

The Hidden Disability: Sex Chromosome Variations (SCV)

By Carol Samango-Sprouse

Tommy is an 18-month-old toddler with mild truncal hypotonia. He has a slow-to-warm temperament with expressive language delay, and he has difficulty with social interactions. He is an attractive little boy who seems quite bright. He has received no medical diagnosis, but his mother wonders, "What is the cause of his developmental problems?"

Robert is a sweet 20-month-old boy with delays in gross motor skills and expressive language skills. His receptive language skills appear age-appropriate, but he rarely babbles and prefers to use only vowel sounds. He often avoids eye-contact and prefers to place his toys in a straight line. He becomes upset if the toys are rearranged but then settles down when comforted by an adult. His mother wonders if there is a link between his motor and language delay. What is the future for Robert and his school performance?

Both children are receiving early intervention services for their delays, but their Individualized Family Service Plans (IFSPs) could be more individualized and more family-centered if the etiology and medical diagnosis were identified. Robert and Tommy both may have a *sex chromosome disorder*.

During early childhood, children with Sex Chromosome Variations (SCV) have minor physical variations. As young children, many are extremely attractive. Because such children often have few major congenital malformations, chromosomal analysis may not be considered in the presence of developmental delay, speech delay, or learning problems. Consequently, many children with these disorders are not being diagnosed during their early years, and the opportunities for syndrome-specific intervention services are missed during a critical period of brain growth and differentiation.

A SEX CHROMOSOME DISORDER

Sex chromosomes anomaly is a result of a chromosomal mistake or aberration that produces an additional X or Y to the normal complement of 46. The resulting total of 47 chromosomes may affect all aspects of the child's developing central nervous system and his or her body condition. Sex chromosome variations occur in 1 in 500-1,000 live births with four different types of chromosomal arrangements.¹ They are XXY (Klinefelter syndrome), XYY, XXX (Triple X), and XO (Turner syndrome). Turner or XO is often identified since there are some physical abnormalities and congenital anomalies. Sex chromosome variations affect boys and girls equally. It is reasonable to speculate that undiagnosed sex chromosome variations may account for the learning disabilities in some children although this hypothesis has not been studied in a large multi-institutional study.²

Recent research findings reveal that 64% to 85% of all children with XXY, XXX, and XYY are undiagnosed with only 10%- 20% identified from amniocentesis performed because of advanced maternal age.³ Although these disorders are associated with learning disabilities, children remain largely undiagnosed because their neurodevelopmental problems are often perceived as "*just a speech delay or motor delay,*" or as children become older, "*merely a learning disability.*"

LANGUAGE PROCESSING

In the last three years at Neurodevelopmental Diagnostic Center for Young Children, a prospective study of fifty boys with XXY has been conducted. Infant boys with XXY have evidence of speech delay as early as 12 months of age. Expressive language delay is the most consistent and pervasive feature of the language dysfunction in these young boys. The severity of their speech delay is often underestimated because they acquire single words and two-word phrases within normal limits. In contrast to the speech delay, most boys with XXY have age-appropriate receptive language skills.

Articulation of specific sounds improves with therapy in preschool years, but language organization issues continue to occur for many years.

COGNITION

Many of the boys tested experienced the greatest problems in organization, planning, and attention for verbal tasks. As early as infancy and continuing throughout preschool, the boys with XXY are very focused and directed for tasks of spatial cognition regardless of the degree of difficulty. When verbal tasks are introduced, attention wanders and compliance is reduced. In many cases, there was a significant increase in oppositional behavior if the language items were not scattered throughout the evaluation. There was significant anxiety generated by the language demands of testing and/or social interactions.

Behaviorally, these infants and toddlers have a wide array of temperamental styles. There was a characteristic response to stress during testing that may be a hallmark for the syndrome. When stressed or overwhelmed during testing or socially, they lowered their heads with a mild head tilt, blushed, and became very shy. This physiological reaction may be mediated by the anxiety of the situation or conversely may be a unique behavior in boys with XXY.

SUMMARY

Sex Chromosome Variations are very common and occur in 1 in 500-1,000 live births. They are more common than Down syndrome and Cystic Fibrosis, yet they are largely underdiagnosed because they are not well understood by the medical community and are not a well-described disorder in the research literature. Therefore, it seems reasonable to consider the possibility of SCV in any child who has developmental delay, speech delay, or behavior problems without a known etiology and discuss these concerns with the primary physician. The diagnosis of SCV can be confirmed through standard chromosomal analysis, a simple blood test. With more effective early identification of young boys with XXY, we may enter the new millennium with more family-centered and syndrome-specific IFSPs for many previously undiagnosed children.

¹National Genetics Foundation, 1987.

²Abramsky, L., and Chapple, J. (1997). 47, XXY and 47, XYY: Estimated rates of and indications for postnatal diagnosis with implications for prenatal counseling. *Prenatal Diagnosis*, 17:363-368.

³Personal Communication with Melissa Alystock, Executive Director of Klinefelter Syndrome and Associates, May 20, 1999.