

Recognize little-known disorder that causes speech, language problems

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Summary: Klinefelter syndrome is a genetic disorder that can greatly impact the ability of children to learn. Lazerson Klinefelter syndrome only affects males occurring in one out of every 500 male conceptions. According to the National Institute of Child Health and Human Development, KS results from a genetic abnormality, an extra sex chromosome, XXY, instead of the typical male arrangement, XY. Although having an extra sex chromosome is widespread, the syndrome itself and the characteristics resulting from having the extra chromosome are not. A speech-language pathologist Nancy Lazerson has spent more than 20 years in her field, many of them working with children and families of children diagnosed with Klinefelter Syndrome.

The following Q & A addresses some of her issues about recognizing and diagnosing the syndrome.

Q. What are the possible speech and language development issues that can impact children who go undiagnosed?

A. One of the defining symptoms of KS is a lack of early expressive language and speech development. Typically, KS boys have intact receptive language skills (up until around age 9, when higher level language processing becomes more important), but are very delayed in their speech and expressive development.

Klinefelter impacts:

- Intelligence.
- Expressive language.
- Receptive language.
- Auditory processing.
- Pragmatics.
- Articulation skills.

Should Klinefelter Syndrome go undiagnosed, these boys can have lifelong language deficits and learning disabilities.

Q. In your work, how often do you encounter people in early intervention or preschool who are familiar with Klinefelter Syndrome?

A. Given the incidence (1 in 500 boys), I run into remarkably few children who are already diagnosed. I have had several kids over the years get diagnosed based on my recommendation for further testing.

Q. What should early childhood officials do to ensure more children are tested for Ks and start receiving treatment as soon as possible?

A. I think any preschooler with severe expressive and speech delays (e.g., no speech or language by age 2), or with global developmental delays, should be at least referred to a geneticist, and perhaps get tested if there is any concern. But some might find that view extreme. Certainly, though I think we are missing the ball with a lot of KS kids.

I have several babies with a diagnosis of autism spectrum who I have referred for KS testing, mostly because they seem to fit more with KS than autism, and I want to rule it out.

Q. You've recommended several babies who are diagnosed with autism receive KS testing. Are the characteristics similar? Is this a common misdiagnosis?

A. We're just starting to see babies under 2 being diagnosed with autism. My suspicion is that a good percentage of these kids may actually have KS. It may be some time before we can tease this out.

Preliminary results in a study being conducted by the National Institute of Mental Health reveal that the brains of children with Klinefelter syndrome may differ in structure from those without chromosomal abnormalities.

So far, the brain scans of 43 KS individuals as young as ages 5 have shown less development in the area of the brain dealing with language, emotions, memory and social cognition. On the other hand, the area that controls spatial processing abilities is better developed than normal.

The findings were presented at the American Association for Klinefelter Syndrome Information and Support's national conference. According to the NIMH, 60 percent to 80 percent of students with KS are diagnosed with learning disabilities and /or attention deficit disorders even if their genetic variation is not discovered.

For more information, visit www.aaksis.org/index.cfm

Comments: One really good resource for accurate information on the web is the National Organization for Rare Disorders www.rarediseases.org. It includes many different disorders.

There is always a question about further testing when a child without a diagnosis has a significant problem. I wonder how many children go undiagnosed because no one ever mentioned other doctors or specialists who could give the family more information about their child's problems.

Many of us refer to Children with Special Health Care Needs but many may not refer to any other providers. This article made me think about how we decide who to refer and who not to refer. I believe that parents have the right to the information so that they can make the decision. It is what we all want for ourselves and what we want all the doctors to do for families. Telling parents that there are others besides a pediatrician who could do further testing will help them to understand what is available.

The other issue is cost and if the parents have the energy to go through the possibility of many normal tests or even worse a positive test that can bring devastating news.

You can tell parents that costly tests may be recommended and results may be normal. The good part of that is that you know the child doesn't have that.

You may need to help parents if they receive a diagnosis that is devastating. Be prepared.

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