



# Neuropsychological Associations of Turner Syndrome

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## Introduction

- Turner syndrome (TS) is a rare neurodevelopmental disorder which occurs in approximately one in 2500 live-born females.<sup>1</sup>
- This genetically inherited condition is typically caused by either meiotic or mitotic chromosomal nondisjunction.
  - Meiotic nondisjunction most often occurs in the paternal gametes and results in complete sex chromosomal monosomy 45,XO.
  - Mitotic nondisjunction of an embryonic cell results in sex chromosomal mosaicism 45,XO/46,XX, and often presents with a milder phenotype.



**Figure 1.** A chromosome picture (karyotype) from a woman with Turner syndrome, meiotic nondisjunction type 45,XO.<sup>2</sup>

## Case Report

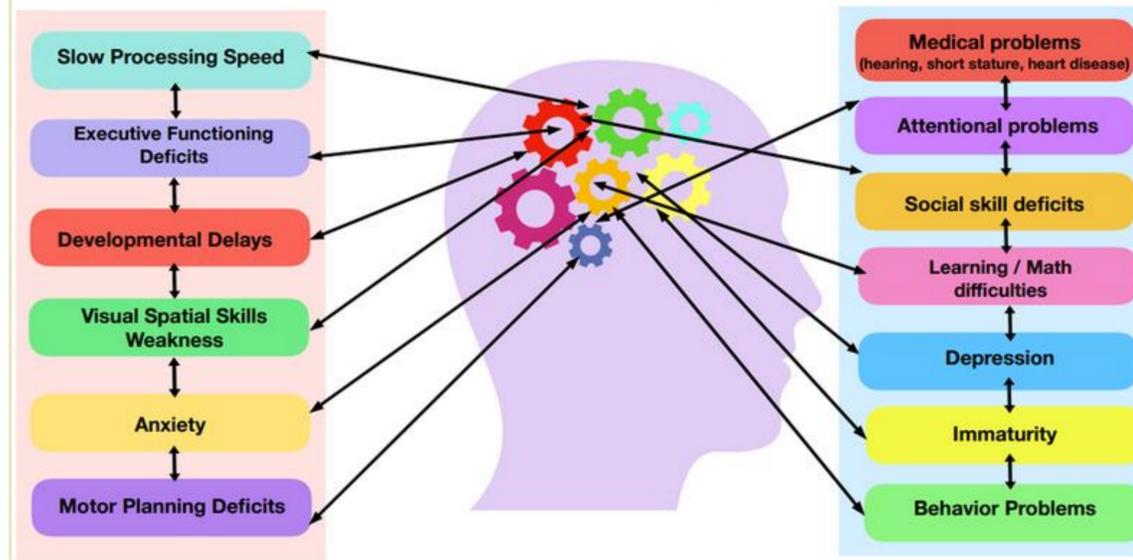
A 7-year-old female patient with a history of Turner syndrome, and learning disability presents with behavioral problems at home and at school in the form of inattention, hyperactivity, disobedience, and throwing fits when frustrated. She is in Kindergarten for the second time, due to failure to meet academic milestones last year. She has an Individualized Education Program (IEP) at school and was recently evaluated for learning disability.

At home she has difficulty completing tasks when asked, and at school she distracts others, has trouble staying focused on assignments, and often leaves her desk without permission.

On exam the patient shows signs of Attention Deficit Hyperactivity disorder (ADHD) including the inability to remain seated, and trouble focusing on the conversation. Her responses were often tangential or unrelated to the questions asked.

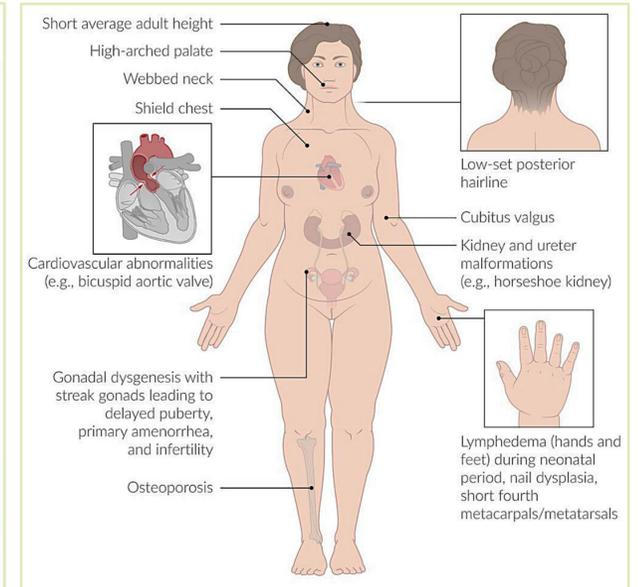
## Neuropsychological Associations of Turner Syndrome

### Turner Syndrome Neuropsychological Risks



**Figure 2.** The interplay between the variety of potential neurodevelopmental, social, and emotional deficits results in a large variety of neuropsychological phenotypes. Taken with permission from Hutaff-Lee, et al.<sup>3</sup>

- Patients with TS are at an increased risk for difficulties with visual-spatial reasoning and memory, executive functioning, and attention.
- An 18-fold increase in prevalence of ADHD was found in girls with TS (24%) compared with girls in the general population (1.3%).<sup>4</sup>
- Girls with TS seem to have an increased risk for the Hyperactive/Impulsive subtype of ADHD, which is otherwise more prevalent in males with ADHD than females.<sup>5</sup>
- TS can also increase the risk of specific learning disability, especially in mathematics.<sup>5</sup>
- Other psychological hardships of higher prevalence in TS patients include anxiety, depression, and social difficulties.<sup>5</sup>



**Figure 3.** Turner Syndrome affects a variety of organ systems. While the cardiovascular, renal and orthopedic symptoms pose the largest health risks overall, the infertility and altered physical appearance can contribute to self image problems and increased rates of depression.<sup>4</sup>

## Discussion

- Given the number of associated complications in Turner syndrome (Figure 3), management requires multidisciplinary care and collaborative efforts across various specialties.
- Patients with Turner syndrome should be evaluated for neuropsychological comorbidities including ADHD, learning disability, anxiety and depression.
- It is imperative that psychiatric conditions not be ignored in patients with TS as they can be exacerbated by their physical conditions and can further deteriorate quality of life.

## References

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