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The Need for Greater Awareness of Sex Chromosome Variations

Health care providers remain ill prepared to recognize these conditions and support patients and their families.

"I was devastated when I looked up my son's diagnosis online." "I don't want anyone to know what my daughter has because once they find out they will judge her."

As a family psychiatric NP at the National Institutes of Health, I heard such sentiments far too often when I started working with families of children with XYY syndrome and other conditions caused by sex chromosome variations. Most of my experience in the past five years has been with the trisomies XYY, XXY, and XXX. Even though XXY syndrome (Klinefelter syndrome) occurs in one in 500 live male births, XYY in one in 1,000 males, and XXX (trisomy X or triple X syndrome) in one in 1,000 females, awareness of these conditions among frontline health care providers is limited and many cases are missed. It is important to raise providers' awareness of sex chromosome variations to increase early diagnosis, help children and families better understand their diagnosis, and decrease the stigma surrounding the condition.

Early diagnosis can help children receive services critical to their health and wellness.

While sex chromosome variations are like most syndromes in that affected carriers can vary greatly, each syndrome has certain common characteristics. Individuals with sex chromosome variations are typically taller than predicted from parental height, can have curvature of their fingers and wider-set eyes, and have low muscle tone. Speech delays are also seen on a spectrum: some children are marginally delayed, while a high percentage require speech and language services. Many of the children I have assessed struggle in school and at home because they have difficulty focusing and trouble remembering tasks they have been asked to complete.

Seeing an individual with behavioral and psychiatric concerns coupled with speech delays and taller stature relative to parental height should alert the practitioner to consider genetic testing for sex chromosome variations. Early intervention is key. Early diagnosis can help children receive services critical to their development and long-term health and wellness.

Unfortunately, many parents find themselves having to educate their providers about these conditions. Even worse, for parents researching the diagnosis online, information is limited and what is available can be inaccurate or outdated. My first internet searches relating to XYY syndrome returned results about "super males" and suggested an association (which has since been disproven) between XYY and high rates of aggression or incarceration. Relatedly, many people have the misconception that XXY males will be more "feminine" and females with an extra X are "super females."

I have also become increasingly aware of the disparities among families of color facing similar diagnoses. Recently I heard stories from families of color on how difficult it was to get their child tested. They reported that various health care professionals had attributed their children's delays or behavioral issues to their race and not to a syndrome, leading some families to not obtain appropriate treatment.

In speaking with other health care providers, I've been struck by the fact that I and many others graduated without any training in sex chromosome variations. Imagining the agony and frustration of being the parent of a child with a new diagnosis who can't go to their health care provider for information, reassurance, or a plan of action inspired me to find ways to educate not only families but providers as well. As frontline providers, it is important that we can detect these syndromes. If in practice you come across a taller individual with a history of speech delays and attention difficulties, genetic testing could provide the missing piece to the puzzle. ▼

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