenders to minimize external noise. The individual should be seated away from windows with open curtains where they can be distracted by what is occurring outside. Homework should also be done in a low-traffic area of the house. Similarly, these principles should apply in the school and work setting (Betker, 2017).

Structure and routine will make it easier for individuals with ADHD and attentional weaknesses to attend to necessary tasks, including homework and chores. This can be done by teaching children how to use checklists for daily chores, steps for getting ready for bed or school, and items needed for school and extracurricular activities (Betker, 2017; Evans et al., 2014). Providing frequent verbal and nonverbal cueing (e.g., visual schedules, pointing to the work/task) to remain on task will also be beneficial (Betker, 2017).

Behavior management techniques, such as the use of reinforcement, can help teach children and adolescents appropriate behaviors (Evans et al., 2018; Zwi et al., 2011). This is done by giving praise and rewards when rules are followed rather than criticizing bad behavior and ignoring behaviors that are not dangerous. Additionally, making eye contact or gently touching the arm or shoulder of the child or adolescent to get their attention can ensure that they are listening. This should be followed by giving brief, simple steps and short commands that are straight to the point. Multiple directions or wordy statement and questions should be avoided. Training for parents in behavior management can be beneficial and have a positive effect on the behavior of children with ADHD. It may also reduce parental stress and enhance parental confidence (Evans et al., 2014; Zwi et al., 2011).

It should be noted that the above recommendations are based on a general ADHD population. It is unclear at this time if individuals with TS would benefit in the same way. Future research should evaluate whether these specific recommendations are also beneficial for individuals diagnosed with TS and co-morbid ADHD.

Recommendations for Conducting Neuropsychological Evaluations of Individuals with TS

There currently is no established neuropsychological test battery available for the TS population. Neuropsychological testing amongst clinicians can look different due to the different approaches of neuropsychological assessment (i.e, level of performance, pathognomonic signs, pattern of performance) and testing batteries (e.g., fixed vs. flexible), as well as the many different neuropsychological tests and measures which are often selected based on clinician preference and availability. Despite these differences, a comprehensive neuropsychological evaluation should generally include tests that measure language, visual-spatial, attention, executive functioning, learning and memory, and processing speed, as well as motor and

sensory-perceptual capacities as needed (Baron, 2018). Given the still developing research on the executive functioning profile in TS, neuropsychological evaluations should examine all aspects of EF, including the following domains: inhibition, selective attention, sustained attention, set-shifting, problem-solving, and working memory.

As cognitive inhibition has been found to be more impaired than other inhibitory control abilities, such as selective or focused attention and response inhibition (Mauger et al., 2018), it will be especially important to include in a neuropsychological test battery. To measure the domain of inhibition, a form of Stroop test is recommended, either the Stroop Color and Word Test (Golden et al., 2002) or the D-KEFS Color Word Interference Test (Delis et al., 2001). The Delis-Kaplan Executive Function System (D-KEFS) Trail Making Test (Delis et al., 2001) and Trail Making Test are recommended to be used as a measure of selective attention. Sustained Attention is best measured by a continuous performance test, such as the Conner's Continuous Performance Test, Third Edition (CPT 3) which evaluates concentration, sustained vigilance, and attention for a simple task over a time interval (Baron, 2018). The Wisconsin Card Sorting Test (Reitan & Davison, 1974) has been found to be strongly related to set-shifting (Miyake et al., 2000). The Halstead Category Test (Reitan & Wolfson, 1985) is recommended to assess problem-solving. To measure verbal working memory, the digit span tests from the WISC-V (Wechsler, 2014) and the WAIS-IV (Wechsler, 2008) can be used to gauge how much information can be held after a single presentation. For individuals ages 6 through 16, a measure of visual working memory can be obtained using the WISC-IV Integrated Spatial Span subtest (Wechsler, 2015).

Finally, it is recommended that a behavioral checklist rating scale be completed by the individual with TS, a parent/guardian, and/or a teacher, which can provide insight into some

behaviors associated with attentional weaknesses and executive dysfunction. A behavioral assessment of executive function using the Behavior Rating Inventory of Executive Function (BRIEF; Gioia et al., 2000) is recommended. Further, either a clinical interview or standardized measures of psychological functioning is recommended to asses for symptoms of anxiety and depression that may be impacting cognitive functioning. Future research will want to continue to determine appropriateness of a neuropsychological test battery on the TS population.

Limitations

A major limitation of the present study was the small sample size and missing karyotype data in the retrospectively completed neuropsychological reports. During the course of this study, this researcher has found the TS research population to often be very helpful and willing to share information. Reaching out to TS clinics across the United States could have allowed for access to a number of different data sets and retrospective neuropsychological reports, which would have expanded this sample size and possibly allowed for a normal distribution of scores across karyotype and age.

A second limitation in the study was that some test performance data gathered from retrospective tests could not be used due to narrative differences on neuropsychological reports, especially related to observations of test performance over time. Some of the retrospective neuropsychological reports dated back to 2008 when test scores were reported differently from current recommendations. For example, qualitative descriptors were used, such as “significantly impaired” or “significantly below the mean,” rather than reporting raw or standard scores. Until recently, no universally accepted system had existed for assigning qualitative descriptors to scores in specific ranges, which invariably resulted in inconsistencies in the use of test score labels and in the use of the term “impairment” (Guilmette et al., 2020).

To solve the problem of inconsistent labeling of test scores across clinicians, the American Academy of Clinical Neuropsychology (AACN) now recommends the application of uniform labels for test performance. In 2018, a consensus conference was held, and a 7-category model derived from the Wechsler system was adopted (Guilmette et al., 2020). It was important that the labels were free from terms that appeared judgmental, biased, or could be viewed as representing a clinical conclusion; instead, the current recommendation is for the examiner to reflect only a score position within the normal distribution. The consensus conference has recommended the following labels: exceptionally high score (>130); above average score (120-129); high average score (110-119); average score (90-109); low average score (80-89); below average score (70-79); and exceptionally low score (<70). The AACN has also recommended that clinicians specify the normative group and any demographic adjustments used for standard score determination (e.g., adjusted for sex, age, education, etc.). Lastly, it is recommended that clinicians include a table or graph within reports explicitly identifying how standard scores are labeled (see Figire 2; Guilmette et al., 2020).

Lastly, the retrospective sample was predominantly White and from the United States, which suggests the current sample may not represent EF differences observed in a more racially or internationally diverse sample. By contrast, the sample was quite diverse in terms parents' education, which may reflect diversity in socioeconomic status. However, demographic information related to socioeconomic status was not available in the present study and limits any claims that can be made.

Directions for Future Research

Methodological differences across studies make it difficult to determine the EF profile of individuals with TS. Many studies, including the present one, use different neuropsychological

tests and measures. In addition, standard neuropsychological measures come with their own limitations, as multiple constructs are often measured simultaneously. Executive tests typically involve different types of EFs, and it is often difficult to determine which executive process contributes most to task achievement. Furthermore, executive tasks are not pure measures of a single skill, such that an individuals' executive performance can be contaminated by non-executive skills required of the task (Burgess, 1997). Additionally, some EF tasks require that the individual provide a response as quickly as possible. The relationship between processing speed and EF is controversial and remains to be specified (Lee et al., 2013). Although decreased processing speed has been described in individuals with TS (Mazzocco, 2006), it remains unclear if poor performance in executive timed tasks reflect specific EF difficulties or if the results have been impacted by deficits in processing speed. With improved knowledge of EF profiles for individuals with TS, future researchers will be aided in their exploration of individual EF difficulties across several tasks requiring organization.

Finally, future research has the opportunity to use more inclusive language when discussing this population. While exploring gender identity in TS was not an area of focus for this dissertation, it was observed that much of the literature and resources refer to individuals with TS as "girls" and/or "woman" and use the gender pronouns "she/her." Using more gender-inclusive language, such as "phenotypically female" and "individuals with TS," shows appreciation for the diversity of the individuals being studied in psychological research, and is recommended per the APA inclusive language guidelines (American Psychological Association, 2021). It further allows individuals with TS who may not identify with a female gender identity to feel like the research is relevant to them. It will also be important, however, to ask individuals with TS how they wish to be addressed as a group (individuals with TS vs a TS individual).

Conclusion

There is little known about the strengths and weaknesses of EF domains in individuals with TS, especially as it relates to karyotype. Given the impact that EF deficits can have on social, emotional, academic, and vocational functioning, it is imperative that we consider implications on the TS population in order to promote and inform earlier interventions. Educating individuals with TS about how use their strengths and overcome their weaknesses will help to develop mastery, empowerment, and build a positive self-concept. This study was met with methodological challenges related to using retrospectively completed neuropsychological reports with an unknown karyotype, as well as qualitative descriptors. Clinicians are encouraged to use the uniform labels for test performance as set out by the AACN (Guilmette et al., 2020). Future research is encouraged to continue to explore what EF domains present as strengths and weaknesses in the TS population, what EF interventions are most beneficial for the TS population, and what neuropsychological EF tests and measures are most appropriate for the TS population.

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APPENDIX A: PERMISSION FORM

Maple Leaf Clinic

[redacted]

[redacted]

March 1, 2021

Antioch University New England

[redacted]

[redacted]

Please note that Sara Scull, AUNE Graduate Student, has the permission of Maple Leaf Clinic to conduct research at our Wallingford, VT location for her study, "Executive Functioning Profiles of Individuals with Turner Syndrome: Exploring the Different Karyotypes and Further Implications for Early Intervention."

Sara Scull's study advertisement for participants will be sent out to clients participating in Turner Syndrome support groups through the Maple Leaf Clinic, as well as on the Maple Leaf Clinic website and Facebook page. She will also be given access to client's neuropsychological test measure protocols from testing that was previously completed by Maple Leaf Clinic. Clients of Maple Leaf Clinic sign an informed consent notifying them that their test data may be used for research at a later date. She will record raw and standard scores from test measures into her own

password protected data collection spreadsheet on her own password protected computer. Sara Scull's on-site research activities will be finished by September 15, 2021.

Sara Scull has agreed to follow all COVID protocols while she is inside the facility and on Maple Leaf Clinic property. Sara Scull has also agreed to provide to my office a copy of the Antioch University IRB-approved stamped consent document before she accesses the data set or participants and will also provide a copy of any aggregate results.

If there are any questions, please contact my office at [redacted].

Signed,

[redacted]

Dean J. M. Mooney, PhD, NCSP

Director – Maple Leaf Clinic

APPENDIX B: UPDATED PERMISSION FORM

Maple Leaf Clinic

[redacted]

[redacted]

March 1, 2022

Antioch University New England

[redacted]

[redacted]

Please note that Sara Scull, AUNE Graduate Student, has the permission of Maple Leaf Clinic to conduct research at our Wallingford, VT location for her study, "Executive Functioning Profiles of Individuals with Turner Syndrome: Exploring the Different Karyotypes and Further Implications for Early Intervention."

Sara Scull's study advertisement for participants will be sent out to clients participating in Turner Syndrome support groups through the Maple Leaf Clinic, as well as on the Maple Leaf Clinic website and Facebook page. She will also be given access to client's neuropsychological test measure protocols from testing that was previously completed by Maple Leaf Clinic. Clients of Maple Leaf Clinic sign an informed consent notifying them that their test data may be used for research at a later date. She will record raw and standard scores from test measures into her own password protected data collection spreadsheet on her own password protected computer. Sara

Scull's on-site research activities have been extended from September 15, 2021 to April 30, 2022.

Sara Scull has agreed to follow all COVID protocols while she is inside the facility and on Maple Leaf Clinic property. Sara Scull has also agreed to provide to my office a copy of the Antioch University IRB-approved stamped consent document before she accesses the data set or participants and will also provide a copy of any aggregate results.

If there are any questions, please contact my office at [redacted].

Signed,

[redacted]

Dean J. M. Mooney, PhD, NCSP

Director – Maple Leaf Clinic

APPENDIX C: RECRUITMENT FLYER IN ENGLISH

Hi! My name is Sara Scull, MS. I am a fifth-year student in the clinical psychology doctoral program at Antioch University New England. For my dissertation, I am collecting neuropsychological reports from individuals with Turner syndrome. If you, or your child/appointee, has a diagnosis of Turner syndrome, has had a neuropsychological evaluation since 2008, and were between the ages of 6 and 40 years old at the time of the neuropsychological evaluation, then you, or your child/appointee, are eligible to participate in my research. It will involve answering a brief questionnaire and uploading or faxing a copy of a neuropsychological report to me. If you would like to participate follow this link https://forms.gle/Uprf7cgszt5RqvDs5 to begin! If you have any questions, please email me, Sara, at [redacted]. Thank you!

APPENDIX D: RECRUITMENT FLYER IN SPANISH – LATIN AMERICAN

¡Hola! Mi nombre es Sara Scull, MS. Estoy estudiando el quinto año del programa de doctorado de psicología clínica en Antioch University New England. Para mi disertación, estoy recabando informes neuropsicológicos de personas con síndrome de Turner. Si usted, o su hijo/persona designada, tiene un diagnóstico de síndrome de Turner, ha sido objeto de una evaluación neuropsicológica desde 2008, y tenía entre 6 y 40 años de edad en el momento de dicha evaluación, entonces usted, o su hijo/persona designada, son elegibles para participar en mi investigación. Consistirá en responder a un breve cuestionario y subir o enviar por fax una copia de un informe neuropsicológico. Si desea participar, ¡siga este enlace https://forms.gle/Uprf7cgszt5RqvDs5 para comenzar! Si tiene alguna pregunta, por favor envíeme un correo electrónico a [redacted]. ¡Gracias!

APPENDIX E: CONSENT FORM FOR PARTICIPANTS 18+ IN ENGLISH

Project Title: Executive Functioning Profiles of Individuals with Turner Syndrome: Exploring the Different Karyotypes and Further Implications for Early Intervention

Project Investigator: Sara Scull, MS

Dissertation Chair: Monique Bowen, PhD

1) This study is of a research nature. It may offer no direct benefit to you.
2) Participation in this study is voluntary. Some people in other studies have reported feeling forced to participate because their doctor has asked them to. You may refuse to be in this study. You may withdraw at any time. There will be no harmful consequences to you. Dr. Dean Mooney will not be told whether or not you participate in this study. Dr. Dean Mooney will not get paid if you participate in this study.
3) You can be removed from the study at any time. This could happen if:
    - You no longer meet the criteria to participate
4) The purpose of this study is to learn about how the brain works in Turner syndrome. Areas looked at will include:

    - how the brain solves problems.
    - how the brain pays attention
    - how impulsive is the brain
    - how the brain organizes information
    - how the brain makes plans

5) As a participant in the study, you will be asked to:

● Fill out a form. The form will include information about you with Turner syndrome. This will include age, gender, education, any tutoring or extra help being received in school, and medical history.

● Share a copy of a report from when you were tested. Participation in the study will take 30 minutes of your time.

6) All of your information will be kept on a flash drive that has a password. Only the researcher will have the password. The flash drive will be kept in a locked file cabinet at Maple Leaf Clinic, [redacted]. Only this researcher will have access to the file cabinet.

7) The risks, discomforts and inconveniences of the above procedures might be:

- No study is completely risk-free. But we do not think that you will be harmed or upset during this study. The internet programs used in this study are secure but may be hacked by an outside party. If your information is stolen, someone other than this researcher may see your information, such as your name, age, and diagnosis. You may stop being in the study at any time if you become uncomfortable.

8) The possible benefits of the study might be:

- Being in this study may not help you. You may learn new strategies or interventions to help you at home, school, or work. Information from this study might help others with Turner syndrome in the future. Learning difficulties may be identified earlier in individuals with Turner syndrome. Accommodations and strategies to learning difficulties may be better tailored to individuals. Accommodations and strategies may be taught at an earlier age. Learning strategies early in school may improve school success.

9) In any written reports or publications, no one will be able to identify you. Information may be used for future research without asking you. This will not include your name or other personal information.
10) If you have any questions, you can call the researcher, Sara Scull, MS, at [redacted] or email at [redacted].

The purpose of this study is for Sara Scull, MS to fulfill requirements to complete a dissertation at Antioch University. However, she also intends to include the data and results of this study in scholarly publications and presentations in the future. In these future publications, no one will be able to identify you. This will not include your name or other personal information.

If you have any questions about the study, you may contact Sara Scull, MS, at telephone # [redacted] or via email at [redacted].

If you have any questions about your rights as a research participant, you may contact Dr. Kevin Lyness, Chair of the Antioch University New England Human Research Committee, [redacted] or Dr. Shawn Fitzgerald, Antioch University New England Provost, [redacted].

Printed Name of Participant _____

Electronic Signature of Participant_____

Date_____

APPENDIX F: CONSENT FORM FOR PARTICIPANTS 18+ IN SPANISH – LATIN AMERICAN

Título del proyecto: Perfiles de funcionamiento ejecutivo en personas con síndrome de Turner: Exploración de diferentes cariotipos y otras consecuencias para la intervención temprana

Investigador del proyecto: Sara Scull, MS

Cátedra de Tesis: Monique Bowen, PhD

1) Este estudio tiene por objeto la investigación. Puede que no le ofrezca ningún beneficio directo.

2) La participación en este estudio es voluntaria. Algunas personas de otros estudios dijeron sentirse obligadas a participar porque su médico se lo pidió. Puede negarse a formar parte de este estudio. Puede retirarse en cualquier momento. No habrá consecuencias perjudiciales para usted. No se le dará aviso al Dr. Dean Mooney sobre si usted participa o no en este estudio. El Dr. Dean Mooney no cobrará si usted participa en este estudio.

3) Puede ser retirado del estudio en cualquier momento. Esto podría ocurrir si:

- Ya no cumple con los criterios de participación

4) El objetivo de este estudio es conocer cómo funciona el cerebro en el síndrome de Turner. Las áreas examinadas incluyen:

- cómo el cerebro resuelve los problemas.
- cómo el cerebro presta atención
- qué tan impulsivo es el cerebro
- cómo el cerebro organiza la información
- cómo el cerebro hace planes

5) Como participante en el estudio, usted:

● Rellene un formulario. El formulario incluirá información sobre usted con el síndrome de Turner. Esto incluirá la edad, el sexo, la educación, cualquier tutoría o ayuda adicional recibida en la escuela, y la historia clínica.

● Compartirá una copia de un informe de cuando se examinó. La participación en el estudio le tomará 30 minutos de su tiempo.

6) Toda su información se almacenará en una unidad flash protegida por contraseña. Solo el investigador tendrá la contraseña. La unidad flash se guardará en un archivador cerrado bajo llave en la Clínica Maple Leaf, [redacted]. Solo este investigador tendrá acceso al archivo.

7) Los riesgos, inconvenientes y desventajas de los procedimientos anteriores podrían ser:

• Ningún estudio está completamente libre de riesgos. Pero no creemos que se vea perjudicado o incómodo durante este estudio. Los programas de Internet utilizados en este estudio son seguros, pero pueden ser pirateados por un tercero. Si su información es robada, alguien que no sea este investigador podría ver dicha información, como su nombre, edad y diagnóstico. Puede dejar de participar en el estudio en cualquier momento si se siente incómodo.

8) Los posibles beneficios del estudio podrían ser:

• Formar parte de este estudio puede no ayudarlo. Puede aprender nuevas estrategias o intervenciones que le ayuden en casa, en la escuela o en el trabajo. La información originada de este estudio puede ayudar a otras personas con síndrome de Turner en el futuro. Los problemas de aprendizaje pueden identificarse con antelación en personas con síndrome de Turner. Las adecuaciones y estrategias para los problemas de aprendizaje pueden adaptarse mejor a las personas. Las adecuaciones y estrategias pueden enseñarse a una edad más temprana. El aprendizaje de estrategias al principio de la escuela puede mejorar el éxito académico.

9) En cualquier informe o publicación escrita, nadie podrá identificarlo. La información puede utilizarse para futuras investigaciones sin que usted lo pida. Estas no incluirán su nombre u otra información de identificación personal.

10) Si tiene alguna pregunta, comuníquese por teléfono con la investigadora, Sara Scull, MS, al [redacted] o envíe un correo electrónico a [redacted].

El propósito de este estudio es que Sara Scull, MS, cumpla con los requisitos para completar una disertación en la Universidad de Antioquía. Sin embargo, también tiene la intención de incluir los datos y resultados de este estudio en futuras publicaciones y presentaciones académicas. En estas futuras publicaciones, nadie podrá identificarlo. Estas no incluirán su nombre u otra información de identificación personal.

Si tiene alguna pregunta sobre el estudio, comuníquese por teléfono con Sara Scull, MS, al [redacted] o por correo electrónico en [redacted].

Si tiene preguntas sobre sus derechos como participante en la investigación, puede ponerse en contacto telefónico con el Dr. Kevin Lyness, Presidente del Comité de Investigación Humana de la Universidad de Antioquía de Nueva Inglaterra, al [redacted], [redacted] o con el Dr. Shawn Fitzgerald, Rector de la Universidad de Antioquía de Nueva Inglaterra, al [redacted].

Nombre en letra imprenta del participante _____

Firma electrónica del participante _____

Fecha_____

APPENDIX G: GUARDIAN CONSENT FORM FOR PARTICIPANTS BELOW 18 YEARS OF AGE IN ENGLISH

Project Title: Executive Functioning Profiles of Individuals with Turner Syndrome: Exploring the Different Karyotypes and Further Implications for Early Intervention

Project Investigator: Sara Scull, MS

Dissertation Chair: Monique Bowen, PhD

1) This study is of a research nature. It may offer no direct benefit to your child/appointee.

2) Participation in this study is voluntary. Some people in other studies have reported feeling forced to participate because their doctor has asked them to. Your child/appointee may refuse to be in this study. Your child/appointee may withdraw at any time. There will be no harmful consequences to your child/appointee. Dr. Dean Mooney will not be told whether or not your child/appointee participates in this study. Dr. Dean Mooney will not get paid for participation in this study.

3) Your child/appointee can be removed from the study at any time. This could happen if:

- Your child/appointee no longer meet the criteria to participate

4) The purpose of this study is to learn about how the brain works in Turner syndrome. Areas looked at will include:

- how the brain solves problems.
- how the brain pays attention
- how impulsive is the brain
- how the brain organizes information
- how the brain makes plans

5) If your child/appointee is a participant in the study, you will be asked to:

● Fill out a form. The form will include information about your child/appointee with Turner syndrome. This will include their age, gender, education, any tutoring or extra help being received in school, and medical history.

● Share a copy of a report from when your child/appointee was tested. Participation in the study will take 30 minutes of your time.

6) All of your child/appointee's information will be kept on a flash drive that has a password. Only the researcher will have the password. The flash drive will be kept in a locked file cabinet at Maple Leaf Clinic, [redacted]. Only this researcher will have access to the file cabinet.

7) The risks, discomforts and inconveniences of the above procedures might be:

• No study is completely risk-free. But we do not think that your child/appointee will be harmed or upset during this study. The internet programs used in this study are secure but may be hacked by an outside party. If your child/appointee's information is stolen, someone other than this researcher may see their information, such as their name, age, and diagnosis. Your child/appointee may stop being in the study at any time if you become uncomfortable.

8) The possible benefits of the study might be:

• Being in this study may not help your child/appointee. You may learn new strategies or interventions to help your child/appointee at home, school, or work. Information from this study might also help others with Turner syndrome in the future. Learning difficulties may be identified earlier in individuals with Turner syndrome. Accommodations for learning difficulties may be better tailored to individuals with Turner syndrome. Accommodations and strategies may be taught at an earlier age. Learning strategies early in school may improve school success.

9) In any written reports or publications, no one will be able to identify your child/appointee. Information may be used for future research without asking you. This will not include your child/appointee's name, age, or other personal information.

10) If you have any questions, you can call the researcher, Sara Scull, MS, at [redacted] or email at [redacted].

The purpose of this study is for Sara Scull, MS to fulfill requirements to complete a dissertation at Antioch University. However, she also intends to include the data and results of this study in scholarly publications and presentations in the future. In these future publications, no one will be able to identify you. This will not include your name or other personal information.

If you have any questions about the study, you may contact Sara Scull, MS, at telephone [redacted] or via email at [redacted].

If you have any questions about your rights as a research participant, you may contact Dr. Kevin Lyness, Chair of the Antioch University New England Human Research Committee, [redacted], [redacted] or Dr. Shawn Fitzgerald, Antioch University New England Provost, [redacted].

Printed Name of Parent/Guardian _____

Electronic Signature of Parent/Guardian_____

Date_____

Who is signing this form? Parent, Guardian

Printed Name of Participant _____

Electronic Signature of Participant________________________________

Date____

APPENDIX H: GUARDIAN CONSENT FORM FOR PARTICIPANTS BELOW 18 YEARS OF AGE IN SPANISH – LATIN AMERICAN

Título del proyecto: Perfiles de funcionamiento ejecutivo en personas con síndrome de Turner: Exploración de diferentes cariotipos y otras consecuencias para la intervención temprana

Investigador del proyecto: Sara Scull, MS

Cátedra de Tesis: Monique Bowen, PhD

1) Este estudio tiene por objeto la investigación. Puede que no ofrezca ningún beneficio directo a su hijo/persona designada.

2) La participación en este estudio es voluntaria. Algunas personas de otros estudios dijeron sentirse obligadas a participar porque su médico se lo pidió. Su hijo o persona designada puede negarse a participar en este estudio. Su hijo o persona designada puede retirarse en cualquier momento. No habrá consecuencias perjudiciales para su hijo/persona designada. No se le dará aviso al Dr. Dean Mooney sobre si su hijo/persona designada participa o no en este estudio. El Dr. Dean Mooney no recibirá ninguna remuneración por su participación en este estudio.

3) Su hijo/persona designada puede ser retirado del estudio en cualquier momento. Esto podría ocurrir si:

- Su hijo o persona designada ya no cumple los criterios de participación

4) El objetivo de este estudio es conocer cómo funciona el cerebro en el síndrome de Turner. Las áreas examinadas incluyen:

- cómo el cerebro resuelve los problemas.
- cómo el cerebro presta atención
- qué tan impulsivo es el cerebro
- cómo el cerebro organiza la información

- cómo el cerebro hace planes

5) Si su hijo/persona designada participa en el estudio, se le pedirá que:

● Rellene un formulario. El formulario incluirá información sobre su hijo/persona designada con síndrome de Turner. Esto incluirá su edad, sexo, educación, cualquier tutoría o ayuda adicional que haya recibido en la escuela y su historial médico.

● Comparta una copia de un informe de cuando su hijo/persona designada fue examinado. La participación en el estudio le tomará 30 minutos de su tiempo.

6) Toda la información de su hijo/persona designada se almacenará en una unidad flash protegida por contraseña. Solo el investigador tendrá la contraseña. La unidad flash se guardará en un archivador cerrado bajo llave en la Clínica Maple Leaf [redacted]. Solo este investigador tendrá acceso al archivo.

7) Los riesgos, inconvenientes y desventajas de los procedimientos anteriores podrían ser:

• Ningún estudio está completamente libre de riesgos. Pero no creemos que su hijo/persona designada se vea perjudicado o incómodo durante este estudio. Los programas de Internet utilizados en este estudio son seguros, pero pueden ser pirateados por un tercero. Si la información de su hijo/persona designada es robada, alguien que no sea este investigador podría ver dicha información, como su nombre, edad y diagnóstico. Su hijo/persona designada puede dejar de participar en el estudio en cualquier momento si se siente incómodo.

8) Los posibles beneficios del estudio podrían ser:

• Formar parte de este estudio puede no ayudar a su hijo/persona designada. Puede aprender nuevas estrategias o intervenciones que ayuden a su hijo/persona designada en casa, en la escuela o en el trabajo. La información de este estudio también puede ayudar a otras personas con síndrome de Turner en el futuro. Los problemas de aprendizaje pueden identificarse con

antelación en personas con síndrome de Turner. Las adaptaciones para los problemas de aprendizaje pueden ser más adecuadas para las personas con síndrome de Turner. Las adecuaciones y estrategias pueden enseñarse a una edad más temprana. El aprendizaje de estrategias al principio de la escuela puede mejorar el éxito académico.

9) En cualquier informe o publicación escrita, nadie podrá identificar a su hijo/persona designada. La información puede utilizarse para futuras investigaciones sin que usted lo pida. Esto no incluirá el nombre, la edad u otra información personal de su hijo/persona designada.

10) Si tiene alguna pregunta, comuníquese por teléfono con la investigadora, Sara Scull, MS, al [redacted] o envíe un correo electrónico a [redacted].

El propósito de este estudio es que Sara Scull, MS, cumpla con los requisitos para completar una disertación en la Universidad de Antioquía. Sin embargo, también tiene la intención de incluir los datos y resultados de este estudio en futuras publicaciones y presentaciones académicas. En estas futuras publicaciones, nadie podrá identificarlo. Estas no incluirán su nombre u otra información de identificación personal.

Si tiene alguna pregunta sobre el estudio, comuníquese por teléfono con Sara Scull, MS, al [redacted] o por correo electrónico en [redacted].

Si tiene preguntas sobre sus derechos como participante en la investigación, puede ponerse en contacto telefónico con el Dr. Kevin Lyness, Presidente del Comité de Investigación Humana de la Universidad de Antioquía de Nueva Inglaterra, al [redacted], [redacted] o con

el Dr. Shawn Fitzgerald, Rector de la Universidad de Antioquía de Nueva Inglaterra, al [redacted].

Nombre en letra imprenta del padre/tutor _____

Firma electrónica del padre/tutor _____

Fecha_____

¿Quiénes firman este formulario? Padres, tutores

Nombre en letra imprenta del participante _____

Firma electrónica del participante _____

Fecha_____

APPENDIX I: ASSENT FORM IN ENGLISH

Study Title: Executive Functioning Profiles of Individuals with Turner Syndrome: Exploring the Different Karyotypes and Further Implications for Early Intervention

Researcher: Sara Scull, MS

Email Address and Telephone Number: [redacted]; [redacted]

Research Supervisor: Monique Bowen, PhD

Email Address: [redacted]

You are invited to be part of a research study. The researcher is a student at Antioch University New England. This information is to help you decide if you want to participate. This form explains what the study is about. This form also describes the risks and benefits of the study. If you have any questions or do not understand something in this form, you should ask the researcher. Do not sign this form unless the researcher has answered your questions. Do not sign this form unless you decide that you want to be part of this study.

WHAT IS THIS STUDY ABOUT?

The researcher wants to learn about how the brain works in Turner syndrome. Areas looked at will include:

- how the brain solves problems.
- how the brain pays attention
- how impulsive is the brain
- how the brain organizes information
- how the brain makes plans

WHY AM I BEING ASKED TO BE IN THE STUDY?

You are invited to be in the study because you are:

- diagnosed with Turner syndrome
- have had neuropsychological testing

WHAT WILL HAPPEN DURING THIS STUDY?

If you decide to be in this study and sign this form, you do not have to do anything else. Your parent or guardian will do the following things:

- Fill out a form. The form will include information about you. This will include your age, gender, your grade, if you get extra help with school, and medical history.
- Share a copy of a report from when you were tested.

WILL BEING IN THIS STUDY HELP ME?

Being in this study may not help you. At the end of this study, you may learn new skills to help you at school. Information from this study might help others with Turner syndrome in the future.

ARE THERE RISKS TO ME BEING IN THIS STUDY?

No study is completely risk-free. But we do not think that you will be harmed or upset during this study. The online systems being used in this study are secure but may be hacked by an outside party. If your information is stolen, someone other than this researcher may see your information, such as your name, age, and diagnosis. You may stop being in the study at any time if you become uncomfortable. The study takes about 30 minutes to complete.

DO I HAVE TO BE IN THIS STUDY?

Your participation in this study is voluntary. Some people in other studies have reported feeling forced to participate because their doctor has asked them to. You can decide not to be in the study. You can change your mind about being in the study at any time. There will be no penalty to you. If you want to stop being in the study, tell the researcher. Dr. Dean Mooney will not be told whether or not you volunteer to participate in this study. Dr. Dean Mooney will not be paid if you participate in this study.

Your parent(s)/guardian(s) have also said that you may participate in this study.

The researcher can remove you from the study at any time. This could happen if:

- You no longer meet the criteria to participate

HOW WILL MY INFORMATION BE USED?

Your name, age, and other personal information will be kept private. In any written reports or publications, no one will be able to identify you. Information may be used for future research without asking you. This will not include your name or other personal information.

The researcher will keep your information on a flash drive that has a password. Only this research will have the password. The flash drive will be locked in a file cabinet. The file cabinet will be locked at all times at Maple Leaf Clinic, [redacted].

FUTURE PUBLICATION

Information may be used for future research without asking you. This will not include your name or other personal information.

RIGHT TO REFUSE OR WITHDRAW

You do not have to be in this study if you do not want to. You may stop being in this study at any time. You will not be punished if you do not want to be in this study.

WHO TO CONTACT

If you have questions, you may contact Sara Scull, MS at telephone [redacted] or email at [redacted].

If you have any questions about your rights as a research participant, you may contact Dr. Kevin Lyness, Chair of the Antioch University New England Human Research Committee, [redacted], [redacted] or Dr. Shawn Fitzgerald, Antioch University New England Provost, [redacted].

DO YOU WANT TO BE IN THIS STUDY?

I have read this form. I have been given information about this study. I have had my questions answered. I voluntarily agree to be in this study. I agree to allow my information to be used as described above.

By signing this form, I have not given up any of my legal rights as a research participant. I will get a signed copy of this consent form for my records.

Printed Name of Participant _____

Electronic Signature of Participant_____

Date_____

APPENDIX J: ASSESNT FORM IN SPANISH – LATIN AMERICAN

Título del estudio: Perfiles de funcionamiento ejecutivo en personas con síndrome de Turner: Exploración de diferentes cariotipos y otras consecuencias para la intervención temprana

Investigador: Sara Scull, MS

Dirección de correo electrónico y número de teléfono: [redacted]; [redacted]

Supervisor de la investigación: Monique Bowen, PhD

Dirección de correo electrónico: [redacted]

Se lo invita a participar en un estudio de investigación. La investigadora es estudiante de la Universidad de Antioquía de Nueva Inglaterra. Esta información es para ayudarle a decidir si quiere participar. Este módulo explica en qué consiste el estudio. Este formulario también describe los riesgos y beneficios del estudio.

Si tiene alguna duda o no entiende alguna parte de este formulario, debe consultarlo con el investigador. No firme este formulario si el investigador no ha respondido a sus preguntas. No firme este formulario a menos que decida que quiere formar parte de este estudio.

¿DE QUÉ TRATA ESTE ESTUDIO?

El investigador quiere comprender cómo funciona el cerebro en el síndrome de Turner. Las áreas examinadas incluyen:

- cómo el cerebro resuelve los problemas.
- cómo el cerebro presta atención
- qué tan impulsivo es el cerebro
- cómo el cerebro organiza la información
- cómo el cerebro hace planes

¿POR QUÉ SE ME PIDE QUE PARTICIPE EN EL ESTUDIO?

Se lo invita a participar en el estudio porque usted:

- está diagnosticado con el síndrome de Turner
- fue sometido a pruebas neuropsicológicas

¿QUÉ OCURRIRÁ DURANTE ESTE ESTUDIO?

Si decide participar en este estudio y firmar este formulario, no tiene que hacer nada más. Su padre o tutor deberá ser quien:

- Rellene un formulario. El formulario incluirá información sobre usted. Se incluirá su edad, sexo, grado, si recibe ayuda adicional en la escuela y su historial médico.
- Compartirá una copia de un informe de cuando se examinó.

¿ME AYUDARÁ PARTICIPAR EN ESTE ESTUDIO?

Formar parte de este estudio puede no ayudarlo. Al final de este estudio, podrás aprender nuevas habilidades que te ayudarán en la escuela. La información originada de este estudio puede ayudar a otras personas con síndrome de Turner en el futuro.

¿EXISTEN RIESGOS PARA MÍ EN ESTE ESTUDIO?

Ningún estudio está completamente libre de riesgos. Pero no creemos que se vea perjudicado o incómodo durante este estudio. Los sistemas en línea utilizados en este estudio son seguros, pero pueden ser pirateados por un tercero. Si su información es robada, alguien que no sea este investigador podría ver dicha información, como su nombre, edad y diagnóstico. Puede dejar de participar en el estudio en cualquier momento si se siente incómodo. El estudio tarda unos 30 minutos en completarse.

¿TENGO QUE PARTICIPAR EN ESTE ESTUDIO?

Su participación en este estudio es voluntaria. Algunas personas de otros estudios dijeron sentirse obligadas a participar porque su médico se lo pidió. Puede optar por no participar en el estudio. Puede cambiar de opinión sobre estar en el estudio en cualquier momento. No habrá ninguna penalización para usted. Si quieres dejar de participar en el estudio, comuníqueselo al investigador. No se le dará aviso al Dr. Dean Mooney sobre si usted se ofrece o no como voluntario para participar en este estudio. El Dr. Dean Mooney no cobrará si usted participa en este estudio.

Su padre(s)/tutor(es) también han dicho que puede participar en este estudio.

El investigador puede retirarlo del estudio en cualquier momento. Esto podría ocurrir si:

- Ya no cumple con los criterios de participación

¿CÓMO SE UTILIZARÁ MI INFORMACIÓN?

Su nombre, edad y otros datos de identificación personal se mantendrán en secreto. En cualquier informe o publicación escrita, nadie podrá identificarlo. La información puede utilizarse para futuras investigaciones sin que usted lo pida. Estas no incluirán su nombre u otra información de identificación personal.

El investigador almacenará su información en una unidad flash protegida por contraseña. Esta investigación será la única con acceso a la contraseña. El pendrive se guardará en un archivador. El archivador se ubicará en Maple Leaf Clinic, [redacted], y permanecerá cerrado en todo momento.

FUTURA PUBLICACIÓN

La información puede utilizarse para futuras investigaciones sin que usted lo pida. Estas no incluirán su nombre u otra información de identificación personal.

DERECHO A NEGARSE O RETIRARSE

No tiene que participar en este estudio si no quiere. Puede dejar de participar en este estudio en cualquier momento. No será castigado si no quiere formar parte de este estudio.

A QUIÉNES PUEDE CONTACTAR

Si tiene alguna pregunta, póngase en contacto con Sara Scull, MS por teléfono al [redacted] o por correo electrónico en [redacted].

Si tiene preguntas sobre sus derechos como participante en la investigación, puede ponerse en contacto telefónico con el Dr. Kevin Lyness, Presidente del Comité de Investigación Humana de la Universidad de Antioquía de Nueva Inglaterra, al [redacted], [redacted] o con el Dr. Shawn Fitzgerald, Rector de la Universidad de Antioquía de Nueva Inglaterra, al [redacted].

¿DESEA PARTICIPAR EN ESTE ESTUDIO?

He leído este formulario. Me dieron información sobre este estudio. Mis preguntas han sido respondidas. Acepto voluntariamente participar en este estudio. Estoy de acuerdo en que mis datos se utilicen como se ha descrito anteriormente.

Al firmar este formulario, no he renunciado a ninguno de mis derechos legales como participante en la investigación. Recibiré una copia firmada de este formulario de consentimiento a fin de que quede en mis registros.

Nombre en letra imprenta del participante _____

Firma electrónica del participante _____

Fecha_____

APPENDIX K: GOOGLE FORMS TURNER SYNDROME EXECUTIVE FUNCTIONING QUESTIONNAIRE IN ENGLISH

Instructions: If you are the individual with Turner syndrome and are over the age of 18, please answer each question to the best of your knowledge.

If the individual with Turner syndrome is under the age of 18, this questionnaire is to be filled out by the parent or guardian.

If the individual with Turner syndrome is under guardianship or is a conservatee (has been determined by a court to lack capacity to make some or all personal decisions and for whom a guardian has been appointed), this questionnaire is to be filled out by the appointed guardian.

Prerequisite Questions

1. Which karyotype does the individual with Turner syndrome have?
    a. Monosomy X (45,X)
    b. Isochromosome X [46,X,i(X)]
    c. Ring Chromosome [46,X,r(X)]
    d. Deletion (Xp)
    e. Deletion (Xq)
    f. Mosaicism (45,X/46,XX)
    g. Mosaicism (45,X/46,XY)
    h. Mosaicism (45,X/47,XXX)
    i. Mosaicism (45,X/46,XX/47,XXX)
    j. Unknown (If the karyotype is unknown, then the individual with Turner syndrome is not eligible to participate in this study. Thank You) [Questionnaire will end here]

2. Has the individual with Turner syndrome ever had a neuropsychological evaluation?
    a. Yes
    b. No (If the individual with Turner syndrome has never had a neuropsychological evaluation, then they are not eligible to participate in this study. Thank You) [Questionnaire will end here]
3. If yes, was the neuropsychological evaluation completed after 2007?
    a. Yes
    b. No (If the neuropsychological evaluation was completed before 2008, then the individual with Turner syndrome is not eligible to participate in this study. Thank You) [Questionnaire ends here]
4. If yes, was the individual with Turner syndrome between the ages of 6 and 40 years old when the neuropsychological evaluation was completed?
    a. Yes
    b. No (If the individual was not between the ages of 6–40 years old at the time of the neuropsychological evaluation, then they are not eligible to participate in this study. Thank You) [Questionnaire ends here]

Demographics

5. Who is filling out this form?
    a. Individual with Turner syndrome
    b. Parent of individual with Turner syndrome
    c. Other relative of individual with Turner syndrome
    d. Guardian of individual with Turner syndrome
    e. Other (free response item)

6. What is the current age of the individual with Turner syndrome?
    a. (free response item)
7. What is the racial/ethnic background of the individual with Turner syndrome? If identify as bi- or multi-racial or ethnic, please check all that apply:
    a. Alaska Native
    b. American Indian
    c. Black or African American
    d. East Asian
    e. Hispanic
    f. Latinx
    g. Native Hawaiian
    h. North African
    i. Middle Eastern
    j. Other Pacific Islander
    k. South Asian
    l. White/European Descent
    m. Prefer not to say
    n. Not Known
    o. Other (free response item)
8. What is the gender identity of the individual with Turner syndrome? (Check all that apply)
    a. Female
    b. Male

c. Genderfluid

d. Genderqueer

e. Non-binary

f. Pangender

g. Queer

h. Trans

i. Two-spirit

j. Prefer not to say

k. Not Known

l. Other (free response item)

9. What Country does the individual with Turner syndrome live in?

a. Canada

b. Mexico

c. United States

d. Other (free response item)

10. Which state, province, or territory does the individual with Turner syndrome live in?

a. (free response item)

Education

11. What is the highest level of education of the individual with Turner syndrome?

a. Current grade if still in school (free response item)

b. Less than a high school diploma

c. High school degree or equivalent

d. Bachelor's degree (BA, BS)

e. Master's degree (e.g., MA, MS, Med, etc.)

f. Doctorate (e.g., PhD, EdD, etc.)

g. Other (free response item)

12. What is the highest level of education of the mother of the individual with Turner syndrome?

    a. Less than a high school diploma

    b. High school degree or equivalent

    c. Bachelor's degree (BA, BS)

    d. Master's degree (e.g., MA, MS, Med, etc.)

    e. Doctorate (e.g., PhD, EdD, etc.)

    f. Other (free response item)

13. What is the highest level of education of the father of the individual with Turner syndrome?

    a. Less than a high school diploma

    b. High school degree or equivalent

    c. Bachelor's degree (BA, BS)

    d. Master's degree (e.g., MA, MS, Med, etc.)

    e. Doctorate (e.g., PhD, EdD, etc.)

    f. Other (free response item)

Learning, Educational Evaluation, and Support History

14. Did the individual with Turner syndrome receive special education services?

    a. Yes

    b. No

15. If yes, were they on an Individual Education Plan (IEP)?
    a. Yes (If yes, what grade did this start? what grade did this stop? What did they receive services for?)
    b. No
    c. Not Known
16. Did the individual with Turner syndrome have a 504 plan?
    a. Yes (If yes, what grade did this start? what grade did this stop? What did they receive services for?)
    b. No
    c. Not Known
17. Has the individual with Turner syndrome been diagnosed with a learning disability?
    a. Yes
    b. No
18. If yes, what kind of learning disability have they been diagnosed with? Please check all that apply
    a. Reading Disorder
    b. Dyslexia
    c. Writing Disorder
    d. Math Disorder
    e. Other (free response item)
19. Has the individual with Turner syndrome been diagnosed with Attention-Deficit/Hyperactivity Disorder (ADHD) or Attention Deficit Disorder (ADD)?
    a. Yes (if yes, at what age was the diagnosis given?)

b. No

c. Not Known

20. Did the individual with Turner syndrome ever receive special education services for executive functioning, organizational, planning, problem-solving, or attention problems?

    a. Yes (If yes, please describe what services they received)

    b. No

    c. Not Known

21. Has the individual with Turner syndrome ever received tutoring?

    a. Yes (if yes, was the tutoring for ADHD/ADD or executive functioning, organizational, planning, problem-solving, or attention problems? If yes, what types of services were received? At what age did tutoring start? How long did tutoring occur?)

    b. No

    c. Not Known

22. Has the individual with Turner syndrome ever received life coaching?

    a. Yes (if yes, at what age did life coaching start? How long did life coaching last? Did life coaching include executive functioning, organizational, planning, problem-solving, or attention skills?)

    b. No

    c. Not Known

23. Has the individual with Turner syndrome ever received coaching for executive functioning, organizational, planning, problem-solving, or attention problems?

a. Yes (if yes, at what age did coaching start? How long did coaching last? What type of skills were focused on?)
b. No
c. Not Known

24. Has the individual with Turner syndrome ever received cognitive rehabilitation (cog rehab, REHABIT, cognitive remediation)?
    a. Yes (if yes, at what age did this start? How long did cognitive rehabilitation last? What skills were worked on?)
    b. No
    c. Not Known

Health and Medical History

25. Medical history of the individual with Turner syndrome. Please check all that apply
    a. Alzheimer's disease (if yes, what age diagnosed)
    b. Autoimmune disorder (if yes, what kind; what age diagnosed)
    c. Brain tumor (if yes, what kind; what age diagnosed)
    d. Cancer (if yes, what kind; what age diagnosed)
    e. COPD (if yes, what age diagnosed)
    f. Dementia (if yes, what kind; what age diagnosed)
    g. Diabetes (if yes, what age diagnosed)
    h. Epilepsy (if yes, what age diagnosed)
    i. Heart attack (if yes, what age diagnosed)
    j. High blood pressure (if yes, what age diagnosed)
    k. High cholesterol (if yes, what age diagnosed)

l. Kidney disease (if yes, what age diagnosed)

m. Liver disease (if yes, what age diagnosed)

n. Lyme disease (if yes, what age diagnosed)

o. Meningitis (if yes, what age diagnosed)

p. Obesity (if yes, what age diagnosed)

q. Seizures (if yes, what age diagnosed)

r. Sleep apnea (if yes, what age diagnosed)

s. Thyroid disorder (if yes, what kind; what age diagnosed)

t. Other (free response item; if yes, what age diagnosed)

26. Please list all surgeries that occurred in the first year of life

    a. (free response item)

27. Please list all surgeries that occurred during childhood (2–12 years old)

    a. (free response item)

28. Please list all surgeries that occurred during adolescence (13-17 years old)

    a. (free response item)

29. Please list all surgeries that occurred after the age of 18

    a. (free response item)

30. Has the individual with Turner syndrome received growth hormone treatment?

    a. Yes

    b. No

    c. Not Known

31. If yes, at what age did the individual with Turner syndrome begin to receive growth hormone treatment?

a. (free response item)

32. Has the individual with Turner syndrome received estrogen replacement therapy?

    a. Yes

    b. No

    c. Not Known

33. If yes, at what age did the individual with Turner syndrome begin to receive estrogen replacement therapy?

    a. (free response item)

Thank you for completing this survey!

Appendix L: GOOGLE FORMS TURNER SYNDROME EXECUTIVE FUNCTIONING QUESTIONNAIRE IN SPANISH – LATIN AMERICAN

Instrucciones: si usted es quién padece el síndrome de Turner y tiene más de 18 años, responda a cada una de las preguntas en la medida que se lo permitan sus conocimientos.

Si la persona con síndrome de Turner es menor de 18 años, este cuestionario debe ser respondido por el padre o tutor.

Si la persona con síndrome de Turner está bajo la protección de un curador o es una persona bajo tutela (es decir, un tribunal determinó que no tiene la capacidad para tomar algunas o todas las decisiones personales y le designó un representante legal), este cuestionario debe ser respondido por el representante legal designado.

Preguntas sobre los requisitos previos

1. ¿Qué cariotipo tiene la persona con síndrome de Turner?
    a. Monosomía X (45,X)
    b. Isocromosoma X [46,X,i(X)]
    c. Cromosoma en anillo [46,X,r(X)]
    d. Deleción (Xp)
    e. Deleción (Xq)
    f. Mosaicismo (45,X/46,XX)
    g. Mosaicismo (45,X/46,XY)
    h. Mosaicismo (45,X/47,XXX)
    i. Mosaicismo (45,X/46,XX/47,XXX)
    j. Desconocido (Si el cariotipo es desconocido, la persona con síndrome de Turner no puede participar en este estudio. Gracias)

2. ¿Fue sometida la persona con síndrome de Turner a una evaluación neuropsicológica?
    a. Sí
    b. No (Si la persona con síndrome de Turner nunca fue sometida a una evaluación neuropsicológica, no podrá participar en este estudio. Gracias)
3. En caso afirmativo, ¿la evaluación neuropsicológica fue realizada después de 2007?
    a. Sí
    b. No (Si la evaluación neuropsicológica se realizó antes de 2008, la persona con síndrome de Turner no puede participar en este estudio. Gracias)
4. En caso afirmativo, ¿tenía la persona con síndrome de Turner entre 6 y 40 años de edad cuando se realizó la evaluación neuropsicológica?
    a. Sí
    b. No (Si la persona no tenía entre 6 y 40 años de edad en el momento de la evaluación neuropsicológica, entonces no puede participar en este estudio. Gracias)

Datos demográficos

5. ¿Quién completa este formulario?
    a. Persona con síndrome de Turner
    b. Padre de la persona con síndrome de Turner
    c. Otro familiar de la persona con síndrome de Turner
    d. Representante legal de la persona con síndrome de Turner
    e. Otros

6. ¿Cuál es la edad actual de la persona con síndrome de Turner?

7. ¿Cuál es el origen racial/étnico de la persona con síndrome de Turner? Si se identifica como birracial o multirracial o con algún grupo étnico determinado, marque todo lo que corresponda
    a. Nativo de Alaska
    b. Indígena Americano
    c. Negro o afroamericano
    d. Asia Oriental
    e. Hispano
    f. Latino
    g. Nativo de Hawái
    h. Norte de África
    i. Medio Oriente
    j. Otros isleños del Pacífico
    k. Asia Meridional
    l. Blanco/descendiente europeo
    m. Prefiero no decirlo
    n. No lo sé
    o. Otros
8. ¿Cuál es la identidad de género de la persona con síndrome de Turner? (Marque todo lo que corresponda)
    a. Mujer

b. Hombre

c. Género fluido

d. Genderqueer

e. No binario

f. Pangénero

g. Queer

h. Transgénero

i. Dos espíritus

j. Prefiero no decirlo

k. No lo sé

l. Otros

9. ¿En qué país vive la persona con síndrome de Turner?

    a. Canadá

    b. México

    c. Estados Unidos

    d. Otros

10. ¿En qué estado, provincia o territorio vive la persona con síndrome de Turner?

Educación

¿Cuál es el nivel de estudios más alto de la persona con síndrome de Turner?

a. Grado actual, si todavía está en la escuela

b. Nivel inferior que el diploma de secundaria

c. Diploma de secundaria o equivalente

d. Licenciatura (BA, BS)

e. Grado de Maestría (por ejemplo, MA, MS, Med, etc.)

f. Doctorado (por ejemplo, PhD, EdD, etc.)

g. Otros

11. ¿Cuál es el nivel de estudios más alto de la madre de la persona con síndrome de Turner?

a. Nivel inferior que el diploma de secundaria

b. Diploma de secundaria o equivalente

c. Licenciatura (BA, BS)

d. Grado de Maestría (por ejemplo, MA, MS, Med, etc.)

e. Doctorado (por ejemplo, PhD, EdD, etc.)

f. Otros

12. ¿Cuál es el nivel de estudios más alto del padre de la persona con síndrome de Turner?

a. Nivel inferior que el diploma de secundaria

b. Diploma de secundaria o equivalente

c. Licenciatura (BA, BS)

d. Grado de Maestría (por ejemplo, MA, MS, Med, etc.)

e. Doctorado (por ejemplo, PhD, EdD, etc.)

f. Otros

Aprendizaje, Historial de apoyo académico y evaluación educativa

13. ¿La persona con síndrome de Turner recibió servicios de educación especial?

a. Sí

b. No

14. En caso afirmativo, ¿estaban en un Plan Educativo Individual (PEI)?
    a. Sí (En caso afirmativo, ¿en qué grado empezó? ¿en qué grado lo dejó/terminó? ¿Para qué recibió los servicios?).
    b. No
    c. No lo sé
15. ¿La persona con síndrome de Turner tenía un plan 504?
    a. Sí (En caso afirmativo, ¿en qué grado empezó? ¿En qué grado lo dejó/terminó? ¿Para qué recibió los servicios?).
    b. No
    c. No lo sé
16. ¿Se le diagnosticó a la persona con síndrome de Turner un problema de aprendizaje?
    a. Sí
    b. No
17. En caso afirmativo, ¿qué tipo de problema de aprendizaje se le diagnosticó? Marque todo lo que corresponda:
    a. Trastorno de la lectura
    b. Dislexia
    c. Trastorno de la expresión escrita
    d. Dificultad en el aprendizaje de las matemáticas
    e. Otros
18. ¿Se diagnosticó a la persona con síndrome de Turner un trastorno por déficit de atención (TDA) o un trastorno por déficit de atención con hiperactividad (TDAH)?
    a. Sí (en caso afirmativo, ¿a qué edad se dio el diagnóstico?)

b. No

c. No lo sé

19. ¿La persona con síndrome de Turner recibió alguna vez servicios de educación especial por problemas de funcionamiento ejecutivo, organización, planificación, resolución de problemas o atención?

    a. Sí (En caso afirmativo, describa los servicios que recibió)

    b. No

    c. No lo sé

20. ¿La persona con síndrome de Turner recibió alguna vez clases particulares?

    a. Sí (en caso afirmativo, ¿la tutoría era para el TDAH/ADD o para problemas de funcionamiento ejecutivo, organización, planificación, resolución de problemas o atención? En caso afirmativo, ¿qué tipo de servicios recibió? ¿A qué edad comenzó a recibir las clases particulares? ¿Cuánto tiempo duraron?)

    b. No

    c. No lo sé

21. ¿La persona con síndrome de Turner recibió alguna vez un entrenamiento de vida?

    a. Sí (en caso afirmativo, ¿a qué edad comenzó el entrenamiento de vida? ¿Cuánto tiempo duró? ¿Incluyó el entrenamiento de vida el funcionamiento ejecutivo, la organización, la planificación, la resolución de problemas o las habilidades de atención)?

    b. No

    c. No sabe

22. ¿La persona con síndrome de Turner recibió alguna vez entrenamiento para problemas de funcionamiento ejecutivo, organización, planificación, resolución de problemas o atención?
    a. Sí (en caso afirmativo, ¿a qué edad empezó este entrenamiento? ¿Cuánto duró? ¿En qué tipo de habilidades se centró?).
    b. No
    c. No sabe
23. ¿La persona con síndrome de Turner recibió alguna vez rehabilitación cognitiva (cog rehab, REHABIT, readaptación cognitiva)?
    a. Sí (en caso afirmativo, ¿a qué edad comenzó? ¿Cuánto duró la rehabilitación cognitiva? ¿Qué habilidades se trabajaron en la rehabilitación?)
    b. No
    c. No lo sé

Salud e historial médico

24. Historial médico de la persona con síndrome de Turner. Marque todo lo que corresponda:
    a. Enfermedad de Alzheimer (en caso afirmativo, a qué edad se le diagnosticó)
    b. Trastorno autoinmune (en caso afirmativo, de qué tipo; a qué edad se le diagnosticó)
    c. Tumor cerebral (en caso afirmativo, de qué tipo; a qué edad se le diagnosticó)
    d. Cáncer (en caso afirmativo, de qué tipo; a qué edad se le diagnosticó)
    e. EPOC (en caso afirmativo, a qué edad se le diagnosticó)
    f. Demencia (en caso afirmativo, de qué tipo; a qué edad se le diagnosticó)
    g. Diabetes (en caso afirmativo, a qué edad se le diagnosticó)

h. Epilepsia (en caso afirmativo, a qué edad se le diagnosticó)

i. Ataque al corazón (en caso afirmativo, a qué edad se le diagnosticó)

j. Hipertensión arterial (en caso afirmativo, a qué edad se le diagnosticó)

k. Colesterol alto (en caso afirmativo, a qué edad se le diagnosticó)

l. Enfermedad renal (en caso afirmativo, a qué edad se le diagnosticó)

m. Enfermedad hepática (en caso afirmativo, a qué edad se le diagnosticó)

n. Enfermedad de Lyme (en caso afirmativo, a qué edad se le diagnosticó)

o. Meningitis (en caso afirmativo, a qué edad se le diagnosticó)

p. Obesidad (en caso afirmativo, a qué edad se le diagnosticó)

q. Convulsiones (en caso afirmativo, a qué edad se le diagnosticó)

r. Apnea del sueño (en caso afirmativo, a qué edad se le diagnosticó)

s. Enfermedad de tiroides (en caso afirmativo, de qué tipo; a qué edad se le diagnosticó)

t. Otros (opción de respuesta libre; en caso afirmativo, a qué edad se le diagnosticó)

25. Enumere todas las cirugías que le realizaron durante su primer año de vida

26. Indique todas las cirugías que le realizaron durante la infancia (2–12 años)

27. Enumere todas las cirugías que le realizaron durante la adolescencia (13-17 años)

28. Indique todas las cirugías que le realizaron después de los 18 años

29. ¿La persona con síndrome de Turner recibió un tratamiento con hormona de crecimiento?

a. Sí

b. No

c. No lo sé

30. En caso afirmativo, ¿a qué edad empezó a recibir la persona con síndrome de Turner el tratamiento con hormona del crecimiento?
31. ¿La persona con síndrome de Turner recibió terapia de reemplazo de estrógenos?
    a. Sí
    b. No
    c. No lo sé
32. En caso afirmativo, ¿a qué edad comenzó la persona con síndrome de Turner a recibir terapia de reemplazo de estrógenos?

Gracias por completar esta encuesta.

Appendix M: TABLES

Table 1

Retrospective Sample Means on EF Measures by Domain

EF Domain	EF Measure	*n*	Retrospective Mean	SD
Short Sustained Attention		13	-0.64	1.77
	Seashore Rhythm	13	-0.64	1.77
Long Sustained Attention		13	-0.59	1.55
	Speech Sounds	13	-0.59	1.55
Goal Setting		18	-0.84	2.33
	Category Test	18	-0.84	2.33
Selective Attention		15	-2.04	3.06
	TMT B	14	-1.97	3.16
	DKEFS Switching	1	-3.00	N/A
Inhibition		11	-0.64	1.12
	Stroop Color-Word	10	-0.48	1.02
	DKEFS Color/Word	1	-2.33	N/A
Digit Span Forward		17	-0.31	0.91
	WISC-IV DSF	9	-0.10	1.07
	WISC-V DSF	3	-0.90	1.01
	WAIS-IV DSF	5	-0.34	0.41
Digit Span Backward		17	-0.82	0.94
	WISC-IV DSB	9	-0.49	0.73
	WISC-V DSB	3	-1.77	1.37

	WAIS-IV DSB	5	-0.86	0.80
Working Memory		20	-0.49	1.05
	WISC-IV WMI	11	-0.34	1.14
	WISC-V WMI	4	-0.73	1.49
	WAIS-IV WMI	5	-0.62	0.46
Spatial Span Forward		12	-1.13	0.76
	WISC-IV Integrated SSF	5	-1.20	0.67
	WISC-V Integrated SSF	1	-2.30	N/A
	Knox Cube	6	-0.87	0.74
Spatial Span Backward		6	-1.07	0.48
	WISC-IV Integrated SSB	5	-0.88	0.16
	WISC-V Integrated SSB	1	-2.00	N/A

Note. Scores are presented as *z* scores

Appendix N: FIGURES

Figure 1

Number of Participants Diagnosed with ADHD and Receiving Special Education Services

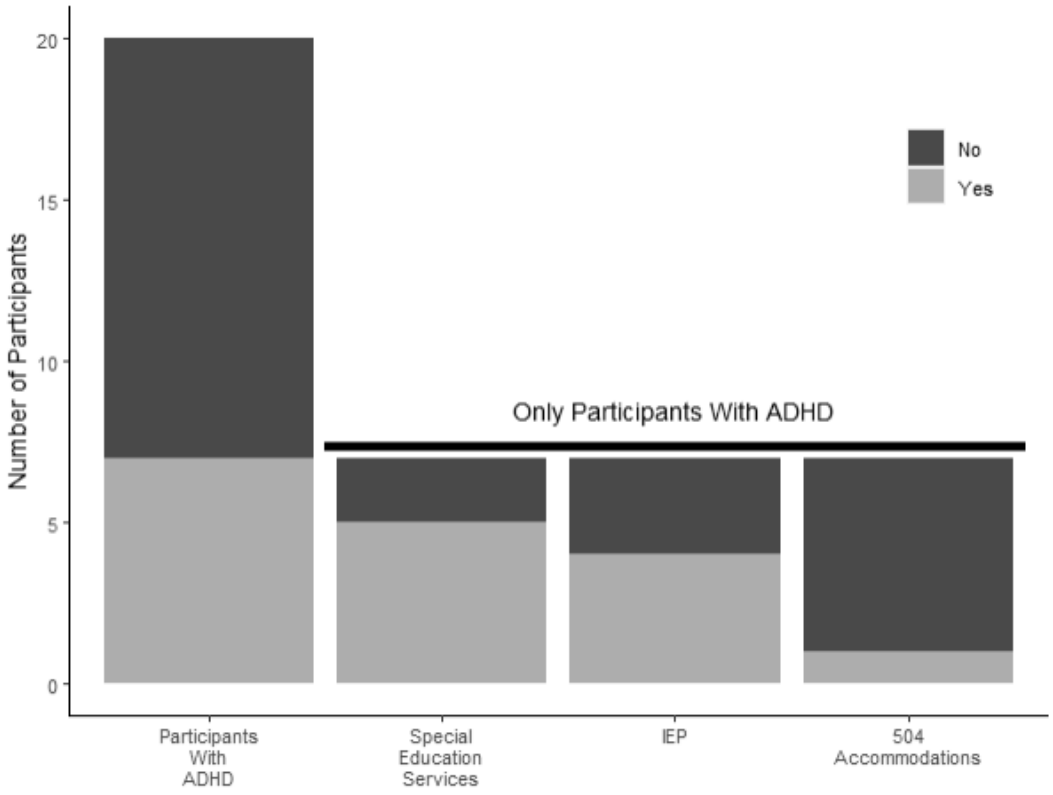
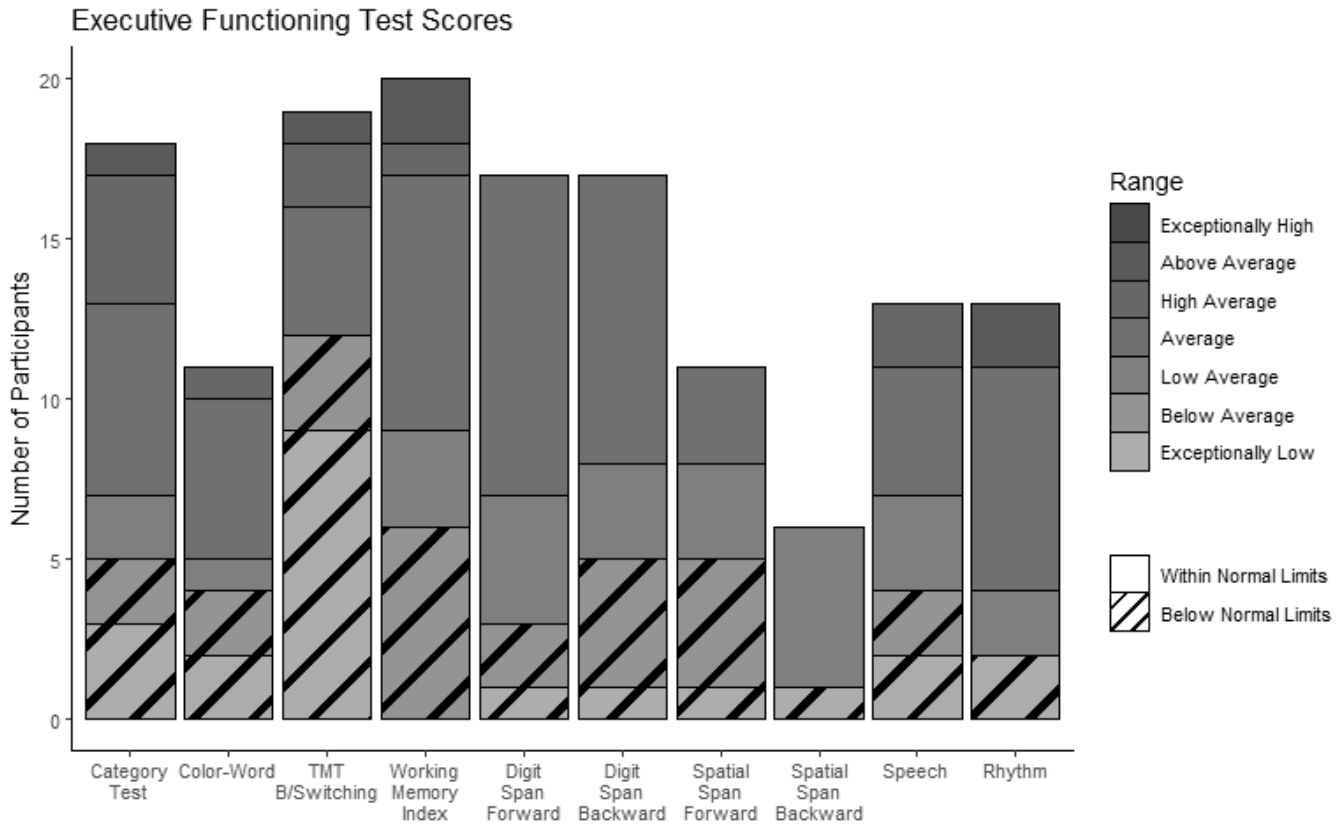


Figure 2

Retrospective Sample Qualitative Descriptors on each EF Neuropsychological Measure



Note. Qualitative descriptors were used as recommended by the American Academy of Clinical Neuropsychology (Guilmette et al., 2020).

Standard Score	Percentile Score	Descriptor
>130	>98	Exceptionally high score
120-129	91-97	Above average score
110-119	75-90	High average score
90-109	25-74	Average Score
80-89	9-24	Low average score
70-79	2-8	Below average score

<70	<2	Exceptionally low score

Figure 3

Retrospective Sample Qualitative Descriptors on each EF Domain

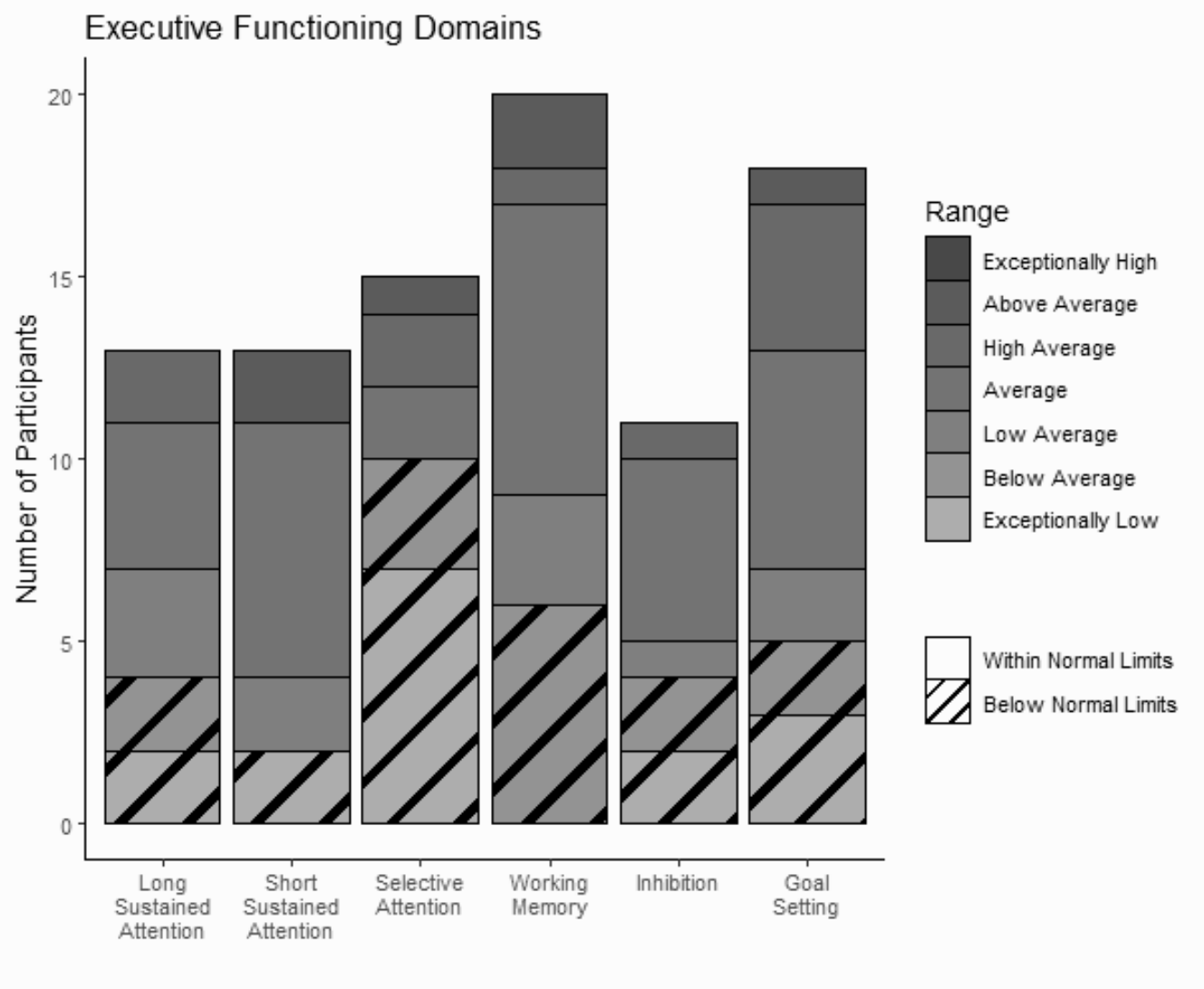


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Figure 4

Retrospective Sample ASEBA Behavioral Checklist Results

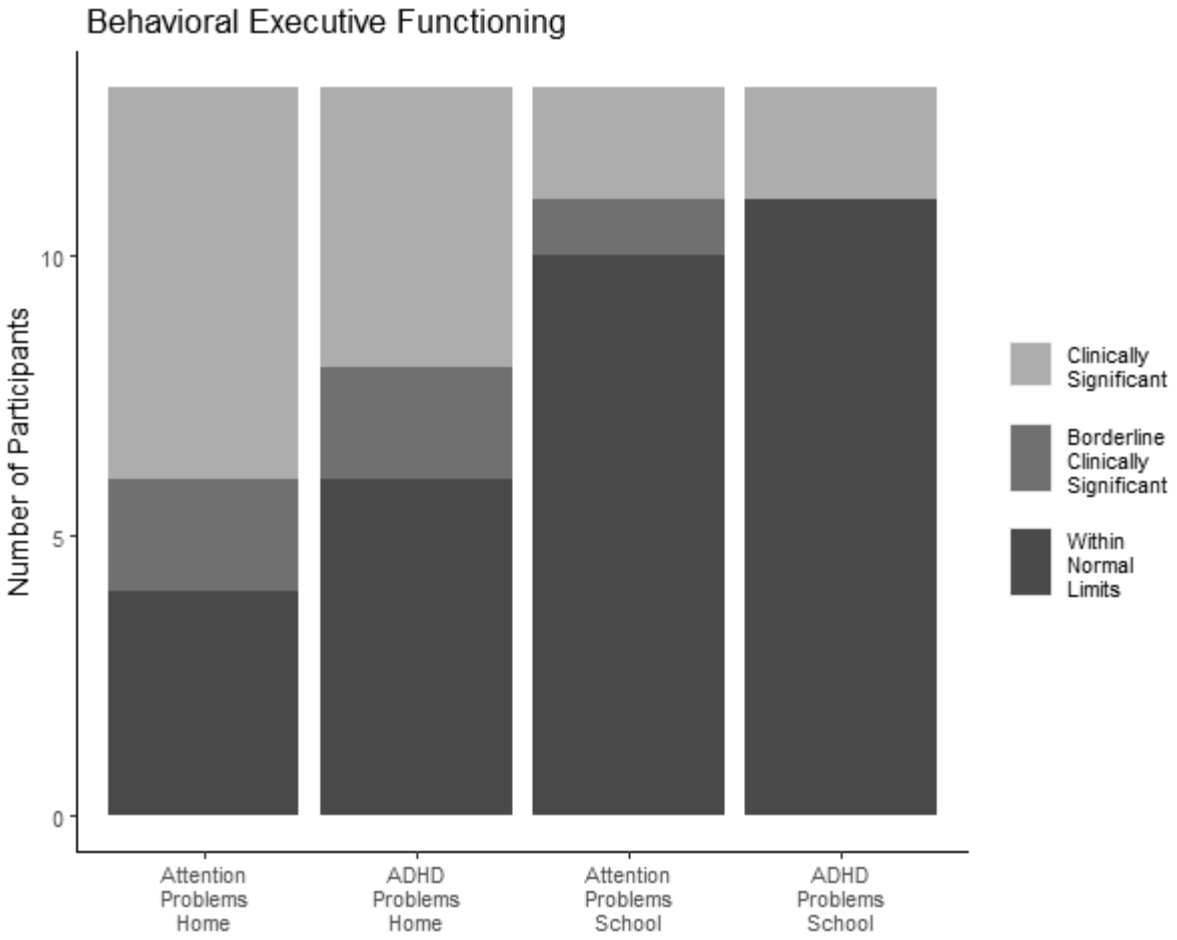

Figure 5

Correlation Between the ASEBA Behavioral Checklist Attention Problems and ADHD Problems Scales at Home vs. at School