

Understanding and Appreciating Turner Syndrome

Adapted from:

Summary of presentations by Dr. Shelli Kesler

Dr. Kesler began her presentation by outlining the age-old debate in science. “How much do nature (genetics) or nurture (environmental factors) affect who we become?” Most researchers would now agree that both nature and nurture affect an individual. In the case of Turner Syndrome (TS), the question becomes to what extent does the missing ‘X’ affect the behavior and thinking (cognition) of a girl or women with TS, and to what extent do environmental factors (prenatal events, parenting, education, peer group, nutrition, etc.) affect behavior and thinking? The position of this research team is that TS is a “risk factor” for certain cognitive-behavioral problems, but TS does not “cause” these problems.

Dr Kesler then posed the question, “When attempting to understand the thinking and behavioral problems in TS, are descriptive diagnoses such as a Nonverbal Learning Disability (NLD) and Attention Deficit Disorder (ADD) helpful? This research team believes these labels are not at all helpful because, in their words, “TS is a diagnosis with clear biological markers and increasingly well understood disease mechanisms...whereas NLD and ADD are descriptively defined” (meaning that a diagnosis of NLD and ADD are made by observing certain behaviors.) The team thinks NLD and ADD may have different neurobiological and neurogenetic factors underlying them, and that using the NLD label to describe the cognitive and behavioral challenges of people with TS distracts from the real issues of TS. To put it another way, the concern is that, although the learning challenges evident in girls with TS may look similar to NLD, effective strategies for dealing with each may be different. Therefore, the approach of these researchers has been to first focus on the neurobiology of TS (what is going on in the brain of TS girls), and then try to identify strategies to enhance their thinking processes based on those observations. This led into the second keynote address by Dr. Kesler.

The second presentation started with a review of the known cognitive features of TS, namely, the girls with TS: 1) nearly always has a normal IQ (individuals with TS with severe learning problems may have unusual genetic characteristics) 2) tend to have a higher verbal IQ than performance IQ. 3) Often have strength in verbal skills and 4) often show a weakness in visual memory, visual-spatial skills and executive functioning (planning and organizing skills). The psychosocial features of girls and women with TS were also summarized. Dr.Kesler noted that physical well-being (how healthy the person is), the pattern of sexual maturation and height, clearly affect behavior. She also noted the literature suggests individuals with TS are at increased risk for 1) attention problems, 2) impaired face and emotion processing (difficulty “reading” social cues), 3) problems with peer relations, and perhaps 4) anxiety and depression- though this last point is not clearly established in current research literature.

Dr. Kesler moved on to pose the question, “What can we do about the thinking and behavior problems of TS girls, if they are present?” She then listed some recognized general strategies for addressing thinking and behavior challenges, namely, early identification of problems, family or parental counseling and guidance, treatment of behavioral problems with therapy and (sometimes) medication and placement in an optimal educational environment. Treatments for psychosocial challenges are not as clear cut, since there has been little research in this area. It appears these strategies may be helpful: 1) general coping and adaptive skill training (focusing on dealing with medical issues) 2) social skills training (focusing on face processing and body language interpretation), 3) stress management training, 4) effort to improve self-esteem, and 5) developing effective learning and organizational strategies to compensate for cognitive weaknesses. The research team that Dr. Kesler is part of believes that much research still needs to be done to find new ways to identify early on whether a girl with TS will have some of the thinking challenges associated with TS, and research also has to be done to find ways to help girls and women with TS optimize their thinking and develop their social skills. They feel the guiding questions for future research are:

- What are the specific genetic, biological and environmental factors that contribute to cognitive and behavioral strengths and weaknesses?
- Can we identify brain correlates of these factors and use them to improve treatment specificity and effectiveness?
- Which of these factors predict response for particular treatments?
- Which treatments are most likely to be of benefit?
- Who is most likely to benefit from early identification and treatment?
- How can we increase the likelihood that each individual with TS will achieve their maximum potential?
- What is the brain structure and function in TS and how can we develop a model for understanding this?

Dr. Kesler went on to describe some of the work her team is doing in Stanford University. Currently the research team at Stanford is using brain imaging (pictures obtained by using magnetic resonance imaging or MRI) to see which parts of the brain are being used when subjects are given certain tasks. They have discovered that people with TS have certain differences in their brains and that they use their brains differently. They have determined that the volume of the parietal lobe (where visual-spatial processing occurs) tends to be smaller, and the temporal lobe (where verbal processing happens) tends to be enlarged in girls with TS. In a study of math functions, TS girls also showed greater activation in frontal parietal regions of the brain during simple math tasks, but less activation during harder tasks (Kesler et al 2006 Cerebral Cortex).

A study of verbal and visual-spatial working memory led to the conclusion that there is some frontal-parietal dysfunction in TS and that active storage of materials in the working memory is impaired. Collectively, the brain imaging results suggest “network” issues-certain

pathways or connections in the brain are not as well-developed in girls with TS. There appears to be a “disconnection” between parietal and frontal regions associated with visual spatial-executive function in TS. People with TS tend to engage the parietal-temporal lobe of the brain (where verbal skills reside) and do not tend to engage the parietal-frontal pathways (where visual skills arise), when doing visual-spatial tasks or executive functioning (organizing) tasks. This way of using the brain shows that they are compensating, but they are less efficient and the compensation may only work for lower-level tasks.

The team is also working to develop cognitive Interventions for girls with TS. They are trying to find out if they can “increase parietal-frontal connectivity in individuals with TS, thus improving planning ability and math skills.” They are doing this by using a cognitive intervention they have developed which involves asking the individual to perform specific skills, and then asking the person to practice the skill in small, increasingly difficult steps. The researchers believe this intervention works by taking advantage of the brain to develop new connections between its components parts by exercising the brain, much as you would exercise a muscle. As the brain encounters mental challenges it learns new things and develops more efficient and effective brain networks, a process that can then be measured with functional MRI images. Early results of this work demonstrates that girls with TS can be taught to achieve much higher accuracy on certain cognitive tasks and improve the efficiency or speed with which they perform correct tasks. MRI comparisons show the girls can actually be taught to activate different parts of their brain! Continued research is being done to determine how long this effect will last.

The team is currently recruiting additional volunteers for their ongoing study of the brain function and TS. A flyer with study inclusion criteria, a brief outline of what is required of participants, and contact information, can be obtained by contacting 1-888-411-2672)