



Math achievement, numerical processing, and executive functions in girls with Turner syndrome: Do girls with Turner syndrome have math learning disability?

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ABSTRACT

Turner syndrome is a common genetic disorder associated with select deficits in executive functions, working memory and mathematics. In Study 1, we examined growth trajectories of skills in these areas, from grades 1 to 6, among girls with or without Turner syndrome. Rates of growth and performance levels at 6th grade, on an untimed math achievement test, did not suggest that girls with Turner syndrome have math learning difficulty (MLD). However, analyses did reveal lower efficiency on timed executive function tasks, among girls with Turner syndrome, who traded accuracy for speed under mild to moderate working memory demands. In Study 2 we compared numerical processing skills of 6th graders who had either Turner syndrome, MLD, low math achievement, or typical achievement in math. A numerical decomposition task revealed numerical processing deficits for the Turner syndrome and MLD groups, relative to typically achieving students. The relative difficulties in how numerical processing vs. working memory demands affected performance accuracy differed among groups, with the former demands leading to more performance difficulties in the Turner syndrome group. Our findings support the notion that girls with Turner syndrome recruit different strategies than their peers during allegedly basic numerical processing, that numerical processing deficits vs. executive function deficits underlie their difficulties with mathematics, and that math difficulties among these girls may not be apparent on untimed tests. These findings have implications for a possible manifestation of MLD.

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1. Introduction

Turner syndrome is a spontaneously occurring chromosomal disorder with a frequency of 1:2000 to 3000 live female births (Lippe, 1991). It results from the complete or partial loss of one of the two X chromosomes typically present in females. The well-documented physical phenotype of Turner syndrome includes short stature, a webbed neck, and the lack of spontaneous development of secondary sexual characteristics due to ovarian dysgenesis (Ross, Roeltgen, Kushner, Wei, & Zinn, 2000). The cognitive phenotype is broadly summarized by relatively superior verbal versus nonverbal skills, specifically above average reading skills, average to above average vocabulary, and weakness in mathematics, sustained attention, visual-spatial, and executive functions (as reviewed by Davenport, Hooper, & Zegar, 2007). These cognitive deficits, which are observed despite average overall intellectual functioning (Mazzocco, 2001; Rovet, Szekely, & Hockenberry, 1994; Temple & Carney, 1993; Temple

& Marriott, 1998), have led researchers to consider Turner syndrome as a potential model of biologically-driven mathematical learning disability (MLD) (e.g., Molko et al., 2003).

1.1. Mathematical and working memory difficulties in girls with Turner syndrome

1.1.1. Mathematics skills in girls with Turner syndrome

It is now well established that girls with Turner syndrome are at increased risk for mathematical learning disabilities (MLD), or dyscalculia (Rovet, 1993; Mazzocco, 2001). These deficits are evident by kindergarten (Mazzocco, 2001), persist through the elementary and middle school years (Murphy, Mazzocco, Gerner, & Henry, 2006; Murphy & Mazzocco, 2008; Rovet et al., 1994; Temple & Marriott, 1998), continue through adolescence (Kesler, Menon, & Reiss, 2005; Mazzocco, 1998), and are also reported in adults with the syndrome (e.g., Bruandet, Molko, Cohen, & Dehaene, 2004). Across reports, the prevalence of MLD in Turner syndrome is approximately 50%, which far exceeds frequencies reported in the general population (~6 to 10%; Barbaresi et al., 2005; Shalev, 2007).

The underpinnings of these broadly defined math deficiencies are not well established, in either the general population or Turner

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syndrome. Competing hypotheses concerning MLD in Turner syndrome have been posed and tested for the last fifteen years, implicated by evidence of specific right hemisphere, frontal-striatal, and/or frontal-parietal abnormalities in this population (as reviewed by Mazzocco & McCloskey, 2005). Initially, the co-occurrence of math and visual spatial deficits in girls with Turner syndrome led to speculation that performance deficits in these two domains were correlated with one another, but this notion was refuted by the researchers who first investigated this potential link (Rovet et al., 1994), on the basis of no discernable association between math achievement scores and performance on a wide range of visual perceptual or reasoning tasks. What few correlations did emerge involved measures that, like the Judgment of Line Orientation (JLO), are also believed to tap effortful processing (e.g., Mazzocco, 1998). Thus, alternative hypotheses linked poor math achievement in Turner syndrome with executive dysfunction (Rovet et al., 1994; Temple & Marriott, 1998), but with equivocal support.

More recent studies have shown inconsistent support for select numerical processing deficits in persons with Turner syndrome. For example, Bruandet et al. (2004) investigated the performance of women with Turner syndrome on tasks that involved “cognitive manipulation of numbers.” In their study, women with Turner syndrome were slower (yet as accurate) when compared with their peers on arithmetic calculations using addition, subtraction, or division, slower and less accurate during a rapidly presented numerical estimation task, yet they were as accurate as their peers while counting two to nine items forwards or backwards and while selecting the largest of two rapidly appearing digits. Women with Turner syndrome, like their peers, showed a classic distance effect during this numerical judgment task (Bruandet et al., 2004), such that response times increased as the numerical distance between the pairs of numbers being compared decreased. Thus, basic number *comprehension* seems preserved, consistent with earlier findings that girls with Turner syndrome are accurate at reading and writing numbers (Rovet et al., 1994; Murphy et al., 2006), and are accurate at symbolic (and untimed) numerical judgments, and problem verification (Kesler et al., 2005; Murphy & Mazzocco, 2008). Still, some features of numerical processing are impaired. This notion received additional support from evidence that school age girls with Turner syndrome are significantly slower (but as accurate) when compared with their peers when enumerating one to eight items (Simon et al., 2008). However, slower numerical processing does not rule out the potential influence of executive functions, including working memory, on math performance among girls with Turner syndrome.

1.1.2. Executive functions in girls with Turner syndrome

Like the mathematics difficulties summarized above (Section 1.1.1), executive dysfunction is a persistent phenotypic feature observed among girls and women with Turner syndrome. Such deficits have been reported in primary school age girls with Turner syndrome (Kirk, Mazzocco, & Kover, 2005), school age girls (Temple & Marriott, 1998), adolescents (Lasker, Mazzocco, & Zee, 2007; Tamm, Menon, & Reiss, 2003), and women with the syndrome (Haberecht et al., 2001; Kesler et al., 2005). Comparable to the uneven profile of mathematics strengths and weaknesses, there is inconsistent evidence regarding the nature of executive dysfunction in Turner syndrome. For instance, Temple and colleagues have reported intact goal-oriented planning and organizing skills on measures such as the Tower of Hanoi, but Romans found impaired performance levels on this task (Romans, Roeltgen, Kushner, & Ross, 1997). Poor search strategies and slowed response times are evident on verbal fluency tasks (Temple, 2002; Temple, Carney, & Mullarkey, 1996), but intact motor response times fail to support the notion of general slowed processing speed (Simon et al., 2008). Stroop-like interference tasks that require inhibition and cognitive set shifting reveal inefficient and inflexible response patterns in girls with Turner syndrome (Temple et al., 1996), particularly as working memory demands increase (Kirk

et al., 2005), although not on verbally mediated tasks that present a minimal cognitive load (Tamm et al., 2003).

Although the specificity of executive function deficits in this population remains unclear, a logical extension of these findings is the notion that executive functions under select working memory demands may interfere with mathematics tasks such as three-operand calculations (Kesler et al., 2005), or may even underlie more basic numerical processing difficulties (Bruandet et al., 2004). As such, girls with Turner syndrome may provide a model of a central executive mediated pathway to MLD, one of several mechanisms proposed by Geary (2004) as underlying some instances of MLD. Consistent with this notion is evidence from neuroimaging studies that females with Turner syndrome show atypical activation of prefrontal and frontoparietal circuitry under conditions of increased working memory demands, such as three-operand vs. two-operand arithmetic (Haberecht et al., 2001; Kesler et al., 2005; Tamm et al., 2003).

1.2. The present study

The goal of our first study was to examine whether girls with vs. without Turner syndrome show parallel developmental trajectories of math and executive skills within groups, and different trajectories between groups, on measures of effortful performance under varying levels of working memory demands. A review of the construct of executive functions and working memory is beyond the scope of this paper, particularly given that herein we focus on one measure of executive function for which inefficient performance among girls with Turner syndrome has already been demonstrated, at least in primary school (Kirk et al., 2005). We selected this task, the Contingency Naming Test (CNT), because it includes varying levels of working memory demands, and is sensitive to both developmental (e.g., Anderson, Anderson, Northam, & Taylor, 2000) and clinical (e.g., Taylor, Albo, Phebus, Sachs, & Bierl, 1987) differences in reactive flexibility and cognitive shifting. We also explored whether performance on the CNT predicts later math achievement or the rate of growth in math achievement from grades 1 to 6 in girls with Turner syndrome.

There is much support for the notion that executive functions influence math performance (e.g., Bull & Scerif, 2001), that MLD is associated with poor executive functioning and working memory (e.g., Geary, Hoard, Byrd-Craven, Nugent, & Numtee, 2007), and that children with low working memory have poor mathematics achievement (Alloway, Gathercole, Kirkwood, & Elliott, 2009). However, an obstacle to revealing the precise nature of this relationship stems from the fact that both “mathematics” and “executive functions” are multifaceted constructs. We address only one potential aspect of this association in the present study, but we approach this study by examining Turner syndrome because it is a population for which deficits in both math and executive functions have been reported.

1.3. Contributions of syndrome research to understanding MLD

Constructs such as “mathematical learning disability” and the equally broad domains of “executive dysfunction” or “visual spatial deficits” skills are likely to have multiple biological or environmental sources of influence. Although no single research program can address — much less identify — the full range of primary or secondary influences, it is useful to diminish such influences on variables of interest within the research context, to the extent possible. Research on syndrome-specific phenotypes helps to narrow the range of biologically mediated factors influencing manifestation of MLD in a study sample. This is the case whenever the known genotype is believed to underlie mathematics-related behaviors, regardless of whether specific genes leading to the outcome have been identified. Moreover, the range of specificity among the components of broad behavioral constructs — such as executive functions or working memory — should also be

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